# Annual Report

# 2014

INTERNATIONAL CLEARINGHOUSE FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH

**INTERNATIONAL CLEARINGHOUSE**
**FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH**
**(ICBDSR)**

A non-governmental organization in official relations with
the World Health Organization

# Annual Report

# 2014

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# INTERNATIONAL CLEARINGHOUSE
# FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH

# ANNUAL REPORT 2014



**Monitoring Systems:**

- Description of the Registry
- Number of births by year and by maternal age
- Number of Terminations of Pregnancy (ToP) for selected malformations
- Table annual data (the most recent available year)
- Birth prevalence rates on previous years
- Time trends

**for the following Monitoring Systems:**

**Monitoring Systems, not contributing with Annual Data:**

# Synopsis of Contributing Monitoring Systems

| Monitoring Program | Coverage | Year Joined ICBDSR | Maximum age at diagnosis | Criteria defining stillbirths | Termination of Pregnancy (ToP) |
|---|---|---|---|---|---|
| **Argentina: RENAC** | Hospital-based National | 2012 | 3 Days | 500 grams | Not permitted |
| **Australia: WARDA** | Population-based, Statewide | 2002 | Up to 6 years | 20 weeks or 400 grams | Permitted, Reported |
| **Canada: Alberta-ACASS** | Population-based Provincial | 1996 | 1 year | 20 weeks or 500 grams | Permitted, Reported |
| **Canada: CCASS** | Population-based National | 1996 | 30 days | 20 weeks or 500 grams | Permitted, Not reported |
| **Chile-Maule: RRMC-SSM** | Hospital-based Regional | 2003 | Hospital discharge | 500 grams | Not permitted, Not reported |
| **Colombia: BCMSP** | Hospital-based Regional | 2011 | Hospital discharge | 500 grams | Permitted only for a few selected cases, Not reported |
| **Costa Rica: CREC** | Population-based National | 2003 | 3 days | 20 weeks or 500 grams | Not permitted |
| **Cuba: RECUMAC** | Hospital-based National | 2003 | Hospital discharge | 500 grams | Permitted, Reported |
| **Czech Republic** | Population-based National | 1974 | Up to 15 years | Non-viable fetuses, 28 weeks or >1000 grams | Permitted, Reported |
| **Finland** | Population-based National | 1974 | 1 year | 22 weeks or 500 grams | Permitted, Reported |
| **France: Paris** | Population-based Regional | 1982 | Hospital discharge | 22 weeks | Permitted, Reported |
| **France: REMERA** | Population-based Regional | 1974 | 1 year | 22 weeks (*) | Permitted, Reported |
| **Germany: Saxony-Anhalt** | Population-based (Federal State) | 2001 | Hospital discharge (almost first week of life) – up to 1 year | >/= 500 grams | Permitted, Reported |
| **Hungary** | Population-based National | 1974 | 1 year | 24 weeks or 500 grams (**) | Permitted, Reported |
| **India: BDRI** | Hospital-based, Regional | 2010 | 1 year | 24 weeks | Permitted, Reported |
| **Iran: TROCA** | Hospital-based Regional | 2006 | 5 year | 20 weeks or 400 grams | Permitted, Reported only for a few selected malformations |
| **Israel: IBDSP** | Hospital-based Regional | 1974 | Hospital discharge 2-5 days | 20weeks or 500 grams | Permitted, Reported |
| **Italy: BDRCam** | Population-based Regional | 1996 | 7 days | 180 days (25 weeks + 5 days) | Permitted, Reported |
| **Italy: IMER** | Population-based Regional | 1985 | 7 days | 180 days (25 weeks + 5 days) | Permitted, Reported |
| **Italy: Lombardy-RMCL** | Population-based Regional | 2007 | 1 year | 180 days (25 weeks + 5 days) | Permitted, Reported |
| **Italy: North East** | Population-based Regional | 1997 | 7 days | 180 days (25 weeks + 5 days) | Permitted, Reported |
| **Italy-Tuscany:RTDC** | Population-based Regional | 1998 | 1 year | 180 days (25 weeks + 5 days) | Permitted, Reported |

| Monitoring Program | Coverage | Year Joined ICBDSR | Maximum age at diagnosis | Criteria defining stillbirths | Termination of Pregnancy (ToP) |
|---|---|---|---|---|---|
| **Japan: JAOG** | Hospital-based , National | 1988 | 7 days | 22 weeks | Permitted, Not reported |
| **Malta: MCAR** | Population-based National | 2000 | 1 year | 20 weeks | Not permitted, Not reported |
| **Mexico: RYVEMCE** | Hospital-based National | 1980 | 72 hours | 20 weeks or 500 grams | Not permitted |
| **Mexico-Nuevo Leon: BDPSP** | Population-based Regional | 2014 | Hospital discharge | 20 weeks | Not permitted |
| **New Zealand** | Population-based National | 1979 | No limit | 20 weeks or 400 grams | Permitted, Reported |
| **Northern Netherlands** | Population-based Regional | 1993 | 10 years | 24 weeks | Permitted, Reported |
| **Norway: MBRN** | Population-based National | 1974 | 1 year | 20 weeks or 300 grams | Permitted, Reported |
| **Russia-Moscow Region: MRRCM** | Population-based Regional | 2001 | 1 year | 28 weeks | Permitted, Reported |
| **Saudi Arabia** | Hospital-based National | 2012 | 2 years | >= 16 weeks | Permitted in few cases. Only major malformations reported. |
| **Slovak Republic** | Population-based Regional | 2003 | 1 year | 28 weeks or 1000 grams | Permitted, Reported |
| **South America: ECLAMC** | Hospital-based Multinational | 1977 | Hospital discharge | 500 grams | Not permitted |
| **Spain: ECEMC** | Hospital-based National | 1979 | 3 days (***) | 24 weeks or 500 grams | Permitted, Not reported on a routine basis by all the participating hospitals |
| **Sweden** | Population-based National | 1974 | 28 days | 22 weeks | Permitted, Reported |
| **United Kingdom: Wales: CARIS** | Population-based Regional | 2005 | 1 year | 24 weeks | Permitted, Reported |
| **Ukraine: OMNI-Net** | Population-based Regional | 2001 | 1 year | >= 500 grams | Permitted, Only selected malformations reported |
| **USA-Arkansas: ARHMS** | Population-based Statewide | 2013 | 2 years | 20 weeks or >=350 grams | Permitted, Reported |
| **USA-Atlanta: MACDP** | Population-based Local | 1974 | 6 years | 20 weeks | Permitted, Reported |
| **USA-Iowa: IRCID** | Population-based Statewide | 2013 | 2 years | 20 weeks or >=350 grams | Permitted, Reported |
| **USA-Texas: BDES** | Population-based Statewide | 2004 | 1 year | 20 weeks (****) | Permitted, Reported |
| **USA-Utah UBDN** | Population-based Statewide | 2005 | No limit | 20 weeks | Permitted, Reported |

(*) Before 1993: 22 weeks; since 1993: 20 weeks
(**) Before 1998: 28 weeks; since 1998: 24 weeks
(***) For some cases a longer follow-up is performed
(****) Before 2001: 20 weeks. Since 2001: all stillbirths with documented birth defects included

# ICBDSR Definitions of the Reported Malformations

The following definitions have been adopted by all monitoring systems except when indicated in the Table "Deviations from ICBDSR Definitions".

**1. Anencephaly:** a congenital malformation characterized by the total or partial absence of the cranial vault, the covering skin, and the brain missing or reduced to small mass. Includes: craniorachischisis and infants with iniencephaly and other neural tube defects as encephalocele or open spina bifida, when associated with anencephaly.Excludes: acephaly, that is, absence of head observed in amorphous acardiac twins.

**2. Spina bifida:** a family of congenital malformation defects in the closure of the spinal column characterized by herniation or exposure of the spinal cord and/or meninges through an incompletely closed spine. Includes: meningocele, meningomyelocele, myelocele, myelomeningocele, rachischisis. Spina bifida is not counted when present with anencephaly. Excludes: spina bifida occulta, sacrococcygeal teratoma without dysraphism .

**3. Encephalocele:** a congenital malformation characterized by herniation of the brain and/or meninges through a defect in the skull. Encephalocele is not counted when present with spina bifida.

**4. Microcephaly:** a congenitally small cranium, defined by an occipito-frontal circumference (OFC) 3 standard deviation below the age- and sex-appropriate distribution curves. [If using a different definition or cut-off point (e.g., 2 standard deviations), report but specify criteria]. Excludes: microcephaly associated with anencephaly or encephalocele.

**5. Holoprosencephaly**: a congenital malformation of the brain, characterized by various degrees of incomplete lobation of the brain hemispheres. Olfactory nerve tract may be absent. Holoprosencephaly includes cyclopia, ethmocephaly, cebocephaly, and premaxillary agenesis.

**6. Hydrocephaly:** a congenital malformation characterized by dilatation of the cerebral ventricles, not associated with a primary brain atrophy, with or without enlargement of the head, and diagnosed at birth. Not counted when present with encephalocele or spina bifida. Excludes: macrocephaly without dilatation of ventricular system, skull of macerated fetus, hydranencephaly, holoprosencephaly, and postnatally acquired hydrocephalus.

**7. Anophthalmos/microphthalmos:** apparently absent or small eyes. Some normal adnexal elements and eyelids are usually present. In microphthalmia, the corneal diameter is usually less than 10 mm. and the antero-posterior diameter of the globe is less than 20 mm.

**8. Anotia/microtia**: a congenital malformation characterized by absent parts of the pinna (with or without atresia of the ear canal) commonly expressed in grades (I-IV) of which the extreme form (grade IV) is anotia, absence of pinna. Excludes: small, normally shaped ears, imperforate auditory meatus with a normal pinna, dysplastic and low set ears.

**9. Transposition of great vessels**: a cardiac defect where the aorta exits from the right ventricle and the pulmonary artery from the left ventricle, with or without other cardiac defects. Includes: double outlet ventricle and the so-called corrected transposition.

**10. Tetralogy of Fallot:** a condition characterized by ventricular septal defect, overriding aorta, infundibular pulmonary stenosis, and often right ventricular hypertrophy.

**11. Hypoplastic left heart syndrome:** a cardiac defect with a hypoplastic left ventricle, associated with aortic and/or mitral valve atresia, with or without other cardiac defect.

**12. Coarctation of the aorta**: an obstruction in the descending aorta, almost invariably at the insertion of the ductus arteriosus

**13. Choanal atresia, bilateral:** congenital obstruction (membraneous or osseous) of the posterior choana or choanae. Excludes: choanal stenosis and congestion of nasal mucosa.

**14. Cleft palate without cleft lip:** a congenital malformation characterized by a closure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip. Includes: submucous cleft palate. Excludes: cleft palate with cleft lip, cleft uvula, functional short palate, and high narrow palate.

**15. Cleft lip with or without cleft palate:** a congenital malformation characterized by partial or complete clefting of the upper lip, with or without clefting of the alveolar ridge or the hard palate. Excludes: midline cleft of upper or lower lip and oblique facial fissure (going towards the eye).

**16. Oesophageal atresia/stenosis:** a congenital malformation characterized by absence of continuity or narrowing of the esophagus, with or without tracheal fistula. Includes: tracheoesophageal fistula with or without mention of atresia or stenosis of oesophagus.

**17**. **Small intestine atresia/stenosis:** complete or partial occlusion of the lumen of a segment of the small intestine. It can involve a single area or multiple areas of the jejunum or ileum. Excludes: duodenal atresia.

**18. Anorectal atresia/stenosis:** a congenital malformation characterized by absence of continuity of the anorectal canal or of communication between rectum and anus, or narrowing of anal canal, with or without fistula to neighboring organs. Excludes: mild stenosis which does not need correction, and ectopic anus.

**19. Undescended testis:** bilateral undescended testes in at term newborn or at least unilateral undescended testis in males more than 1 year of age. Excludes: retractile testis.

**20. Hypospadias:** a congenital malformation characterized by the opening of the urethra on the ventral side of the penis, distally to the sulcus. Includes: penile, scrotal, and perineal hypospadias. Excludes: glandular or first-degree hypospadias and ambiguous genitalia (intersex or pseudohermaphroditism).

**21**. **Epispadias:** a congenital malformation characterized by the opening of the urethra on the dorsal surface of the penis. Not counted when part of exstrophy of the bladder.

**22. Indeterminate sex:** genital ambiguity at birth that does not readily allow for phenotypic sex determination. Includes: male or female, true or pseudohermaphroditism.

**23. Renal agenesis:** a congenital malformation characterized by complete absence of kidneys bilaterally or severely dysplastic kidneys.

**24. Cystic kidney:** a congenital malformation characterized by multiple cysts in the kidney. Includes: infantile polycystic kidney, multicystic kidney, other forms of cystic kidney and unspecified cystic kidney. Excludes: single kidney cyst.

**25. Bladder exstrophy:** complex malformation characterized by a defect in the closure of the lower abdominal wall and bladder. Bladder opens in the ventral wall of the abdomen between the umbilicus and the symphysis pubis. It is often associated with epispadias and structural anomalies of the pubic bones.

**26. Polydactyly, preaxial**: extra digit(s) on the radial side of the upper limb or the tibial side of the lower limb. It can affect the hand, the foot, or both.

**27. Limb reduction defects:** a congenital malformation characterized by total or partial absence or severe hypoplasia of skeletal structures of the limbs. Includes: femoral hypoplasia. Excludes: mild hypoplasia with normal shape of skeletal parts, brachydactyly, finger or toe reduction directly associated with syndactyly, general skeletal dysplasia and sirenomelia.

**28. Diaphragmatic hernia:** a congenital malformation characterized by herniation into the thorax of abdominal contents through a defect of the diaphragm. Includes: total absence of the diaphragm. Excludes: hiatus hernia, eventration and phrenic palsy.

**29. Abdominal wall defects:** cases specified as omphalocele and/or gastroschisis plus unspecified cases.

**30. Omphalocele:** a congenital malformation characterized by herniation of abdominal contents through the umbilical insertion and covered by a membrane which may or may not be intact. Excludes: gastroschisis (para-umbilical hernia), a - or hypoplasia of abdominal muscles, skin-covered umbilical hernia.

**31. Gastroschisis:** a congenital malformation characterized by visceral herniation usually through a right side abdominal wall defect to an intact umbilical cord and not covered by a membrane. Excludes: a-or hypoplasia of abdominal muscles, skin-covered umbilical hernia, omphalocele.

**32. Prune belly sequence:** a complex congenital malformation characterized by deficient abdominal muscle and urinary obstruction/distension. It can be caused by urethral obstruction secondary to posterior urethral valves or urethral atresia. In the affected fetus the deficiency of the abdominal muscle may not be evident. It can be associated with undescended testes, clubfoot, and limb deficiencies.

**33. Trisomy 13:** a congenital chromosomal malformation syndrome associated with extra chromosome 13 material. Includes: translocation and mosaic trisomy 13.

**34. Trisomy 18:** a congenital chromosomal malformation syndrome associated with extra chromosome 18 material.
Includes: translocation and mosaic trisomy 18

**35. Down syndrome:** a congenital chromosomal malformation syndrome characterized by a well known pattern of minor and major anomalies and associated with excess chromosomal 21 material. Includes: trisomy mosaicism and translocations of chromosome 21.

# Deviations from the ICBDSR Definitions by Registry

| | Encephalocele | Microcephaly | Arhinencephaly / Holoprosencephaly | Hydrocephaly | Anophthalmos / Microphthalmos | Anotia | Transposition of great vessels | Tetralogy of Fallot | Choanal atresia, bilateral | Cleft palate without cleft lip | Cleft lip with or without cleft palate | Oesophageal atresia / stenosis | Small intestine atresia / stenosis | Anorectal atresia / stenosis | Undescended testis | Hypospadias | Epispadias | Indeterminate sex | Renal agenesis | Cystic kidney | Polydactyly, preaxial | Limb reduction defects | Prune belly sequence | Trisomy 13 | Trisomy 18 | Down syndrome |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| **Argentina: RENAC** | 1 | 3 | | | 2 | | | | | | | | | | | | | | | | | | | | | 2 |
| **Australia: WARDA** | | | | | | | | | 11 | | | | | | | 25 | | | 28 | | 35 | | | | | |
| **Canada: Alberta ACASS** | | 2 | | | 2 | 7 | 8 | | 11,12 | | | | | | | 25 | | | | | 35 | | | | | 2 |
| **Canada: CCASS** | 1 | 2 | | 6 | 2 | | | | 11,12 | 14 | | | 18 | 21 | 23 | 25 | 26 | | 28 | 31 | 35 | | 40 | 2 | 2 | 2 |
| **Chile-Maule: RRMC-SSM** | 1 | 2 | | 24 | | | | | 11 | | 15,16 | | | | | | | | | | | | | 2 | 2 | 2 |
| **Colombia: BCMSP** | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| **Costa Rica: CREC** | | | | 6 | | | 9 | | 11,12 | | | | | | | | 26 | 27 | 28 | 31 | 35 | | | 2 | 2 | 2 |
| **Cuba: RECUMAC** | 1 | 2 | | 6 | 2 | 7 | | | 11 | 14 | 15 | | 18 | | | 25 | 26 | 27 | 28 | 32 | 35 | 37 | | 2 | 2 | 2 |
| **Czech Republic** | | | | | | | | | | | | | | | | 25 | | | | | 35 | | | | | |
| **Finland** | | 2 | | | 2 | 42 | 8 | | 11,12 | | | | | | | 25 | | 27 | | 32 | | | | 2 | 2 | 2 |
| **France: REMERA** | | | | | | | | | | | | | | | | 25 | | | | | | | | | | 2 |
| **France: Paris** | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| **Germany: Saxony-Anhalt** | | 2,3 | | | | | 9 | | 11 | | | | 19 | | | 25 | | | | 32 | 36 | 38 | | 2 | 2 | 2 |
| **Hungary** | 1 | 2 | | | 2 | | 9 | | | | | | | | | 25 | 26 | | | | 35 | 38,39 | | 2 | 2 | 2 |
| **Iran: TRoCA** | 1 | | 4 | 6 | | | 9 | | | | | | 18 | 21 | | | | | | | 35 | 38 | | | | 2 |
| **Israel: IBDMS** | | | | | | | 8 | | | | | | | | | 25 | | | | 33 | | | | | | |
| **Italy: BDRCam** | | | | | | | | | | | | | | | | | | | | | | | | 2 | 2 | 2 |
| **Italy: IMER** | | | | | | | | | | | | | | | | 25 | | | | | 35 | | | | | |
| **Italy: North East** | | | 5 | | 2 | | | | | 13 | 15 | 17 | 18,20 | 22 | | | | | 29 | | 35 | | | | | 2 |
| **Italy-Tuscany: RTDC** | | | | | | | 8 | | | | | | | | | | | | | | | | | | | |
| **Italy-Lombardy:CMLR** | | 3 | | | | | | | 11 | | | | 18 | | | 25 | | | 28 | | 35 | | | | | |
| **Japan: JAOG** | | 2 | | | 2 | | | | | | | | | | | | | | | 31 | | | | | | |
| **Malta** | | 2 | | | 2 | | 9 | | 11 | | | | | | | | | 27 | | 31 | 35 | 37 | | 2 | 2 | 2 |
| **Mexico: RYVEMCE** | | 2 | | | 2 | | | | 11,12 | | | | 18 | | | | | 27 | 28 | 30 | 35 | | | 2 | 2 | 2 |
| **Mexico-Nuevo Leon: BDSP** | | | | 6 | 2 | 7 | 9 | | 11 | 14 | 15 | | | | | 25 | | 27 | 29 | | 35 | | | 2 | 2 | 2 |
| **New Zealand** | | | | | 2 | | | | | | | | | | | 25 | 26 | | | | | | | 2 | 2 | 2 |
| **Northern Netherlands** | | | | | | | | | | | | | | | 24 | 25 | | | | | 35 | | | | | |
| **Norway: MBRN** | | | | | | | | | | | | | | | | | | | | | | | | | | |
| **Saudi Arabia Kingdom: MSD-BDR** | | 3 | | | | | 9 | | | | | | 19,3 | | 23 | 25 | | | 29 | 32,3 | 35 | 37,4 | | | | 2 |
| **Slovak Republic** | | | | | | | | | | | 15 | | | | | 25 | | | | | 35 | | | | | 2 |
| **South America: ECLAMC** | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| **Spain: ECEMC** | | 3 | | | 2 | | | | | | | | | | | | | 27 | | | | 37 | | | | 2 |
| **Sweden** | | 2 | | | 2 | | | | 11 | | | | | | | 25 | | | 28 | 32 | | | | | | 2 |
| **Ukraine** | | 41 | | 6 | | | 9 | | | | 16 | | | | | | | 27 | | | | | | 2 | 2 | 2 |
| **United Kingdom - Wales: CARIS** | 1 | 2 | | | 2 | 7 | 8 | | | | | | | | 24 | 25 | | | | | | | | 2 | 2 | 2 |
| **USA-Arkansas: ARHMS** | | | | | | | | | | | | | | | | | | | | | | | | | | |
| **USA: Atlanta: MACDP** | | | | | | | | | 12 | | 16 | | | | | | | | | | | | | | | |
| **USA-Iowa: IRCID** | | | | | | | | | | | | | | | | | | | | | | | | | | |
| **USA-Texas: BDES** | | | | | | 7 | | | 11,12 | | 15,16 | | | | | | | 27 | | | | | | | | |
| **USA-Utah UBDN** | | 43 | | | | | | | | | | | | | 24 | | | 24 | | | 24 | | | 2 | 2 | 2 |

1 = when present with spina bifida counted

2 = clinical diagnosis included

3 = OCF below 3rd percentile

4 = there may be other defects with the same code

5 = only cyclopia included

6 = hydranencephaly included

7 = absence of auricle

8 = double outlet right ventricle excluded

9 = all kind of transposition included

10 = Trilogy of Fallot included

11 = unilateral cases included

12 = stenosis included

13 = submucous cleft palate excluded

14 = cleft uvula included

15 = midline and oblique facial clefts included

16 = clefts of the alveolar ridge without cleft lip included

17 = stenosis excluded

18 = doudenal  atresia included

19 = doudenal stenosis excluded

20 = intestinal stenosis excluded

21 = large intestine atresia/stenosis included

22 = stenosis excluded

23 = no gestational age information (cases at all gestational age collected)

24 = registered when it is combined with other defects

25 = all types included

26 = epispadias counted with hypospadias

27 = genital ambiguity and absent genitalia included

28 = unilateral defects included

29 = severely dysplastic kidneys excluded

30 = single cyst included

31 = all kind of cystic kidney included

32 = all cystic kidneys are included except for single renal cysts

33 = AR polycystic kidney excluded

34 = some autosomalrecessive polycystyc kidneys are not excluded

35 = any type of polydactyly included

36 = polysyndactyly preaxial excluded

37 = any hypoplasia of skeletal limb structures included except branchydactyly and hypoplasia as part of skeletal dysplasia

38 = any hypoplasia of skeletal structures included

39 = sirenomelia included

40 = Prune belly sequence counted with Total abdominal wall defects

41 = includes congenital and postnatally diagnosed microcephaly (up to 1 year of age)

42 = anotia and microtia are reported without specification

43 =  Isolated cases not reported (from January 1, 2011)

# Argentina: RENAC

National Registry of Congenital Anomalies of Argentina

**History:**
The Programme started in November 2009 in 4 provinces of Argentina as a hospital-based registry. Since then it has grown in size and coverage, reaching all 24 provinces of the country. It was created with two main objectives: the classical one of generating epidemiological knowledge about distribution and determinants of birth defects, but also with the objective of improving care of affected newborns.

**Size and coverage:**
The number of participating hospitals has grown from 4 in 2009 to 120 at the present time. RENAC covers 300,000 annual births, approximately 70% of births in the public sector and 40% of births of the whole country. The registry works by now in public maternity hospitals with more than 1,000 annual births per year. In the next years it will includes maternity hospitals from de the nonpublic sector

**Legislation and funding:**
The Programme is funded by the National Center of Medical Genetics (CNGM) and the Programme of Medical Genetics National Network, under the National Ministry of Health. Information is disseminated to the stakeholders, including the participating neonatologists who feel empowered when using locally their own processed data. The dissemination is performed through a printed annual report, an annual meeting funded by the NMoH (attended by neonatologists, members of other health programs, clinical geneticists and authorities); and the electronic sending of reports to the stakeholders.

**Sources of ascertainment:**
Reporting is made by collaborating neonatologists at the maternity hospitals. The detection period lasts until discharge from the hospital, including live birth and stillbirths (with more than 500 grams) with major morphological birth defects. The neonatologists describe birth defects in an open field with a verbatim description. Each month, they send data to the Coordination through a restricted access website (a forum platform) that allows data sending, resolution of operational issues and discussion of clinical cases. Forum interaction allows social cohesion among all participants who feel themselves as members of the same team. To achieve a high and homogeneous coding quality, coding is performed by medical geneticists of the Coordination. To allow comparisons with other sources we use the ICD-10 with the British Pediatrics Association modification

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# Argentina: RENAC, 2012

| | |
|---|---|
| Live births (LB) | 237,728 |
| Stillbirths (SB) | 2,243 |
| Total births | 239,971 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 53 | 34 | nr | 3.63 |
| Spina bifida | 114 | 7 | nr | 5.04 |
| Encephalocele | 25 | 5 | nr | 1.25 |
| Microcephaly | 74 | 4 | nr | 3.25 |
| Holoprosencephaly | 68 | 8 | nr | 3.17 |
| Hydrocephaly | 176 | 7 | nr | 7.63 |
| Anophthalmos | 11 | 2 | nr | 0.54 |
| Microphthalmos | 27 | 2 | nr | 1.21 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 7 | 1 | nr | 0.33 |
| Microtia | 40 | 0 | nr | 1.67 |
| Unspecified Anotia/Microtia | 54 | 2 | nr | 2.33 |
| Transposition of great vessels | 50 | 1 | nr | 2.13 |
| Tetralogy of Fallot | 38 | 0 | nr | 1.58 |
| Hypoplastic left heart syndrome | 34 | 1 | nr | 1.46 |
| Coarctation of aorta | 36 | 0 | nr | 1.50 |
| Choanal atresia, bilateral | 8 | 0 | nr | 0.33 |
| Cleft palate without cleft lip | 77 | 5 | nr | 3.42 |
| Cleft lip with or without cleft palate | 295 | 8 | nr | 12.63 |
| Oesophageal atresia/stenosis with or without fistula | 72 | 0 | nr | 3.00 |
| Small intestine atresia/stenosis | 25 | 0 | nr | 1.04 |
| Anorectal atresia/stenosis | 61 | 4 | nr | 2.71 |
| Undescended testis (36 weeks of gestation or later) | 31 | 0 | nr | 1.29 |
| Hypospadias | 34 | 0 | nr | 1.42 |
| Epispadias | 5 | 0 | nr | 0.21 |
| Indeterminate sex | 43 | 13 | nr | 2.33 |
| Renal agenesis | 8 | 6 | nr | 0.58 |
| Cystic kidney | 92 | 6 | nr | 4.08 |
| Bladder exstrophy | 3 | 2 | nr | 0.21 |
| Polydactyly, preaxial | 39 | 0 | nr | 1.63 |
| Total Limb reduction defects (include unspecified) | 122 | 6 | nr | 5.33 |
| Transverse | 54 | 2 | nr | 2.33 |
| Preaxial | 23 | 2 | nr | 1.04 |
| Postaxial | 4 | 1 | nr | 0.21 |
| Intercalary | 2 | 0 | nr | 0.08 |
| Mixed | 20 | 2 | nr | 0.92 |
| Unspecified | 52 | 1 | nr | 2.21 |
| Diaphragmatic hernia | 78 | 4 | nr | 3.42 |
| Omphalocele | 42 | 7 | nr | 2.04 |
| Gastroschisis | 178 | 14 | nr | 8.00 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 6 | 1 | nr | 0.29 |
| Trisomy 13 | 8 | 1 | nr | 0.38 |
| Trisomy 18 | 22 | 8 | nr | 1.25 |
| Down syndrome, all ages (include age unknown) | 406 | 11 | nr | 17.38 |
| <20 | 49 | 2 | nr | 10.17 |
| 20-24 | 58 | 1 | nr | 8.22 |
| 25-29 | 50 | 0 | nr | 9.56 |
| 30-34 | 54 | 2 | nr | 14.94 |
| 35-39 | 93 | 3 | nr | 49.00 |
| 40-44 | 88 | 2 | nr | 151.41 |
| 45+ | 11 | 1 | nr | 269.06 |
| unknown | 3 | 0 | nr | --- |

nr = data not reported or not available

# Australia: WARDA

## Western Australian Register of Developmental Anomalies

**History:**
The Register is located in a teaching obstetric hospital. In January, 2011, notification to the Register became statutory and the Western Australian Cerebral Palsy Register was combined with the Western Australian Birth Defects Registry, to become the Western Australian Register of Developmental Anomalies (WARDA). The objectives of the Register remain the same: to establish how often birth defects and cerebral palsy occur, to conduct research into their causes and prevention, to provide health professionals and the public with information about birth defects and cerebral palsy, and to monitor and evaluate screening, treatment and prevention programs.

**Size and coverage:**
Population-based in the state of Western Australia. 30,000 birth a year, ~6% reported with a birth defect; 2.5 per 1000 with cerebral palsy. Birth defects diagnosed prenatally and up to the age of 6 years, in stillbirths, terminations of pregnancy and live births are included. Cerebral palsy of all types and severity, including postnatal causes and diagnosed up to 5 years of age is now also included. The Register covers births from 1980 for birth defects and from 1956 for cerebral palsy.

**Legislation and funding:**
Following a period of short term funding from both Federal and State sources, the Register is now wholly funded by the Western Australian Department of Health. Notification to the Register by medical practitioners was made statutory in January 2011.

**Sources of ascertainment:**
Statutory sources: Midwives' Notification of Birth Forms (all births over 20 weeks gestation), Death Certificates (perinatal, infant and childhood); Hospital Morbidity (all hospital discharges in Western Australia); medical practitioners and hospitals. The latter two sources include notifications from maternity and paediatric hospitals, obstetricians, paediatricians, orthopaedic surgeons, cytogenetic laboratories, pathology services (including prenatal screening services), child development services, ultrasound practices and genetic services.

**Exposure information:**
No exposure information is routinely collected.

**Background information:**
The data on WARDA are routinely linked to the linked dataset of all births, deaths and hospital admissions for Western Australia. This linkage provides information on variables such as maternal and paternal age, labour and delivery data, and maternal illnesses, for both cases of developmental anomalies (numerators) and all births in Western Australia (denominators). Data from the Register are provided to the National Perinatal Statistics and Epidemiology Unit and the Australia Cerebral Palsy Register. Further information is available on the WARDA website:
http://kemh.health.wa.gov.au/services/register_developmental_anomalies/

**Addresses and Staff:**
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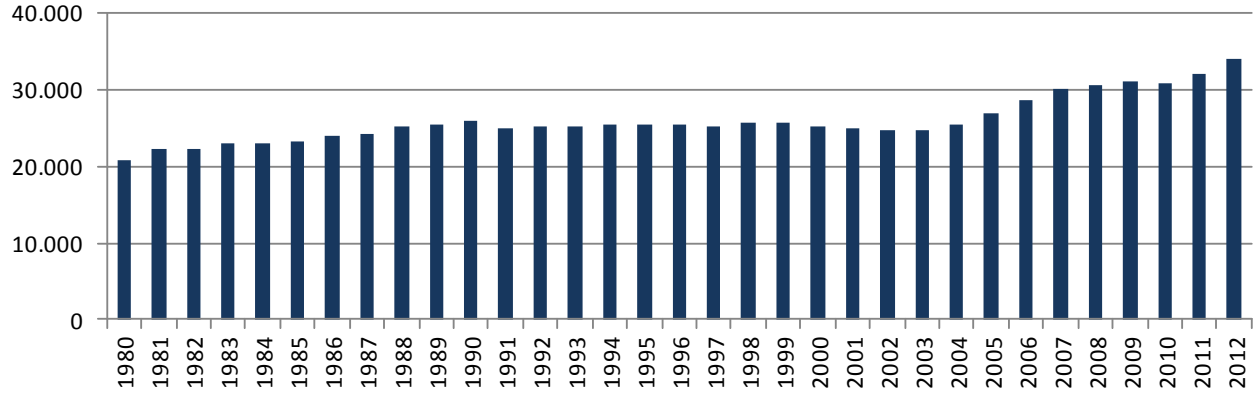
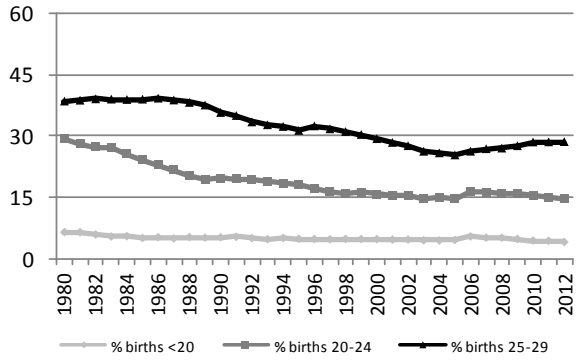
**Phone:** 618 9340 2721
**Fax:** 618 9340 2636
**E-mail:** caroline.bower@health.wa.gov.au
**Website:**
http://kemh.health.wa.gov.au/services/register_developmental_anomalies/

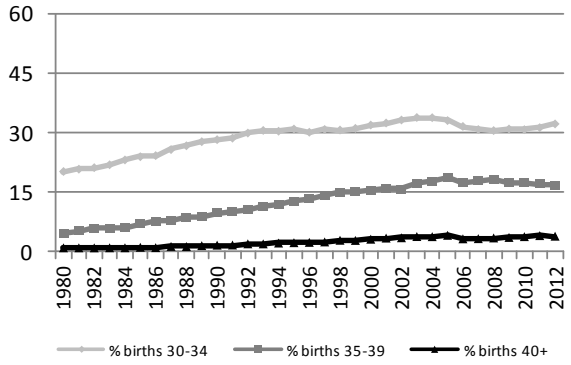
# Australia: WARDA

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 41 | 93.2 | Cystic kidney | 25 | 26.9 |
| Spina bifida | 43 | 75.4 | Limb reduction defects | 28 | 45.9 |
| Encephalocele | 14 | 82.3 | Diaphragmatic hernia | 8 | 22.8 |
| Holoprosencephaly | 5 | 62.5 | Omphalocele | 26 | 74.3 |
| Hydrocephaly | 48 | 65.7 | Gastroschisis | 2 | 5.5 |
| Hypoplastic left heart syndrome | 11 | 57.9 | Trisomy 13 | 17 | 73.9 |
| Cleft palate without cleft lip | 3 | 3.6 | Trisomy 18 | 65 | 82.3 |
| Cleft lip with or without cleft palate | 11 | 12.3 | Down syndrome | 215 | 73.6 |
| Renal agenesis | 18 | 29.0 | | | |

Total ToPs with births defects = 689 (Ratio ToPs/Births: 7.09 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Australia: WARDA, 2012

| | |
|---|---|
| Live births (LB) | 33,862 |
| Stillbirths (SB) | 237 |
| Total births | 34,099 |
| Number of terminations of pregnancy (ToP) for birth defects | 224 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 16 | 4.69 |
| Spina bifida | 3 | 0 | 13 | 4.69 |
| Encephalocele | 0 | 0 | 4 | 1.17 |
| Microcephaly | 10 | 0 | 3 | 3.81 |
| Holoprosencephaly | 0 | 0 | 1 | 0.29 |
| Hydrocephaly | 5 | 0 | 18 | 6.75 |
| Anophthalmos | 0 | 0 | 2 | 0.59 |
| Microphthalmos | 7 | 0 | 0 | 2.05 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.29 |
| Microtia | 3 | 0 | 0 | 0.88 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 12 | 1 | 3 | 4.69 |
| Tetralogy of Fallot | 6 | 0 | 2 | 2.35 |
| Hypoplastic left heart syndrome | 3 | 0 | 3 | 1.76 |
| Coarctation of aorta | 5 | 0 | 0 | 1.47 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 24 | 2 | 0 | 7.62 |
| Cleft lip with or without cleft palate | 30 | 0 | 3 | 9.68 |
| Oesophageal atresia/stenosis with or without fistula | 6 | 0 | 1 | 2.05 |
| Small intestine atresia/stenosis | 8 | 1 | 0 | 2.64 |
| Anorectal atresia/stenosis | 15 | 0 | 3 | 5.28 |
| Undescended testis (36 weeks of gestation or later) | 85 | 0 | 0 | 24.93 |
| Hypospadias | 104 | 0 | 0 | 30.50 |
| Epispadias | 1 | 0 | 0 | 0.29 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 11 | 0 | 5 | 4.69 |
| Cystic kidney | 22 | 0 | 7 | 8.50 |
| Bladder exstrophy | 1 | 0 | 0 | 0.29 |
| Polydactyly, preaxial | 31 | 1 | 3 | 10.26 |
| Total Limb reduction defects (include unspecified) | 15 | 0 | 7 | 6.45 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 7 | 0 | 0 | 2.05 |
| Omphalocele | 0 | 0 | 5 | 1.47 |
| Gastroschisis | 12 | 0 | 1 | 3.81 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 3 | 5 | 2.35 |
| Trisomy 18 | 2 | 0 | 25 | 7.92 |
| Down syndrome, all ages (include age unknown) | 25 | 0 | 73 | 28.74 |
| <20 | 1 | 0 | 1 | 14.84 |
| 20-24 | 1 | 0 | 3 | 8.05 |
| 25-29 | 7 | 0 | 6 | 13.44 |
| 30-34 | 5 | 0 | 15 | 18.36 |
| 35-39 | 8 | 0 | 27 | 61.71 |
| 40-44 | 3 | 0 | 16 | 153.72 |
| 45+ | 0 | 0 | 5 | 746.27 |
| unknown | 0 | 0 | 1 | --- |

nr = data not reported or not available

# Australia: WARDA, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **43,036** | **115,671** | **125,934** | **127,109** | **126,876** | **130,651** | **155,021** |
| Anencephaly | | 7.67 | 9.60 | 7.86 | 8.58 | 6.78 | 5.43 | 5.48 |
| Spina bifida | | 8.37 | 8.65 | 9.61 | 8.18 | 6.70 | 7.42 | 5.68 |
| Encephalocele | | 2.09 | 1.73 | 1.83 | 1.73 | 1.42 | 1.22 | 2.06 |
| Microcephaly | | 5.58 | 5.71 | 5.24 | 5.35 | 6.54 | 6.12 | 3.81 |
| Holoprosencephaly | | 0.93 | 1.47 | 1.75 | 2.28 | 2.21 | 1.99 | 1.55 |
| Hydrocephaly | | 8.13 | 6.66 | 6.83 | 9.91 | 8.35 | 7.58 | 7.74 |
| Anophthalmos | | 0.70 | 0.61 | 0.40 | 0.24 | 0.95 | 0.31 | 0.26 |
| Microphthalmos | | 1.63 | 1.64 | 2.14 | 1.81 | 2.44 | 1.38 | 0.97 |
| Unspecified Anophthalmos/Microphthalmos | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | | 1.86 | 1.56 | 1.83 | 2.44 | 2.36 | 1.91 | 1.29 |
| Microtia | | 1.16 | 0.61 | 0.87 | 1.57 | 1.73 | 1.00 | 1.03 |
| Unspecified Anotia/Microtia | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | 2.09 | 4.58 | 4.29 | 4.96 | 4.18 | 5.43 | 3.87 |
| Tetralogy of Fallot | | 2.09 | 3.63 | 3.26 | 3.70 | 3.23 | 3.44 | 2.90 |
| Hypoplastic left heart syndrome | | 2.32 | 1.64 | 2.06 | 2.44 | 1.26 | 1.68 | 2.19 |
| Coarctation of aorta | | 5.34 | 5.45 | 4.84 | 5.59 | 5.28 | 6.74 | 4.26 |
| Choanal atresia, bilateral | | 0.70 | 1.64 | 1.03 | 0.71 | 0.87 | 0.61 | 0.45 |
| Cleft palate without cleft lip | | 7.67 | 9.34 | 8.66 | 10.86 | 12.53 | 12.09 | 10.19 |
| Cleft lip with or without cleft palate | | 11.15 | 13.40 | 11.99 | 11.09 | 12.77 | 12.86 | 10.84 |
| Oesophageal atresia/stenosis with or without fistula | | 4.18 | 2.25 | 3.65 | 2.91 | 3.15 | 4.52 | 4.06 |
| Small intestine atresia/stenosis | | 3.49 | 2.59 | 2.70 | 2.12 | 3.07 | 2.68 | 2.90 |
| Anorectal atresia/stenosis | | 6.04 | 4.75 | 6.03 | 7.08 | 6.23 | 6.51 | 4.64 |
| Undescended testis (36 weeks of gestation or later) | | 65.06 | 64.84 | 69.48 | 61.99 | 55.41 | 44.47 | 39.54 |
| Hypospadias | | 23.47 | 30.43 | 29.22 | 36.03 | 38.62 | 34.90 | 34.64 |
| Epispadias | | 0.46 | 0.17 | 0.40 | 0.24 | 0.24 | 0.15 | 0.19 |
| Indeterminate sex | | 0.00 | 0.17 | 0.32 | 0.24 | 0.24 | 0.15 | 0.06 |
| Renal agenesis | | 2.32 | 4.24 | 3.65 | 4.17 | 4.81 | 5.43 | 5.61 |
| Cystic kidney | | 2.79 | 3.29 | 5.08 | 7.79 | 9.30 | 8.65 | 9.48 |
| Bladder exstrophy | | 0.00 | 0.17 | 0.24 | 0.08 | 0.47 | 0.15 | 0.13 |
| Polydactyly, preaxial | | 8.13 | 10.37 | 10.72 | 12.27 | 11.35 | 12.32 | 12.06 |
| Total Limb reduction defects (include unspecified) | | 5.11 | 5.79 | 6.35 | 7.47 | 10.56 | 9.34 | 6.97 |
| Transverse | | nr | nr | nr | nr | nr | nr | nr |
| Preaxial | | nr | nr | nr | nr | nr | nr | nr |
| Postaxial | | nr | nr | nr | nr | nr | nr | nr |
| Intercalary | | nr | nr | nr | nr | nr | nr | nr |
| Mixed | | nr | nr | nr | nr | nr | nr | nr |
| Unspecified | | nr | nr | nr | nr | nr | nr | nr |
| Diaphragmatic hernia | | 4.88 | 2.42 | 3.02 | 4.01 | 4.02 | 3.83 | 3.42 |
| Omphalocele | | 1.86 | 2.51 | 3.02 | 3.30 | 3.47 | 4.98 | 4.32 |
| Gastroschisis | | 1.16 | 1.64 | 1.99 | 2.91 | 3.94 | 3.75 | 3.81 |
| Unspecified Omphalocele/Gastroschisis | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Prune belly sequence | | 0.46 | 0.69 | 0.40 | 0.63 | 0.16 | 0.00 | 0.06 |
| Trisomy 13 | | 0.46 | 0.86 | 0.95 | 1.81 | 2.36 | 3.29 | 2.90 |
| Trisomy 18 | | 1.63 | 1.47 | 2.86 | 3.93 | 6.62 | 7.65 | 7.74 |
| Down syndrome, all ages (include age unknown) | | 10.46 | 13.66 | 15.33 | 19.43 | 21.52 | 27.25 | 29.16 |
| <20 | | 0.00 | 9.71 | 6.21 | 8.15 | 8.40 | 13.14 | 4.12 |
| 20-24 | | 4.86 | 4.43 | 8.72 | 6.86 | 6.44 | 7.56 | 8.25 |
| 25-29 | | 9.03 | 8.86 | 7.51 | 8.98 | 11.50 | 9.93 | 10.02 |
| 30-34 | | 13.63 | 15.13 | 14.21 | 19.19 | 17.10 | 19.94 | 18.00 |
| 35-39 | | 33.07 | 49.51 | 42.22 | 42.02 | 44.10 | 57.06 | 64.64 |
| 40-44 | | 81.52 | 183.04 | 183.49 | 183.67 | 149.00 | 160.93 | 215.92 |
| 45+ | | 833.33 | 483.87 | 447.76 | 408.16 | 397.73 | 516.43 | 387.93 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

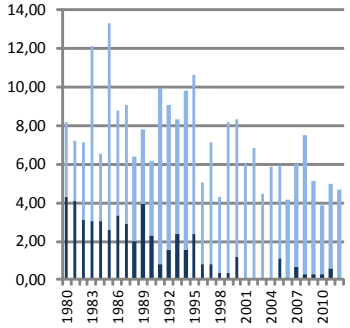
nr = data not reported or not available

* data include less than 5 years

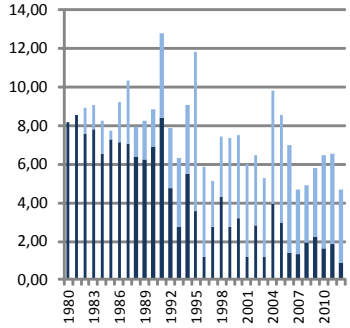
# Australia: WARDA, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

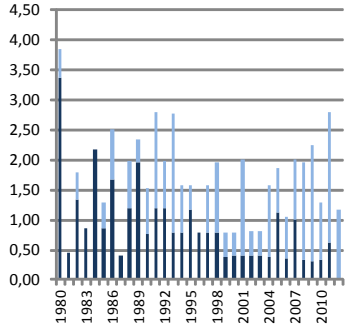
**Anencephaly**



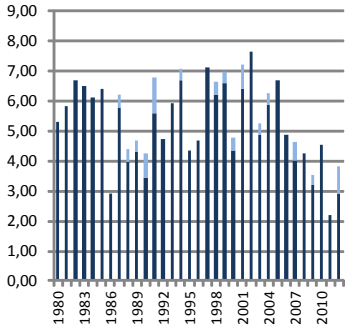

**Spina Bifida**



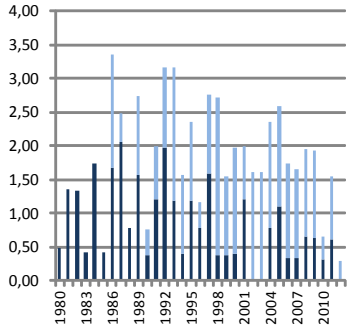

**Encephalocele**



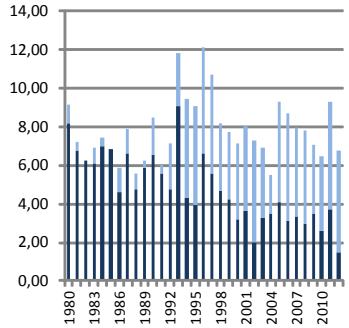

**Microcephaly**



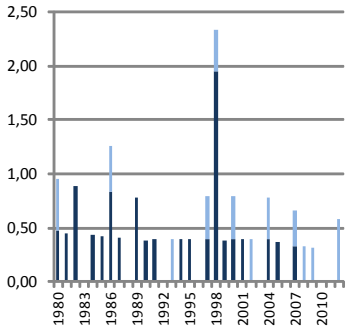

**Holoprosencephaly**



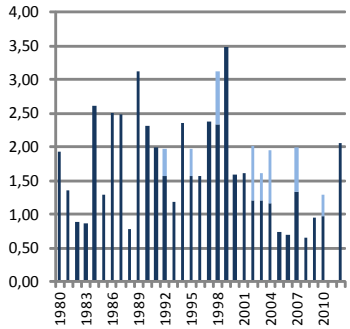

**Hydrocephaly**



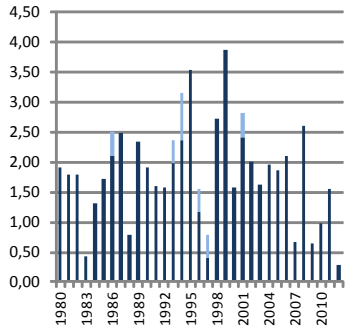

**Anophthalmos**



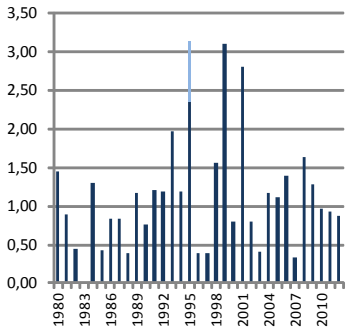

**Microphthalmos**



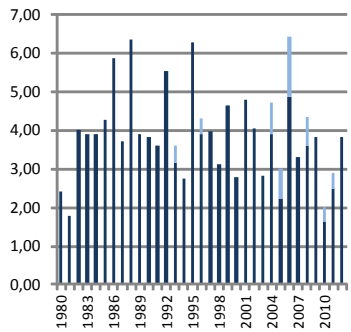

**Anotia**



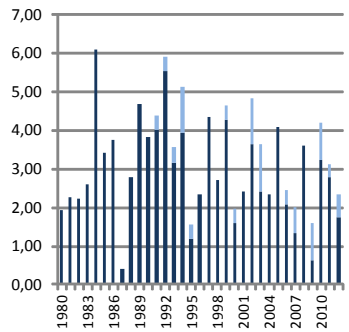

**Microtia**




**Transposition of great vessels**




**Tetralogy of Fallot**

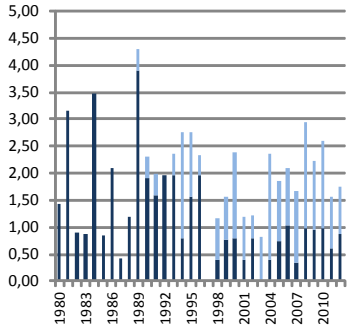



**L + S rates** **ToP rates**

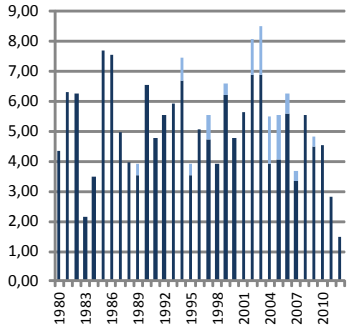
# Australia: WARDA, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

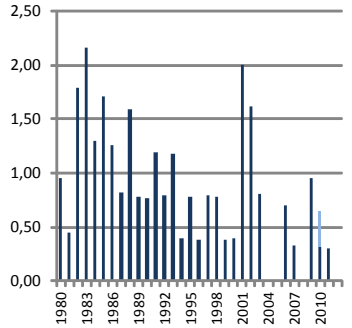
**Hypoplastic left heart syndrome**



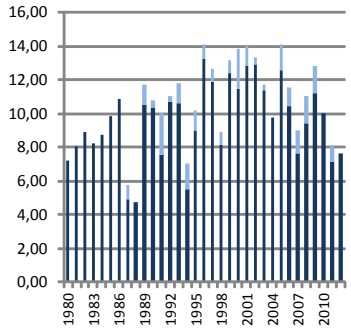

**Coarctation of aorta**



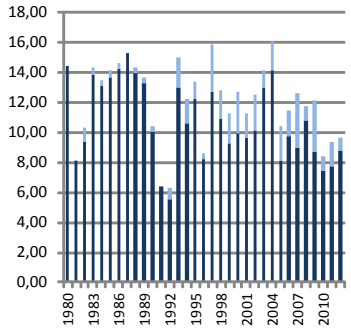

**Choanal atresia, bilateral**



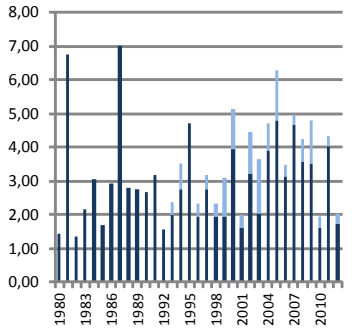

**Cleft palate without cleft lip**



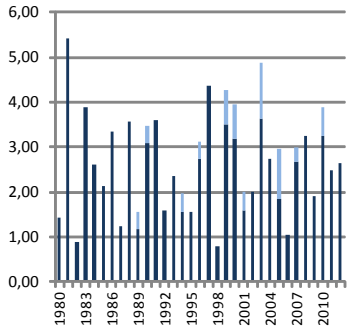

**Cleft lip with or without cleft palate**



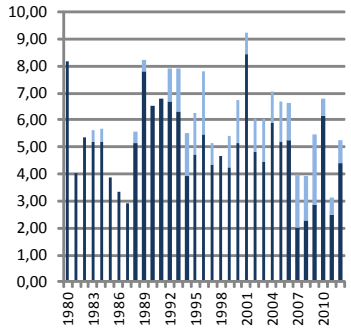

**phageal atresia/stenosis with or without f**



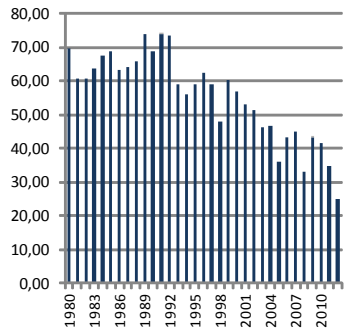

**Small intestine atresia/stenosis**



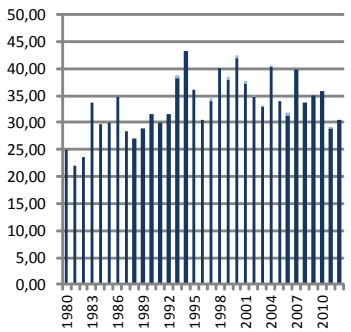

**Anorectal atresia/stenosis**



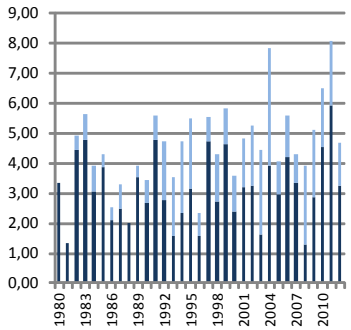

**Undescended testis**



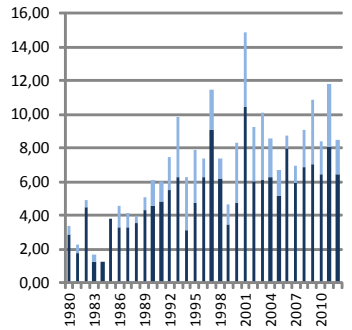

**Hypospadias**




**Renal agenesis**




**Cystic kidney**

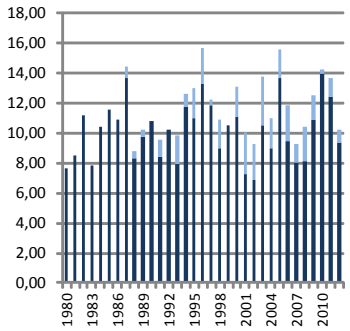



**L + S rates** **ToP rates**

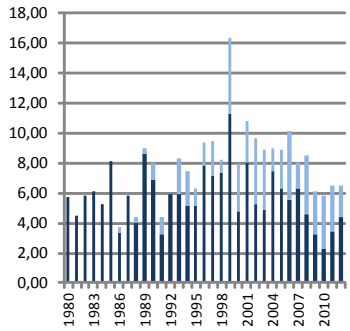
# Australia: WARDA, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

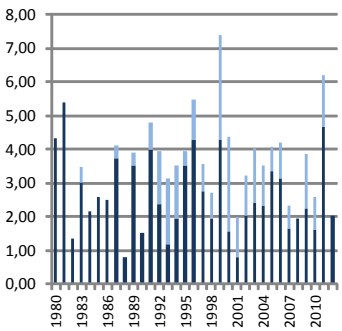
**Polydactyly, preaxial**



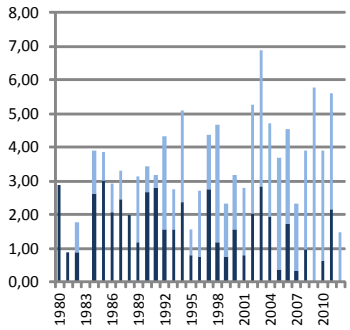

**Limb reduction defects**



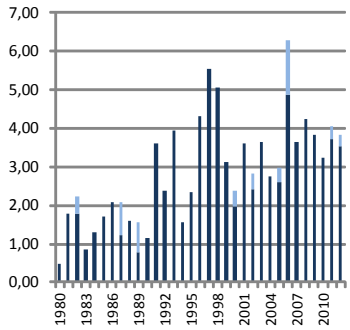

**Diaphragmatic hernia**



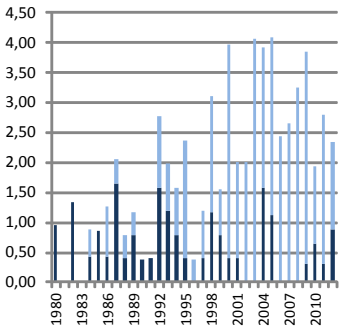

**Omphalocele**



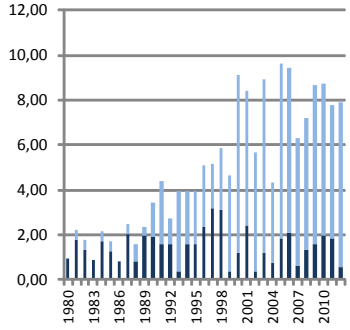

**Gastroschisis**



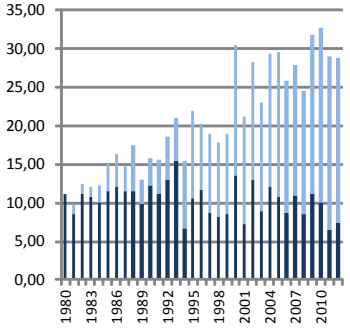

**Trisomy 13**



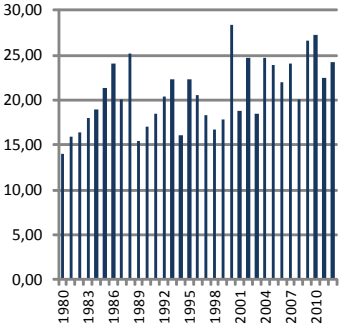

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Canada-Alberta: ACASS

## Alberta Congenital Anomalies Surveillance System

**History:**
The programme began in 1963 as a general Registry for Handicapped Children. This was disbanded in 1980 and continued as a surveillance system for live and stillborn infants with congenital anomalies who were born in the province of Alberta.

**Size and coverage:**
All live and stillbirths in the province are covered which at present comprise about 50,000 births per year. The definition of stillbirth is 20 weeks or more gestation or 500 grams or more birth weight. The vast majority of births occur in hospital (approximately 97%). Since 1997, fetuses with congenital anomalies who were either spontaneously lost before 20 weeks or where there was a termination of pregnancy as a result of prenatal diagnosis have been included.

**Legislation and funding:**
Although reporting is voluntary, the Health Information Act allows ACASS to receive and others to submit data on infants with congenital anomalies. The system is run by members of the Department of Medical Genetics, Alberta Children's Hospital - University of Calgary, reporting to Alberta Health, Surveillance and Assessment. Funding is from the Alberta Ministry of Health.

**Sources of ascertainment:**
Reports are obtained from physicians' notices of birth, live birth and stillbirth registrations, death registrations and a special congenital anomalies reporting form (CARF) from hospitals. The latter is based on discharge diagnosis, including readmissions for any reason up to one year of age. Additional sources include cytogenetics laboratories, the provincial metabolic screening programme and specialty clinics such as medical genetics.

**Exposure information:**
None is routine.

**Background information:**
Linkage studies are possible with other statistical data from Alberta Health.

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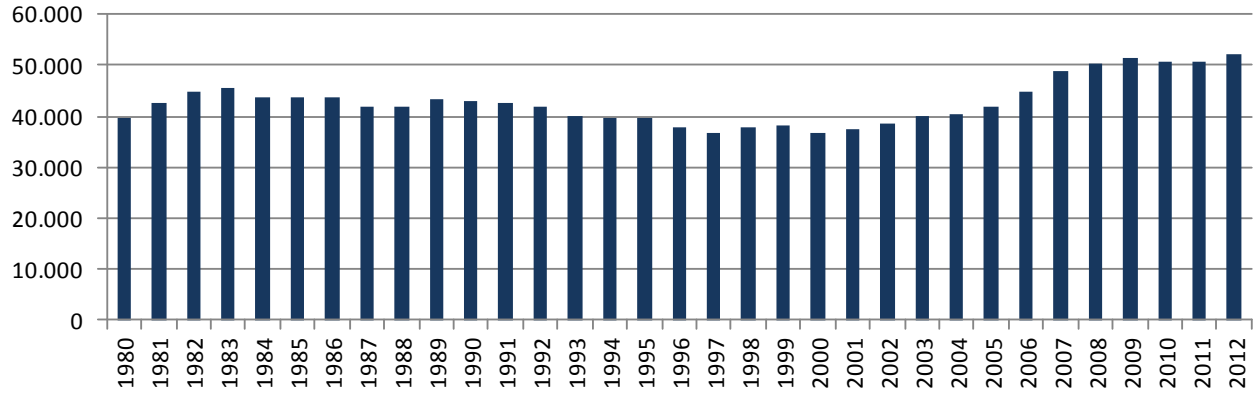
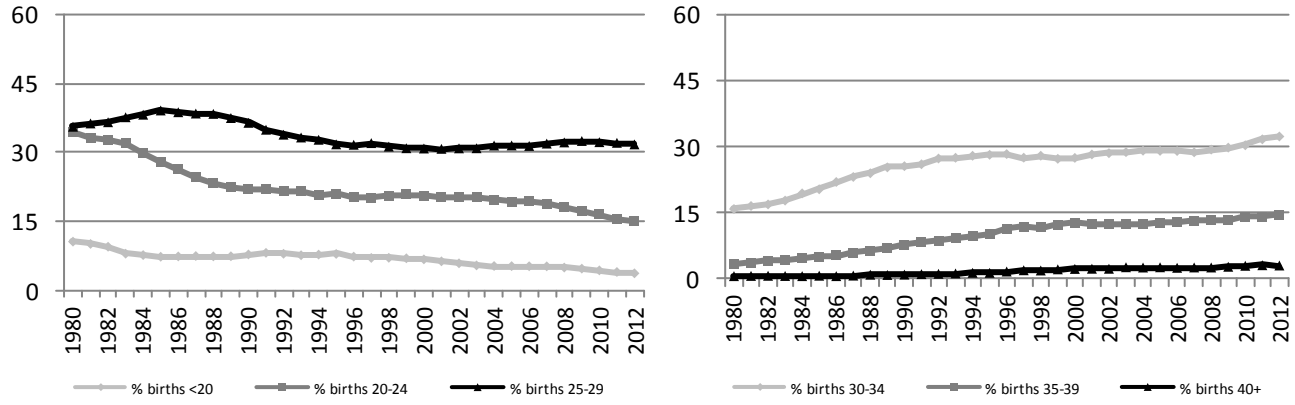
barbara.sibbald@albertahealthservices.ca

# Canada-Alberta: ACASS

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 15 | 45.5 | Cystic kidney | 9 | 7.3 |
| Spina bifida | 7 | 10.6 | Limb reduction defects | 40 | 21.6 |
| Encephalocele | 3 | 15.8 | Diaphragmatic hernia | 1 | 1.7 |
| Holoprosencephaly | 15 | 48.4 | Omphalocele | 29 | 41.4 |
| Hydrocephaly | 6 | 6.5 | Gastroschisis | 3 | 4.4 |
| Hypoplastic left heart syndrome | 4 | 8.7 | Trisomy 13 | 17 | 47.2 |
| Cleft palate without cleft lip | 5 | 3.7 | Trisomy 18 | 44 | 50.0 |
| Cleft lip with or without cleft palate | 12 | 6.5 | Down syndrome | 126 | 34.0 |
| Renal agenesis | 3 | 13.0 | | | |

Total ToPs with births defects = 392 (Ratio ToPs/Births: 2.55 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Canada-Alberta: ACASS, 2012

| | |
|---|---|
| Live births (LB) | 51,994 |
| Stillbirths (SB) | 324 |
| Total births | 52,318 |
| Number of terminations of pregnancy (ToP) for birth defects | 122 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 3 | 2 | 3 | 1.53 |
| Spina bifida | 18 | 4 | 0 | 4.21 |
| Encephalocele | 2 | 3 | 1 | 1.15 |
| Microcephaly | 16 | 6 | 0 | 4.21 |
| Holoprosencephaly | 4 | 3 | 4 | 2.10 |
| Hydrocephaly | 17 | 6 | 1 | 4.59 |
| Anophthalmos | 1 | 0 | 1 | 0.38 |
| Microphthalmos | 4 | 1 | 0 | 0.96 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 12 | 0 | 1 | 2.48 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 19 | 1 | 0 | 3.82 |
| Tetralogy of Fallot | 13 | 2 | 0 | 2.87 |
| Hypoplastic left heart syndrome | 14 | 2 | 1 | 3.25 |
| Coarctation of aorta | 24 | 3 | 0 | 5.16 |
| Choanal atresia, bilateral (*) | 7 | 0 | 0 | 1.34 |
| Cleft palate without cleft lip | 34 | 3 | 3 | 7.65 |
| Cleft lip with or without cleft palate | 45 | 6 | 4 | 10.51 |
| Oesophageal atresia/stenosis with or without fistula | 9 | 2 | 0 | 2.10 |
| Small intestine atresia/stenosis | 9 | 0 | 0 | 1.72 |
| Anorectal atresia/stenosis | 12 | 4 | 2 | 3.44 |
| Undescended testis (36 weeks of gestation or later) | 159 | 0 | 0 | 30.39 |
| Hypospadias | 128 | 0 | 0 | 24.47 |
| Epispadias | 1 | 0 | 0 | 0.19 |
| Indeterminate sex | 4 | 0 | 0 | 0.76 |
| Renal agenesis | 1 | 6 | 1 | 1.53 |
| Cystic kidney | 37 | 5 | 2 | 8.41 |
| Bladder exstrophy | 3 | 0 | 1 | 0.76 |
| Polydactyly (**) | 85 | 3 | 6 | 17.97 |
| Total Limb reduction defects (include unspecified) | 34 | 14 | 15 | 12.04 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 18 | 5 | 0 | 4.40 |
| Omphalocele | 9 | 4 | 8 | 4.01 |
| Gastroschisis | 16 | 1 | 2 | 3.63 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 1 | 0 | 0.19 |
| Trisomy 13 | 2 | 6 | 5 | 2.48 |
| Trisomy 18 | 6 | 8 | 14 | 5.35 |
| Down syndrome, all ages (include age unknown) | 76 | 9 | 41 | 24.08 |
| <20 | 2 | 0 | 0 | 10.57 |
| 20-24 | 9 | 0 | 0 | 11.39 |
| 25-29 | 9 | 1 | 4 | 8.44 |
| 30-34 | 22 | 2 | 11 | 20.74 |
| 35-39 | 26 | 4 | 16 | 61.07 |
| 40-44 | 7 | 2 | 10 | 131.12 |
| 45+ | 1 | 0 | 0 | 119.05 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

(*) Excludes stenosis but cannot distinguish whether uni - or bilateral;

(**) All included - axis often not reported.

# Canada-Alberta: ACASS, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **82,118** | **221,364** | **212,703** | **199,308** | **186,870** | **206,076** | **251,895** |
| Anencephaly | | 3.53 | 3.61 | 2.68 | 1.86 | 3.00 | 2.28 | 2.46 |
| Spina bifida | | 4.26 | 5.38 | 4.94 | 5.27 | 3.85 | 3.64 | 4.17 |
| Encephalocele | | 0.97 | 1.13 | 0.80 | 1.00 | 1.34 | 1.31 | 1.27 |
| Microcephaly | | 3.41 | 3.48 | 3.67 | 2.71 | 3.69 | 3.88 | 4.53 |
| Holoprosencephaly | | 0.37 | 0.54 | 1.03 | 1.15 | 1.61 | 2.04 | 2.38 |
| Hydrocephaly | | 6.45 | 6.14 | 4.61 | 5.12 | 5.14 | 6.11 | 6.23 |
| Anophthalmos | | 0.12 | 0.36 | 0.42 | 0.35 | 0.37 | 0.29 | 0.36 |
| Microphthalmos | | 1.10 | 0.86 | 1.22 | 0.90 | 1.93 | 1.21 | 1.03 |
| Unspecified Anophthalmos/Microphthalmos | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | | 0.00 | 0.18 | 0.19 | 0.30 | 0.54 | 0.49 | 0.24 |
| Microtia | | 0.12 | 0.45 | 0.66 | 1.40 | 1.12 | 1.80 | 2.34 |
| Unspecified Anotia/Microtia | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | 2.44 | 2.94 | 3.34 | 3.16 | 3.37 | 4.27 | 2.86 |
| Tetralogy of Fallot | | 1.22 | 2.21 | 3.20 | 2.71 | 2.09 | 2.09 | 2.90 |
| Hypoplastic left heart syndrome | | 2.31 | 2.26 | 2.16 | 2.11 | 3.05 | 2.81 | 3.10 |
| Coarctation of aorta | | 2.92 | 4.25 | 4.09 | 5.42 | 2.84 | 3.40 | 4.68 |
| Choanal atresia, bilateral | | 0.85 | 1.40 | 1.60 | 1.66 | 1.66 | 1.84 | 1.15 |
| Cleft palate without cleft lip | | 6.45 | 6.01 | 8.51 | 7.53 | 9.15 | 7.23 | 7.78 |
| Cleft lip with or without cleft palate | | 10.11 | 10.39 | 12.74 | 11.94 | 11.34 | 11.94 | 13.85 |
| Oesophageal atresia/stenosis with or without fistula | | 1.46 | 2.94 | 3.34 | 2.06 | 2.46 | 2.38 | 2.58 |
| Small intestine atresia/stenosis | | 0.61 | 0.81 | 1.41 | 1.30 | 2.09 | 1.36 | 1.59 |
| Anorectal atresia/stenosis | | 2.80 | 3.70 | 5.69 | 4.92 | 5.99 | 5.82 | 3.85 |
| Undescended testis (36 weeks of gestation or later) | | 25.21 | 26.79 | 30.32 | 23.78 | 23.60 | 25.96 | 27.31 |
| Hypospadias | | 16.68 | 19.20 | 25.43 | 21.57 | 19.37 | 20.19 | 23.30 |
| Epispadias | | 0.61 | 0.41 | 0.38 | 0.40 | 0.54 | 0.73 | 0.83 |
| Indeterminate sex | | 0.24 | 0.41 | 0.89 | 0.90 | 1.66 | 1.31 | 1.31 |
| Renal agenesis | | 2.19 | 2.48 | 2.49 | 1.56 | 1.61 | 1.46 | 1.47 |
| Cystic kidney | | 0.85 | 3.12 | 4.51 | 5.17 | 5.78 | 8.49 | 7.19 |
| Bladder exstrophy | | 0.12 | 0.36 | 0.28 | 0.25 | 0.48 | 0.34 | 0.36 |
| Polydactyly, preaxial | | 11.08 | 9.67 | 16.27 | 14.55 | 12.31 | 17.47 | 18.82 |
| Total Limb reduction defects (include unspecified) | | 6.09 | 7.18 | 9.92 | 9.63 | 12.20 | 10.92 | 12.39 |
| Transverse | | nr | nr | nr | nr | nr | nr | nr |
| Preaxial | | nr | nr | nr | nr | nr | nr | nr |
| Postaxial | | nr | nr | nr | nr | nr | nr | nr |
| Intercalary | | nr | nr | nr | nr | nr | nr | nr |
| Mixed | | nr | nr | nr | nr | nr | nr | nr |
| Unspecified | | nr | nr | nr | nr | nr | nr | nr |
| Diaphragmatic hernia | | 3.29 | 3.57 | 2.87 | 2.61 | 3.80 | 3.35 | 3.69 |
| Omphalocele | | 0.97 | 1.90 | 2.59 | 1.51 | 2.57 | 2.62 | 4.09 |
| Gastroschisis | | 1.22 | 1.54 | 1.46 | 1.91 | 2.68 | 4.56 | 4.84 |
| Unspecified Omphalocele/Gastroschisis | | 0.61 | 0.59 | 0.42 | 0.25 | 0.00 | 0.00 | 0.00 |
| Prune belly sequence | | 0.61 | 0.41 | 0.28 | 0.05 | 0.48 | 0.49 | 0.24 |
| Trisomy 13 | | 0.73 | 0.72 | 1.03 | 1.10 | 1.66 | 2.18 | 3.10 |
| Trisomy 18 | | 1.34 | 1.63 | 1.93 | 2.51 | 4.44 | 4.80 | 5.64 |
| Down syndrome, all ages (include age unknown) | | 10.11 | 8.81 | 10.86 | 10.44 | 17.61 | 21.35 | 22.47 |
| <20 | | nr | 3.14 | 6.21 | 3.29 | 7.93 | 10.03 | 6.97 |
| 20-24 | | nr | 6.98 | 5.76 | 5.76 | 4.98 | 7.16 | 8.09 |
| 25-29 | | nr | 6.52 | 6.34 | 7.26 | 10.32 | 10.44 | 10.14 |
| 30-34 | | nr | 6.28 | 15.01 | 11.62 | 16.08 | 16.01 | 18.19 |
| 35-39 | | nr | 39.46 | 31.13 | 25.99 | 42.05 | 55.95 | 51.22 |
| 40-44 | | nr | 186.92 | 90.25 | 67.17 | 162.27 | 179.95 | 184.17 |
| 45+ | | nr | nr | 307.69 | 266.67 | 260.87 | 377.36 | 323.45 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

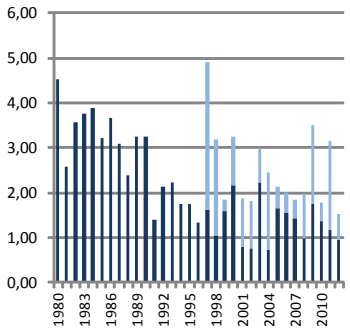
nr = data not reported or not available

* data include less than 5 years

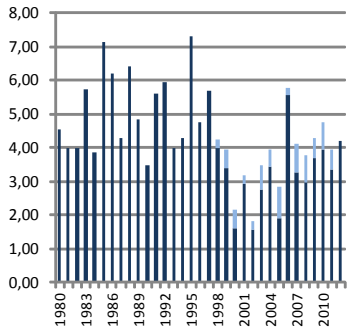
# Canada-Alberta: ACASS, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

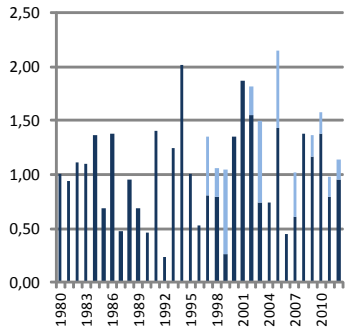
**Anencephaly**



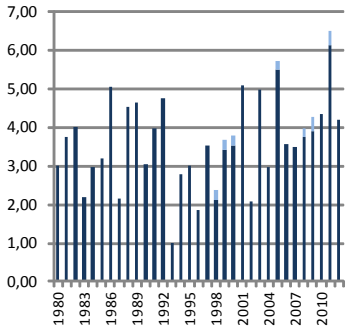

**Spina Bifida**



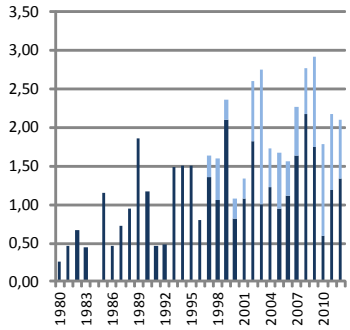

**Encephalocele**



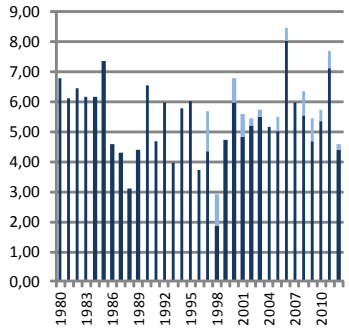

**Microcephaly**



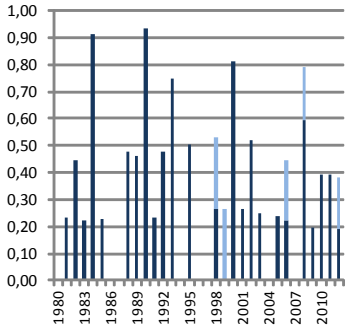

**Holoprosencephaly**



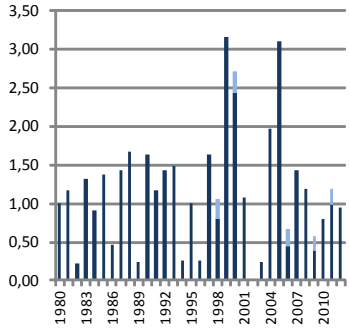

**Hydrocephaly**



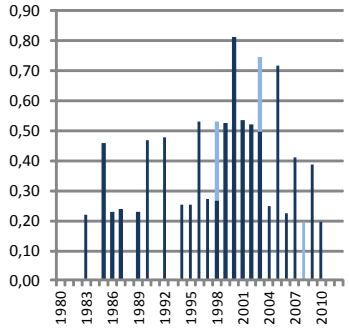

**Anophthalmos**



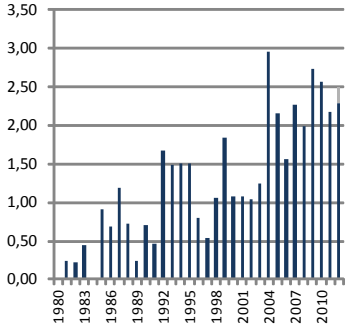

**Microphthalmos**



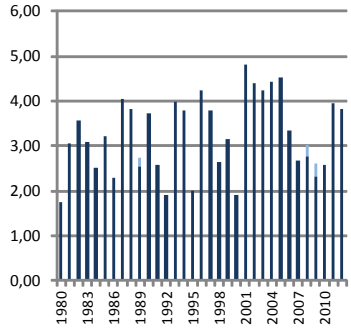

**Anotia**



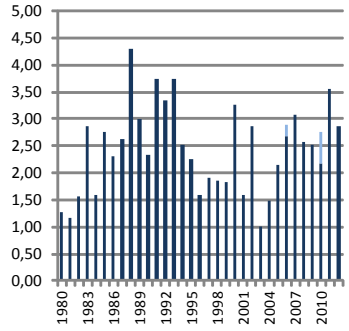

**Microtia**




**Transposition of great vessels**




**Tetralogy of Fallot**

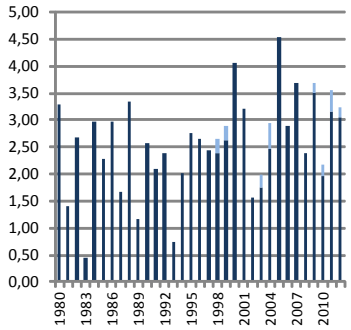



**L + S rates** **ToP rates**

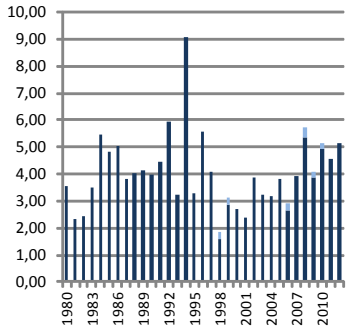
# Canada-Alberta: ACASS, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

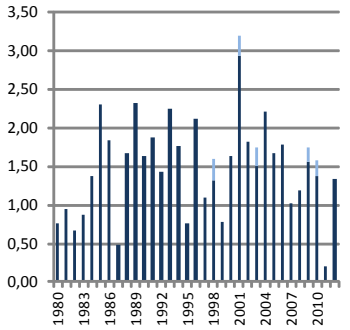
**Hypoplastic left heart syndrome**



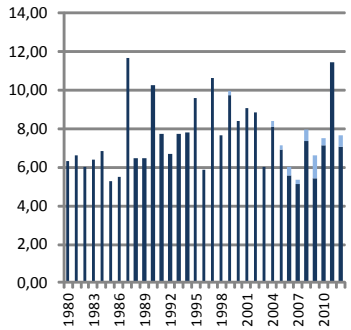

**Coarctation of aorta**



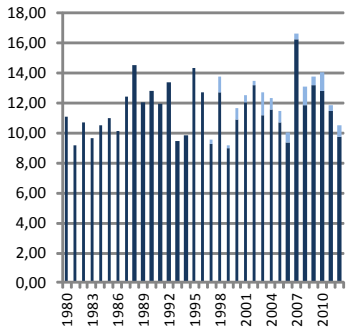

**Choanal atresia, bilateral**



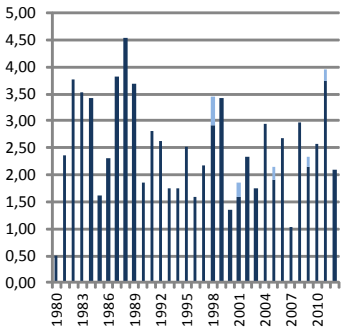

**Cleft palate without cleft lip**



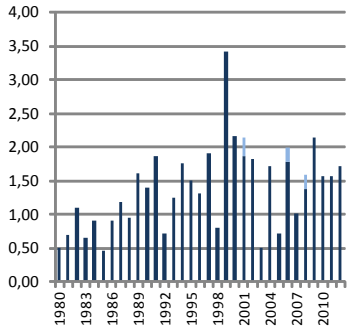

**Cleft lip with or without cleft palate**



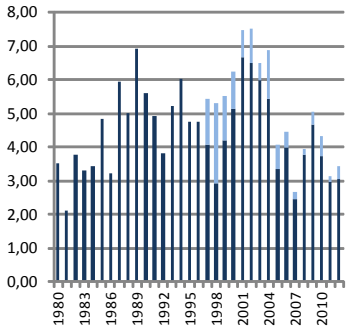

**phageal atresia/stenosis with or without f**



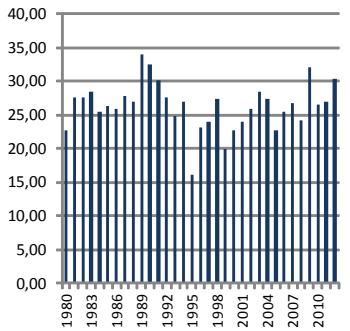

**Small intestine atresia/stenosis**



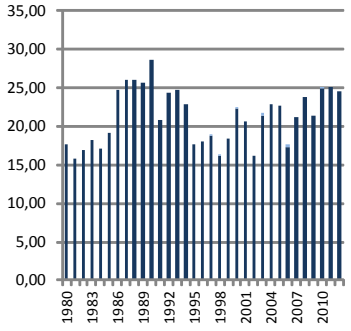

**Anorectal atresia/stenosis**



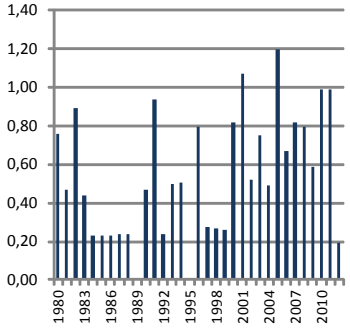

**Undescended testis**



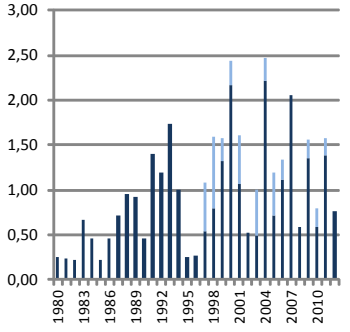

**Hypospadias**




**Epispadias**




**Indeterminate sex**

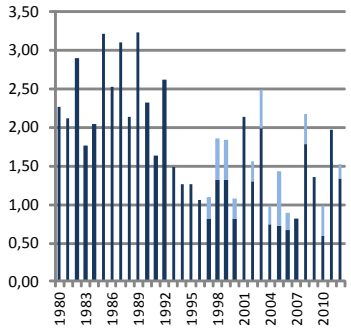



**L + S rates** **ToP rates**

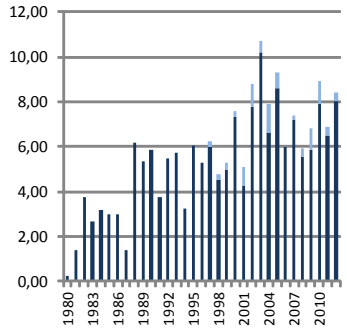
# Canada-Alberta: ACASS, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

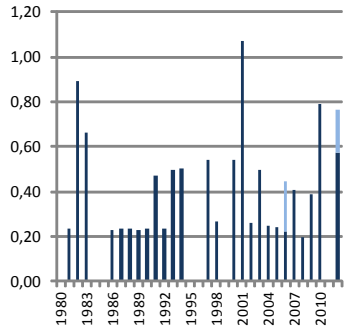
**Renal agenesis**



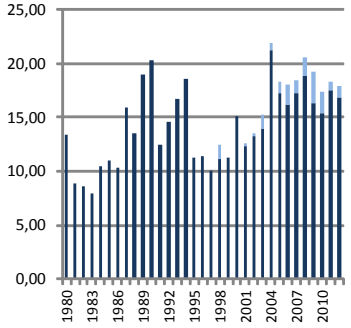

**Cystic kidney**



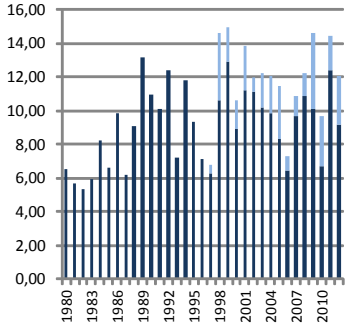

**Bladder exstrophy**



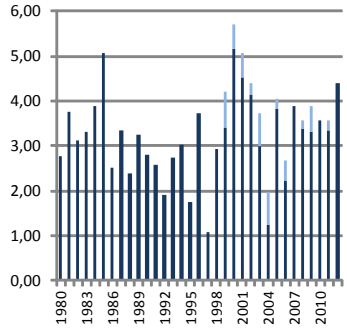

**Polydactyly, preaxial**



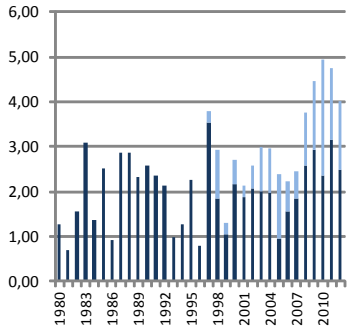

**Limb reduction defects**



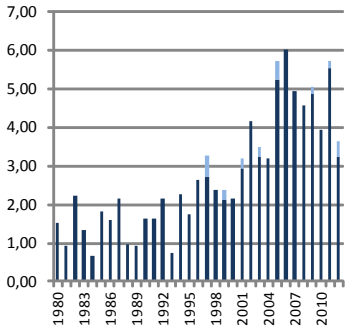

**Diaphragmatic hernia**



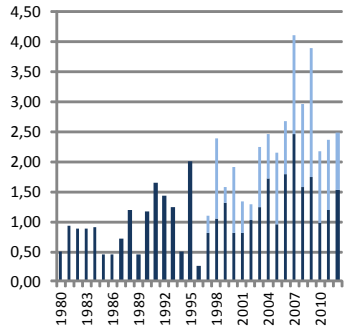

**Omphalocele**



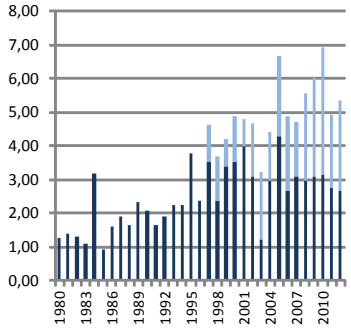

**Gastroschisis**



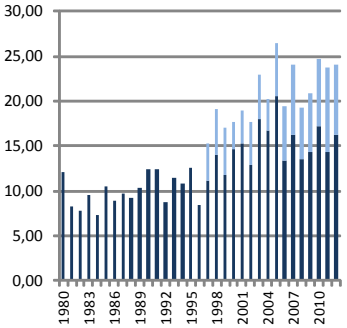

**Trisomy 13**



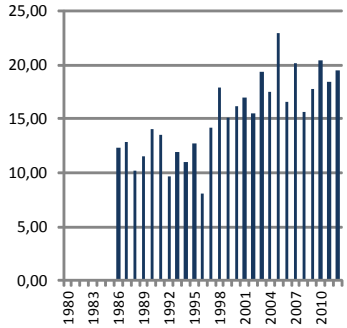

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Canada National: CCASS

## Canadian Congenital Anomalies Surveillance System

**History:**
The Programme was started in 1966. The Programme was a full member until 1987, when it became an associate member. The Programme was discontinued as an associate member of the ICBDSR in the early 1990s, and reinstated its member status in 1996.

**Size and coverage:**
This system presently monitors about 30,000 births annually, which captures virtually all live births and registered stillbirths (a birth weight of greater or equal to 500 grams, or greater than or equal to 20 weeks in pregnancy) in the 10 provinces and 3 territories of Canada.

**Legislation and funding:**
Reporting is done by the Public Health Agency of Canada (PHAC) as part of its national surveillance mandate. For congenital anomalies reporting, PHAC uses hospitalization data obtained through the Canadian Institute for Health Information (CIHI). Med-Echo (Système de maintenance et d'exploitation des données pour l'étude de la clientèle hospitalière) for the province of Québec provides their data separately up to 2007.

**Sources of ascertainment:**
Cases from most provinces and territories are ascertained from hospital admission - separation summary records collected by CIHI and Med-Echo. The Alberta Congenital Anomalies Surveillance System provides its own separate provincial data. All data sources had a one year follow-up period until 2000. Since 2001, all data provided by CIHI only include a 30-day follow-up period.

**Exposure information:**
Currently no exposure information is routinely collected.

**Background information:**
Background information is based on hospital admission/separation summary records from CIHI and Med-Echo. Alberta Congenital Anomalies Surveillance provides its own background information. Interpretation of trends should be done cautiously, since 2001 an increasing percentage of records are being coded using ICD-10 CA and may cause discrepancies from previously used ICD-9 coding. Also, as mentioned previously the variation in the follow-up period is another factor which may alter reporting of trends.

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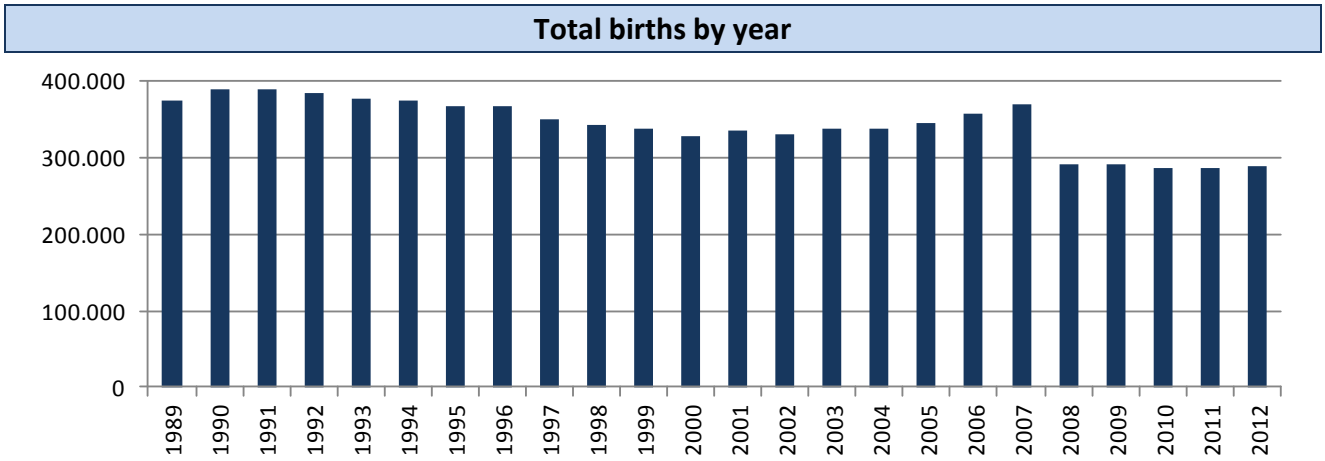
## Canada National: CCASS



# Canada National: CCASS, 2012 (*)

| | |
|---|---|
| Live births (LB) | 287,667 |
| Stillbirths (SB) | 2,206 |
| Total births | 289,873 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 9 | 18 | nr | 0.93 |
| Spina bifida | 77 | 19 | nr | 3.31 |
| Encephalocele | 18 | 6 | nr | 0.83 |
| Microcephaly | 137 | 3 | nr | 4.83 |
| Holoprosencephaly | 13 | 14 | nr | 0.93 |
| Hydrocephaly | 105 | 18 | nr | 4.24 |
| Anophthalmos | 2 | 0 | nr | 0.07 |
| Microphthalmos | 20 | 0 | nr | 0.69 |
| Unspecified Anophthalmos/Microphthalmos | 21 | 0 | nr | 0.72 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 29 | 0 | nr | 1.00 |
| Unspecified Anotia/Microtia | 29 | 0 | nr | 1.00 |
| Transposition of great vessels | 139 | 7 | nr | 5.04 |
| Tetralogy of Fallot | 101 | 16 | nr | 4.04 |
| Hypoplastic left heart syndrome | 56 | 9 | nr | 2.24 |
| Coarctation of aorta | 155 | 1 | nr | 5.38 |
| Choanal atresia, bilateral | 69 | 0 | nr | 2.38 |
| Cleft palate without cleft lip | 189 | 1 | nr | 6.55 |
| Cleft lip with or without cleft palate | 252 | 19 | nr | 9.35 |
| Oesophageal atresia/stenosis with or without fistula | 72 | 1 | nr | 2.52 |
| Small intestine atresia/stenosis | 105 | 2 | nr | 3.69 |
| Anorectal atresia/stenosis | 107 | 0 | nr | 3.69 |
| Undescended testis (**) | 1003 | 0 | nr | 34.60 |
| Hypospadias | 846 | 0 | nr | 29.19 |
| Epispadias | 22 | 0 | nr | 0.76 |
| Indeterminate sex | 42 | 1 | nr | 1.48 |
| Renal agenesis | 150 | 27 | nr | 6.11 |
| Cystic kidney | 227 | 10 | nr | 8.18 |
| Bladder exstrophy | 10 | 1 | nr | 0.38 |
| Polydactyly, preaxial | 398 | 1 | nr | 13.76 |
| Total Limb reduction defects (include unspecified) | 80 | 1 | nr | 2.79 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 87 | 11 | nr | 3.38 |
| Omphalocele | 54 | 12 | nr | 2.28 |
| Gastroschisis | 97 | 7 | nr | 3.59 |
| Unspecified Omphalocele/Gastroschisis | 151 | 18 | nr | 5.83 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13 | 16 | 17 | nr | 1.14 |
| Trisomy 18 | 34 | 37 | nr | 2.45 |
| Down syndrome, all ages (include age unknown) | 406 | 53 | nr | 15.83 |
| <20 | nr | nr | nr | nr |
| 20-24 | nr | nr | nr | nr |
| 25-29 | nr | nr | nr | nr |
| 30-34 | nr | nr | nr | nr |
| 35-39 | nr | nr | nr | nr |
| 40-44 | nr | nr | nr | nr |
| 45+ | nr | nr | nr | nr |
| unknown | nr | nr | nr | nr |

nr = data not reported or not available
(*) Province of Quebec excluded
(**) Any gestational age

# Canada National: CCASS, Previous years rates 1989 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991* | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | **1,156,605** | **1,872,269** | **1,698,115** | **1,710,260** | **1,529,025** |
| Anencephaly | | | | 2.10 | 1.66 | 1.14 | 0.98 | 1.16 |
| Spina bifida | | | | 7.58 | 6.30 | 4.08 | 2.94 | 3.24 |
| Encephalocele | | | | 1.38 | 1.29 | 0.87 | 0.58 | 0.74 |
| Microcephaly | | | | 5.78 | 5.11 | 5.65 | 4.65 | 3.66 |
| Holoprosencephaly | | | | nr | nr | nr | 0.36* | 0.94 |
| Hydrocephaly | | | | 7.31 | 6.80 | 6.60 | 4.95 | 4.70 |
| Anophthalmos | | | | 0.34 | 0.28 | 0.26 | 0.19 | 0.20 |
| Microphthalmos | | | | 1.15 | 0.87 | 1.07 | 0.69 | 0.69 |
| Unspecified Anophthalmos/Microphthalmos | | | | nr | nr | nr | nr | 0.35* |
| Anotia | | | | nr | nr | nr | 0.11* | 0.03 |
| Microtia | | | | nr | nr | nr | 0.59* | 0.97 |
| Unspecified Anotia/Microtia | | | | nr | nr | nr | nr | 0.37* |
| Transposition of great vessels | | | | 4.51 | 4.96 | 5.45 | 4.72 | 4.74 |
| Tetralogy of Fallot | | | | 4.87 | 4.62 | 5.02 | 3.94 | 3.39 |
| Hypoplastic left heart syndrome | | | | 3.03 | 2.78 | 3.02 | 2.47 | 2.44 |
| Coarctation of aorta | | | | 5.20 | 5.74 | 6.13 | 4.75 | 4.53 |
| Choanal atresia, bilateral | | | | 2.08 | 2.06 | 2.89 | 2.52 | 2.13 |
| Cleft palate without cleft lip | | | | 7.20 | 6.96 | 7.41 | 7.13 | 6.42 |
| Cleft lip with or without cleft palate | | | | 11.54 | 11.05 | 10.38 | 9.03 | 9.42 |
| Oesophageal atresia/stenosis with or without fistula | | | | 3.57 | 3.18 | 3.46 | 2.75 | 2.42 |
| Small intestine atresia/stenosis | | | | 3.55 | 3.42 | 3.75 | 3.87 | 3.77 |
| Anorectal atresia/stenosis | | | | 5.48 | 4.86 | 4.90 | 4.10 | 3.72 |
| Undescended testis (36 weeks of gestation or later) | | | | 35.69 | 32.86 | 34.72 | 38.67 | 34.43 |
| Hypospadias | | | | 27.11 | 26.36 | 28.01 | 25.19 | 27.49 |
| Epispadias | | | | nr | nr | nr | 0.61* | 0.69 |
| Indeterminate sex | | | | 0.78 | 0.60 | 0.75 | 1.13 | 1.43 |
| Renal agenesis | | | | 4.98 | 4.94 | 5.11 | 5.19 | 4.94 |
| Cystic kidney | | | | 4.44 | 5.43 | 6.50 | 7.26 | 6.89 |
| Bladder exstrophy | | | | 0.45 | 0.38 | 0.35 | 0.39 | 0.24 |
| Polydactyly, preaxial | | | | 12.44 | 11.39 | 12.70 | 14.38 | 13.15 |
| Total Limb reduction defects (include unspecified) | | | | 4.80 | 4.49 | 3.95 | 3.68 | 3.29 |
| Transverse | | | | nr | nr | nr | nr | nr |
| Preaxial | | | | nr | nr | nr | nr | nr |
| Postaxial | | | | nr | nr | nr | nr | nr |
| Intercalary | | | | nr | nr | nr | nr | nr |
| Mixed | | | | nr | nr | nr | nr | nr |
| Unspecified | | | | nr | nr | nr | nr | nr |
| Diaphragmatic hernia | | | | 3.59 | 3.75 | 3.67 | 3.17 | 3.15 |
| Omphalocele | | | | 4.51 | 6.19* | nr | 1.79* | 2.13 |
| Gastroschisis | | | | nr | nr | nr | 3.69* | 4.23 |
| Unspecified Omphalocele/Gastroschisis | | | | nr | 6.62* | 5.90* | nr | 5.99* |
| Prune belly sequence | | | | nr | nr | nr | nr | nr |
| Trisomy 13 | | | | 1.23 | 1.09 | 1.21 | 1.13 | 1.18 |
| Trisomy 18 | | | | 2.16 | 2.21 | 2.40 | 2.39 | 2.34 |
| Down syndrome, all ages (include age unknown) | | | | 13.65 | 12.57 | 14.25 | 14.64 | 14.64 |
| <20 | | | | nr | nr | nr | nr | nr |
| 20-24 | | | | nr | nr | nr | nr | nr |
| 25-29 | | | | nr | nr | nr | nr | nr |
| 30-34 | | | | nr | nr | nr | nr | nr |
| 35-39 | | | | nr | nr | nr | nr | nr |
| 40-44 | | | | nr | nr | nr | nr | nr |
| 45+ | | | | nr | nr | nr | nr | nr |
| unknown | | | | --- | --- | --- | --- | --- |

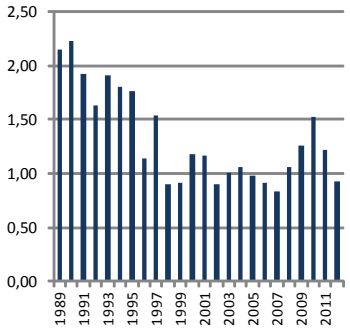
nr = data not reported or not available

* data include less than 5 years

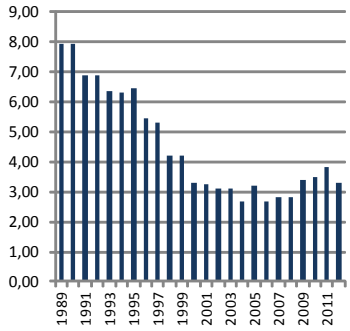
# Canada National: CCASS, Time trends 1989 – 2012

(Birth prevalence rates per 10,000)

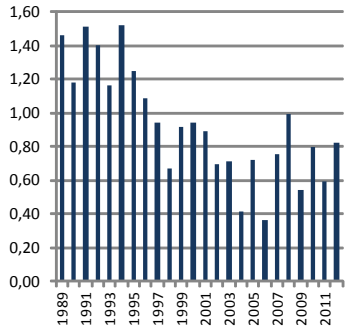
**Anencephaly**



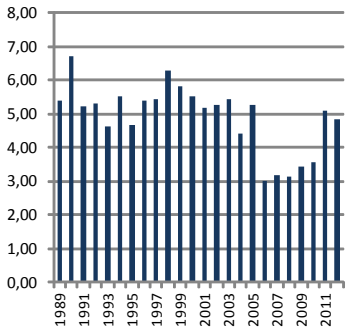

**Spina Bifida**



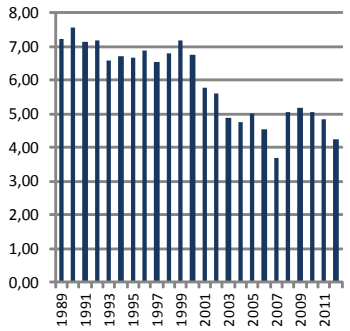

**Encephalocele**



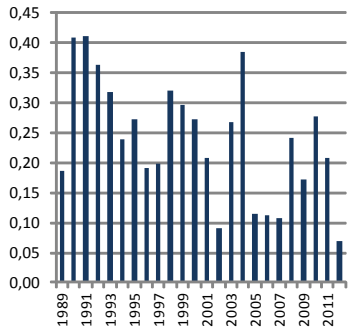

**Microcephaly**



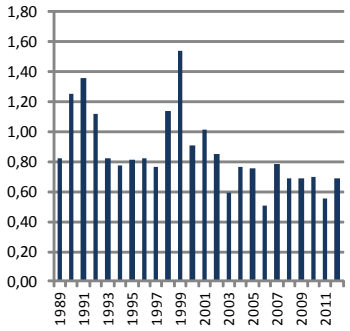

**Hydrocephaly**



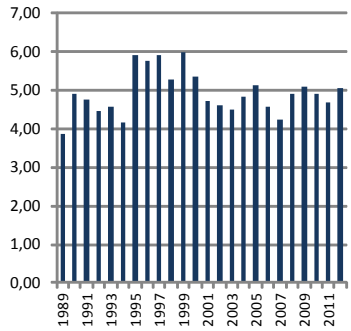

**Anophthalmos**



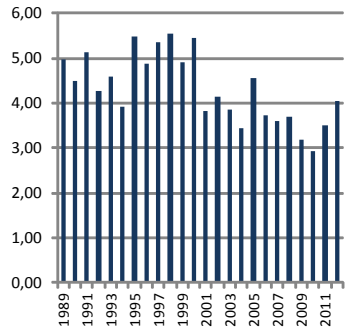

**Microphthalmos**



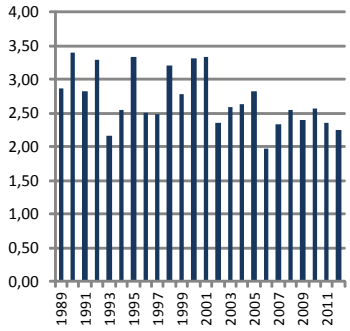

**Transposition of great vessels**



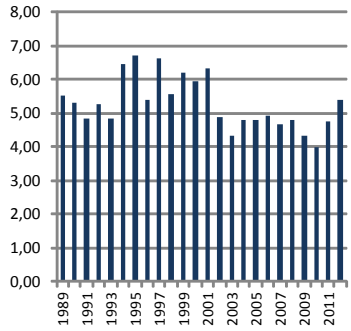

**Tetralogy of Fallot**



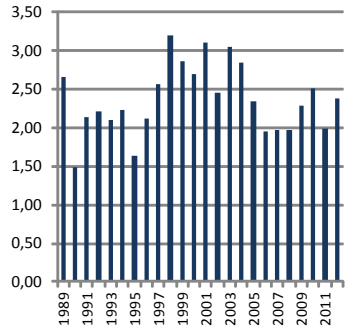

**Hypoplastic left heart syndrome**




**Coarctation of aorta**




**Choanal atresia, bilateral**

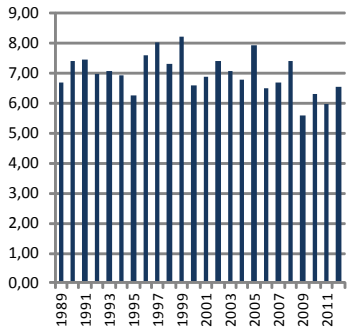



**L + S rates**

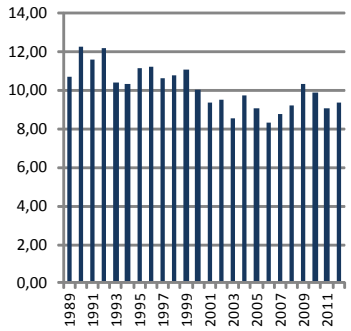
# Canada National: CCASS, Time trends 1989 – 2012

(Birth prevalence rates per 10,000)

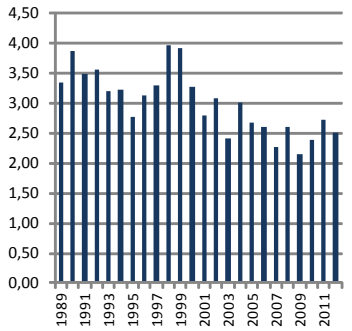
**Cleft palate without cleft lip**



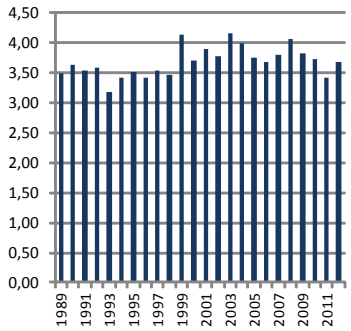

**Cleft lip with or without cleft palate**



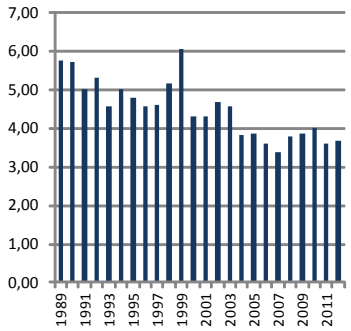

**phageal atresia/stenosis with or without f**



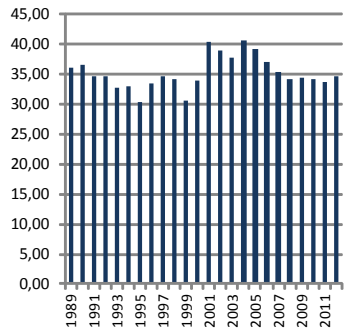

**Small intestine atresia/stenosis**



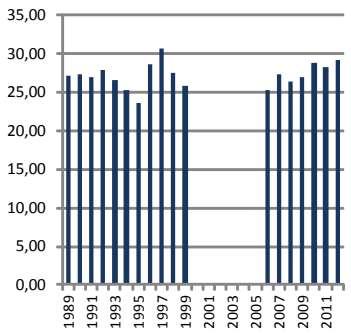

**Anorectal atresia/stenosis**



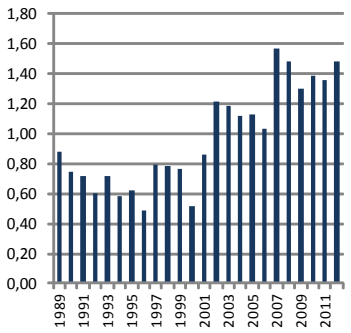

**Undescended testis**



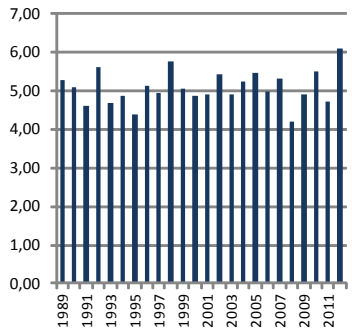

**Hypospadias**



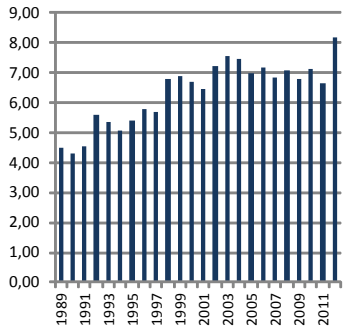

**Indeterminate sex**



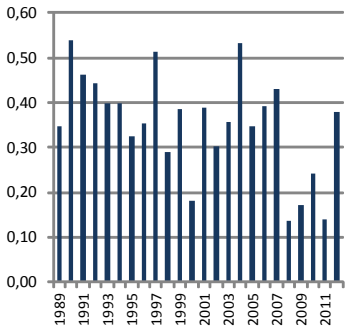

**Renal agenesis**



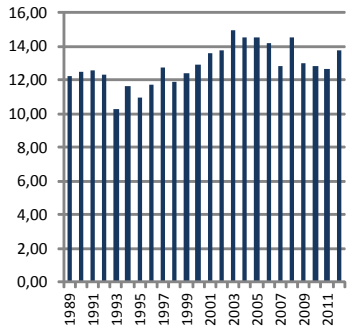

**Cystic kidney**




**Bladder exstrophy**




**Polydactyly, preaxial**

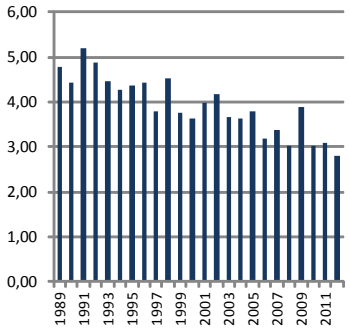



**L + S rates**

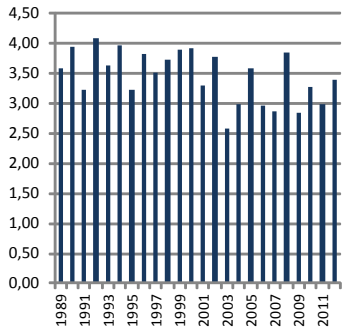
# Canada National: CCASS, Time trends 1989 – 2012

(Birth prevalence rates per 10,000)

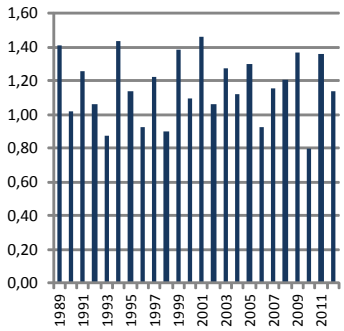
**Limb reduction defects**



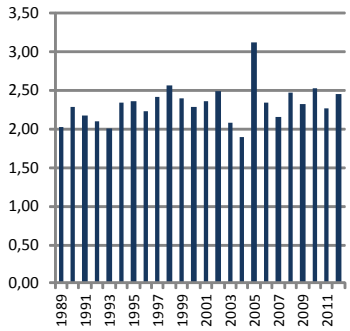

**Diaphragmatic hernia**



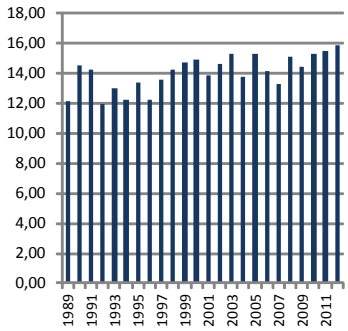

**Trisomy 13**




**Trisomy 18**




**Down Syndrome**




**L + S rates**

# Costa Rica: CREC

## Costa Rican Birth Defects Register Centre

**History:**
The registry was created in 1986, based in a government decret by which birth defects became subject of obligatory notification. The program became an ICBDSR member in September 2003.

**Size and coverage:**
The program is population based. Includes all births from the National Security System (CCSS) which covers about 98% of all births occurred in the country, and births of private hospitals. There are approximately 75000 annual births in Costa Rica.

**Legislation and funding:**
The Registry is financed by the government as a program of the Costa Rican Institute of Research and Training in Nutrition and Health (INCIENSA), Institute that depends from the Ministry of Health.

**Sources of ascertainment:**
Until 2008 reporting was made only by neonatologists, pediatricians and general physicians before newborns discharge from maternity services, with biostatistics personal collaboration. In 2009 the age of obligatory notification was extended to children under one year of age.

**Exposure information:**
In 2009 began rubella vaccine exposure information collect in order to support the performance of Congenital Rubella Syndrome surveillance.

**Background information:**
Linkage studies are possible with other statistical data from the National Statistics Center and the National Security System Statistical Center.

**Addresses and Staff:**
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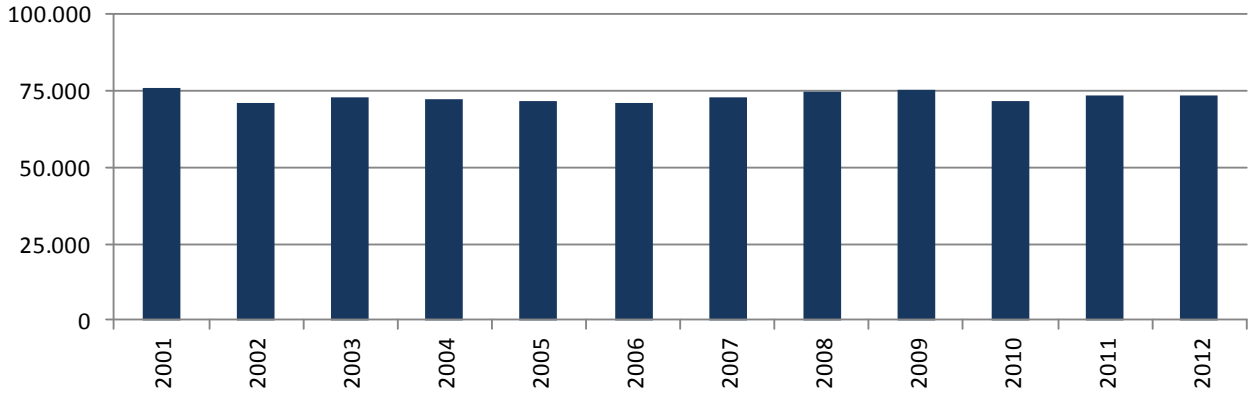
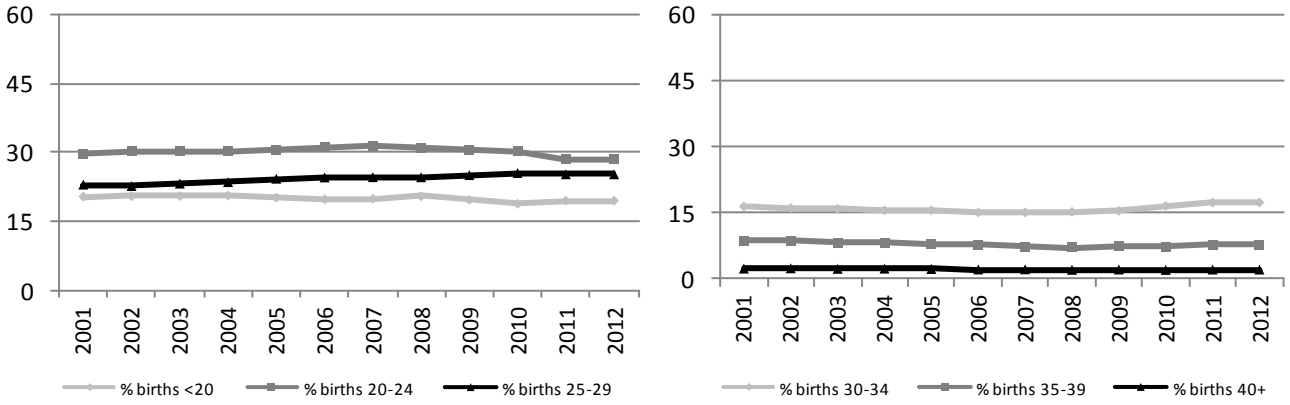
**E-mail:** abenavides@inciensa.sa.cr

# Costa Rica: CREC

## Total births by year




## Percentage of births by year and maternal age



# Costa Rica: CREC, 2012

| | |
|---|---|
| Live births (LB) | 73,326 |
| Stillbirths (SB) | 465 |
| Total births | 73,791 |
| Number of terminations of pregnancy (ToP) for birth defects | not permitted |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB(*) | ToP | Total rate |
| Anencephaly | 12 | 1 | | 1.76 |
| Spina bifida | 29 | 0 | | 3.93 |
| Encephalocele | 5 | 0 | | 0.68 |
| Microcephaly | 17 | 1 | | 2.44 |
| Holoprosencephaly | 3 | 0 | | 0.41 |
| Hydrocephaly | 37 | 1 | | 5.15 |
| Anophthalmos | 7 | 0 | | 0.95 |
| Microphthalmos | 7 | 0 | | 0.95 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 4 | 0 | | 0.54 |
| Microtia | 13 | 0 | | 1.76 |
| Unspecified Anotia/Microtia | 0 | 0 | | 0.00 |
| Transposition of great vessels | 13 | 0 | | 1.76 |
| Tetralogy of Fallot | 11 | 0 | | 1.49 |
| Hypoplastic left heart syndrome | 4 | 0 | | 0.54 |
| Coarctation of aorta | 18 | 0 | | 2.44 |
| Choanal atresia, bilateral | 8 | 0 | | 1.08 |
| Cleft palate without cleft lip | 23 | 0 | | 3.12 |
| Cleft lip with or without cleft palate | 66 | 0 | | 8.94 |
| Oesophageal atresia/stenosis with or without fistula | 21 | 1 | | 2.98 |
| Small intestine atresia/stenosis | 20 | 0 | | 2.71 |
| Anorectal atresia/stenosis | 30 | 1 | | 4.20 |
| Undescended testis (36 weeks of gestation or later) | 107 | 1 | | 14.64 |
| Hypospadias | 46 | 0 | | 6.23 |
| Epispadias | 2 | 0 | | 0.27 |
| Indeterminate sex | 12 | 1 | | 1.76 |
| Renal agenesis | 7 | 1 | | 1.08 |
| Cystic kidney | 4 | 0 | | 0.54 |
| Bladder exstrophy | 0 | 0 | | 0.00 |
| Polydactyly(**) | 91 | 1 | | 12.47 |
| Total Limb reduction defects (include unspecified) | 44 | 2 | | 6.23 |
| Transverse | nr | nr | | nr |
| Preaxial | nr | nr | | nr |
| Postaxial | nr | nr | | nr |
| Intercalary | nr | nr | | nr |
| Mixed | nr | nr | | nr |
| Unspecified | nr | nr | | nr |
| Diaphragmatic hernia | 23 | 0 | | 3.12 |
| Omphalocele | 14 | 0 | | 1.90 |
| Gastroschisis | 30 | 0 | | 4.07 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | | 0.00 |
| Prune belly sequence | 2 | 0 | | 0.27 |
| Trisomy 13 | 0 | 0 | | 0.00 |
| Trisomy 18 | 16 | 1 | | 2.30 |
| Down syndrome, all ages (include age unknown) | 85 | 0 | | 11.52 |
| <20 | 9 | 0 | | 6.34 |
| 20-24 | 11 | 0 | | 5.27 |
| 25-29 | 9 | 0 | | 4.90 |
| 30-34 | 14 | 0 | | 11.12 |
| 35-39 | 14 | 0 | | 25.43 |
| 40-44 | 19 | 0 | | 136.89 |
| 45+ | 2 | 0 | | 273.97 |
| unknown | 7 | 0 | | --- |

nr = data not reported or not available

(*) Birth defects under-reported in stillbirths

(**) All cases, no specification available

# Costa Rica: CREC, Previous years rates 2001 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991* | 1992-1996 | 1997-2001* | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | **75,991** | **359,168** | **368,578** |
| Anencephaly | | | | | | 1.45 | 1.31 | 1.00 |
| Spina bifida | | | | | | 3.95 | 3.26 | 2.55 |
| Encephalocele | | | | | | 0.26 | 0.64 | 0.79 |
| Microcephaly | | | | | | 0.79 | 1.45 | 2.98 |
| Holoprosencephaly | | | | | | 0.66 | 0.22 | 0.62 |
| Hydrocephaly | | | | | | 2.90 | 2.34 | 2.93 |
| Anophthalmos | | | | | | 0.26 | 0.22 | 0.16 |
| Microphthalmos | | | | | | 0.13 | 0.64 | 0.35 |
| Unspecified Anophthalmos/Microphthalmos | | | | | | 0.00 | 0.00 | 0.00 |
| Anotia | | | | | | 0.26 | 0.42 | 1.06 |
| Microtia | | | | | | 1.84 | 2.51 | 1.93 |
| Unspecified Anotia/Microtia | | | | | | 0.00 | 0.00 | 0.17 |
| Transposition of great vessels | | | | | | 0.13 | 0.08 | 0.79 |
| Tetralogy of Fallot | | | | | | 0.66 | 0.92 | 0.76 |
| Hypoplastic left heart syndrome | | | | | | 0.39 | 0.33 | 0.79 |
| Coarctation of aorta | | | | | | 0.39 | 0.28 | 0.81 |
| Choanal atresia, bilateral | | | | | | 0.39 | 0.28 | 0.38 |
| Cleft palate without cleft lip | | | | | | 2.50 | 2.70 | 2.82 |
| Cleft lip with or without cleft palate | | | | | | 7.24 | 6.77 | 8.44 |
| Oesophageal atresia/stenosis with or without fistula | | | | | | 1.71 | 1.28 | 1.95 |
| Small intestine atresia/stenosis | | | | | | 0.66 | 0.70 | 0.38 |
| Anorectal atresia/stenosis | | | | | | 2.76 | 2.70 | 2.63 |
| Undescended testis (36 weeks of gestation or later) | | | | | | 10.26 | 9.52 | 11.34 |
| Hypospadias | | | | | | 5.92 | 6.15 | 6.92 |
| Epispadias | | | | | | 0.13 | 0.06 | 0.19 |
| Indeterminate sex | | | | | | 1.45 | 1.78 | 1.19 |
| Renal agenesis | | | | | | 0.53 | 0.81 | 0.87 |
| Cystic kidney | | | | | | 0.26 | 0.28 | 1.74 |
| Bladder exstrophy | | | | | | 0.13 | 0.06 | 0.00 |
| Polydactyly, preaxial | | | | | | 2.11 | 9.30 | 11.40 |
| Total Limb reduction defects (include unspecified) | | | | | | 4.87 | 4.87 | 4.83 |
| Transverse | | | | | | nr | nr | nr |
| Preaxial | | | | | | nr | nr | nr |
| Postaxial | | | | | | nr | nr | nr |
| Intercalary | | | | | | nr | nr | nr |
| Mixed | | | | | | nr | nr | nr |
| Unspecified | | | | | | nr | nr | nr |
| Diaphragmatic hernia | | | | | | 1.84 | 1.61 | 1.36 |
| Omphalocele | | | | | | 0.13 | 0.86 | 1.33 |
| Gastroschisis | | | | | | 2.24 | 1.34 | 2.36 |
| Unspecified Omphalocele/Gastroschisis | | | | | | 0.00 | 0.17 | 0.00 |
| Prune belly sequence | | | | | | 0.13 | 0.42 | 0.22 |
| Trisomy 13 | | | | | | 2.24 | 0.86 | 0.41 |
| Trisomy 18 | | | | | | 0.39 | 1.11 | 0.84 |
| Down syndrome, all ages (include age unknown) | | | | | | 9.47 | 8.10 | 9.85 |
| <20 | | | | | | 5.17 | 4.84 | 4.32 |
| 20-24 | | | | | | 4.43 | 4.87 | 3.87 |
| 25-29 | | | | | | 4.59 | 4.27 | 4.49 |
| 30-34 | | | | | | 4.83 | 5.58 | 9.33 |
| 35-39 | | | | | | 31.17 | 26.28 | 36.88 |
| 40-44 | | | | | | 121.48 | 66.12 | 95.04 |
| 45+ | | | | | | 79.37 | 146.14 | 207.85 |
| unknown | | | | | | --- | 59.43 | 150.38 |

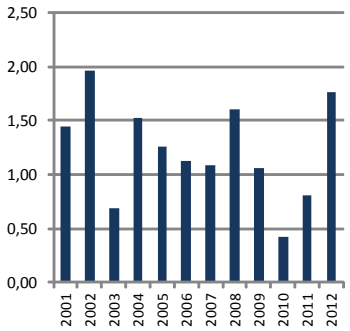
nr = data not reported or not available

* data include less than 5 years

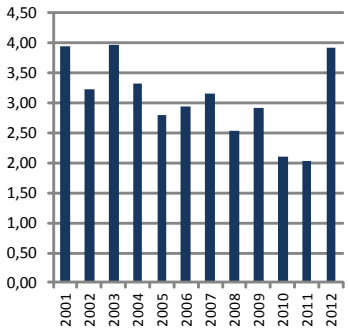
# Costa Rica: CREC, Time trends 2001 – 2012

(Birth prevalence rates per 10,000)

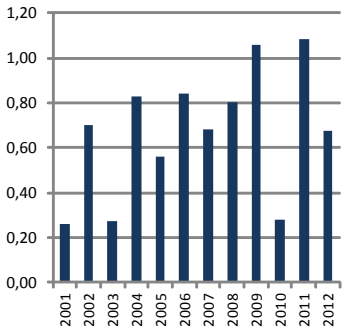
**Anencephaly**



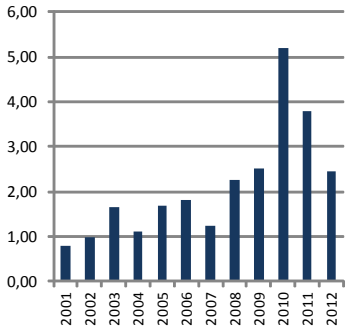

**Spina Bifida**



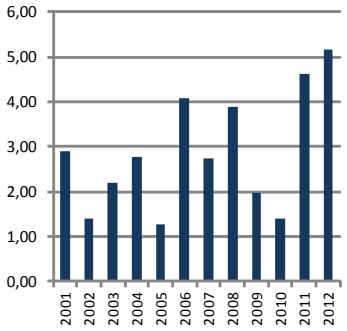

**Encephalocele**



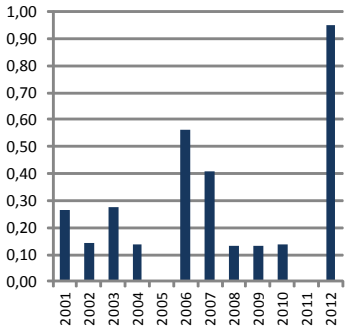

**Microcephaly**



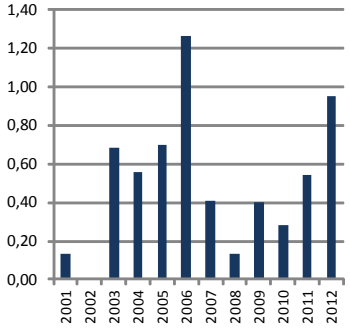

**Hydrocephaly**



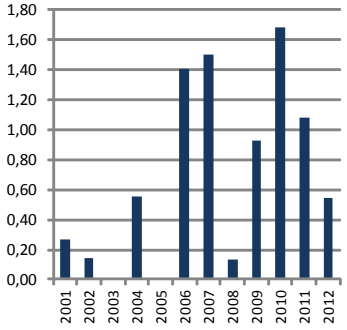

**Anophthalmos**



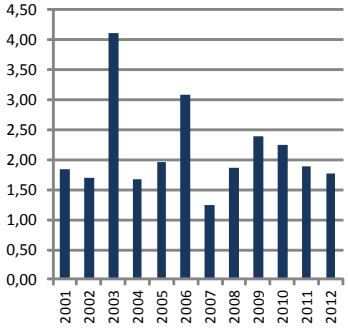

**Microphthalmos**



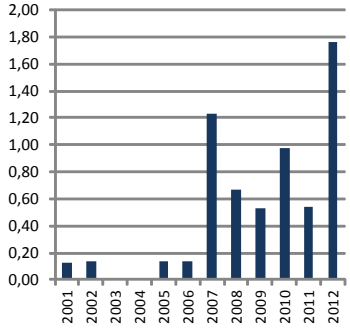

**Anotia**



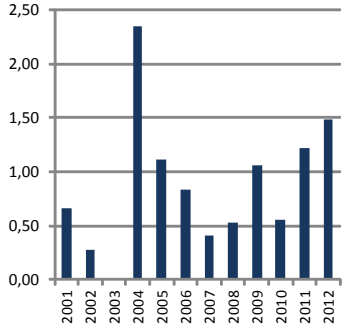

**Microtia**



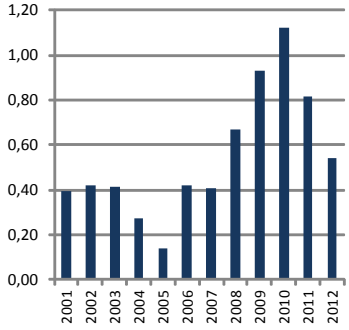

**Transposition of great vessels**




**Tetralogy of Fallot**




**Hypoplastic left heart syndrome**

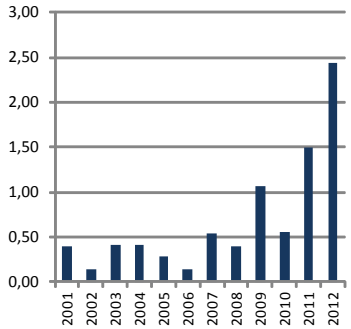



**L + S rates**

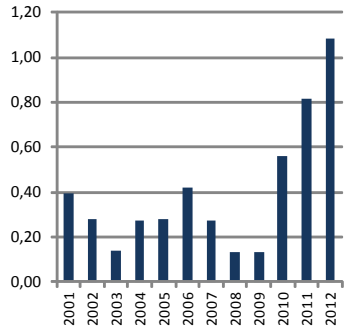
# Costa Rica: CREC, Time trends 2001 – 2012

(Birth prevalence rates per 10,000)

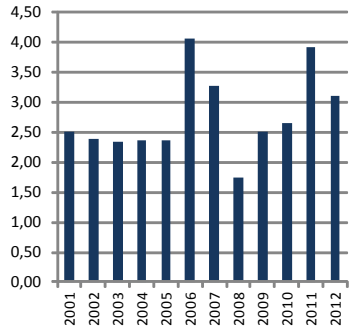
**Coarctation of aorta**



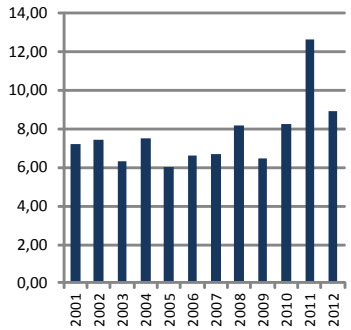

**Choanal atresia, bilateral**



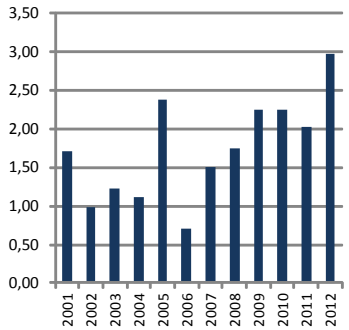

**Cleft palate without cleft lip**



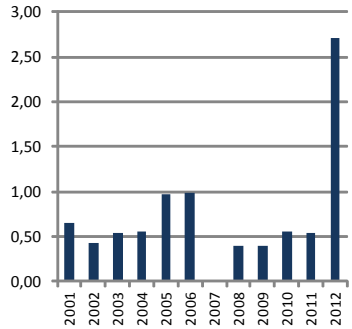

**Cleft lip with or without cleft palate**



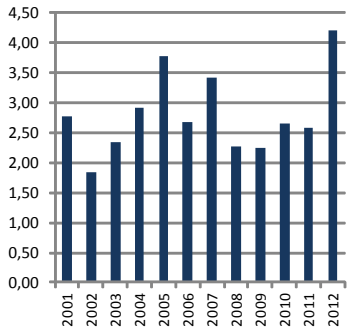

**phageal atresia/stenosis with or without f**



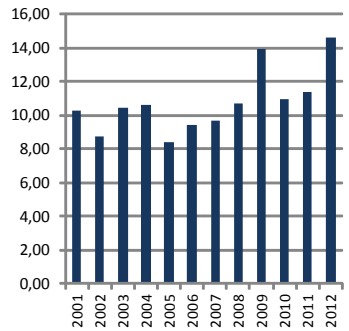

**Small intestine atresia/stenosis**



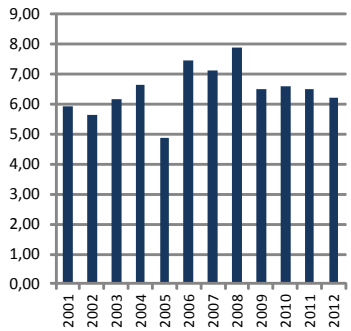

**Anorectal atresia/stenosis**



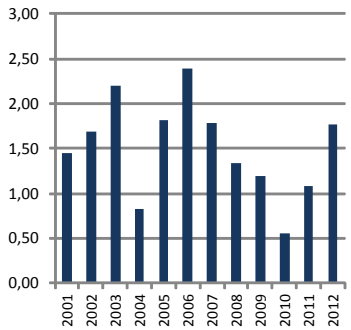

**Undescended testis**



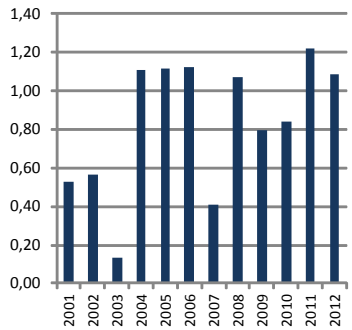

**Hypospadias**



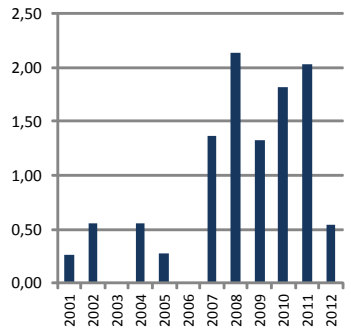

**Indeterminate sex**




**Renal agenesis**




**Cystic kidney**

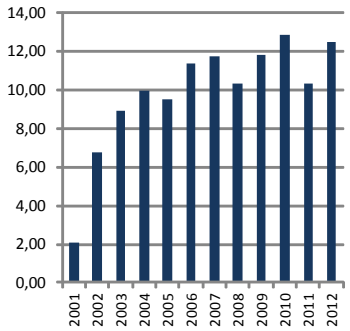



**L + S rates**

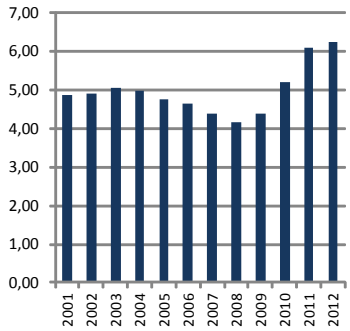
# Costa Rica: CREC, Time trends 2001 – 2012

(Birth prevalence rates per 10,000)

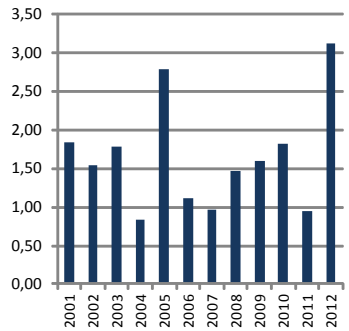
**Polydactyly, preaxial**



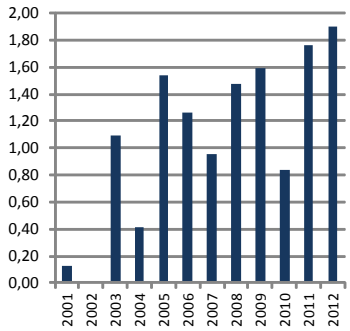

**Limb reduction defects**



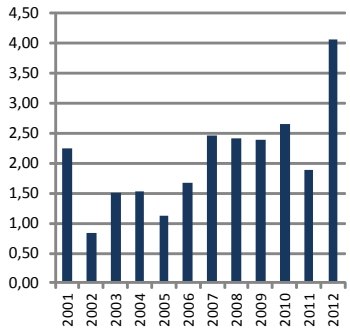

**Diaphragmatic hernia**



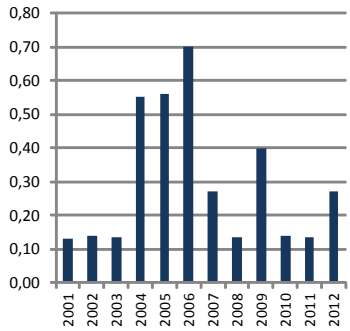

**Omphalocele**



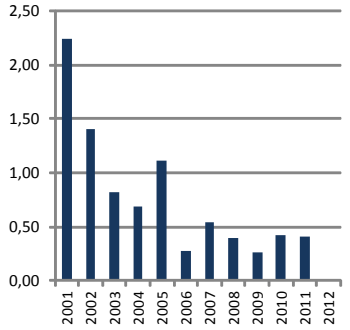

**Gastroschisis**



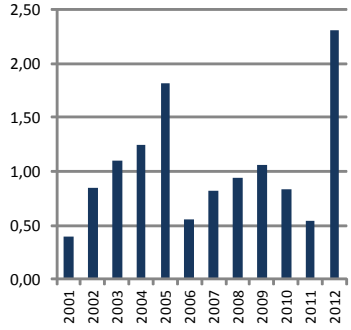

**Prune belly sequence**



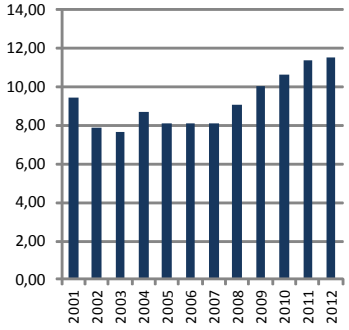

**Trisomy 13**



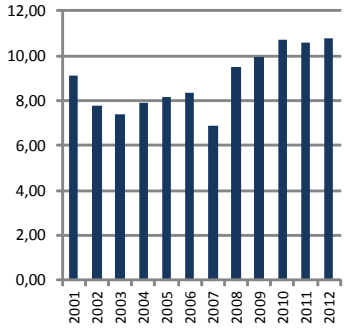

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates**

## Czech Republic

National Registry of Congenital Anomalies of the Czech Republic

**History:**
A registration of congenital malformation began in 1961 and regular monitoring started in 1964. The programme was a founding member of the Clearinghouse and is a full member.

**Size and coverage:**
All births in the Czech Republic (Bohemia, Moravia and Silesia regions) are covered, at present comprising approximately 110,000 annual births. Stillbirths weighting at least 1,000g are included. The information about prenatally diagnosed cases is available from 1994.

**Legislation and funding:**
Reporting is compulsory. The registration is financed and run by the government in the Institute of Health Information and Statistics of the Czech Republic. Analysis of data is supported by Grant projects (currently none available).

**Sources of ascertainment:**
Reports are obtained from delivery units, neonatal, paediatric, child surgery, pathology departments and cytogenetic laboratories. Reporting to the central registry occurs via Regional Department of Institute of Health Information and Statistics.

**Exposure information:**
Some exposure information is available on malformed infants, at present none on controls.

**Background information:**
Information's on all births are available in the Institute of Health Information and Statistics of the Czech Republic.

**Addresses and Staff:**
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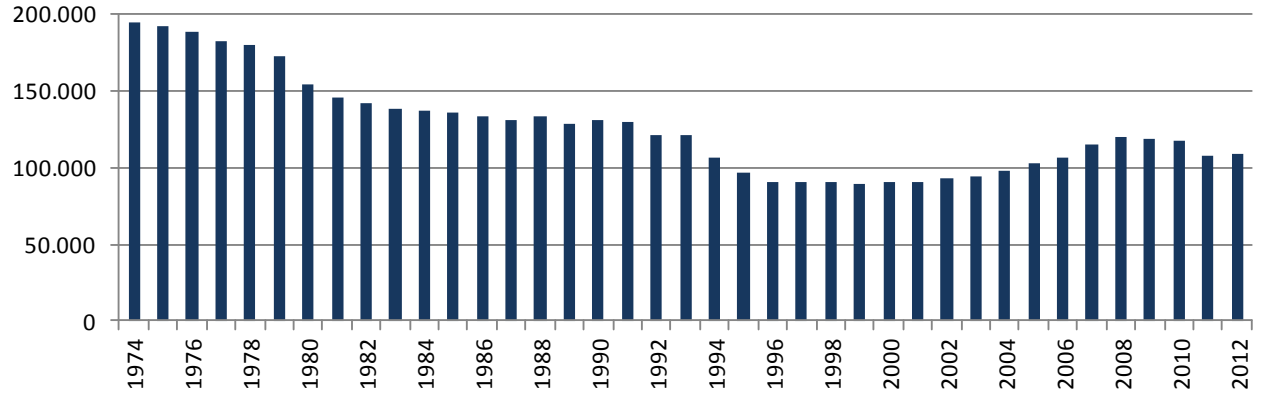
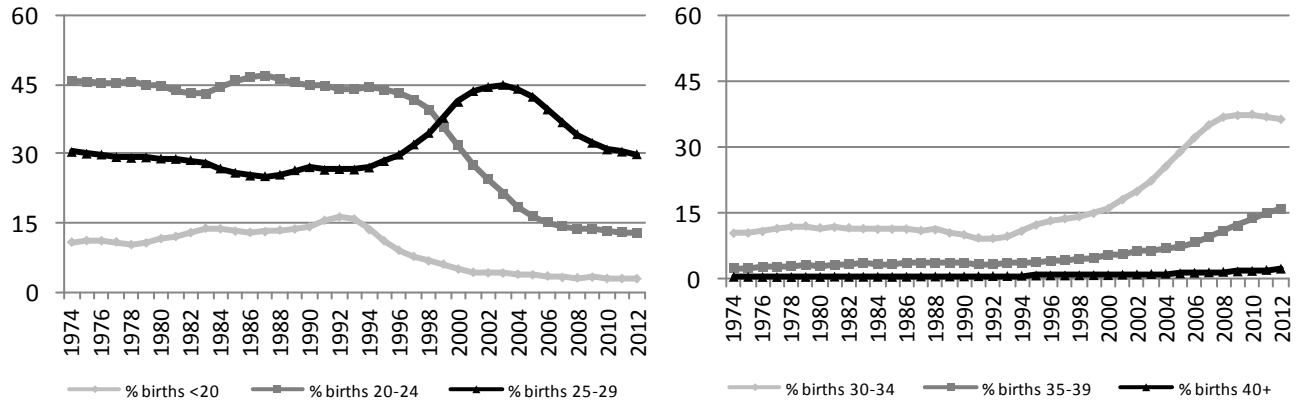
**Websites:** www.vrozene-vady.cz
http://www.uzis.cz/

# Czech Republic

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 110 | 96.5 | Cystic kidney | 22 | 9.2 |
| Spina bifida | 93 | 72.7 | Limb reduction defects | 71 | 28.9 |
| Encephalocele | 36 | 87.8 | Diaphragmatic hernia | 40 | 31.3 |
| Holoprosencephaly | 25 | 67.6 | Omphalocele | 75 | 59.5 |
| Hydrocephaly | 127 | 63.2 | Gastroschisis | 80 | 62.5 |
| Hypoplastic left heart syndrome | 110 | 76.4 | Trisomy 13 | 57 | 95.0 |
| Cleft palate without cleft lip | 4 | 1.5 | Trisomy 18 | 189 | 88.7 |
| Cleft lip with or without cleft palate | 51 | 13.0 | Down syndrome | 681 | 81.9 |
| Renal agenesis | 42 | 14.4 | | | |

Total ToPs with births defects = 2,743 (Ratio ToPs/Births: 8.20 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Czech Republic, 2012

| | |
|---|---|
| Live births (LB) | 108,576 |
| Stillbirths (SB) | 379 |
| Total births | 108,955 |
| Number of terminations of pregnancy (ToP) for birth defects | 970 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 0 | 45 | 4.22 |
| Spina bifida | 11 | 0 | 36 | 4.31 |
| Encephalocele | 1 | 0 | 17 | 1.65 |
| Microcephaly | 11 | 0 | 0 | 1.01 |
| Holoprosencephaly | 3 | 0 | 16 | 1.74 |
| Hydrocephaly | 26 | 2 | 61 | 8.17 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 2 | 0 | 0 | 0.18 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 10 | 0 | 0 | 0.92 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 49 | 0 | 0 | 4.50 |
| Transposition of great vessels | 32 | 1 | 18 | 4.68 |
| Tetralogy of Fallot | 40 | 0 | 4 | 4.04 |
| Hypoplastic left heart syndrome | 14 | 0 | 22 | 3.30 |
| Coarctation of aorta | 45 | 0 | 2 | 4.31 |
| Choanal atresia, bilateral | 9 | 0 | 0 | 0.83 |
| Cleft palate without cleft lip | 97 | 0 | 1 | 8.99 |
| Cleft lip with or without cleft palate | 106 | 0 | 17 | 11.29 |
| Oesophageal atresia/stenosis with or without fistula | 42 | 0 | 0 | 3.85 |
| Small intestine atresia/stenosis | 26 | 1 | 2 | 2.66 |
| Anorectal atresia/stenosis | 52 | 0 | 2 | 4.96 |
| Undescended testis (36 weeks of gestation or later) | 460 | 0 | 0 | 42.22 |
| Hypospadias | 346 | 0 | 0 | 31.76 |
| Epispadias | 11 | 0 | 0 | 1.01 |
| Indeterminate sex | 5 | 0 | 0 | 0.46 |
| Renal agenesis | 81 | 0 | 14 | 8.72 |
| Cystic kidney | 85 | 0 | 12 | 8.90 |
| Bladder exstrophy | 5 | 0 | 2 | 0.64 |
| Polydactyly, preaxial | 215 | 3 | 3 | 20.28 |
| Total Limb reduction defects (include unspecified) | 48 | 0 | 40 | 8.08 |
| Transverse | 25 | 0 | 22 | 4.31 |
| Preaxial | 0 | 0 | 4 | 0.37 |
| Postaxial | 0 | 0 | 4 | 0.37 |
| Intercalary | 0 | 0 | 2 | 0.18 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 23 | 0 | 8 | 2.85 |
| Diaphragmatic hernia | 29 | 0 | 14 | 3.95 |
| Omphalocele | 16 | 0 | 19 | 3.21 |
| Gastroschisis | 13 | 1 | 27 | 3.76 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 1 | 0 | 15 | 1.47 |
| Trisomy 18 | 6 | 0 | 54 | 5.51 |
| Down syndrome, all ages (include age unknown) | 50 | 2 | 230 | 25.88 |
| <20 | 1 | 0 | 1 | 6.45 |
| 20-24 | 7 | 0 | 1 | 5.77 |
| 25-29 | 12 | 0 | 21 | 10.15 |
| 30-34 | 10 | 1 | 60 | 17.95 |
| 35-39 | 11 | 1 | 90 | 58.59 |
| 40-44 | 7 | 0 | 53 | 251.57 |
| 45+ | 1 | 0 | 4 | 390.63 |
| unknown | 1 | 0 | 0 | --- |

nr = data not reported or not available

# Czech Republic, Previous years rates 1974 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | **576,818** | **835,818** | **688,659** | **654,036** | **536,751** | **453,680** | **493,651** | **579,058** |
| Anencephaly | 2.96 | 3.49 | 2.74 | 3.65 | 2.85 | 3.04 | 2.53 | 2.76 |
| Spina bifida | 4.09 | 3.88 | 3.94 | 3.96 | 3.56 | 4.01 | 3.73 | 4.04 |
| Encephalocele | 0.33 | 0.66 | 0.62 | 1.07 | 0.84 | 0.88 | 1.42 | 1.47 |
| Microcephaly | 1.20 | 1.01 | 0.89 | 0.93 | 0.80 | 0.95 | 1.80 | 1.33 |
| Holoprosencephaly | nr | nr | nr | nr | 0.11* | 0.26 | 1.28 | 0.71 |
| Hydrocephaly | 2.03 | 2.64 | 2.96 | 4.45 | 4.79 | 4.34 | 4.62 | 4.87 |
| Anophthalmos | nr | nr | nr | nr | nr | 0.06* | 0.05* | 0.32* |
| Microphthalmos | nr | nr | nr | nr | nr | 0.22* | 0.36* | 0.54* |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr | nr | 0.00* | 0.04 | 0.15* |
| Anotia | nr | nr | nr | nr | nr | 1.02* | 0.41* | 0.22* |
| Microtia | nr | nr | nr | nr | nr | 0.36* | 0.46* | 0.32* |
| Unspecified Anotia/Microtia | nr | nr | nr | nr | nr | 7.03* | 0.10 | 0.60 |
| Transposition of great vessels | 3.17 | 2.26 | 1.86 | 1.47* | 1.87* | 3.95 | 4.01 | 4.32 |
| Tetralogy of Fallot | nr | nr | nr | nr | 2.04* | 3.53 | 3.42 | 3.85 |
| Hypoplastic left heart syndrome | 0.73 | 0.45 | 0.74 | 0.65* | 2.01* | 2.34 | 2.33 | 3.80 |
| Coarctation of aorta | nr | nr | nr | nr | 3.57* | 4.12 | 5.00 | 5.15 |
| Choanal atresia, bilateral | nr | nr | nr | nr | 0.31* | 0.22 | 0.33* | 0.84* |
| Cleft palate without cleft lip | 5.18 | 6.28 | 6.53 | 5.61 | 5.83 | 6.79 | 7.50 | 8.08 |
| Cleft lip with or without cleft palate | 9.43 | 10.39 | 10.47 | 10.24 | 10.10 | 10.10 | 11.34 | 12.02 |
| Oesophageal atresia/stenosis with or without fistula | 1.18 | 1.21 | 1.19 | 1.01 | 1.92 | 2.67 | 2.94 | 3.42 |
| Small intestine atresia/stenosis | nr | nr | nr | nr | 1.87* | 2.38 | 3.44 | 3.38 |
| Anorectal atresia/stenosis | 1.42 | 1.24 | 1.12 | 0.73 | 2.40 | 3.11 | 3.79 | 4.18 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | 4.76* | 16.36 | 25.18* | 33.53* |
| Hypospadias | 19.57 | 18.15 | 20.52 | 23.48 | 23.88 | 27.27 | 32.65 | 32.05 |
| Epispadias | nr | nr | nr | nr | 0.31* | 0.53 | 0.38 | 0.50* |
| Indeterminate sex | nr | nr | nr | nr | 0.34* | 0.53 | 0.36* | 0.37* |
| Renal agenesis | 1.77 | 1.52 | 1.29 | 1.42 | 2.29 | 3.13 | 7.45 | 8.39 |
| Cystic kidney | 2.36 | 2.61 | 2.50 | 2.80 | 2.85 | 4.39 | 6.00 | 6.98 |
| Bladder exstrophy | 0.12 | 0.19 | 0.06 | 0.02* | 0.11* | 0.22 | 0.15* | 0.24* |
| Polydactyly, preaxial | nr | nr | nr | 12.29* | 13.34 | 12.19 | 14.89 | 15.56 |
| Total Limb reduction defects (include unspecified) | 3.80 | 4.75 | 5.33 | 5.17 | 5.03 | 5.40 | 5.85 | 7.27 |
| Transverse | nr | nr | nr | nr | nr | nr | nr | 1.98* |
| Preaxial | nr | nr | nr | nr | nr | nr | nr | 0.19* |
| Postaxial | nr | nr | nr | nr | nr | nr | nr | 0.13* |
| Intercalary | nr | nr | nr | nr | nr | nr | nr | 0.06* |
| Mixed | nr | nr | nr | nr | nr | nr | nr | 1.74* |
| Unspecified | nr | nr | nr | nr | nr | nr | nr | 2.46* |
| Diaphragmatic hernia | 2.51 | 2.57 | 2.58 | 1.85 | 1.88 | 2.40 | 2.92 | 3.25 |
| Omphalocele | 2.22 | 2.32 | 2.29 | 2.43 | 2.38 | 2.40 | 2.39 | 3.44 |
| Gastroschisis | 0.97 | 1.18 | 1.38 | 0.86 | 1.49 | 2.95 | 3.04 | 3.38 |
| Unspecified Omphalocele/Gastroschisis | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.02 | 0.00 | 0.11* |
| Prune belly sequence | nr | nr | nr | nr | nr | nr | 0.13* | 0.17* |
| Trisomy 13 | nr | nr | nr | 0.08* | 0.71 | 1.01 | 2.13 | 1.90 |
| Trisomy 18 | nr | nr | 0.45* | 0.58 | 1.96 | 3.15 | 4.98 | 5.63 |
| Down syndrome, all ages (include age unknown) | 8.60 | 8.36 | 8.20 | 8.16 | 12.28 | 15.45 | 18.52 | 22.81 |
| <20 | 5.69 | 3.70 | 5.92 | 3.52 | 5.42 | 8.24 | 5.88 | 5.76 |
| 20-24 | 6.32 | 4.75 | 4.88 | 2.95 | 5.90 | 8.51 | 8.86 | 7.81 |
| 25-29 | 9.58 | 7.33 | 8.11 | 5.16 | 8.74 | 10.56 | 10.19 | 9.44 |
| 30-34 | 14.85 | 9.19 | 8.43 | 8.24 | 15.11 | 20.40 | 18.42 | 19.65 |
| 35-39 | 30.92 | 31.08 | 31.22 | 25.62 | 54.43 | 56.23 | 67.63 | 59.31 |
| 40-44 | 92.63 | 144.59 | 77.10 | 57.84 | 256.77 | 212.36 | 199.57 | 181.55 |
| 45+ | 79.37 | 368.10 | 200.00 | 404.04 | 791.37 | 625.00 | 504.59 | 580.47 |
| unknown | --- | --- | --- | --- | --- | --- | --- | --- |

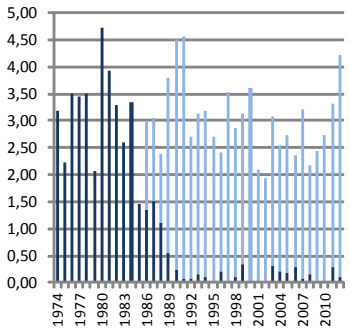
nr = data not reported or not available

* data include less than 5 years

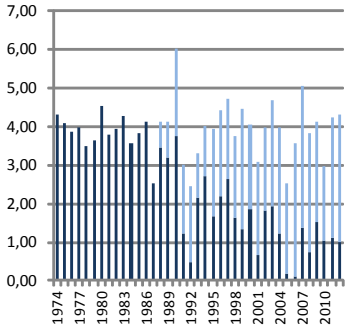
# Czech Republic, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

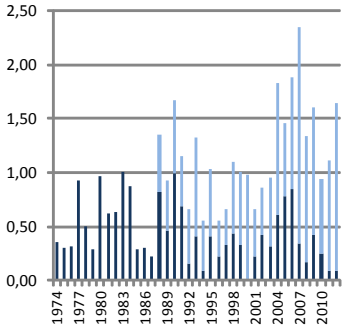
**Anencephaly**



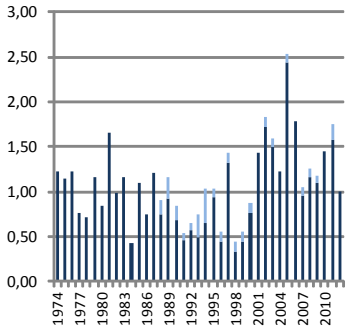

**Spina Bifida**



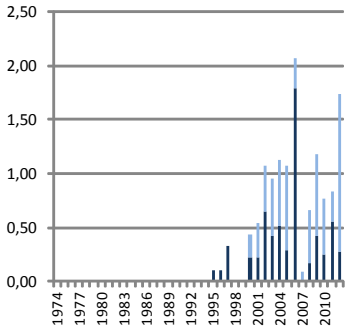

**Encephalocele**



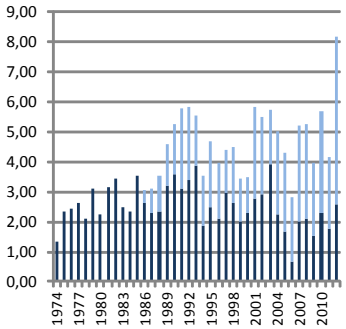

**Microcephaly**



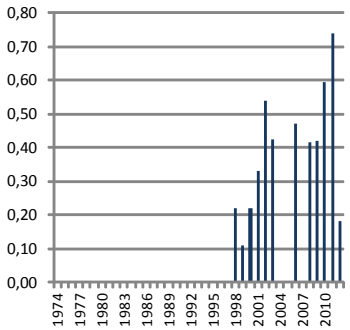

**Holoprosencephaly**



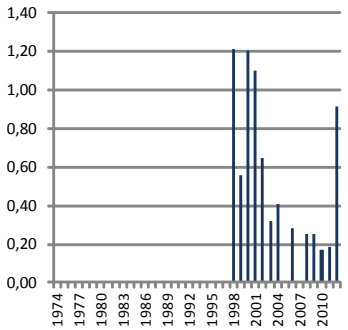

**Hydrocephaly**



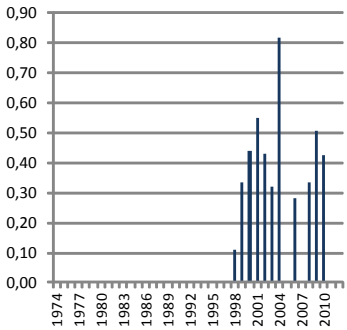

**Microphthalmos**



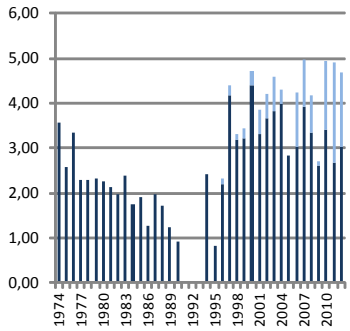

**Anotia**



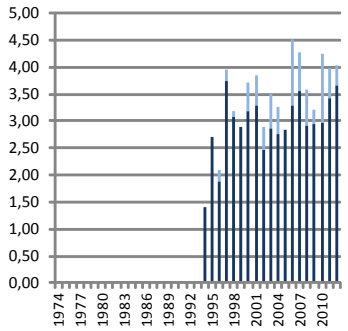

**Microtia**



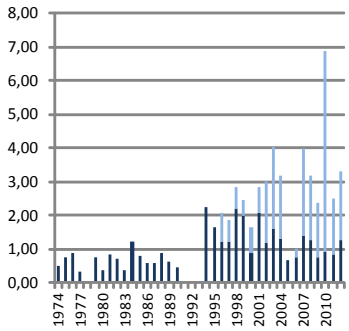

**Transposition of great vessels**




**Tetralogy of Fallot**




**Hypoplastic left heart syndrome**

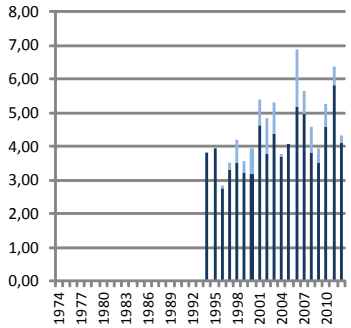



**L + S rates** **ToP rates**

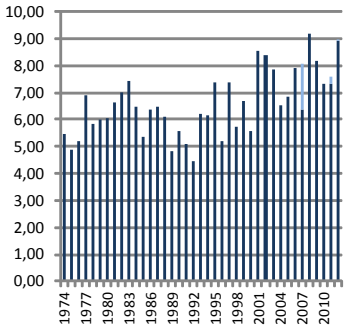
# Czech Republic, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

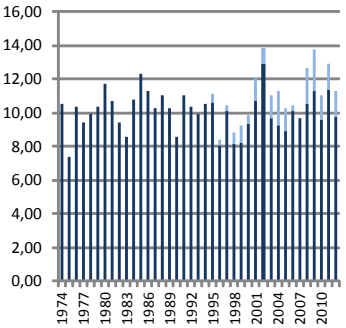
**Coarctation of aorta**



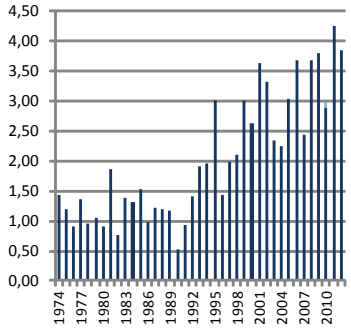

**Cleft palate without cleft lip**



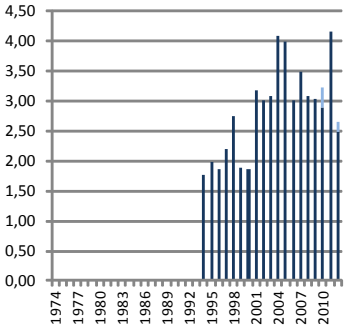

**Cleft lip with or without cleft palate**



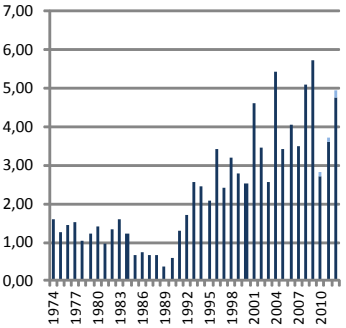

**phageal atresia/stenosis with or without f**



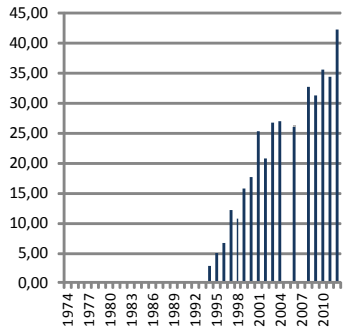

**Small intestine atresia/stenosis**



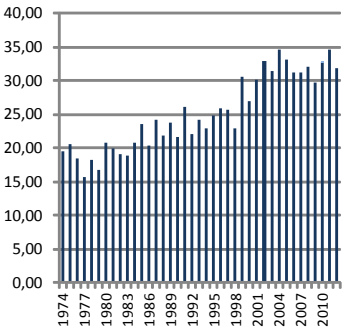

**Anorectal atresia/stenosis**



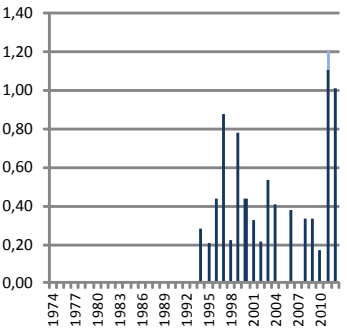

**Undescended testis**



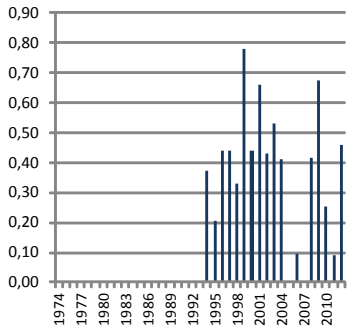

**Hypospadias**



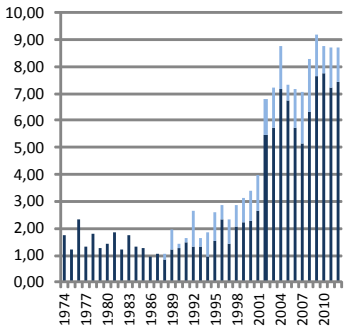

**Epispadias**



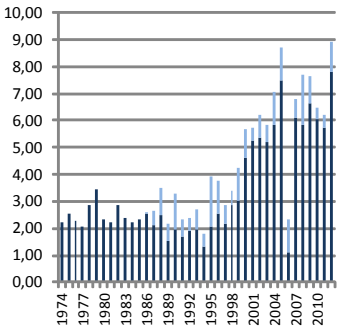

**Indeterminate sex**




**Renal agenesis**




**Cystic kidney**

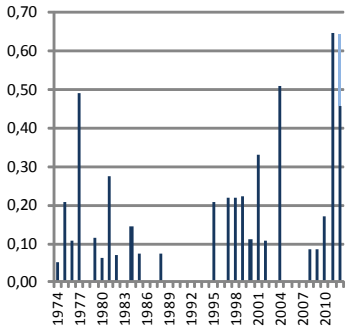



**L + S rates** **ToP rates**

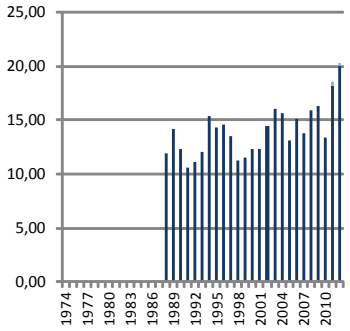
# Czech Republic, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

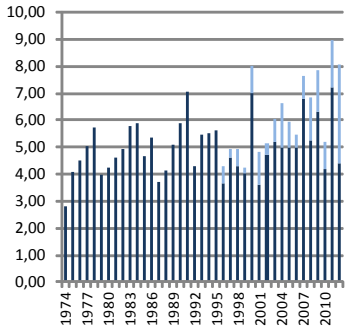
**Bladder, exstrophy**



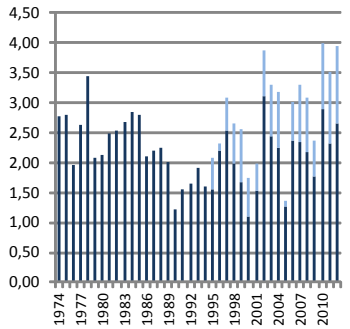

**Polydactyly, preaxial**



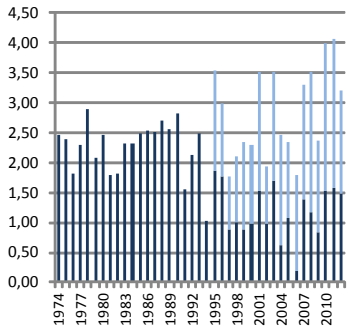

**Limb reduction defects**



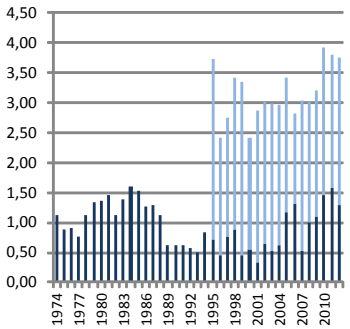

**Diaphragmatic hernia**



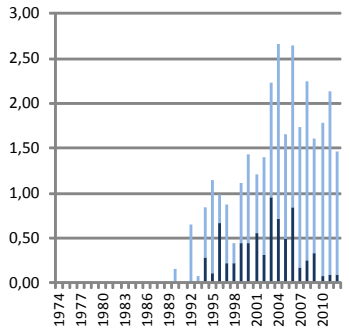

**Omphalocele**



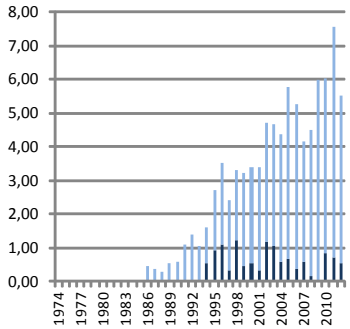

**Gastroschisis**



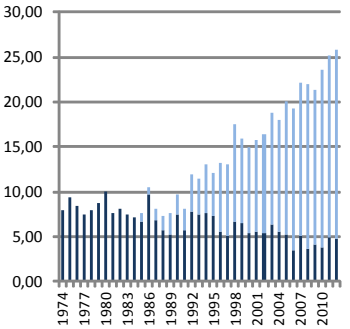

**Trisomy 13**



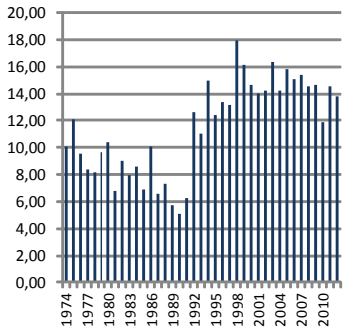

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# France: Paris

**History:**
The Programme was initiated in 1975, but the registry really started in 1981. It became an associate member of the Clearinghouse in 1982. It is also a member of EUROCAT.

**Size and coverage:**
The registry covers 38.000 annual births (about 5% of all births in France), that is all births (live and still births of 22 weeks or more) and terminations of pregnancy in the population of Greater Paris delivering in Paris maternity units. The estimation of the coverage of the registry is around 95%.

**Legislation and funding:**
The registry has been officially recognised by the French National Committee of Registries, and regularly renewed, most recently in 2008 for four years (2009-2012). The activities of the Registry are partially supported by an annual grant from INSERM and Institut de la Veille Sanitaire (Institute for Health Surveillance).

**Sources of ascertainment:**
Reports are actively collected from maternity units, pediatric departments, cytogenetic laboratories, pathology departments. Terminations of pregnancy are included. Case information is also received from the health certificates of the first week.

**Exposure information:**
Information on maternal drug use, maternal and paternal diseases and occupations, outcome of previous pregnancies, is available for the malformed cases. Data about techniques of prenatal screening (ultrasound, serum markers) and prenatal diagnosis are systematically collected.

**Background information:**
Background data on births are available from the National Institute of Statistics (INSEE).

**Addresses and Staff:**
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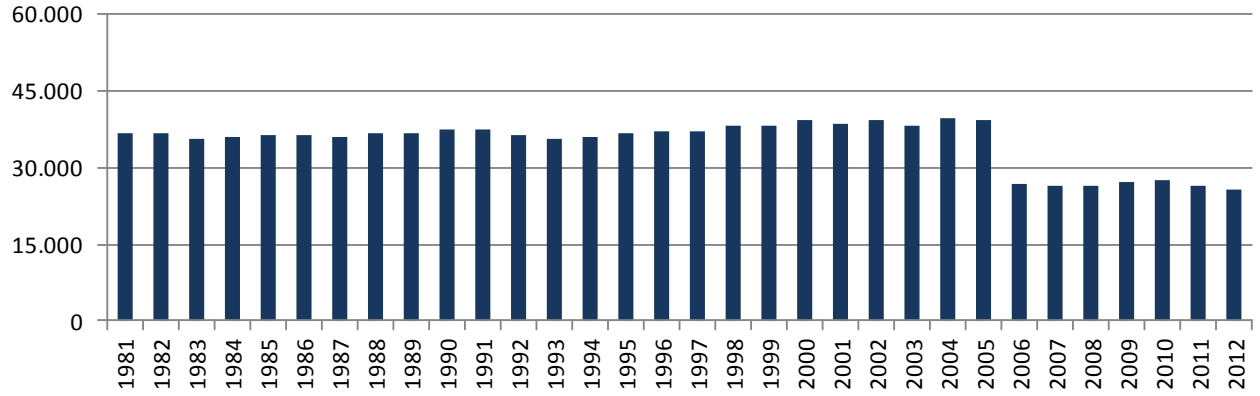
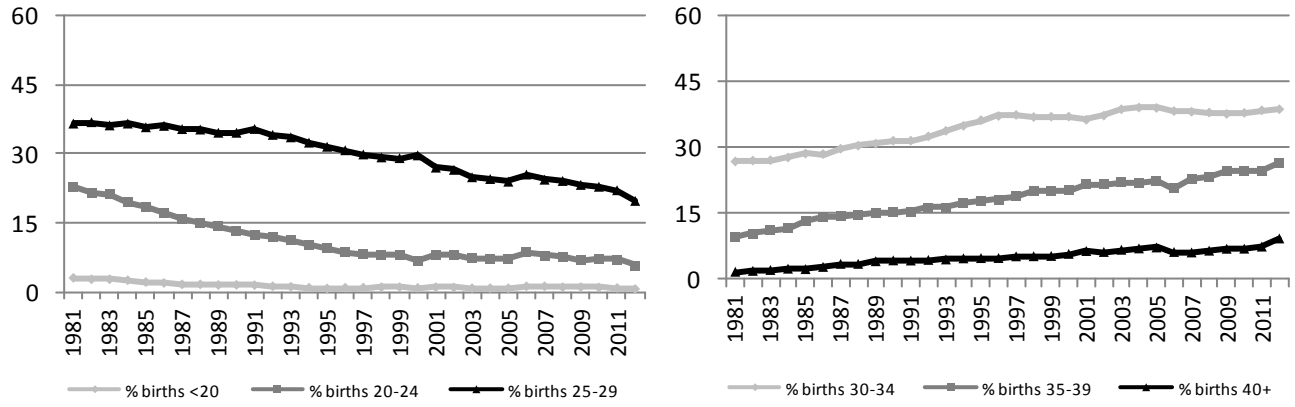
Nathalie Lelong
**E-mail:** Nathalie.lelong@inserm.fr

# France: Paris

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 32 | 94.1 | Cystic kidney | 20 | 24.7 |
| Spina bifida | 30 | 78.9 | Limb reduction defects | 28 | 38.9 |
| Encephalocele | 15 | 88.2 | Diaphragmatic hernia | 8 | 44.4 |
| Holoprosencephaly | 18 | 94.7 | Omphalocele | 40 | 74.1 |
| Hydrocephaly | 30 | 34.9 | Gastroschisis | 2 | 15.4 |
| Hypoplastic left heart syndrome | 16 | 80.0 | Trisomy 13 | 25 | 96.2 |
| Cleft palate without cleft lip | 13 | 27.1 | Trisomy 18 | 93 | 93.9 |
| Cleft lip with or without cleft palate | 14 | 24.1 | Down syndrome | 288 | 81.1 |
| Renal agenesis | 11 | 68.8 | | | |

Total ToPs with births defects = 1,468 (Ratio ToPs/Births: 18.43 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# France: Paris, 2012

| | |
|---|---|
| Live births (LB) | 25,858 |
| Stillbirths (SB) | 11 |
| Total births | 25,869 |
| Number of terminations of pregnancy (ToP) for birth defects | 896 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 9 | 3.48 |
| Spina bifida | 3 | 0 | 12 | 5.80 |
| Encephalocele | 0 | 0 | 6 | 2.32 |
| Microcephaly | 13 | 0 | 7 | 7.73 |
| Holoprosencephaly | 1 | 0 | 11 | 4.64 |
| Hydrocephaly | 15 | 2 | 13 | 11.60 |
| Anophthalmos | 1 | 0 | 0 | 0.39 |
| Microphthalmos | 1 | 0 | 1 | 0.77 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 3 | 0 | 0 | 1.16 |
| Microtia | 1 | 0 | 0 | 0.39 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 3 | 0 | 1 | 1.55 |
| Tetralogy of Fallot | 6 | 0 | 3 | 3.48 |
| Hypoplastic left heart syndrome | 1 | 0 | 5 | 2.32 |
| Coarctation of aorta | 4 | 0 | 1 | 1.93 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 13 | 0 | 5 | 6.96 |
| Cleft lip with or without cleft palate | 15 | 0 | 5 | 7.73 |
| Oesophageal atresia/stenosis with or without fistula | 9 | 1 | 0 | 3.87 |
| Small intestine atresia/stenosis | 4 | 0 | 0 | 1.55 |
| Anorectal atresia/stenosis | 4 | 0 | 5 | 3.48 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 43 | 0 | 3 | 17.78 |
| Epispadias | 1 | 0 | 1 | 0.77 |
| Indeterminate sex | 1 | 0 | 0 | 0.39 |
| Renal agenesis | 4 | 0 | 6 | 3.87 |
| Cystic kidney | 16 | 0 | 4 | 7.73 |
| Bladder exstrophy | 1 | 0 | 2 | 1.16 |
| Polydactyly, preaxial | 7 | 0 | 2 | 3.48 |
| Total Limb reduction defects (include unspecified) | 21 | 0 | 11 | 12.37 |
| Transverse | 3 | 0 | 3 | 2.32 |
| Preaxial | 0 | 0 | 5 | 1.93 |
| Postaxial | 1 | 0 | 0 | 0.39 |
| Intercalary | 14 | 0 | 2 | 6.19 |
| Mixed | 3 | 0 | 1 | 1.55 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 6 | 0 | 4 | 3.87 |
| Omphalocele | 3 | 0 | 15 | 6.96 |
| Gastroschisis | 3 | 0 | 0 | 1.16 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | 10 | 4.25 |
| Trisomy 18 | 1 | 2 | 27 | 11.60 |
| Down syndrome, all ages (include age unknown) | 24 | 2 | 93 | 46.00 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 0 | 0 | 1 | 6.80 |
| 25-29 | 4 | 0 | 3 | 13.73 |
| 30-34 | 6 | 1 | 20 | 27.04 |
| 35-39 | 10 | 1 | 37 | 70.70 |
| 40-44 | 4 | 0 | 31 | 162.56 |
| 45+ | 0 | 0 | 1 | 45.05 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

# France: Paris, Previous years rates 1981 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **36,917** | **181,346** | **184,461** | **182,327** | **191,748** | **183,099** | **133,860** |
| Anencephaly | | 2.17 | 0.88 | 0.65 | 3.07 | 5.48 | 6.17 | 4.93 |
| Spina bifida | | 5.42 | 2.70 | 1.30 | 3.89 | 4.95 | 4.48 | 5.15 |
| Encephalocele | | 0.00 | 0.72 | 0.81 | 1.54 | 2.14 | 2.02 | 2.17 |
| Microcephaly | | 2.17 | 2.48 | 1.95 | 2.85 | 3.13 | 3.06 | 2.39 |
| Holoprosencephaly | | 0.00 | 0.44 | 0.27 | 1.59 | 2.66 | 3.66 | 2.24 |
| Hydrocephaly | | 5.15 | 3.36 | 3.20 | 9.27 | 12.20 | 15.07 | 14.42 |
| Anophthalmos | | 0.27 | 0.22 | 0.22 | 0.38 | 0.31 | 0.22 | 0.07 |
| Microphthalmos | | 0.00 | 0.99 | 1.14 | 1.65 | 1.62 | 0.93 | 0.82 |
| Unspecified Anophthalmos/Microphthalmos | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | | 0.00 | 0.28 | 0.38 | 0.71 | 0.78 | 0.98 | 1.64 |
| Microtia | | 0.00 | 0.39 | 0.65 | 0.55 | 0.78 | 0.71 | 0.60 |
| Unspecified Anotia/Microtia | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | 0.81 | 2.65 | 2.33 | 4.39 | 6.15 | 4.64 | 3.51 |
| Tetralogy of Fallot | | 1.08 | 1.05 | 1.73 | 2.19 | 4.17 | 4.21 | 4.11 |
| Hypoplastic left heart syndrome | | 1.08 | 1.76 | 1.41 | 3.07 | 3.29 | 4.10 | 3.06 |
| Coarctation of aorta | | 0.54 | 1.43 | 2.28 | 3.02 | 3.81 | 3.00 | 3.81 |
| Choanal atresia, bilateral | | 0.81 | 0.50 | 0.76 | 0.44 | 0.52 | 0.33 | 0.22 |
| Cleft palate without cleft lip | | 2.17 | 4.30 | 5.04 | 6.20 | 6.99 | 5.68 | 6.35 |
| Cleft lip with or without cleft palate | | 6.23 | 6.45 | 8.94 | 8.99 | 9.39 | 7.92 | 8.22 |
| Oesophageal atresia/stenosis with or without fistula | | 2.17 | 2.37 | 3.58 | 3.57 | 3.81 | 4.04 | 2.99 |
| Small intestine atresia/stenosis | | 0.00 | 0.50 | 1.41 | 2.41 | 2.09 | 4.26 | 1.34 |
| Anorectal atresia/stenosis | | 2.71 | 3.09 | 2.11 | 3.95 | 3.18 | 3.50 | 3.44 |
| Undescended testis (36 weeks of gestation or later) | | 6.77 | 10.15 | 12.69 | 10.91 | 5.22 | 6.63* | nr |
| Hypospadias | | 10.29 | 10.09 | 12.96 | 13.05 | 11.32 | 16.00 | 17.26 |
| Epispadias | | 0.00 | 0.33 | 0.60 | 0.33 | 0.42 | 0.44 | 0.85* |
| Indeterminate sex | | 1.90 | 1.32 | 1.36 | 1.26 | 1.36 | 1.31 | 0.97 |
| Renal agenesis | | 1.08 | 1.10 | 0.76 | 2.63 | 2.71 | 2.89 | 1.20 |
| Cystic kidney | | 0.81 | 2.15 | 3.47 | 6.80 | 10.17 | 10.76 | 10.76 |
| Bladder exstrophy | | 0.00 | 0.39 | 0.22 | 0.82 | 0.47 | 0.49 | 0.97 |
| Polydactyly, preaxial | | 0.27 | 0.72 | 1.08 | 2.03 | 2.40 | 1.37 | 1.87 |
| Total Limb reduction defects (include unspecified) | | nr | nr | nr | 6.07* | 7.56 | 7.32 | 6.80 |
| Transverse | | nr | nr | nr | 1.62* | 3.91 | 4.21 | 4.18 |
| Preaxial | | nr | nr | nr | 0.40* | 1.36 | 1.20 | 1.05 |
| Postaxial | | nr | nr | nr | 0.40* | 0.42 | 0.87 | 0.07 |
| Intercalary | | nr | nr | nr | 0.40* | 0.57 | 0.33 | 0.52 |
| Mixed | | nr | nr | nr | 0.13* | 0.68 | 0.44 | 0.67 |
| Unspecified | | nr | nr | nr | 0.00* | 0.10 | 0.27 | 0.30 |
| Diaphragmatic hernia | | 1.90 | 2.48 | 2.82 | 4.44 | 5.37 | 5.30 | 2.17 |
| Omphalocele | | 0.81 | 1.76 | 2.01 | 3.24 | 5.68 | 5.79 | 6.42 |
| Gastroschisis | | 0.00 | 0.61 | 1.25 | 2.58 | 3.29 | 2.51 | 1.72 |
| Unspecified Omphalocele/Gastroschisis | | 0.00 | 0.55 | 0.22 | 0.55 | 1.15 | 0.98 | 0.37 |
| Prune belly sequence | | 0.00 | 0.17 | 0.05 | 0.11 | 0.05 | 0.27 | 0.00 |
| Trisomy 13 | | 0.81 | 0.39 | 0.60 | 1.86 | 3.81 | 4.64 | 4.11 |
| Trisomy 18 | | 0.81 | 1.43 | 1.19 | 4.94 | 9.28 | 13.60 | 14.42 |
| Down syndrome, all ages (include age unknown) | | 10.84 | 12.02 | 12.14 | 25.83 | 36.56 | 42.27 | 42.73 |
| <20 | | 9.03 | 11.59 | 10.77 | 5.81 | 16.67 | 12.78 | 7.66 |
| 20-24 | | 10.74 | 6.22 | 6.53 | 10.75 | 12.14 | 11.63 | 14.37 |
| 25-29 | | 3.71 | 7.46 | 6.82 | 10.66 | 14.62 | 15.08 | 14.77 |
| 30-34 | | 8.12 | 12.15 | 12.53 | 16.06 | 23.51 | 23.77 | 23.46 |
| 35-39 | | 17.11 | 28.49 | 24.81 | 48.26 | 55.22 | 67.05 | 58.77 |
| 40-44 | | 90.25 | 31.81 | 25.14 | 155.68 | 203.17 | 187.15 | 232.28 |
| 45+ | | 810.81 | 91.74 | 129.45 | 243.31 | 284.55 | 443.29 | 151.72 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

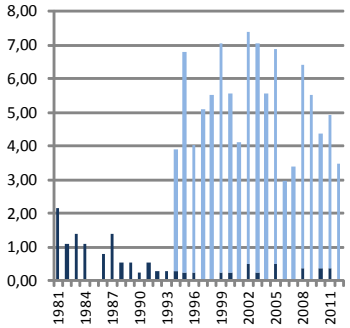
nr = data not reported or not available

* data include less than 5 years

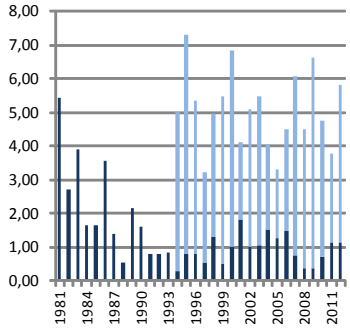
# France: Paris, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

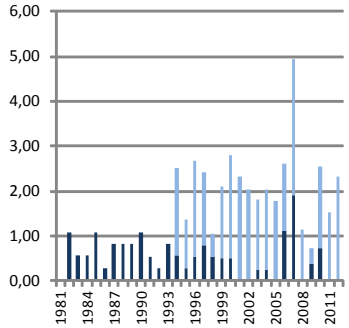
**Anencephaly**



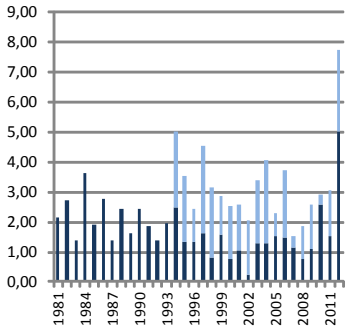

**Spina Bifida**



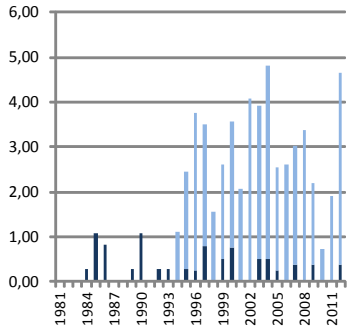

**Encephalocele**



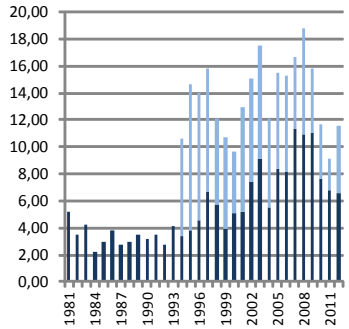

**Microcephaly**



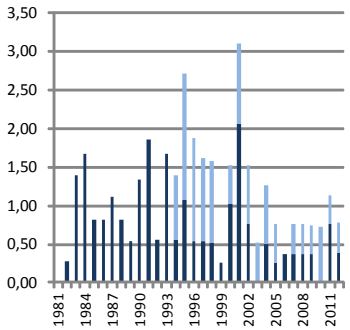

**Holoprosencephaly**



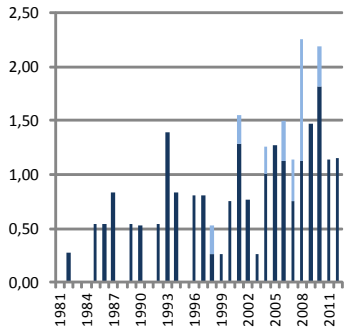

**Hydrocephaly**



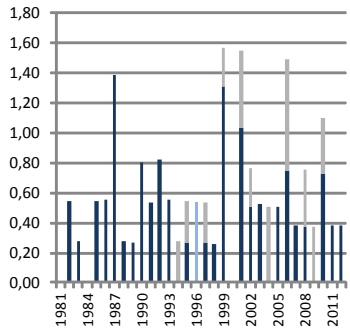

**Microphthalmos**



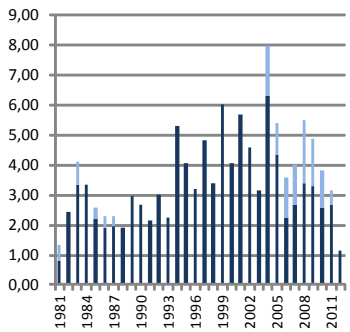

**Anotia**



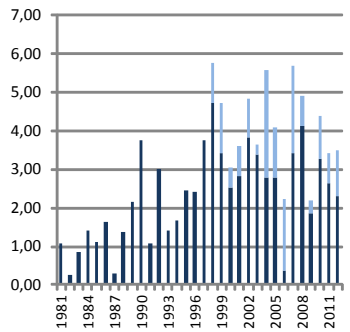

**Microtia**



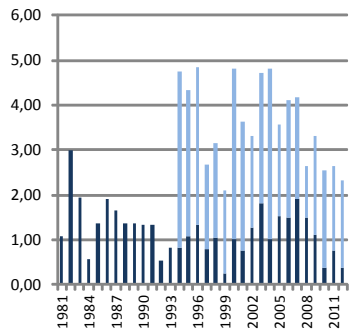

**Transposition of great vessels**




**Tetralogy of Fallot**




**Hypoplastic left heart syndrome**

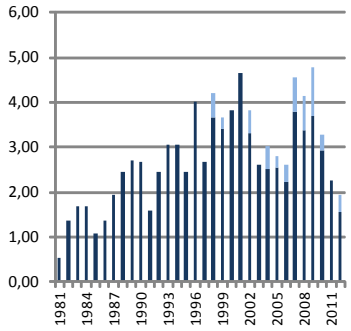



**L + S rates** **ToP rates**

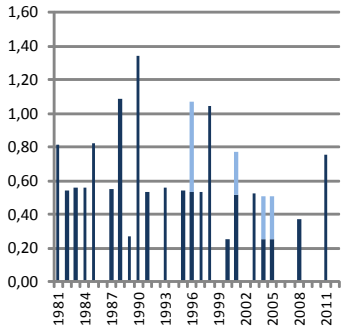
# France: Paris, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

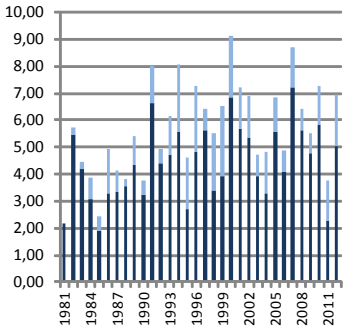
**Coarctation of aorta**



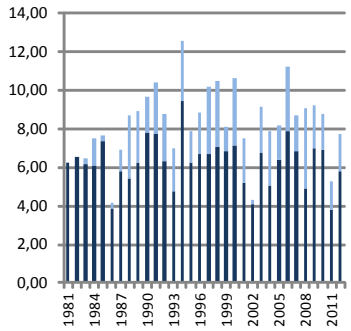

**Choanal atresia, bilateral**



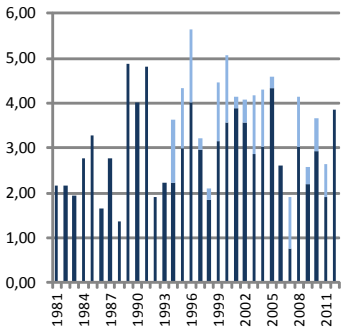

**Cleft palate without cleft lip**



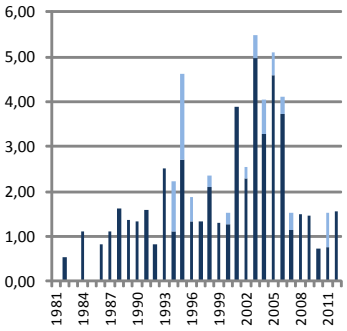

**Cleft lip with or without cleft palate**



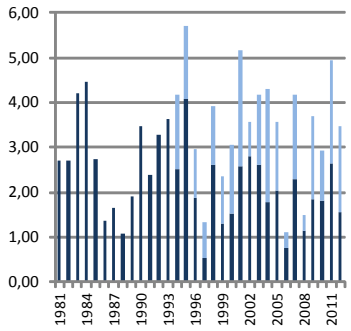

**phageal atresia/stenosis with or without f**



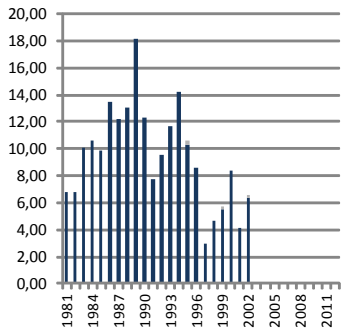

**Small intestine atresia/stenosis**



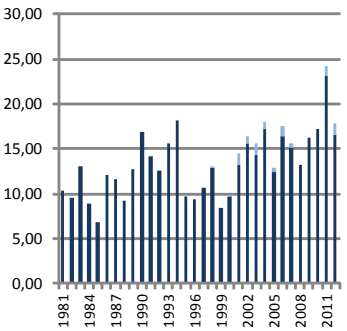

**Anorectal atresia/stenosis**



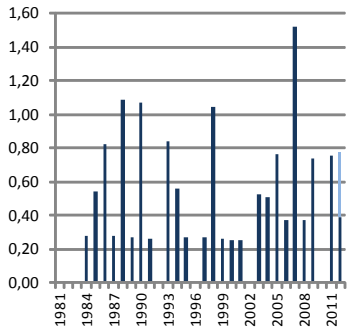

**Undescended testis**



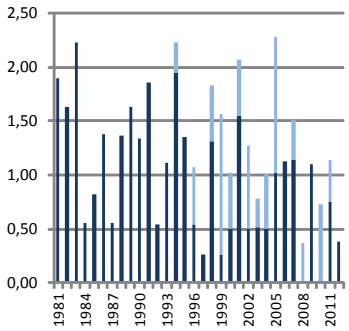

**Hypospadias**



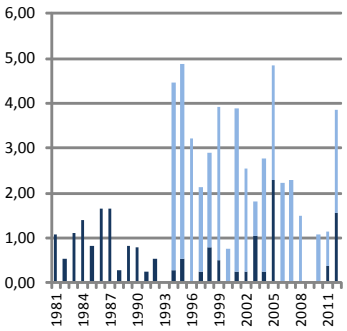

**Epispadias**




**Indeterminate sex**




**Renal agenesis**

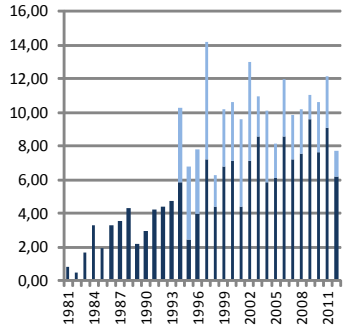



**L + S rates** **ToP rates**

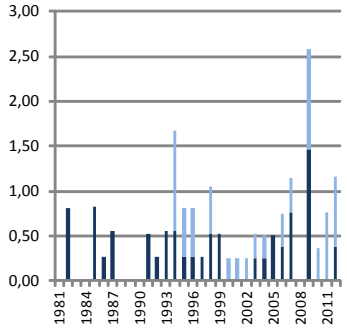
# France: Paris, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

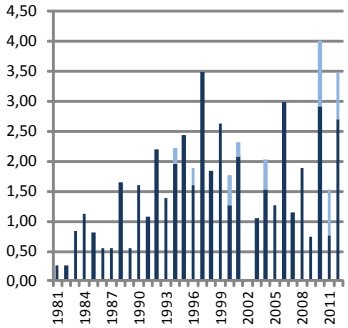
**Cystic kidney**



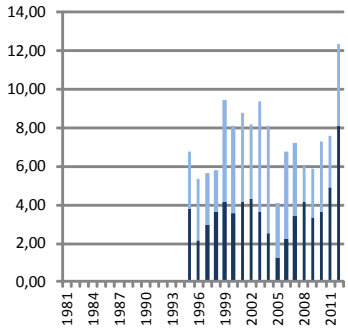

**Bladder exstrophy**



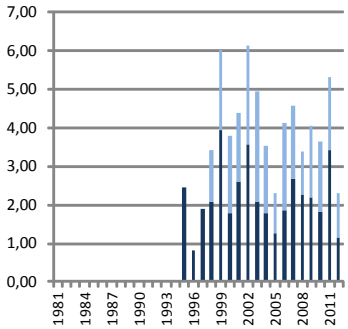

**Polydactyly, preaxial**



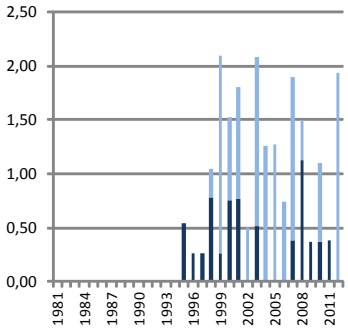

**Limb reduction defects**



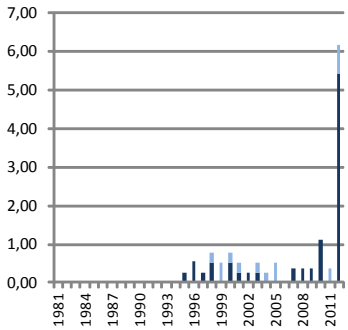

**Limb reduction defects - transverse**



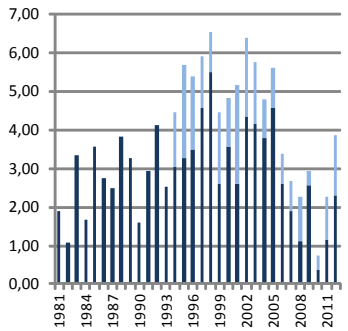

**Limb reduction defects - preaxial**



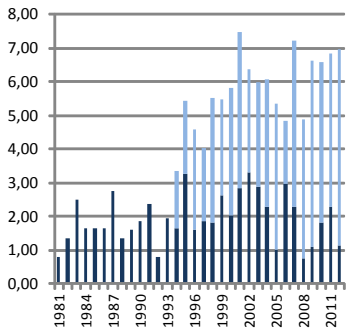

**Limb reduction defects - intercalary**



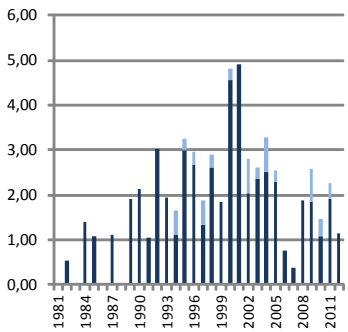

**Diaphragmatic hernia**



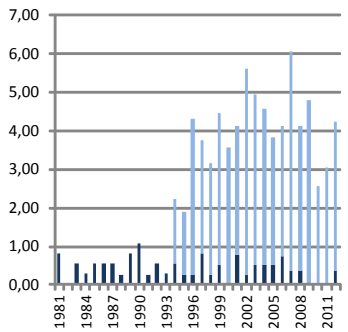

**Omphalocele**



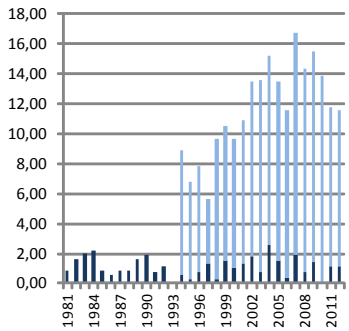

**Gastroschisis**




**Trisomy 13**



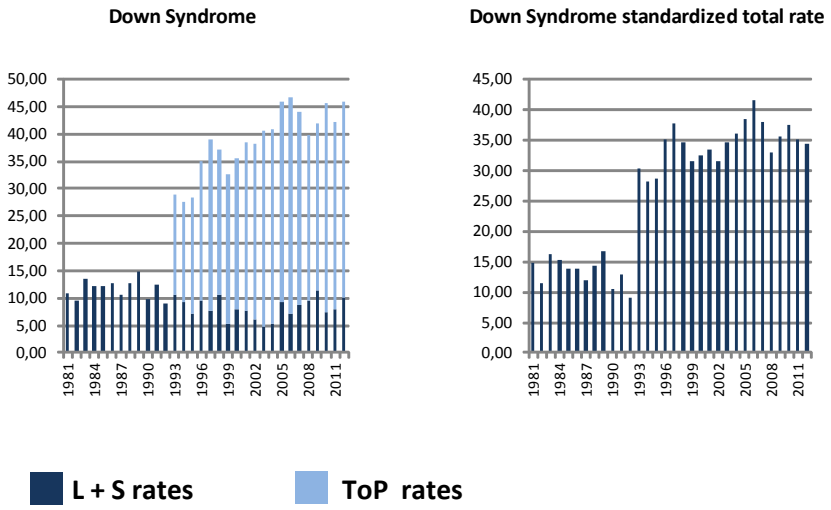

**Trisomy 18**




**L + S rates** **ToP rates**

# France: Paris, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)



# France: REMERA

## Central-East France Register of Congenital Malformation (until 2006)
## Registre des Malformations en Rhône Alpes

**History**:
The registry began in 1973 within the Rhone-Alps area -the Auvergne region was added in 1983, the Jura area in 1985, the Côte d'Or & Nièvre in 1989 and Saône-et-Loire in 1990. The Programme was a founding member of the ICBDSR and is a full member. In 1998 the registry was split up and the Auvergne region became financially independent, under the responsibility of Christine Francannet. The collaboration between Auvergne and the rest of the FCE-registry is maintained. In December 2006, France Central-East Register was closed. A new register (REMERA) was created, covering part of the previous one.

**Size and coverage:**
The registry covers all births in the area approximately 58,000 births annually, which represents about 7% of all births in France.

**Legislation and funding:**
Since January 2007, the financial support is provided only by public funds: by the Rhone-Alps Region authority, the French Institute of disease Monitoring (InVS), the National Institute of Health and Medical Research (INSERM) and The French National Agency for Medicines and Health Products Safety (ANSM)

**Sources of ascertainment:**
The registry is population based and covers 4 French counties of Rhône-Alpes region: Rhône, Loire, Isère, Savoie. Since 2012, Savoie has been replaced by Ain.
Data collection is actively performed in all the private and public maternity wards and paediatric and surgery units. Other sources of information include genetic counselling wards, cytogenetic laboratories, pathology laboratories, departments of medical genetics, birth certificates and data set called "Résumé Standardisé de Sortie" (similar to a "Standardized Discharge Summary"). Data is registered on a dedicated and secured server.

**Inclusion criteria:**
All birth defects are collected. Are excluded from registration: balanced chromosomal anomalies, pyloric stenosis, metabolic disorders, minor malformations (small angiomas or naevi, hip subdislocation, small foot deformities, ill-defined facial anomalies, inguinal and umbilical hernias).
The maximum age at postnatal diagnosis is 1 year. For children born in year x, notifications are taken into account until March x+2.
Stillbirths from 20 weeks' gestation are included. The official stillbirth definition is 22 weeks after LMP (28 weeks before 1997), which is the lower gestational age limit to include early fetal deaths/spontaneous abortions. Terminations of pregnancies (ToP) are registered since 1985. ToP can be performed up to full term in case of lethal or severe foetal abnormalities.

**Exposure information:**
Information on maternal and paternal occupation, medications, chemicals and physical exposures (tobacco, alcohol, drugs, infectious agents, place of residence, environmental factors), lifestyle.

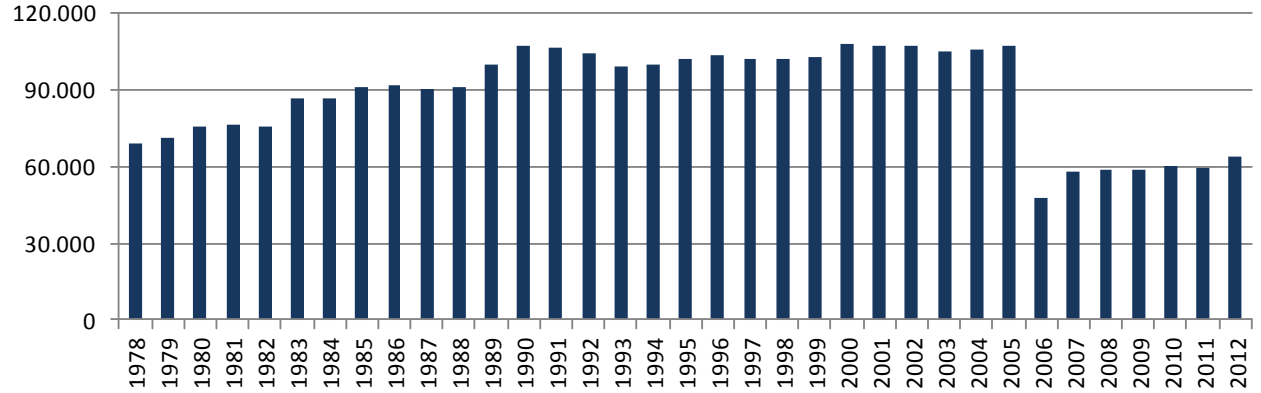
**Prenatal diagnosis Information:**
Data about times and techniques of prenatal diagnosis screening and prenatal diagnosis are systematically collected.

**Background information:**
Denominators information is obtained from National institute of Statistics. Some background information is available from the general population statistics.

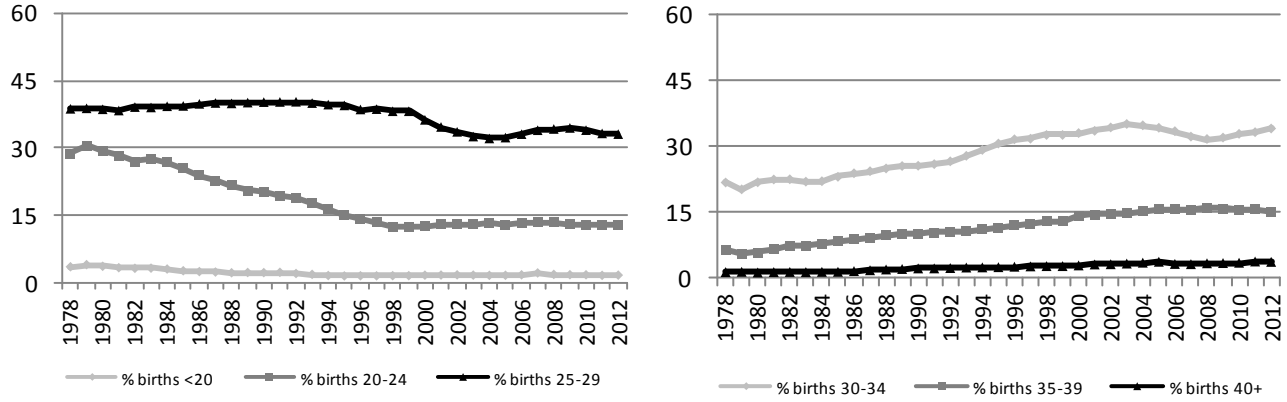
**Addresses and Staff:**
Emmanuelle Amar, Programme Director
Registre Des Malformations en Rhône Alpes
7 rue Sainte Catherine
69001 LYON - FRANCE
**Phone**: 33-4-78583484
**E-mail** : emmanuelle.amar@remera.fr

# France: REMERA

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 86 | 96.6 | Cystic kidney | 75 | 37.3 |
| Spina bifida | 80 | 75.5 | Limb reduction defects | 49 | 49.5 |
| Encephalocele | 21 | 95.5 | Diaphragmatic hernia | 19 | 20.2 |
| Holoprosencephaly | 40 | 85.1 | Omphalocele | 47 | 67.1 |
| Hydrocephaly | 90 | 51.1 | Gastroschisis | 8 | 19.0 |
| Hypoplastic left heart syndrome | 62 | 65.3 | Trisomy 13 | 42 | 89.4 |
| Cleft palate without cleft lip | 26 | 19.7 | Trisomy 18 | 124 | 92.5 |
| Cleft lip with or without cleft palate | 34 | 19.7 | Down syndrome | 379 | 77.5 |
| Renal agenesis | 37 | 26.8 | | | |

Total ToPs with births defects = 1,490 (Ratio ToPs/Births: 18.13 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# France: REMERA, 2012

| | |
|---|---|
| Live births (LB) | 63,447 |
| Stillbirths (SB) | 649 |
| Total births | 64,096 |
| Number of terminations of pregnancy (ToP) for birth defects | 532 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 24 | 3.74 |
| Spina bifida | 14 | 1 | 23 | 5.93 |
| Encephalocele | 1 | 0 | 6 | 1.09 |
| Microcephaly | 6 | 0 | 12 | 2.81 |
| Holoprosencephaly | 0 | 1 | 16 | 2.65 |
| Hydrocephaly | 36 | 4 | 28 | 10.61 |
| Anophthalmos | 0 | 1 | 0 | 0.16 |
| Microphthalmos | 1 | 0 | 5 | 0.94 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 2 | 0 | 0 | 0.31 |
| Microtia | 6 | 0 | 7 | 2.03 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 35 | 1 | 8 | 6.86 |
| Tetralogy of Fallot | 20 | 0 | 7 | 4.21 |
| Hypoplastic left heart syndrome | 9 | 0 | 19 | 4.37 |
| Coarctation of aorta | 20 | 1 | 1 | 3.43 |
| Choanal atresia, bilateral | 8 | 0 | 2 | 1.56 |
| Cleft palate without cleft lip | 36 | 1 | 13 | 7.80 |
| Cleft lip with or without cleft palate | 47 | 2 | 13 | 9.67 |
| Oesophageal atresia/stenosis with or without fistula | 15 | 0 | 2 | 2.65 |
| Small intestine atresia/stenosis | 7 | 0 | 1 | 1.25 |
| Anorectal atresia/stenosis | 4 | 0 | 0 | 0.62 |
| Undescended testis (36 weeks of gestation or later) | 15 | 1 | 3 | 2.96 |
| Hypospadias | 88 | 1 | 5 | 14.67 |
| Epispadias | 4 | 0 | 0 | 0.62 |
| Indeterminate sex | 1 | 1 | 0 | 0.31 |
| Renal agenesis | 44 | 3 | 12 | 9.20 |
| Cystic kidney | 58 | 0 | 24 | 12.79 |
| Bladder exstrophy | 0 | 0 | 4 | 0.62 |
| Polydactyly, preaxial | 74 | 0 | 10 | 13.11 |
| Total Limb reduction defects (include unspecified) | 21 | 0 | 13 | 5.30 |
| Transverse | 15 | 0 | 8 | 3.59 |
| Preaxial | 1 | 0 | 1 | 0.31 |
| Postaxial | 4 | 0 | 2 | 0.94 |
| Intercalary | 1 | 0 | 0 | 0.16 |
| Mixed | 0 | 0 | 2 | 0.31 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 22 | 1 | 6 | 4.52 |
| Omphalocele | 8 | 2 | 19 | 4.52 |
| Gastroschisis | 13 | 0 | 2 | 2.34 |
| Unspecified Omphalocele/Gastroschisis | 1 | 0 | 7 | 1.25 |
| Prune belly sequence | 0 | 1 | 0 | 0.16 |
| Trisomy 13 | 2 | 2 | 20 | 3.74 |
| Trisomy 18 | 3 | 3 | 43 | 7.64 |
| Down syndrome, all ages (include age unknown) | 37 | 2 | 140 | 27.93 |
| <20 | 0 | 0 | 1 | 9.97 |
| 20-24 | 3 | 0 | 7 | 12.23 |
| 25-29 | 4 | 0 | 20 | 11.33 |
| 30-34 | 10 | 0 | 25 | 16.07 |
| 35-39 | 9 | 1 | 44 | 55.99 |
| 40-44 | 8 | 1 | 41 | 227.27 |
| 45+ | 2 | 0 | 0 | 172.41 |
| unknown | 1 | 0 | 2 | --- |

nr = data not reported or not available

# France: REMERA, Previous years rates 1978 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **291,826** | **431,925** | **495,329** | **508,996** | **523,282** | **472,493** | **294,910** |
| Anencephaly | | 0.82 | 0.81 | 0.36 | 1.30 | 1.61 | 1.93 | 4.71 |
| Spina bifida | | 4.04 | 3.40 | 2.06 | 3.44 | 3.46 | 4.89 | 5.76 |
| Encephalocele | | 0.69 | 0.74 | 1.03 | 1.36 | 1.61 | 1.84 | 1.39 |
| Microcephaly | | 1.40 | 2.43 | 2.20 | 1.85 | 1.91 | 2.10 | 1.90 |
| Holoprosencephaly | | 0.31 | 0.51 | 0.91 | 1.43 | 1.40 | 1.69 | 2.34 |
| Hydrocephaly | | 1.78 | 2.99 | 2.99 | 3.93 | 5.20 | 6.35 | 8.54 |
| Anophthalmos | | 0.31 | 0.05 | 0.24 | 0.18 | 0.15 | 0.25 | 0.17 |
| Microphthalmos | | 0.99 | 1.00 | 1.23 | 1.14 | 1.15 | 0.80 | 0.98 |
| Unspecified Anophthalmos/Microphthalmos | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.30* |
| Anotia | | 0.24 | 0.30 | 0.50 | 0.37 | 0.46 | 0.25 | 0.44 |
| Microtia | | 0.14 | 0.25 | 0.28 | 0.47 | 0.46 | 0.53 | 0.78 |
| Unspecified Anotia/Microtia | | 0.38 | 0.56 | 0.79 | 0.69 | 0.92 | 0.13 | 0.30* |
| Transposition of great vessels | | 2.95 | 3.13 | 3.69 | 3.20 | 3.06 | 4.06 | 4.00 |
| Tetralogy of Fallot | | 1.85 | 2.41 | 2.42 | 2.20 | 2.27 | 3.03 | 3.46 |
| Hypoplastic left heart syndrome | | 1.51 | 1.97 | 2.38 | 2.14 | 2.90 | 2.92 | 5.22 |
| Coarctation of aorta | | 1.95 | 2.76 | 2.95 | 2.50 | 2.33 | 2.46 | 2.51 |
| Choanal atresia, bilateral | | 0.65 | 0.67 | 0.85 | 0.53 | 1.03 | 0.87 | 0.54 |
| Cleft palate without cleft lip | | 4.15 | 5.07 | 4.64 | 6.74 | 6.19 | 4.99 | 5.93 |
| Cleft lip with or without cleft palate | | 6.82 | 6.11 | 6.48 | 8.11 | 7.59 | 7.37 | 9.80 |
| Oesophageal atresia/stenosis with or without fistula | | 2.02 | 2.36 | 3.15 | 3.06 | 2.96 | 2.79 | 3.39 |
| Small intestine atresia/stenosis | | 1.64 | 1.53 | 1.78 | 2.02 | 2.77 | 2.79 | 1.53 |
| Anorectal atresia/stenosis | | 2.09 | 3.19 | 3.15 | 3.71 | 3.71 | 3.15 | 0.27 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | nr | nr | 0.00* | 1.29 |
| Hypospadias | | 6.23 | 6.90 | 10.20 | 9.69 | 12.15 | 11.94 | 11.56 |
| Epispadias | | 0.17 | 0.19 | 0.32 | 0.14 | 0.29 | 0.15 | 0.09* |
| Indeterminate sex | | 0.55 | 0.81 | 0.77 | 0.71 | 0.52 | 0.80 | 0.47 |
| Renal agenesis | | 0.48 | 0.90 | 0.46 | 1.06 | 1.41 | 1.78 | 5.70 |
| Cystic kidney | | 0.65 | 1.46 | 2.73 | 3.91 | 4.55 | 5.21 | 9.09 |
| Bladder exstrophy | | 0.17 | 0.23 | 0.38 | 0.33 | 0.38 | 0.25 | 0.47 |
| Polydactyly, preaxial | | 0.82 | 0.86 | 1.59 | 2.08 | 2.06 | 2.20 | 9.90 |
| Total Limb reduction defects (include unspecified) | | 4.69 | 4.07 | 4.06 | 4.99 | 4.97 | 5.23 | 5.56 |
| Transverse | | 2.36 | 2.06 | 2.42 | 2.30 | 2.56 | 2.52 | 2.90* |
| Preaxial | | 0.62 | 0.76 | 0.52 | 0.69 | 0.94 | 1.23 | 1.02* |
| Postaxial | | 0.31 | 0.25 | 0.52 | 0.26 | 0.42 | 0.55 | 0.60* |
| Intercalary | | 0.55 | 0.49 | 0.32 | 0.55 | 0.40 | 0.47 | 0.26* |
| Mixed | | 0.62 | 0.49 | 0.26 | 0.29 | 0.36 | 0.42 | 0.26* |
| Unspecified | | 0.24 | 0.02 | 0.00 | 0.06 | 0.08 | 0.05* | 0.17* |
| Diaphragmatic hernia | | 1.92 | 2.80 | 2.28 | 3.20 | 2.58 | 3.32 | 4.20 |
| Omphalocele | | 1.06 | 1.09 | 1.27 | 1.69 | 2.54 | 2.62 | 3.66 |
| Gastroschisis | | 0.55 | 0.74 | 1.03 | 1.20 | 1.26 | 1.74 | 2.37 |
| Unspecified Omphalocele/Gastroschisis | | 0.00 | 0.00 | 0.00 | 0.04 | 0.08 | 0.08* | 2.41 |
| Prune belly sequence | | 0.27 | 0.16 | 0.38 | 0.47 | 0.40 | 0.17 | 0.31 |
| Trisomy 13 | | 0.41 | 0.58 | 1.09 | 1.14 | 1.93 | 2.35 | 2.37 |
| Trisomy 18 | | 0.89 | 1.02 | 2.30 | 3.14 | 4.64 | 4.53 | 7.15 |
| Down syndrome, all ages (include age unknown) | | 11.38 | 11.11 | 10.96 | 16.70 | 20.51 | 23.41 | 26.41 |
| <20 | | 7.79 | 3.32 | 7.46 | 4.94 | 11.56 | 6.75 | 3.95 |
| 20-24 | | 6.47 | 6.40 | 5.74 | 7.33 | 7.97 | 7.83 | 8.31 |
| 25-29 | | 5.59 | 5.95 | 6.60 | 6.80 | 7.88 | 8.31 | 8.10 |
| 30-34 | | 11.80 | 10.23 | 9.05 | 10.74 | 14.30 | 14.53 | 20.99 |
| 35-39 | | 26.78 | 29.09 | 22.03 | 39.30 | 45.07 | 49.31 | 58.19 |
| 40-44 | | 102.73 | 60.73 | 51.43 | 125.36 | 142.63 | 149.30 | 161.67 |
| 45+ | | 91.46 | 109.89 | 127.93 | 358.31 | 203.76 | 238.41 | 294.74 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

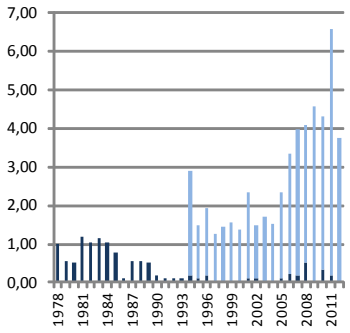
nr = data not reported or not available

* data include less than 5 years

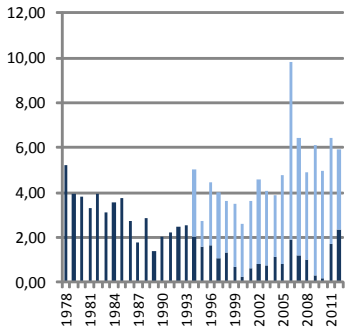
# France: REMERA, Time trends 1978 – 2012

(Birth prevalence rates per 10,000)

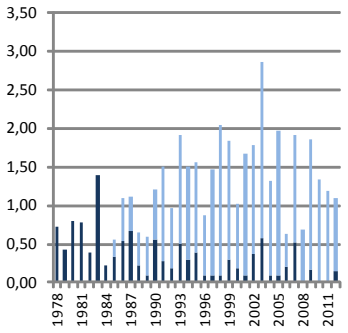
**Anencephaly**



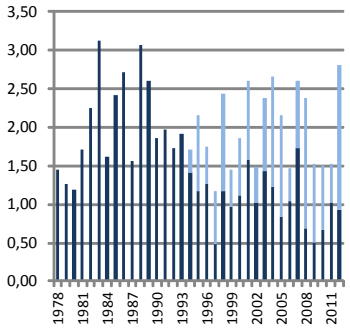

**Spina Bifida**



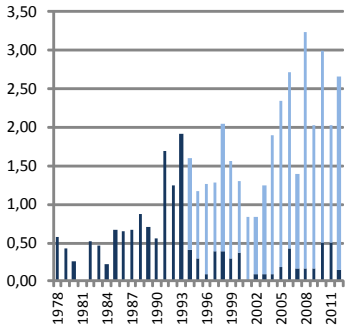

**Encephalocele**



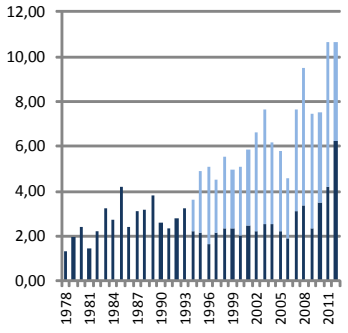

**Microcephaly**



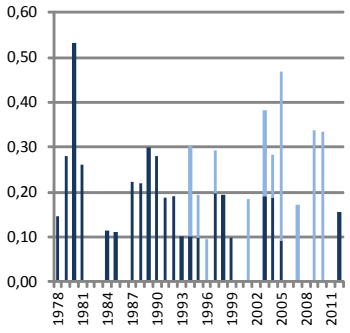

**Holoprosencephaly**



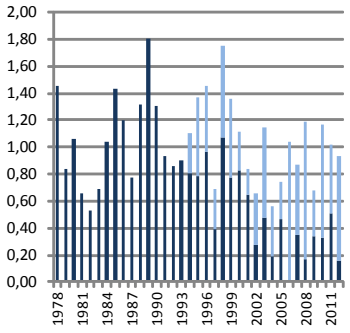

**Hydrocephaly**



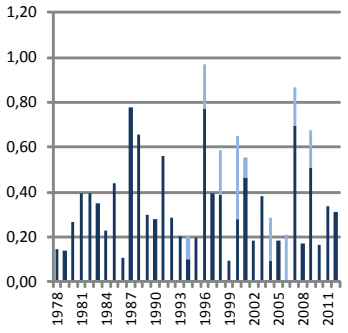

**Anophthalmos**



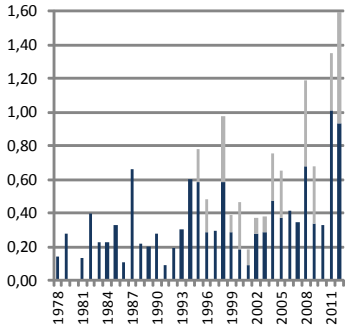

**Microphthalmos**



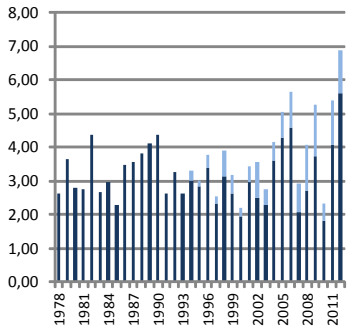

**Anotia**



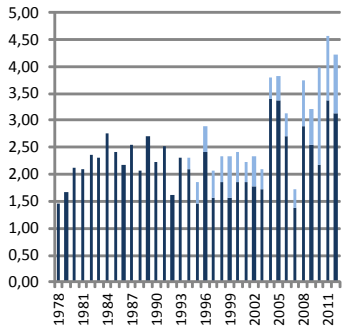

**Microtia**




**Transposition of great vessels**



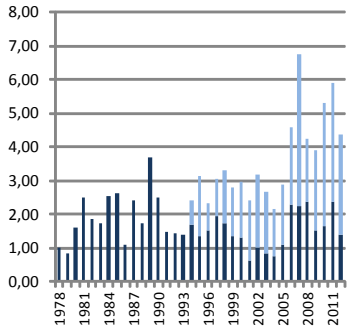

**Tetralogy of Fallot**



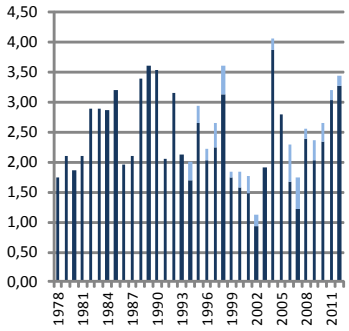
# France: REMERA, Time trends 1978 – 2012

(Birth prevalence rates per 10,000)

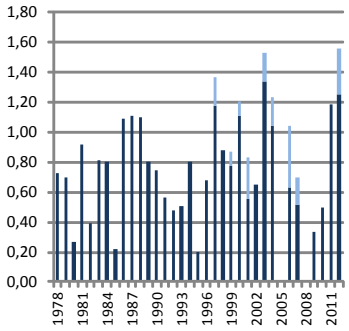
**Hypoplastic left heart syndrome**



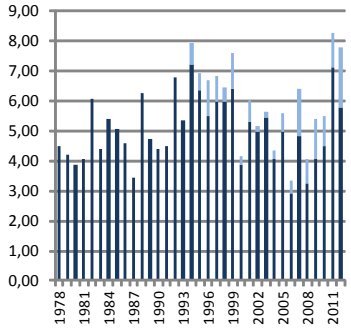

**Coarctation of aorta**



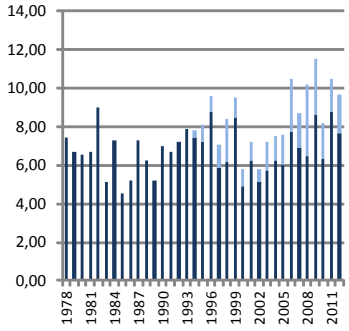

**Choanal atresia, bilateral**



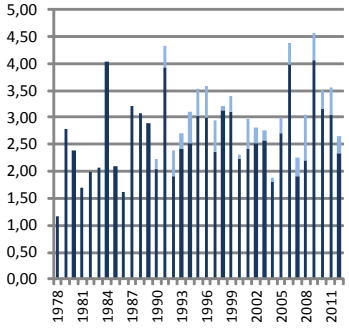

**Cleft palate without cleft lip**



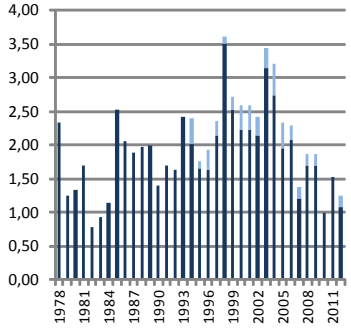

**Cleft lip with or without cleft palate**



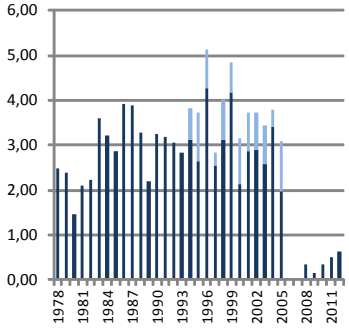

**phageal atresia/stenosis with or without f**



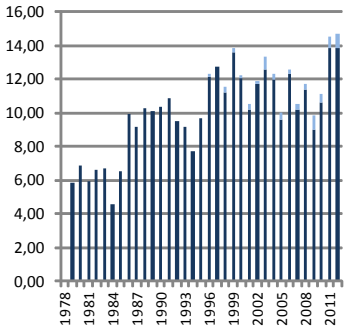

**Small intestine atresia/stenosis**



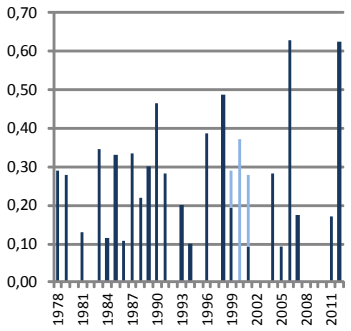

**Anorectal atresia/stenosis**



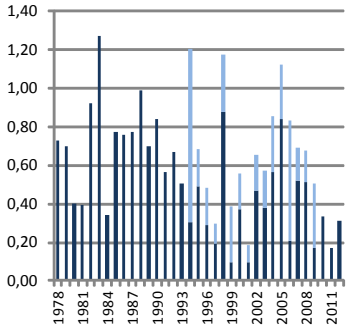

**Hypospadias**



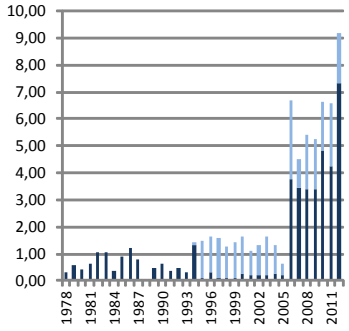

**Epispadias**




**Indeterminate sex**



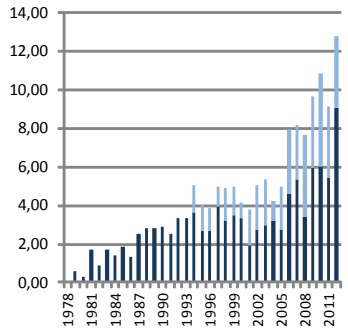

**Renal agenesis**



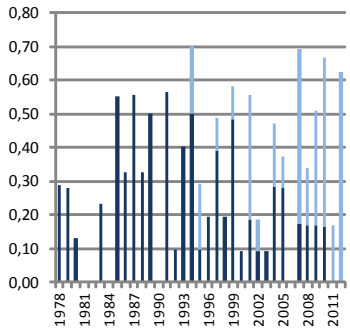
# France: REMERA, Time trends 1978 – 2012

(Birth prevalence rates per 10,000)

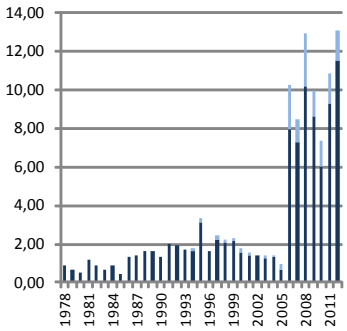
**Cystic kidney**



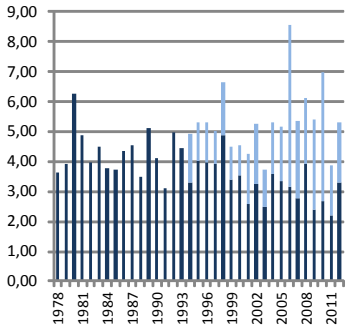

**Bladder, exstrophy**



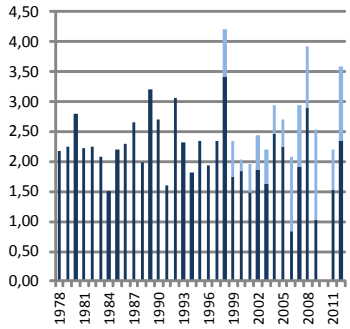

**Polydactyly, preaxial**



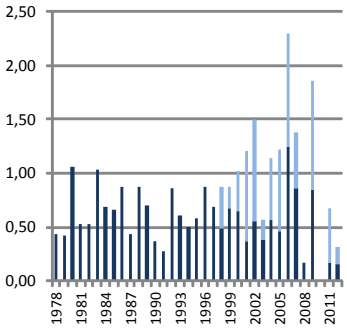

**Limb reduction defects**



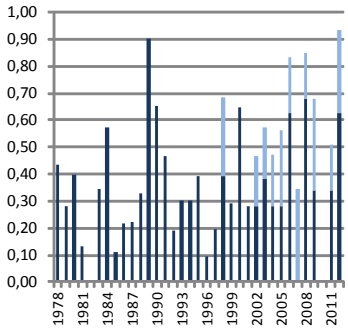

**Limb reduction defects - trasverse**



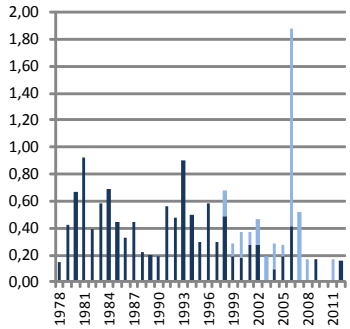

**Limb reduction defects - preaxial**



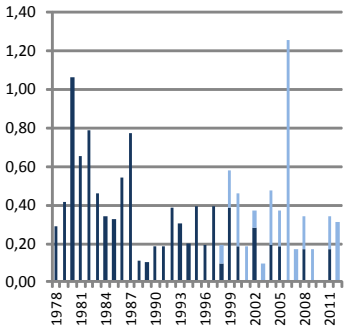

**Limb reduction defects - postaxial**



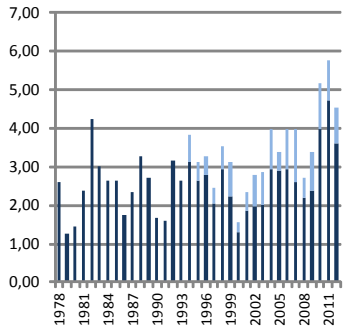

**Limb reduction defects - intercalary**



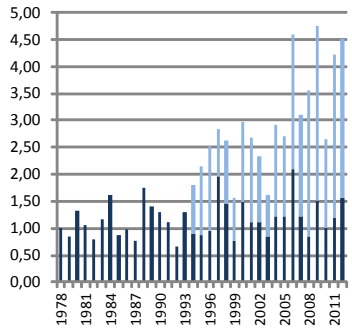

**Limb reduction defects - mixed**



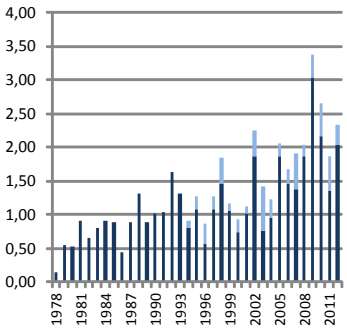

**Diaphragmatic hernia**




**Omphalocele**



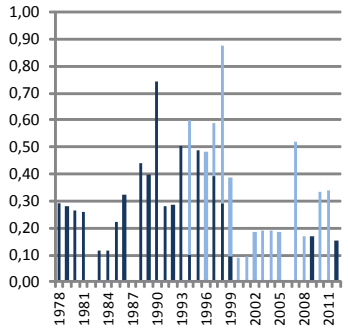

**Gastroschisis**



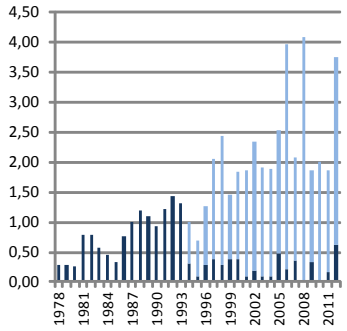
# France: REMERA, Time trends 1978 – 2012

(Birth prevalence rates per 10,000)

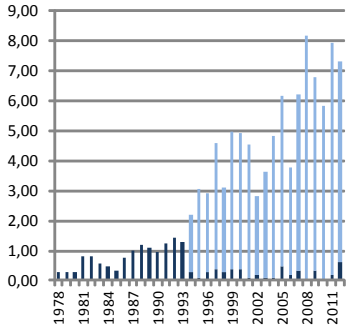
**Prune belly sequence**



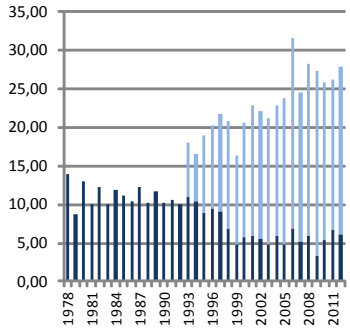

**Trisomy 13**



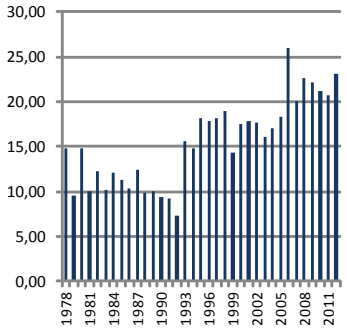

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Germany: Saxony-Anhalt

## Malformation Monitoring Centre Saxony-Anhalt

**History:**
The birth defect registry started in 1980 in the city of Magdeburg with 4000 Births per year. After that, there was a successive enlargement of the registry from 1981-1987. Until 1987 we registered the whole area of the former "District of Magdeburg" (abput 17.000 biths per year). In 1990 there was a dramatic political change, the reunification of Germany. There has been a two-third decrease in the number of births in the registry region. So a similar process of successive territorial enlargement of the surveillance system took place. Since 2000 the system included the whole Federal State of Saxony-Anhalt (up to date 11 districts and 3 major cities). Saxony-Anhalt has currently 2.3 million inhabitants (whole Germany 81.7 million) and a birth rate 17.144 live births in 2009 (2.6% of all live born children in Germany 2009). Additional work: since 2006 the Malformation Monitoring Centre Saxony-Anhalt is collecting and tracking the results of the newborn hearing screening in Saxony-Anhalt. The test is regular performed in the delivery units.

**Legislation and funding:**
1980 to 1989: Ministry of Health of the former GDR 1990 to 1992: Medical Faculty, Otto-von-Guericke University, Magdeburg 1993 to 1995: Ministry of Health, Germany since 1995: Ministry of Labor and Social Affairs of the Federal State of Saxony-Anhalt, Germany. In addition since 2009 a new act concerning the birth defect surveillance and the primary and secondary prevention was adopted by the parliament of Saxony-Anhalt (§ 7).

**Population Coverage:**
The survey system is multi-centric and populationbased, including all mothers resident in Saxony-Anhalt. We exclude non-residents and it is estimated that only a few percent of resident mothers would give birth outside the registry area. Saxony-Anhalt has 2.331 million inhabitants (28.03.2011) and annual births at a rate of 17.300 children (2010).

**Sources of Ascertainment:**
Children and fetuses with congenital anomalies diagnosed before or after birth up to one year of live are eligible for registration at the registry if the mother was resident at time of birth in Saxony-Anhalt. Notification comes from 27 maternity units, 24 paediatric departments, 10 prenatal diagnostic centres, 8 pathology services, and 3 genetic units.

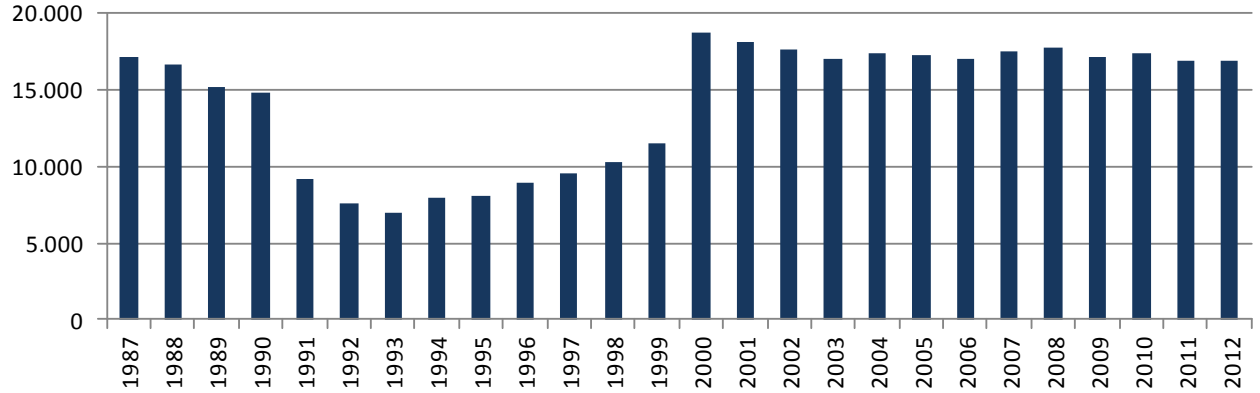
**Exposure information:**
Maternal and paternal occupation (in groups); occupation risk; drugs in pregnancy (ATC-code); alcohol, nicotine, drug abuse.

**Background information:**
Population based registry (region: Federal State Saxony-Anhalt); written informed consent of the parents are necessary. Two healthy "controls" per one malformed child are registered. Termination of pregnancy after prenatal diagnosis is legal and their are registered. Also registered are spontaneous abortions after 16th week of gestation, live and stillborn babies. Definition of stillbirth: >/= 500 grams. The maximum of age of diagnosis is 1 year of live. We do announce an annual report (see www.angeborene-fehlbildungen.com).

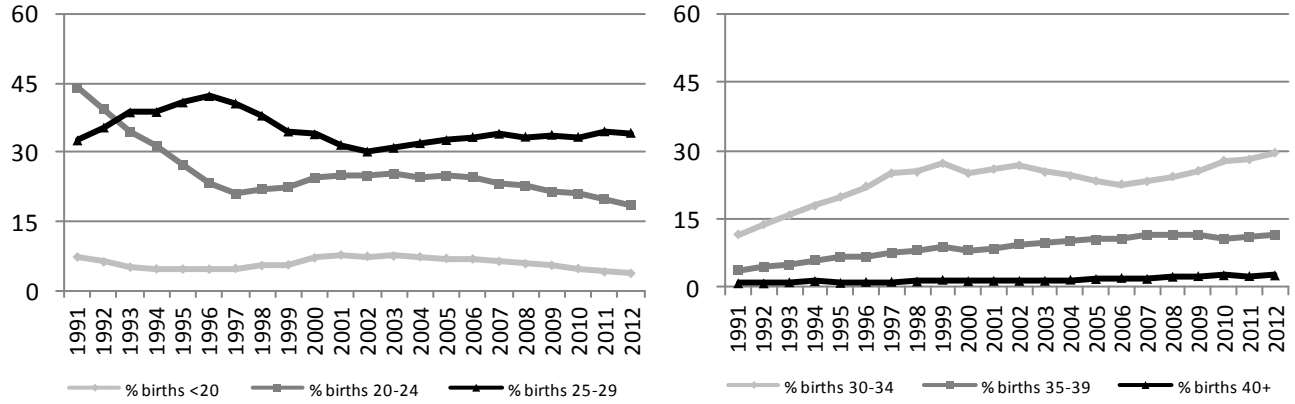
**Addresses and Staff:**
Simone Poetzsch, Program Director, until March 31, 2010 Anke Rißmann, Program Director, from April 1, 2010 Nephrology/ Neonatology Head of Malformation Monitoring Center Saxony-Anhalt Otto-von-Guericke University Leipziger Strasse 44 D-39120 Magdeburg, Germany
**E-mail:** Anke.Rissmann@med.ovgu.de
**Web:**www.angeborene-fehlbildungen.com

# Germany: Saxony-Anhalt

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 3 | 100.0 | Cystic kidney | 7 | 16.3 |
| Spina bifida | 14 | 66.7 | Limb reduction defects | 14 | 29.8 |
| Encephalocele | 5 | 62.5 | Diaphragmatic hernia | 2 | 20.0 |
| Holoprosencephaly | 10 | 76.9 | Omphalocele | 9 | 64.3 |
| Hydrocephaly | 8 | 28.6 | Gastroschisis | 4 | 18.2 |
| Hypoplastic left heart syndrome | 3 | 30.0 | Trisomy 13 | 4 | 80.0 |
| Cleft palate without cleft lip | 2 | 6.3 | Trisomy 18 | 19 | 86.4 |
| Cleft lip with or without cleft palate | 12 | 17.1 | Down syndrome | 52 | 59.1 |
| Renal agenesis | 5 | 62.5 | | | |

Total ToPs with births defects = 256 (Ratio ToPs/Births: 5.00 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Germany: Saxony-Anhalt, 2012

| | |
|---|---|
| Live births (LB) | 16,888 |
| Stillbirths (SB) | 63 |
| Total births | 16,951 |
| Number of terminations of pregnancy (ToP) for birth defects | 76 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 0 | 0.00 |
| Spina bifida | 1 | 0 | 11 | 7.08 |
| Encephalocele | 2 | 0 | 1 | 1.77 |
| Microcephaly | 33 | 2 | 1 | 21.24 |
| Holoprosencephaly | 1 | 0 | 2 | 1.77 |
| Hydrocephaly | 4 | 1 | 3 | 4.72 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 0 | 0 | 0 | 0.00 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.59 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 8 | 0 | 0 | 4.72 |
| Tetralogy of Fallot | 6 | 0 | 0 | 3.54 |
| Hypoplastic left heart syndrome | 4 | 0 | 1 | 2.95 |
| Coarctation of aorta | 8 | 0 | 1 | 5.31 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 15 | 0 | 0 | 8.85 |
| Cleft lip with or without cleft palate | 23 | 0 | 2 | 14.75 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | 0 | 4.72 |
| Small intestine atresia/stenosis | 6 | 0 | 0 | 3.54 |
| Anorectal atresia/stenosis | 4 | 0 | 3 | 4.13 |
| Undescended testis (36 weeks of gestation or later) | 14 | 0 | 0 | 8.26 |
| Hypospadias (*) | 8 | 0 | 0 | 4.72 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 0 | 0 | 2 | 1.18 |
| Renal agenesis | 0 | 0 | 2 | 1.18 |
| Cystic kidney | 6 | 0 | 2 | 4.72 |
| Bladder exstrophy | 1 | 0 | 1 | 1.18 |
| Polydactyly, preaxial | 6 | 0 | 0 | 3.54 |
| Total Limb reduction defects (include unspecified) | 16 | 0 | 8 | 14.16 |
| Transverse | 1 | 0 | 2 | 1.77 |
| Preaxial | 4 | 0 | 1 | 2.95 |
| Postaxial | 4 | 0 | 0 | 2.36 |
| Intercalary | 1 | 0 | 4 | 2.95 |
| Mixed | 5 | 0 | 0 | 2.95 |
| Unspecified | 1 | 0 | 1 | 1.18 |
| Diaphragmatic hernia | 1 | 1 | 1 | 1.77 |
| Omphalocele | 0 | 0 | 3 | 1.77 |
| Gastroschisis | 5 | 0 | 2 | 4.13 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 2 | 1.18 |
| Trisomy 13 | 0 | 1 | 0 | 0.59 |
| Trisomy 18 | 0 | 0 | 6 | 3.54 |
| Down syndrome, all ages (include age unknown) | 12 | 0 | 12 | 14.16 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 1 | 6.43 |
| 25-29 | 4 | 0 | 1 | 8.66 |
| 30-34 | 1 | 0 | 3 | 8.02 |
| 35-39 | 4 | 0 | 1 | 25.60 |
| 40-44 | 2 | 0 | 6 | 188.24 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = data not reported or not available
(*) Only without 1st degree

# Germany: Saxony-Anhalt, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **35,995** | **86,498** | **72,876** | **39,442** | **68,295** | **86,294** | **86,715** |
| Anencephaly | | 1.11 | 2.20 | 3.70 | 3.04 | 1.76 | 2.78 | 1.96 |
| Spina bifida | | 3.06 | 6.82 | 9.88 | 5.58 | 6.30 | 6.03 | 5.19 |
| Encephalocele | | 0.28 | 0.81 | 1.23 | 0.76 | 2.49 | 1.04 | 1.38 |
| Microcephaly | | nr | nr | 2.06 | 4.31 | 7.03 | 14.60 | 11.65 |
| Holoprosencephaly | | nr | nr | 1.51 | 0.51 | 1.02 | 1.74 | 1.61 |
| Hydrocephaly | | nr | nr | 4.94 | 8.62 | 9.52 | 6.37 | 4.61 |
| Anophthalmos | | nr | nr | 0.00 | 1.01 | 0.15 | 0.23 | 0.35 |
| Microphthalmos | | nr | nr | 1.10 | 1.27 | 0.59 | 0.70 | 0.46 |
| Unspecified Anophthalmos/Microphthalmos | | nr | nr | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | | nr | nr | 0.00 | 0.25 | 0.15 | 0.23 | 0.81 |
| Microtia | | nr | nr | 0.14 | 0.00 | 0.73 | 1.04 | 0.46 |
| Unspecified Anotia/Microtia | | nr | nr | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | nr | nr | 2.47 | 3.80 | 5.71 | 5.21 | 3.69 |
| Tetralogy of Fallot | | nr | nr | 0.69 | 2.03 | 2.93 | 3.82 | 2.77 |
| Hypoplastic left heart syndrome | | nr | nr | 3.98 | 3.55 | 4.39 | 2.78 | 2.65 |
| Coarctation of aorta | | nr | nr | 1.65 | 2.03 | 2.78 | 3.94 | 5.42 |
| Choanal atresia, bilateral | | nr | nr | 0.96 | 1.27 | 0.88 | 0.46 | 0.46 |
| Cleft palate without cleft lip | | nr | nr | 5.08 | 5.58 | 10.69 | 9.62 | 5.88 |
| Cleft lip with or without cleft palate | | nr | nr | 13.86 | 13.18 | 18.16 | 14.72 | 12.11 |
| Oesophageal atresia/stenosis with or without fistula | | nr | nr | 2.61 | 2.54 | 2.64 | 2.55 | 2.77 |
| Small intestine atresia/stenosis | | nr | nr | 1.51 | 2.79 | 1.76 | 2.20 | 1.38 |
| Anorectal atresia/stenosis | | nr | nr | 3.84 | 2.54 | 2.64 | 3.71 | 6.80 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | 12.35 | 17.24 | 10.84 | 11.59 | 4.73 |
| Hypospadias | | nr | nr | 13.31 | 21.04 | 13.03 | 8.69 | 6.92 |
| Epispadias | | nr | nr | 0.27 | 0.76 | 0.29 | 0.35 | 0.46 |
| Indeterminate sex | | nr | nr | 0.41 | 0.00 | 1.17 | 0.58 | 0.23 |
| Renal agenesis | | nr | nr | 1.51 | 1.27 | 2.64 | 2.09 | 2.42 |
| Cystic kidney | | nr | nr | 1.92 | 4.56 | 3.66 | 8.11 | 8.65 |
| Bladder exstrophy | | nr | nr | 0.69 | 0.76 | 0.00 | 0.23 | 0.12 |
| Polydactyly, preaxial | | nr | nr | 0.41 | 3.04 | 4.83 | 2.67 | 5.54 |
| Total Limb reduction defects (include unspecified) | | nr | nr | 5.63 | 5.83 | 8.93 | 6.49 | 7.73 |
| Transverse | | nr | nr | nr | nr | 3.52* | 2.43 | 1.73 |
| Preaxial | | nr | nr | nr | nr | 0.27* | 0.46 | 1.27 |
| Postaxial | | nr | nr | nr | nr | 0.00* | 0.00 | 0.58 |
| Intercalary | | nr | nr | nr | nr | 2.71* | 0.93 | 0.23 |
| Mixed | | nr | nr | nr | nr | 1.62* | 1.85 | 2.77 |
| Unspecified | | nr | nr | nr | nr | 0.00* | 0.81 | 1.27 |
| Diaphragmatic hernia | | nr | nr | 1.92 | 0.00 | 1.90 | 2.90 | 2.88 |
| Omphalocele | | nr | nr | 4.80 | 1.77 | 2.93 | 3.94 | 2.65 |
| Gastroschisis | | nr | nr | 1.51 | 2.79 | 3.07 | 4.87 | 3.46 |
| Unspecified Omphalocele/Gastroschisis | | nr | nr | nr | nr | 0.27* | 0.00 | 1.15 |
| Prune belly sequence | | nr | nr | 0.27 | 0.76 | 1.02 | 0.81 | 0.92 |
| Trisomy 13 | | 0.00 | 0.35 | 0.55 | 0.76 | 1.90 | 1.04 | 1.04 |
| Trisomy 18 | | 0.28 | 1.16 | 0.96 | 1.27 | 1.90 | 3.71 | 4.38 |
| Down syndrome, all ages (include age unknown) | | 7.22 | 9.13 | 9.06 | 12.17 | 17.42 | 15.64 | 17.18 |
| <20 | | nr | nr | nr | nr | 3.71* | 4.88 | 0.00 |
| 20-24 | | nr | nr | nr | nr | 6.60* | 7.50 | 6.45 |
| 25-29 | | nr | nr | nr | nr | 11.62* | 7.32 | 7.88 |
| 30-34 | | nr | nr | nr | nr | 11.73* | 13.76 | 17.50 |
| 35-39 | | nr | nr | nr | nr | 71.73* | 43.95 | 45.09 |
| 40-44 | | nr | nr | nr | nr | 119.28* | 178.19 | 137.20 |
| 45+ | | nr | nr | nr | nr | 526.32* | 344.83 | 487.80 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

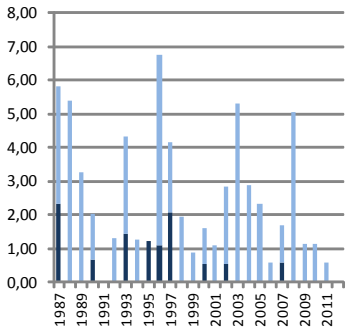
nr = data not reported or not available

* data include less than 5 years

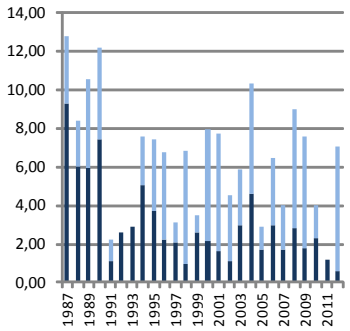
# Germany: Saxony-Anhalt, Time trends 1987 – 2012

(Birth prevalence rates per 10,000)

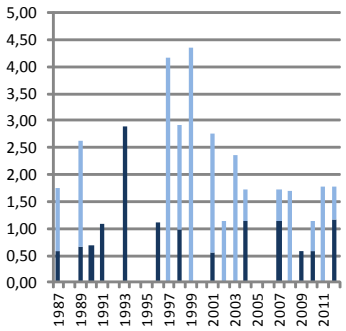
**Anencephaly**



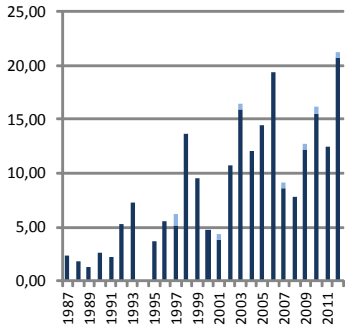

**Spina Bifida**



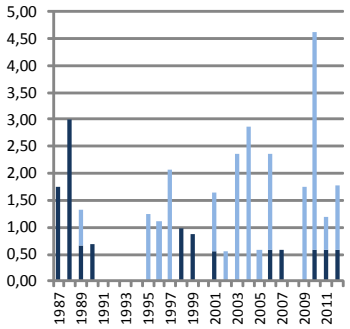

**Encephalocele**



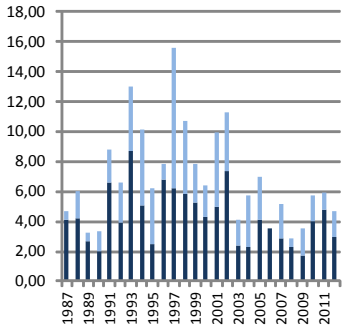

**Microcephaly**



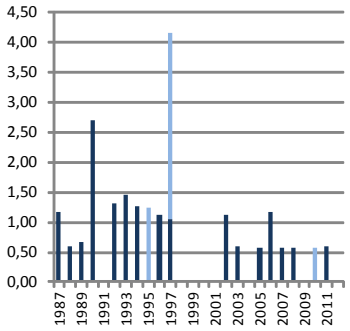

**Holoprosencephaly**



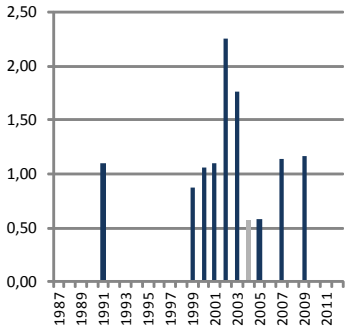

**Hydrocephaly**



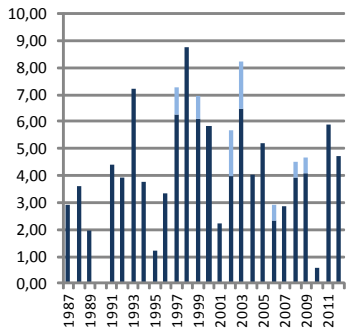

**Microphthalmos**



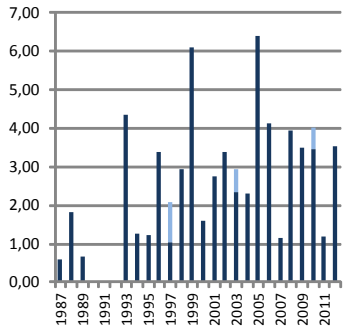

**Microtia**



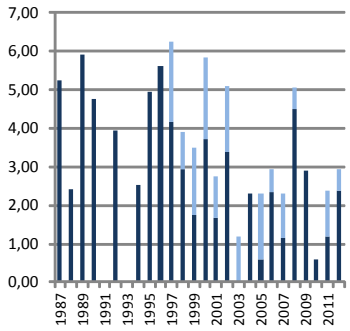

**Transposition of great vessels**



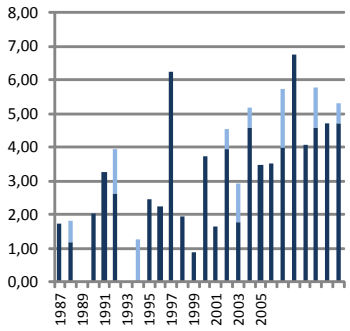

**Tetralogy of Fallot**




**Hypoplastic left heart syndrome**



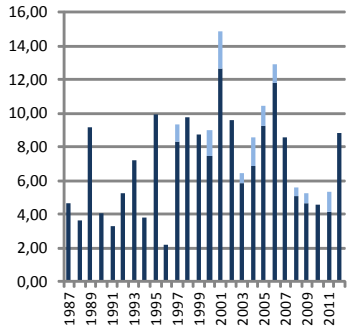

**Coarctation of aorta**



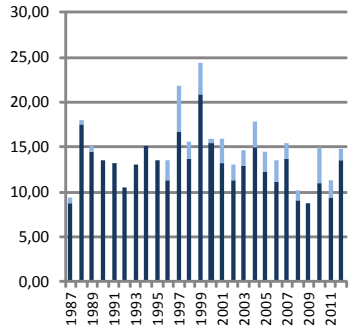
# Germany: Saxony-Anhalt, Time trends 1987 – 2012

(Birth prevalence rates per 10,000)

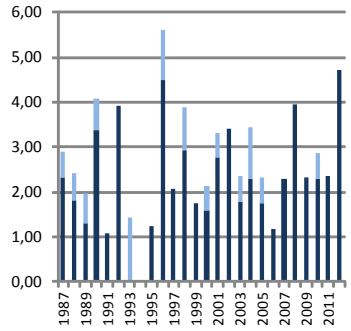
**Cleft palate without cleft lip**



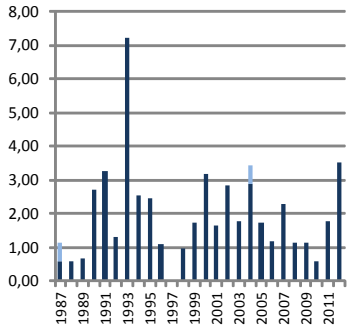

**Cleft lip with or without cleft palate**



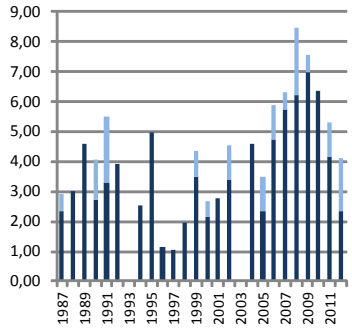

**phageal atresia/stenosis with or without f**



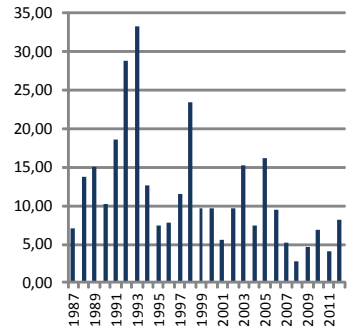

**Small intestine atresia/stenosis**



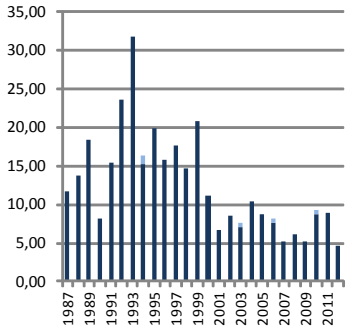

**Anorectal atresia/stenosis**



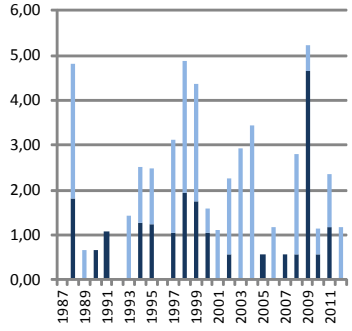

**Undescended testis**



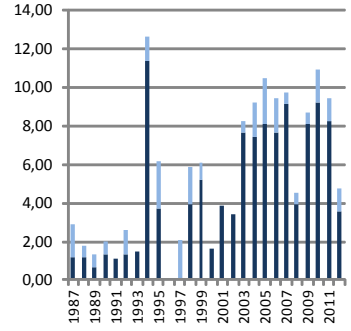

**Hypospadias**



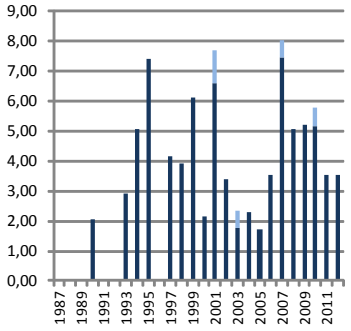

**Renal agenesis**



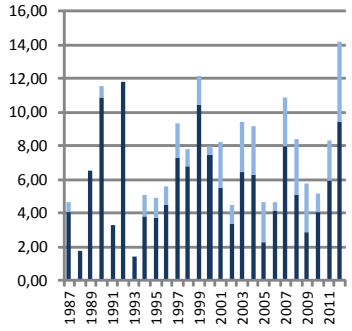

**Cystic kidney**



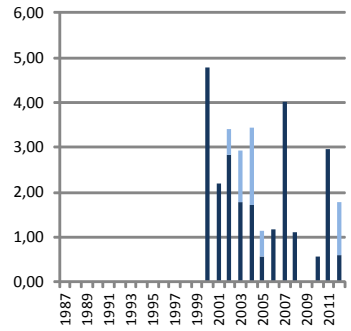

**Polydactyly, preaxial**




**Limb reduction defects**



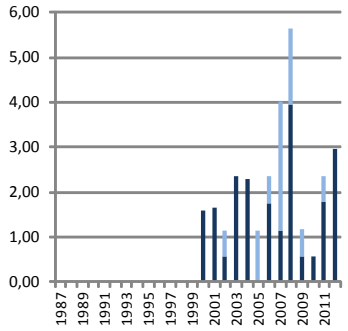

**Limb reduction defects - transverse**



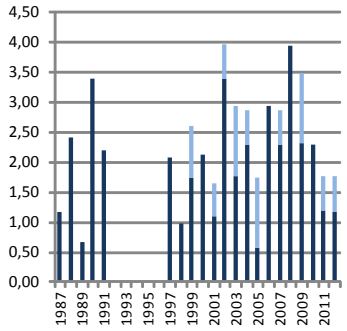
# Germany: Saxony-Anhalt, Time trends 1987 – 2012

(Birth prevalence rates per 10,000)

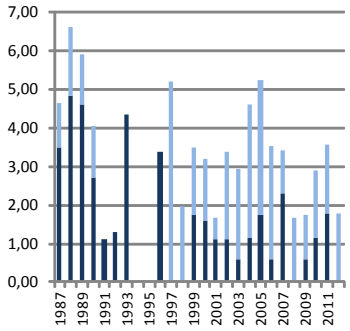
**Limb reduction defects - mixed**



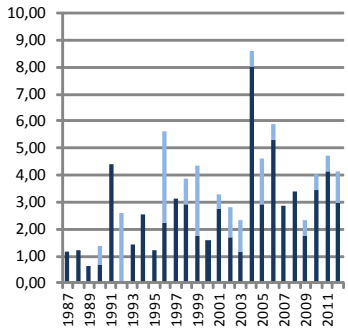

**Diaphragmatic hernia**



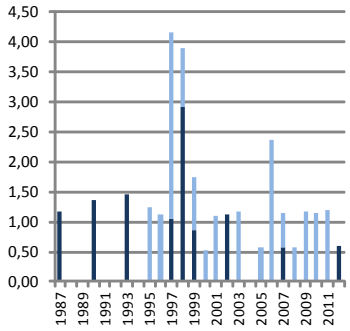

**Omphalocele**



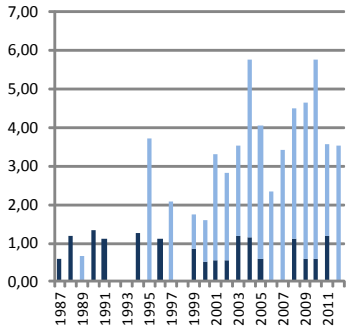

**Gastroschisis**



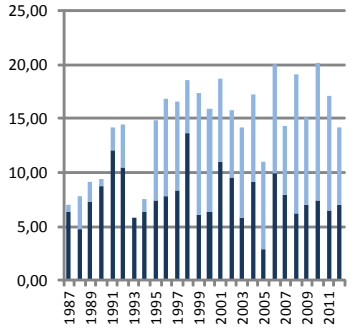

**Trisomy 13**



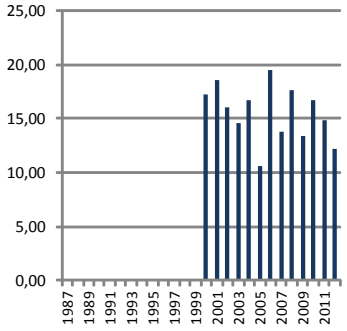

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Hungary

## Hungarian Congenital Abnormality Registry

**History**:
Centralized registration of congenital abnormalities began in Hungary in 1962, and came under our co-ordination in 1970. Monitoring began in 1973. The Programme was a founding member of the International Clearinghouse.

**Size and coverage:**
The registry covers all births in Hungary, approximately 100,000 annually. Criteria to define stillbirth was changed in 1998. At present, stillbirths of at least 24 weeks gestation or 500 grams are registered. Prenatally diagnosed and terminated fetuses are also registered.

**Legislation and funding**:
Reporting is compulsory. The registry is currently run and financed by the National Center for Healthcare Audit and Improvement; formerly by the National Center for Epidemiology, and the National Institute of Public Health.

**Sources of ascertainment:**
Reports are obtained from multiple sources, such as delivery units, neonatal and pediatric surgery, pathology, and prenatal diagnostic centers. Abnormalities detected before the age of one are reported Variations in figures (especially in the1990s) may reflect incomplete notification.

**Exposure information**:
Exposure information has been available since 1980, when a case-control system was initiated. Mothers of selected malformed infants and controls are interviewed by community nurses to collect information.

**Background information**:
General background information on all births is available from central statistics. The online notification (instead of paper-based) has started since 15th of October 2009.

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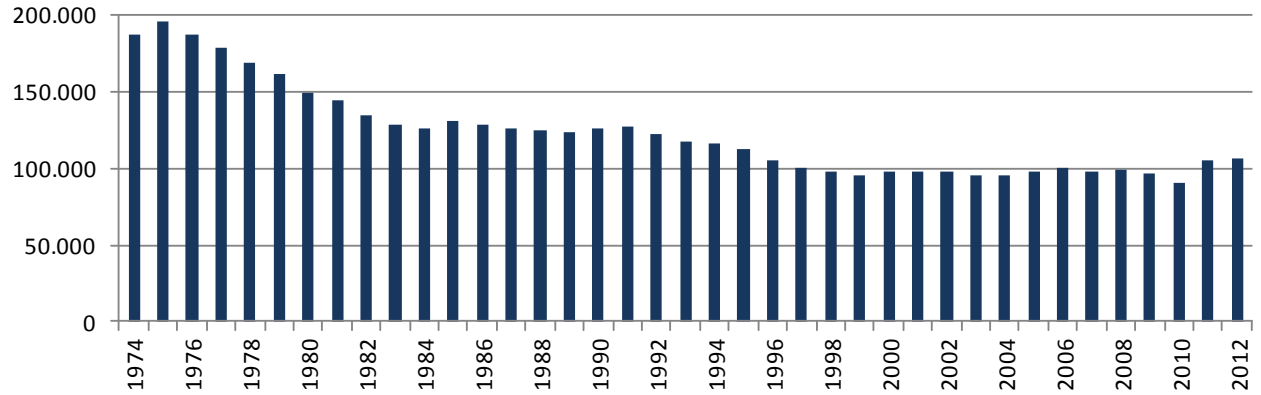
Inactive Staff (Maternity leave)

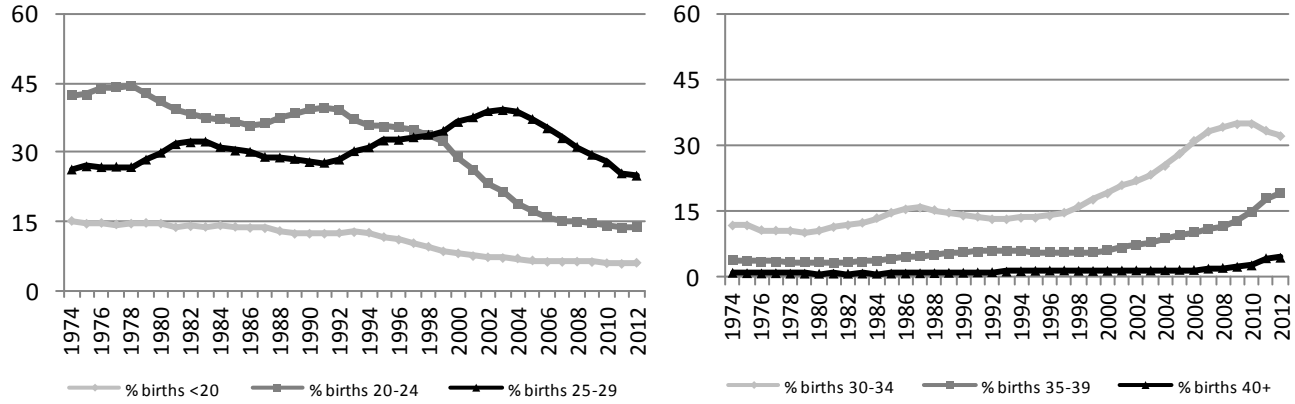
Erzsebet Horvath-Puho, PhD
Melinda Csaky-Szunyogh, MSc

# Hungary

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 16 | 48.5 | Cystic kidney | 11 | 13.9 |
| Spina bifida | 40 | 42.1 | Limb reduction defects | 14 | 15.9 |
| Encephalocele | 14 | 51.9 | Diaphragmatic hernia | 12 | 16.7 |
| Holoprosencephaly | 3 | 37.5 | Omphalocele | 17 | 38.6 |
| Hydrocephaly | 36 | 25.5 | Gastroschisis | 13 | 48.1 |
| Hypoplastic left heart syndrome | 15 | 26.8 | Trisomy 13 | 24 | 64.9 |
| Cleft palate without cleft lip | 17 | 11.2 | Trisomy 18 | 82 | 84.5 |
| Cleft lip with or without cleft palate | 22 | 11.0 | Down syndrome | 360 | 60.3 |
| Renal agenesis | 5 | 8.8 | | | |

Total ToPs with births defects = 440 (Ratio ToPs/Births: 1.45 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Hungary, 2012

| | |
|---|---|
| Live births (LB) | 90,269 |
| Stillbirths (SB) | 16,450 |
| Total births | 106,719 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 15 | 0 | 1 | 1.50 |
| Spina bifida | 33 | 0 | 4 | 3.47 |
| Encephalocele | 10 | 0 | 0 | 0.94 |
| Microcephaly | 24 | 0 | 1 | 2.34 |
| Holoprosencephaly | 4 | 0 | 1 | 0.47 |
| Hydrocephaly | 62 | 0 | 5 | 6.28 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 4 | 0 | 0 | 0.37 |
| Unspecified Anophthalmos/Microphthalmos | 1 | 0 | 1 | 0.19 |
| Anotia | 4 | 0 | 0 | 0.37 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 23 | 0 | 1 | 2.25 |
| Tetralogy of Fallot | 45 | 0 | 0 | 4.22 |
| Hypoplastic left heart syndrome | 15 | 0 | 4 | 1.78 |
| Coarctation of aorta | 29 | 0 | 0 | 2.72 |
| Choanal atresia, bilateral | 18 | 0 | 1 | 1.78 |
| Cleft palate without cleft lip | 42 | 0 | 6 | 4.50 |
| Cleft lip with or without cleft palate | 69 | 0 | 8 | 7.22 |
| Oesophageal atresia/stenosis with or without fistula | 18 | 0 | 0 | 1.69 |
| Small intestine atresia/stenosis | 27 | 0 | 3 | 2.81 |
| Anorectal atresia/stenosis | 30 | 0 | 3 | 3.09 |
| Undescended testis (36 weeks of gestation or later) | 154 | 0 | 0 | 14.43 |
| Hypospadias | 246 | 0 | 31 | 25.96 |
| Epispadias | 51 | 0 | 4 | 5.15 |
| Indeterminate sex | 4 | 0 | 0 | 0.37 |
| Renal agenesis | 24 | 0 | 1 | 2.34 |
| Cystic kidney | 16 | 0 | 5 | 1.97 |
| Bladder exstrophy | 2 | 0 | 0 | 0.19 |
| Polydactyly, preaxial | 16 | 0 | 0 | 1.50 |
| Total Limb reduction defects (include unspecified) | 32 | 0 | 4 | 3.37 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 27 | 0 | 1 | 2.62 |
| Omphalocele | 16 | 0 | 3 | 1.78 |
| Gastroschisis | 8 | 0 | 0 | 0.75 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 7 | 0 | 8 | 1.41 |
| Trisomy 18 | 8 | 0 | 25 | 3.09 |
| Down syndrome, all ages (include age unknown) | 97 | 1 | 109 | 19.40 |
| <20 | 3 | 0 | 2 | 7.86 |
| 20-24 | 8 | 0 | 8 | 10.97 |
| 25-29 | 26 | 1 | 16 | 16.20 |
| 30-34 | 37 | 0 | 27 | 18.69 |
| 35-39 | 18 | 0 | 36 | 26.56 |
| 40-44 | 5 | 0 | 20 | 57.20 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = data not reported or not available

# Hungary, Previous years rates 1974 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | **570,720** | **804,244** | **648,937** | **629,899** | **573,813** | **489,535** | **486,479** | **490,630** |
| Anencephaly | 8.45 | 6.06 | 4.41 | 1.37 | 0.47 | 1.84 | 1.77 | 1.57 |
| Spina bifida | 12.86 | 8.42 | 7.87 | 4.52 | 1.59 | 3.29 | 3.37 | 3.93 |
| Encephalocele | nr | 1.54* | 1.62 | 1.05 | 0.37 | 0.63 | 0.56 | 0.84 |
| Microcephaly | nr | 1.67* | 1.17 | 0.71 | 0.64 | 0.57 | 1.05 | 1.81 |
| Holoprosencephaly | nr | 0.14* | 0.29 | 0.16 | 0.07 | 0.80 | 0.72 | 0.67 |
| Hydrocephaly | 8.13 | 5.46 | 3.76 | 2.87 | 1.13 | 2.68 | 4.52 | 5.28 |
| Anophthalmos | 0.14 | 0.09 | 0.08 | 0.05 | 0.05 | 0.04 | 0.04 | 0.20 |
| Microphthalmos | 0.09 | 0.25 | 0.18 | 0.05 | 0.09 | 0.10 | 0.21 | 0.86 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr | nr | nr | nr | 0.09* |
| Anotia | 0.11 | 0.09 | 0.18 | 0.21 | 0.19 | 0.61 | 0.60 | 0.49 |
| Microtia | 0.04 | 0.05 | 0.02 | 0.02 | 0.03 | 0.04 | 0.12 | 0.10 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr | nr | nr | nr | 0.00* |
| Transposition of great vessels | nr | 0.98* | 1.66 | 1.79 | 1.12 | 1.33 | 1.83 | 3.22 |
| Tetralogy of Fallot | 1.05 | 1.52 | 1.11 | 0.98 | 0.94 | 1.74 | 2.38 | 3.10 |
| Hypoplastic left heart syndrome | Nr | 0.38* | 0.46 | 0.92 | 0.44 | 0.92 | 1.48 | 2.45 |
| Coarctation of aorta | 0.93 | 1.43 | 2.48 | 2.25 | 1.74 | 1.55 | 2.10 | 3.89 |
| Choanal atresia, bilateral | nr | 0.20* | 0.09 | 0.21 | 0.09 | 0.02 | 0.14 | 1.12 |
| Cleft palate without cleft lip | 3.77 | 4.28 | 4.30 | 3.59 | 2.95 | 3.02 | 3.35 | 5.10 |
| Cleft lip with or without cleft palate | 10.74 | 11.84 | 10.14 | 9.21 | 7.89 | 6.54 | 7.28 | 7.28 |
| Oesophageal atresia/stenosis with or without fistula | 2.41* | 1.89 | 1.66 | 1.79 | 1.03 | 0.88 | 1.38 | 2.45 |
| Small intestine atresia/stenosis | nr | 1.50* | 1.42 | 1.16 | 0.82 | 0.53 | 1.13 | 2.41 |
| Anorectal atresia/stenosis | nr | 2.37* | 2.08 | 1.76 | 1.20 | 0.84 | 1.42 | 2.71 |
| Undescended testis (36 weeks of gestation or later) | nr | 14.97* | 18.12 | 16.27 | 13.80 | 10.07 | 16.34 | 19.77 |
| Hypospadias | 15.68 | 16.95 | 22.04 | 21.00 | 20.32 | 20.02 | 25.16 | 26.90 |
| Epispadias | nr | nr | nr | nr | nr | nr | nr | 1.27* |
| Indeterminate sex | nr | 0.27* | 0.31 | 0.37 | 0.10 | 0.14 | 0.51 | 0.37 |
| Renal agenesis | nr | 1.36* | 0.76 | 1.27 | 0.38 | 0.20 | 0.56 | 1.67 |
| Cystic kidney | nr | 0.00* | 0.06 | 0.35 | 0.40 | 1.92 | 3.47 | 4.28 |
| Bladder exstrophy | nr | 0.14* | 0.51 | 0.27 | 0.03 | 0.06 | 0.12 | 0.39 |
| Polydactyly, preaxial | nr | 0.00* | 1.74 | 1.86 | 1.19 | 7.86 | 8.18 | 7.79 |
| Total Limb reduction defects (include unspecified) | nr | nr | 4.44 | 3.62 | 2.75 | 3.06 | 3.27 | 3.28 |
| Transverse | nr | nr | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr | nr | nr | nr | nr |
| Diaphragmatic hernia | 2.75 | 1.55 | 2.30 | 2.16 | 0.94 | 0.84 | 1.15 | 2.43 |
| Omphalocele | nr | nr | 1.99 | 0.95 | 0.64 | 0.94 | 1.21 | 1.57 |
| Gastroschisis | nr | nr | 0.54 | 0.56 | 0.44 | 0.84 | 0.86 | 1.08 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr | nr | nr | nr | 0.00* |
| Prune belly sequence | nr | nr | nr | nr | 0.00* | 0.10 | 0.04 | 0.04 |
| Trisomy 13 | nr | nr | 0.26 | 0.16 | 0.16 | 0.37 | 0.82 | 1.26 |
| Trisomy 18 | nr | nr | 0.25 | 0.33 | 0.19 | 1.04 | 1.85 | 3.20 |
| Down syndrome, all ages (include age unknown) | 8.92 | 8.95 | 7.77 | 8.54 | 7.62 | 10.05 | 14.68 | 19.06 |
| <20 | nr | nr | 1.56 | 2.52 | 1.30 | 3.05 | 8.26 | 9.51 |
| 20-24 | nr | nr | 2.04 | 3.03 | 1.95 | 4.32 | 8.22 | 5.52 |
| 25-29 | nr | nr | 3.46 | 4.25 | 2.37 | 6.22 | 7.34 | 8.77 |
| 30-34 | nr | nr | 4.80 | 5.64 | 4.17 | 9.28 | 13.46 | 13.49 |
| 35-39 | nr | nr | 11.04 | 18.96 | 17.57 | 26.56 | 43.02 | 49.46 |
| 40-44 | nr | nr | 56.53 | 64.97 | 70.18 | 145.25 | 156.14 | 120.70 |
| 45+ | nr | nr | nr | nr | nr | nr | nr | 237.58* |
| unknown | --- | --- | --- | --- | --- | --- | --- | --- |

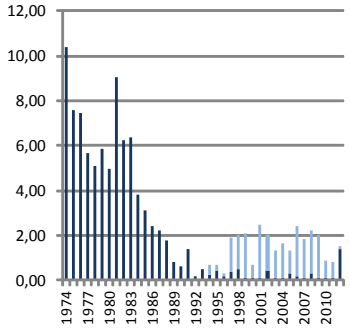
nr = data not reported or not available

* data include less than 5 years

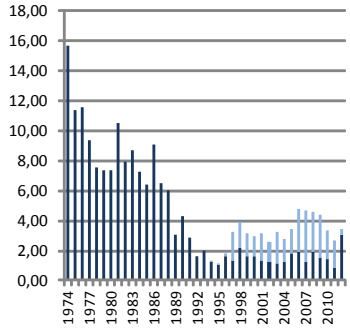
# Hungary, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

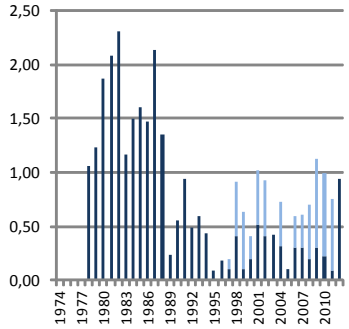
**Anencephaly**



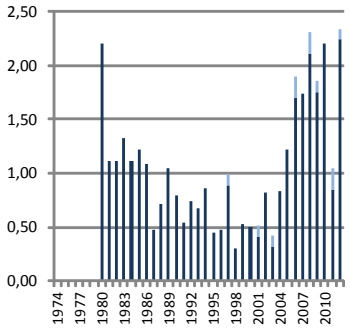

**Spina Bifida**



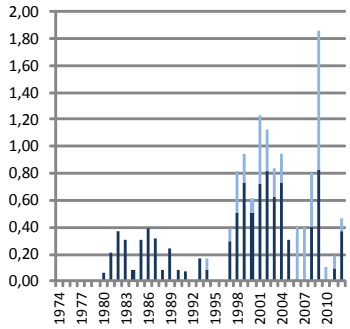

**Encephalocele**



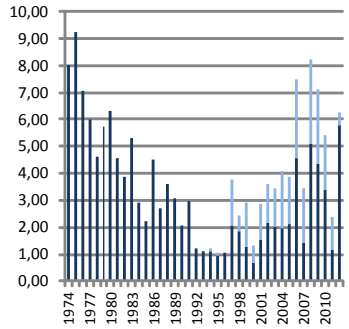

**Microcephaly**



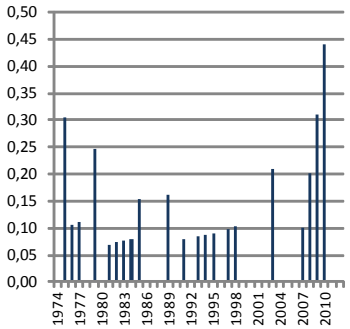

**Holoprosencephaly**



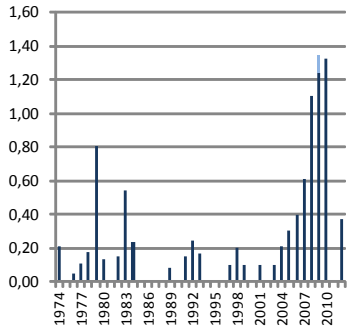

**Hydrocephaly**



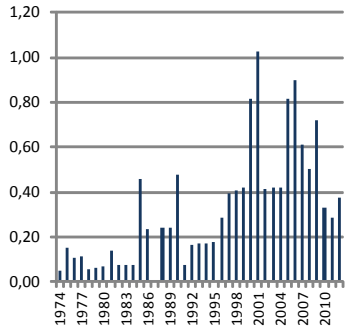

**Anophthalmos**



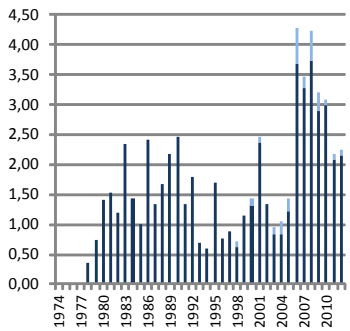

**Microphthalmos**



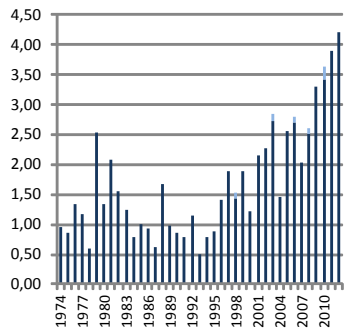

**Anotia**



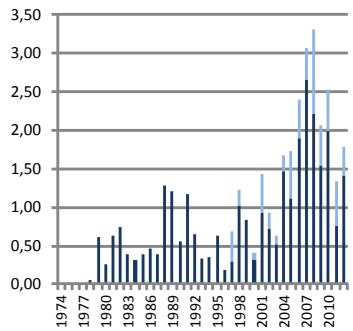

**Transposition of great vessels**




**Tetralogy of Fallot**



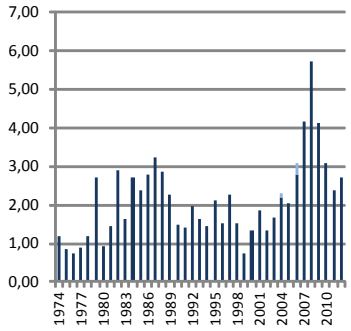

**Hypoplastic left heart syndrome**



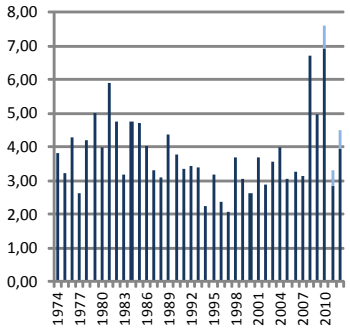
# Hungary, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

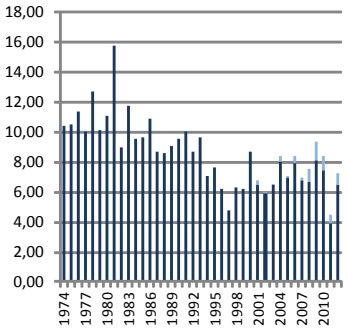
**Coarctation of aorta**



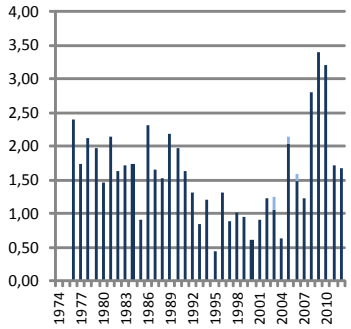

**Cleft palate without cleft lip**



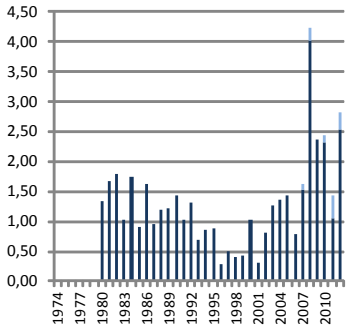

**Cleft lip with or without cleft palate**



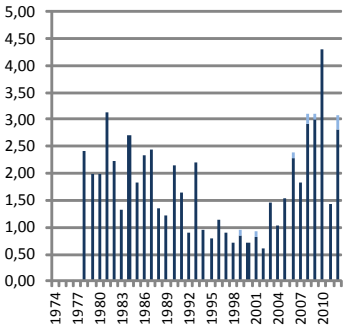

**phageal atresia/stenosis with or without f**



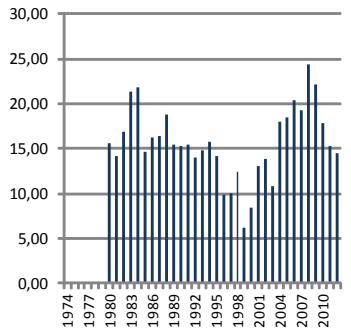

**Small intestine atresia/stenosis**



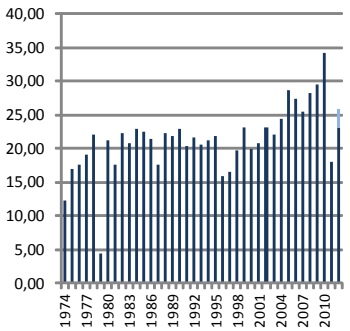

**Anorectal atresia/stenosis**



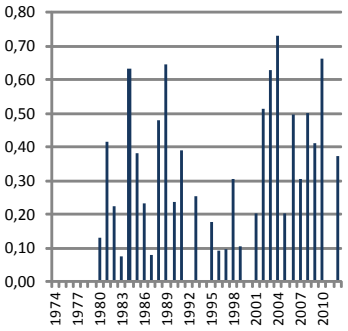

**Undescended testis**



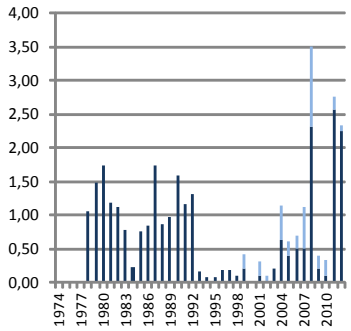

**Hypospadias**



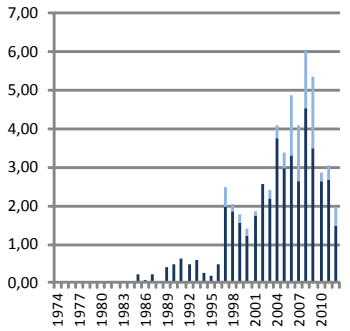

**Indeterminate sex**



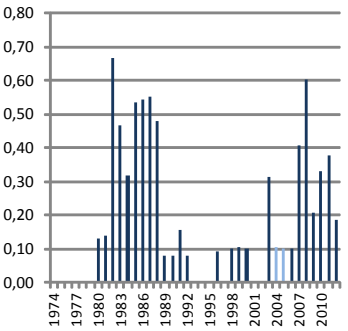

**Renal agenesis**




**Cystic kidney**



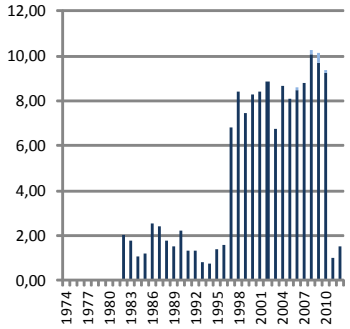

**Bladder, exstrophy**



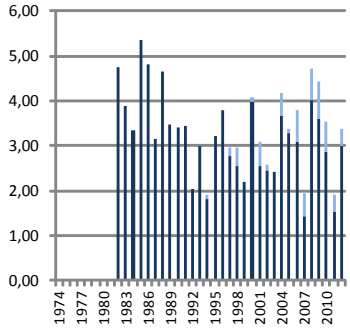
# Hungary, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

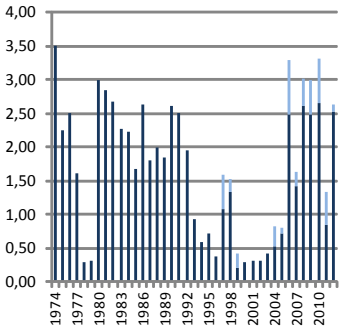
**Polydactyly, preaxial**



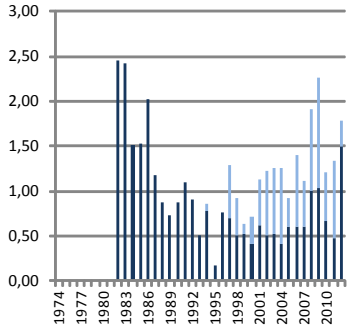

**Limb reduction defects**



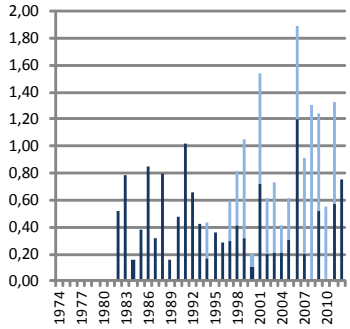

**Diaphragmatic hernia**



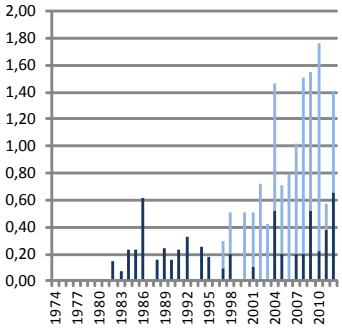

**Omphalocele**



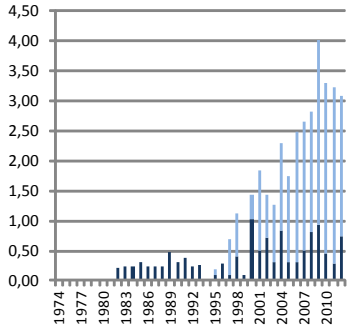

**Gastroschisis**



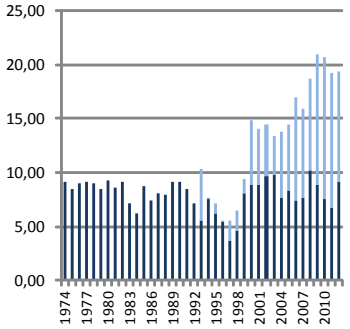

**Trisomy 13**



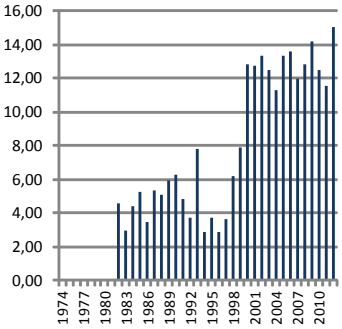

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Iran: TRoCA

## Tabriz Registry of Congenital Anomalies

**History:**
The programme was initiated in 2000, but the registry started in 2003. It was then accepted as a member of the ICBDSR in the 2006 annual meeting in Uppsala, Sweden. Since 2012, the registry is also a "World Affiliate" member of the European network of registries for the epidemiologic surveillance of congenital anomalies (EUROCAT).

**Size and coverage:**
TROCA is a hospital-based registry and situated in the northwest of Iran covering all births and children in three university hospitals in the city of Tabriz. This city is one of the three major cities in the country. The programme is based on approximately 60-70% of all births (20000 births per year) in the area.

**Legislation and funding:**
The programme has been financially supported by the National Public Health Management Centre (a WHO collaborating centre) in Tabriz University of Medical Sciences.

**Exposure information:**
Some exposure information is currently available of mothers of all malformed infants. Other women giving births in all university hospitals with normal newborns routinely complete a similar form. They might be considered as matched control group.

**Background information:**
General epidemiological data and basic characteristic information are available for all births.

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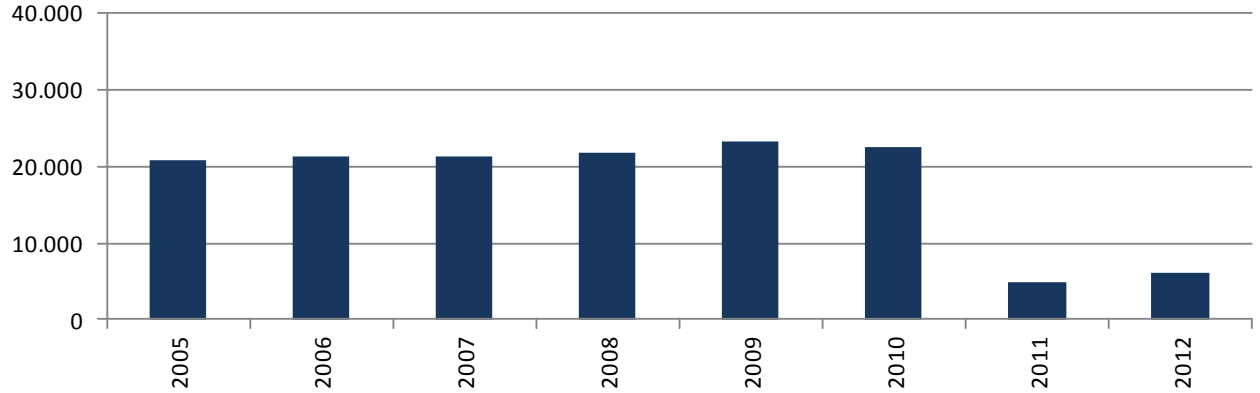
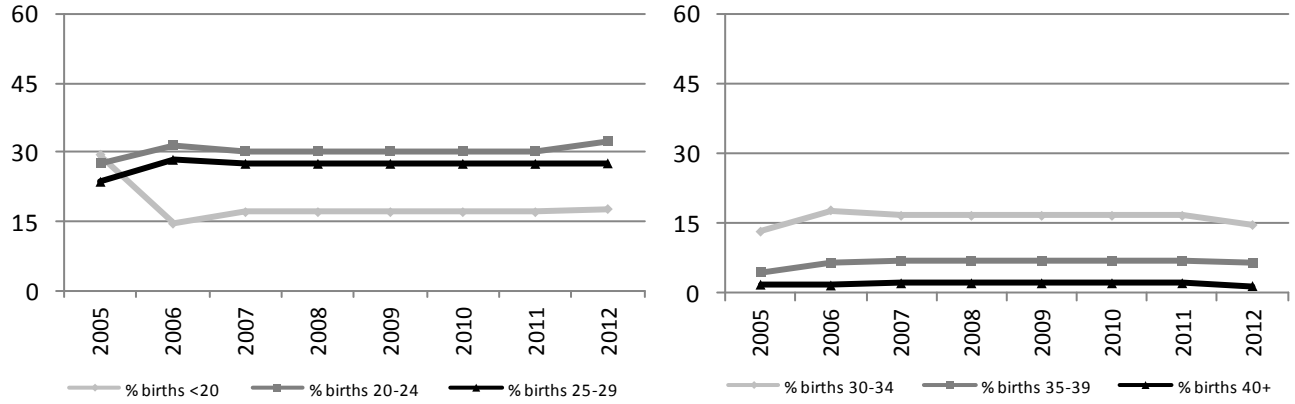
Tabriz, Iran
**Phone:** 98-914 415 7039
**Fax:** 98-411 336 4668
**E-mail:** saeed.dastgiri@gmail.com
**Website:** http://troca.tbzmed.ac.ir

# Iran: TRoCA

## Total births by year




## Percentage of births by year and maternal age



# Iran: TRoCA, 2012(*)

| | |
|---|---|
| Live births (LB) | 6,093 |
| Stillbirths (SB) | 59 |
| Total births | 6,152 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | nr | 2 | nr | 3.25 |
| Spina bifida | 2 | nr | nr | 3.25 |
| Encephalocele | nr | nr | nr | nr |
| Microcephaly | 3 | nr | nr | 4.88 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | 6 | 1 | nr | 11.38 |
| Anophthalmos | 1 | 1 | nr | 3.25 |
| Microphthalmos | 4 | nr | nr | 6.50 |
| Unspecified Anophthalmos/Microphthalmos | 10 | 1 | nr | 17.88 |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | 14 | 2 | nr | 26.01 |
| Transposition of great vessels | 3 | nr | nr | 4.88 |
| Tetralogy of Fallot | nr | nr | nr | nr |
| Hypoplastic left heart syndrome | 4 | nr | nr | 6.50 |
| Coarctation of aorta | nr | nr | nr | nr |
| Choanal atresia, bilateral | 11 | 1 | nr | 19.51 |
| Cleft palate without cleft lip | 7 | nr | nr | 11.38 |
| Cleft lip with or without cleft palate | 16 | nr | nr | 26.01 |
| Oesophageal atresia/stenosis with or without fistula | 2 | nr | nr | 3.25 |
| Small intestine atresia/stenosis | 5 | nr | nr | 8.13 |
| Anorectal atresia/stenosis | 8 | nr | nr | 13.00 |
| Undescended testis (36 weeks of gestation or later) | 31 | nr | nr | 50.39 |
| Hypospadias | 21 | nr | nr | 34.14 |
| Epispadias | 9 | nr | nr | 14.63 |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 2 | 1 | nr | 4.88 |
| Cystic kidney | 3 | nr | nr | 4.88 |
| Bladder exstrophy | nr | nr | nr | nr |
| Polydactyly, preaxial | 17 | nr | nr | 27.63 |
| Total Limb reduction defects (include unspecified) | 92 | 2 | nr | 152.80 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 4 | nr | nr | 6.50 |
| Omphalocele | 3 | 1 | nr | 6.50 |
| Gastroschisis | 1 | nr | nr | 1.63 |
| Unspecified Omphalocele/Gastroschisis | 3 | nr | nr | 4.88 |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 1 | nr | nr | 1.63 |
| Trisomy 18 | nr | nr | nr | nr |
| Down syndrome, all ages (include age unknown) | 13 | nr | nr | 21.13 |
| <20 | nr | nr | nr | nr |
| 20-24 | 6 | nr | nr | 30.05 |
| 25-29 | 2 | nr | nr | 11.81 |
| 30-34 | 5 | nr | nr | 55.87 |
| 35-39 | nr | nr | nr | nr |
| 40-44 | nr | nr | nr | nr |
| 45+ | nr | nr | nr | nr |
| unknown | nr | nr | nr | nr |

nr = data not reported or not available

# Iran: TRoCA, Previous years rates 2005 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006* | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | | **42,048** | **88,676** |
| Anencephaly | | | | | | | 9.28 | 10.71* |
| Spina bifida | | | | | | | 0.71 | 1.39* |
| Encephalocele | | | | | | | 1.90 | 1.00* |
| Microcephaly | | | | | | | 1.43 | 4.49* |
| Holoprosencephaly | | | | | | | nr | 2.05* |
| Hydrocephaly | | | | | | | 12.13 | 13.79* |
| Anophthalmos | | | | | | | 0.47 | nr |
| Microphthalmos | | | | | | | 0.48 | 2.05* |
| Unspecified Anophthalmos/Microphthalmos | | | | | | | 0.71 | 5.55* |
| Anotia | | | | | | | nr | Nr |
| Microtia | | | | | | | nr | 1.81* |
| Unspecified Anotia/Microtia | | | | | | | 0.94 | 9.57* |
| Transposition of great vessels | | | | | | | 0.48 | 7.94* |
| Tetralogy of Fallot | | | | | | | 0.47 | 1.36* |
| Hypoplastic left heart syndrome | | | | | | | nr | 3.67* |
| Coarctation of aorta | | | | | | | nr | 2.92* |
| Choanal atresia, bilateral | | | | | | | nr | nr |
| Cleft palate without cleft lip | | | | | | | 3.09 | 8.44 |
| Cleft lip with or without cleft palate | | | | | | | 3.57 | 11.86* |
| Oesophageal atresia/stenosis with or without fistula | | | | | | | 2.85 | 17.76* |
| Small intestine atresia/stenosis | | | | | | | nr | 12.83* |
| Anorectal atresia/stenosis | | | | | | | 1.19 | 10.94* |
| Undescended testis (36 weeks of gestation or later) | | | | | | | 21.64 | 9.83* |
| Hypospadias | | | | | | | 9.28 | 19.75* |
| Epispadias | | | | | | | nr | 0.68* |
| Indeterminate sex | | | | | | | nr | 1.78* |
| Renal agenesis | | | | | | | 0.97 | 1.37* |
| Cystic kidney | | | | | | | nr | 2.78* |
| Bladder exstrophy | | | | | | | nr | nr |
| Polydactyly, preaxial | | | | | | | 7.61 | 8.02* |
| Total Limb reduction defects (include unspecified) | | | | | | | 20.29 | 50.98 |
| Transverse | | | | | | | nr | nr |
| Preaxial | | | | | | | nr | nr |
| Postaxial | | | | | | | nr | nr |
| Intercalary | | | | | | | nr | nr |
| Mixed | | | | | | | nr | nr |
| Unspecified | | | | | | | 20.29 | nr |
| Diaphragmatic hernia | | | | | | | 0.94 | 8.38* |
| Omphalocele | | | | | | | 0.48 | 1.81* |
| Gastroschisis | | | | | | | nr | nr |
| Unspecified Omphalocele/Gastroschisis | | | | | | | nr | 2.90* |
| Prune belly sequence | | | | | | | nr | nr |
| Trisomy 13 | | | | | | | nr | 1.13* |
| Trisomy 18 | | | | | | | 0.71 | 0.93* |
| Down syndrome, all ages (include age unknown) | | | | | | | 3.57 | 11.86 |
| <20 | | | | | | | 1.09 | 6.66* |
| 20-24 | | | | | | | 1.75 | 6.00 |
| 25-29 | | | | | | | 0.00 | 17.10* |
| 30-34 | | | | | | | 3.08 | 12.93 |
| 35-39 | | | | | | | 8.65 | 22.20 |
| 40-44 | | | | | | | 17.57 | 72.46* |
| 45+ | | | | | | | 0.00 | 95.69* |
| unknown | | | | | | | --- | --- |

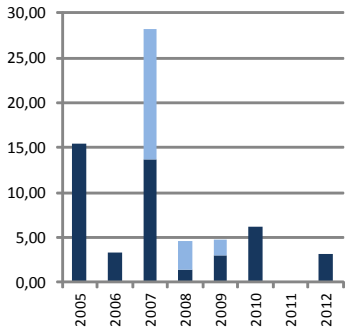
nr = data not reported or not available

* data include less than 5 years

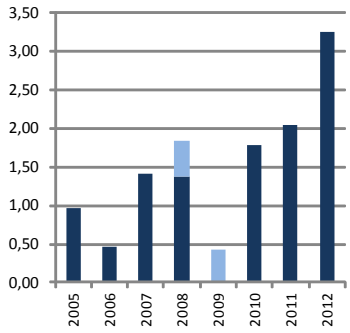
# Iran: TRoCA, Time trends 2005 – 2012

(Birth prevalence rates per 10,000)

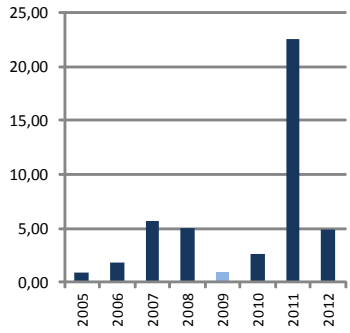
**Anencephaly**



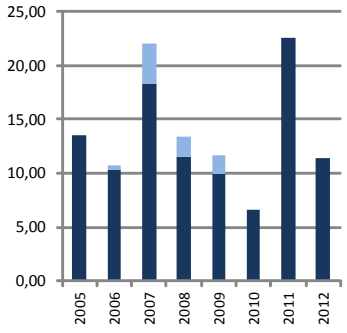

**Spina Bifida**



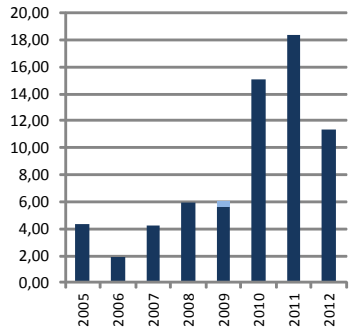

**Microcephaly**



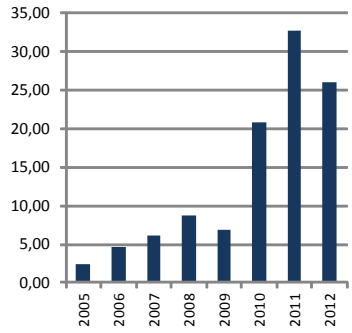

**Hydrocephaly**



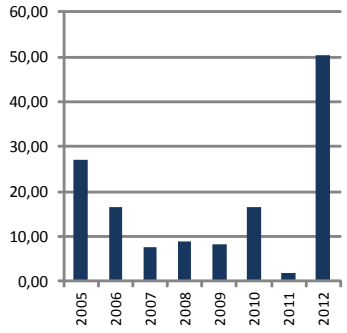

**Cleft palate without cleft lip**



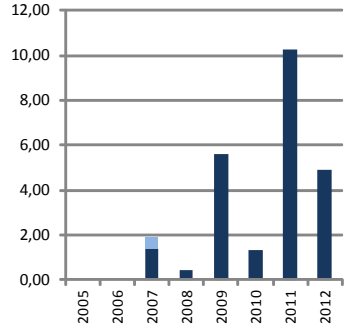

**Cleft lip with or without cleft palate**



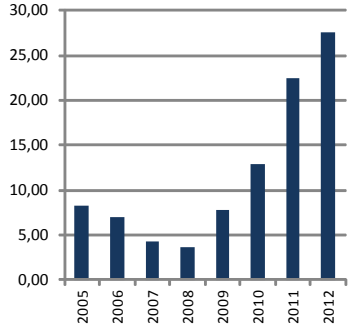

**Undescended testis**



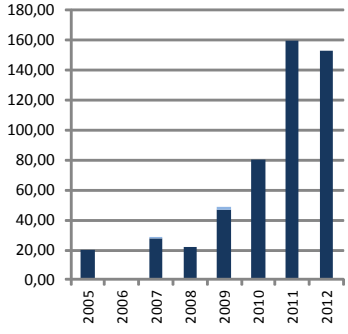

**Cystic kidney**



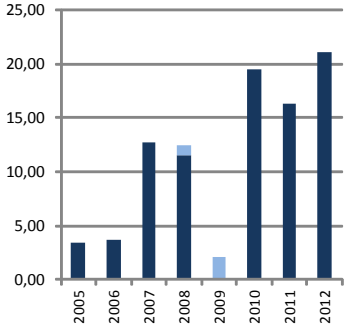

**Polydactyly, preaxial**



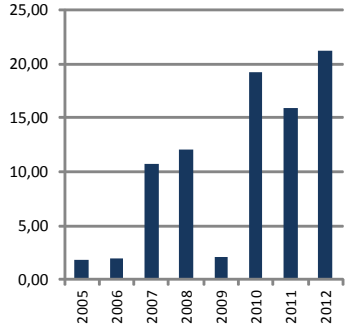

**Limb reduction defects**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

## Italy - Lombardy: LBDR

Lombardy Birth Defects Registry

**History:**
The Registry started in 2000 and is located in National Cancer Institute of Milan. The Registry is full member of ICBDSR since 2007.

**Size and Coverage:**
The Registry is population-based and registers about 14590 births annually, constituting 100% of the total annual births in the Provinces of Sondrio, Varese, Como . This is about 16.4% of the total annual births in the Region of Lombardy, and the 2.8 % of total births in Italy**.**

**Legislation and Funding:**
The Registry is a research programme approved by the Italian Ministry of Health and supported by funding from the Italian National Cancer Institute.

**Source of Ascertainment:**
The registry uses active data collection methods from multiple sources (death certificates, hospital discharge records, pathology reports, birth certificates, outpatient drug prescription records, outpatient records, the social security list of the Region of Lombardy and clinical records). The registry data are routinely cross-checked with the social security list of the Lombardy Region to up-date case (vital status) and parent information (age, vital status, etc.).

**Exposure Information:**
Information on exposure is not collected routinely can be collected on specific indications.

**Addresses and Staff:**
Programme Directors:
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# Italy – Lombardy: LBDR, 2012

| | |
|---|---|
| Live births (LB) | 7,928 |
| Stillbirths (SB) | 11 |
| Total births | 7,939 |
| Number of terminations of pregnancy (ToP) for birth defects | 73 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | 0 | 1.26 |
| Spina bifida | 0 | 0 | 4 | 5.04 |
| Encephalocele | 0 | 1 | 0 | 1.26 |
| Microcephaly | 1 | 0 | 0 | 1.26 |
| Holoprosencephaly | 0 | 0 | 0 | 0.00 |
| Hydrocephaly | 1 | 1 | 2 | 5.04 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 0 | 0 | 0 | 0.00 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 1.26 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 1 | 0 | 1 | 2.52 |
| Tetralogy of Fallot | 3 | 0 | 0 | 3.78 |
| Hypoplastic left heart syndrome | 1 | 0 | 0 | 1.26 |
| Coarctation of aorta | 0 | 0 | 0 | 0.00 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 5 | 0 | 0 | 6.30 |
| Cleft lip with or without cleft palate | 5 | 0 | 0 | 6.30 |
| Oesophageal atresia/stenosis with or without fistula | 3 | 0 | 0 | 3.78 |
| Small intestine atresia/stenosis | 5 | 0 | 0 | 6.30 |
| Anorectal atresia/stenosis | 1 | 0 | 0 | 1.26 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 28 | 0 | 0 | 35,27 |
| Epispadias | 0 | 0 | 0 | 0,00 |
| Indeterminate sex | 0 | 0 | 0 | 0,00 |
| Renal agenesis | 3 | 0 | 1 | 5,04 |
| Cystic kidney | 1 | 0 | 0 | 1,26 |
| Bladder exstrophy | 0 | 0 | 0 | 0,00 |
| Polydactyly, preaxial | 1 | 0 | 1 | 2,52 |
| Total Limb reduction defects (include unspecified) | 2 | 1 | 0 | 3,78 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 3 | 0 | 0 | 3.78 |
| Omphalocele | 1 | 1 | 1 | 3.78 |
| Gastroschisis | 2 | 0 | 0 | 2.52 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 4 | 5.04 |
| Trisomy 18 | 0 | 1 | 4 | 6.30 |
| Down syndrome, all ages (include age unknown) | 23 | 0 | 8 | 39.05 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 0 | 14.49 |
| 25-29 | 1 | 0 | 1 | 12.11 |
| 30-34 | 3 | 0 | 2 | 18.50 |
| 35-39 | 10 | 0 | 3 | 59.25 |
| 40-44 | 7 | 0 | 2 | 166.67 |
| 45+ | 1 | 0 | 0 | 322.58 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

## Italy- Tuscany: RTDC

Tuscany Registry of Congenital Defects

**History:**
The Registry started in 1979 in the province of Florence and from 1992 in the whole Tuscany region. The Programme became a full member of the Clearinghouse in 1998.

**Size and coverage:**
The Programme is population based, involves all the regional hospitals and the coverage is around 95% of all births in the Tuscany region (approximately 3.5 million inhabitants and 30,000 births per year). Stillbirths of 20 weeks or more gestation and induced abortions after prenatal diagnosis of birth defects are systematically included. Malformed babies diagnosed within the first year of life are also registered.

**Legislation and funding:**
The Registry is a surveillance Programme included in the Regional Statistics System; it is formally recognised and supported by the Tuscany Region Health Authority.

**Sources and ascertainment:**
Multiple sources are used to ascertain malformed infants; records are obtained from all obstetrical and maternity units, pediatric departments, paediatric cardiology departments, paediatric cardiac surgery units, prenatal diagnostic centres and medical genetics units.
Cytogenetic laboratories only confirm karyotype for cases already known. Mothers are interviewed by using a standardised questionnaire. Malformed babies diagnosed within the first year of life are also registered.

**Exposure information**:
Maternal and paternal occupation, life-style and socio-economic characteristics are obtained by interviews of mothers of malformed infants.

**Background information**:
Vital statistics and other epidemiological information is obtained by the birth medical records collected by the Regional Bureau of Statistics.

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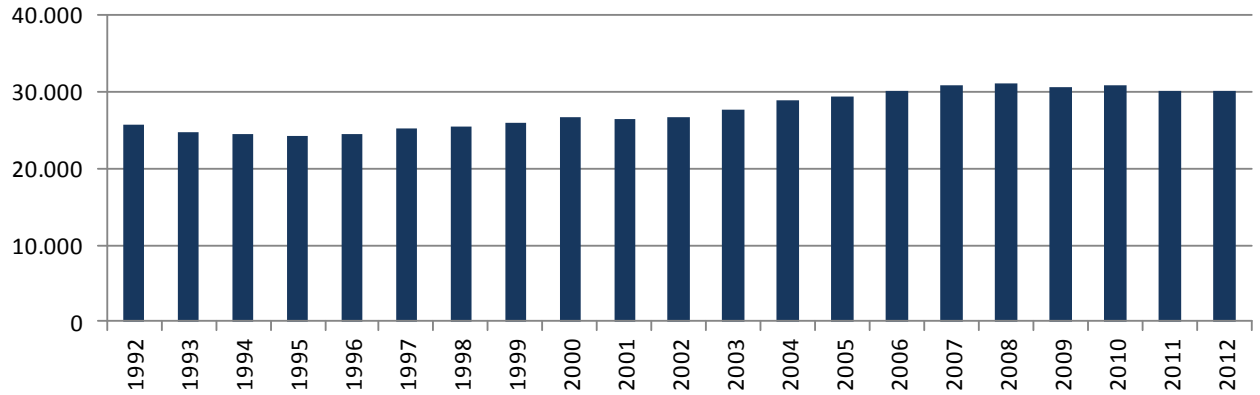
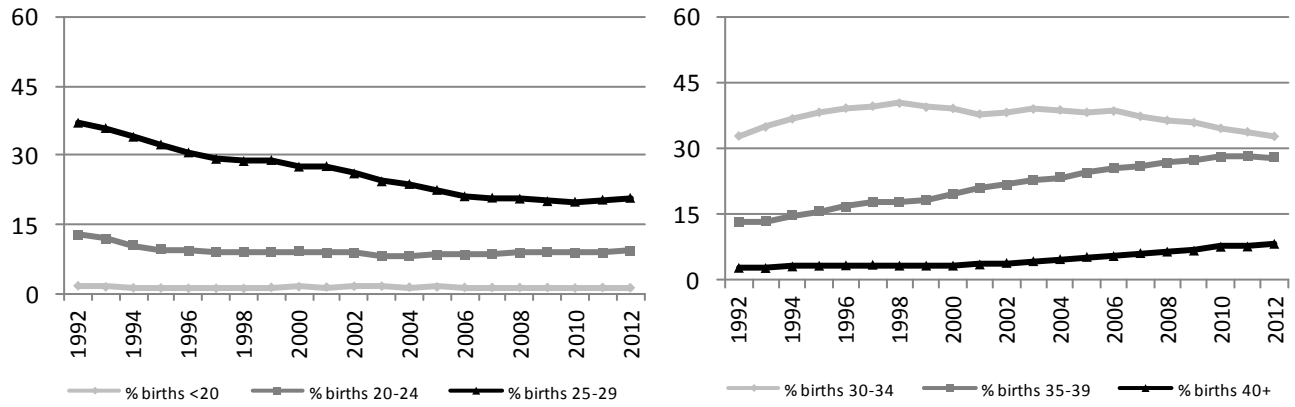
# Italy- Tuscany: RTDC

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 21 | 100.0 | Cystic kidney | 7 | 14.9 |
| Spina bifida | 27 | 79.4 | Limb reduction defects | 15 | 36.6 |
| Encephalocele | 3 | 37.5 | Diaphragmatic hernia | 5 | 20.0 |
| Holoprosencephaly | 9 | 100.0 | Omphalocele | 10 | 71.4 |
| Hydrocephaly | 19 | 51.4 | Gastroschisis | 3 | 27.3 |
| Hypoplastic left heart syndrome | 14 | 58.3 | Trisomy 13 | 13 | 92.9 |
| Cleft palate without cleft lip | 2 | 7.7 | Trisomy 18 | 58 | 95.1 |
| Cleft lip with or without cleft palate | 7 | 17.9 | Down syndrome | 181 | 74.8 |
| Renal agenesis | 6 | 75.0 | | | |

Total ToPs with births defects = 522 (Ratio ToPs/Births: 5.73 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Italy- Tuscany: RTDC, 2012

| | |
|---|---|
| Live births (LB) | 29,934 |
| Stillbirths (SB) | 81 |
| Total births | 30,015 |
| Number of terminations of pregnancy (ToP) for birth defects | 188 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 12 | 4.00 |
| Spina bifida | 0 | 0 | 5 | 1.67 |
| Encephalocele | 0 | 0 | 1 | 0.33 |
| Microcephaly | 2 | 0 | 0 | 0.67 |
| Holoprosencephaly | 0 | 0 | 0 | 0.00 |
| Hydrocephaly | 4 | 0 | 8 | 4.00 |
| Anophthalmos | 1 | 0 | 1 | 0.67 |
| Microphthalmos | 0 | 0 | 1 | 0.33 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.33 |
| Microtia | 3 | 0 | 0 | 1.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 2 | 0 | 1 | 1.00 |
| Tetralogy of Fallot | 7 | 0 | 0 | 2.33 |
| Hypoplastic left heart syndrome | 1 | 0 | 4 | 1.67 |
| Coarctation of aorta | 3 | 0 | 0 | 1.00 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.67 |
| Cleft palate without cleft lip | 10 | 0 | 1 | 3.66 |
| Cleft lip with or without cleft palate | 13 | 1 | 3 | 5.66 |
| Oesophageal atresia/stenosis with or without fistula | 5 | 0 | 0 | 1.67 |
| Small intestine atresia/stenosis | 5 | 1 | 0 | 2.00 |
| Anorectal atresia/stenosis | 4 | 0 | 0 | 1.33 |
| Undescended testis (36 weeks of gestation or later) | 32 | 0 | 0 | 10.66 |
| Hypospadias | 43 | 0 | 1 | 14.66 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 1 | 0 | 1 | 0.67 |
| Renal agenesis | 0 | 0 | 1 | 0.33 |
| Cystic kidney | 22 | 0 | 2 | 8.00 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 2 | 0 | 0 | 0.67 |
| Total Limb reduction defects (include unspecified) | 9 | 0 | 9 | 6.00 |
| Transverse | 6 | 0 | 4 | 3.33 |
| Preaxial | 1 | 0 | 1 | 0.67 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 1 | 0 | 3 | 1.33 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 1 | 0 | 1 | 0.67 |
| Diaphragmatic hernia | 9 | 0 | 0 | 3.00 |
| Omphalocele | 1 | 0 | 3 | 1.33 |
| Gastroschisis | 2 | 0 | 1 | 1.00 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 4 | 1.33 |
| Trisomy 18 | 0 | 0 | 24 | 8.00 |
| Down syndrome, all ages (include age unknown) | 17 | 0 | 69 | 28.65 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 0 | 3.62 |
| 25-29 | 1 | 0 | 1 | 3.22 |
| 30-34 | 4 | 0 | 3 | 7.13 |
| 35-39 | 7 | 0 | 36 | 51.33 |
| 40-44 | 4 | 0 | 27 | 134.90 |
| 45+ | 0 | 0 | 1 | 60.24 |
| unknown | 0 | 0 | 1 | 322.58 |

nr = data not reported or not available

# Italy- Tuscany, Previous years rates 1992 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | **123,738** | **129,710** | **142,822** | **153,650** |
| Anencephaly | | | | | 2.51 | 2.31 | 1.68 | 1.50 |
| Spina bifida | | | | | 2.42 | 3.47 | 2.52 | 3.90 |
| Encephalocele | | | | | 1.13 | 0.62 | 0.42 | 1.04 |
| Microcephaly | | | | | 1.29 | 0.77 | 0.84 | 0.59 |
| Holoprosencephaly | | | | | 0.57 | 0.85 | 1.05 | 1.37 |
| Hydrocephaly | | | | | 3.07 | 2.78 | 4.13 | 3.38 |
| Anophthalmos | | | | | 0.00 | 0.23 | 0.21 | 0.00 |
| Microphthalmos | | | | | 0.40 | 0.62 | 0.56 | 0.72 |
| Unspecified Anophthalmos/Microphthalmos | | | | | 0.00 | 0.00 | 0.07 | 0.00 |
| Anotia | | | | | 0.48 | 0.15 | 0.35 | 0.39 |
| Microtia | | | | | 0.57 | 0.46 | 0.42 | 0.59 |
| Unspecified Anotia/Microtia | | | | | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | | | | 1.86 | 2.62 | 3.01 | 3.32 |
| Tetralogy of Fallot | | | | | 2.02 | 2.93 | 2.80 | 2.28 |
| Hypoplastic left heart syndrome | | | | | 1.94 | 2.16 | 2.17 | 2.60 |
| Coarctation of aorta | | | | | 2.42 | 3.16 | 1.89 | 2.34 |
| Choanal atresia, bilateral | | | | | 0.08 | 0.31 | 0.49 | 0.59 |
| Cleft palate without cleft lip | | | | | 3.64 | 3.55 | 4.13 | 3.32 |
| Cleft lip with or without cleft palate | | | | | 6.79 | 7.17 | 5.32 | 5.21 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 2.26 | 2.47 | 2.31 | 2.21 |
| Small intestine atresia/stenosis | | | | | 0.97 | 0.46 | 1.40 | 1.30 |
| Anorectal atresia/stenosis | | | | | 1.54 | 2.54 | 2.38 | 1.95 |
| Undescended testis (36 weeks of gestation or later) | | | | | 3.80 | 8.87 | 7.35 | 6.64 |
| Hypospadias | | | | | 4.93 | 3.85 | 7.21 | 13.93 |
| Epispadias | | | | | 0.24 | 0.23 | 0.28 | 0.13 |
| Indeterminate sex | | | | | 0.89 | 0.62 | 0.42 | 0.91 |
| Renal agenesis | | | | | 1.62 | 1.31 | 0.70 | 0.98 |
| Cystic kidney | | | | | 3.23 | 3.39 | 4.69 | 4.04 |
| Bladder exstrophy | | | | | 0.32 | 0.15 | 0.14 | 0.33 |
| Polydactyly, preaxial | | | | | 0.81 | 1.31 | 0.91 | 1.11 |
| Total Limb reduction defects (include unspecified) | | | | | 5.41 | 5.32 | 5.39 | 4.23 |
| Transverse | | | | | 3.96 | 2.85 | 3.15 | 3.25 |
| Preaxial | | | | | 0.24 | 0.46 | 0.56 | 0.52 |
| Postaxial | | | | | 0.16 | 0.39 | 0.21 | 0.26 |
| Intercalary | | | | | 0.24 | 0.77 | 0.42 | 0.07 |
| Mixed | | | | | 0.48 | 0.46 | 0.00 | 0.07 |
| Unspecified | | | | | 0.00 | 0.23 | 1.47 | 0.46 |
| Diaphragmatic hernia | | | | | 1.37 | 2.00 | 1.75 | 2.15 |
| Omphalocele | | | | | 2.02 | 1.46 | 1.54 | 2.21 |
| Gastroschisis | | | | | 0.40 | 0.46 | 0.77 | 1.04 |
| Unspecified Omphalocele/Gastroschisis | | | | | 0.32 | 0.46 | 0.00 | 0.13 |
| Prune belly sequence | | | | | 0.16 | 0.08 | 0.00 | 0.26 |
| Trisomy 13 | | | | | 0.65 | 0.85 | 1.47 | 1.69 |
| Trisomy 18 | | | | | 2.67 | 3.08 | 2.87 | 5.08 |
| Down syndrome, all ages (include age unknown) | | | | | 14.87 | 16.27 | 16.31 | 21.74 |
| <20 | | | | | 0.00 | 0.00 | 0.00 | 10.67 |
| 20-24 | | | | | 7.70 | 3.49 | 5.08 | 6.70 |
| 25-29 | | | | | 9.24 | 6.60 | 3.01 | 7.06 |
| 30-34 | | | | | 12.76 | 12.13 | 6.40 | 9.52 |
| 35-39 | | | | | 27.07 | 28.08 | 26.57 | 32.47 |
| 40-44 | | | | | 76.34 | 125.28 | 124.94 | 97.17 |
| 45+ | | | | | 182.65 | 0.00 | 183.49 | 159.86 |
| unknown | | | | | --- | --- | --- | --- |

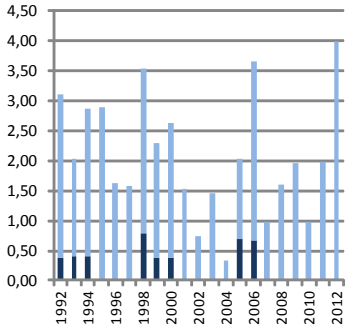
nr = data not reported or not available

* data include less than 5 years

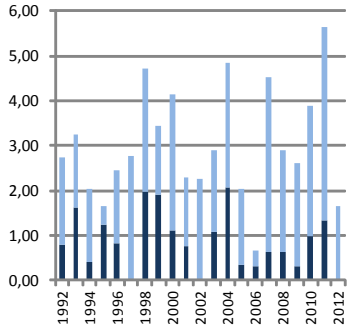
# Italy- Tuscany, Time trends 1992 – 2012

(Birth prevalence rates per 10,000)

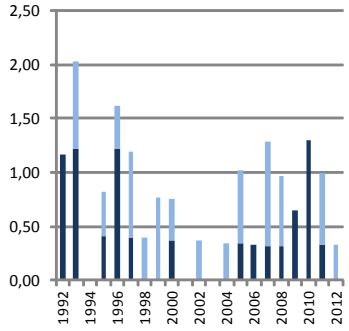
**Anencephaly**



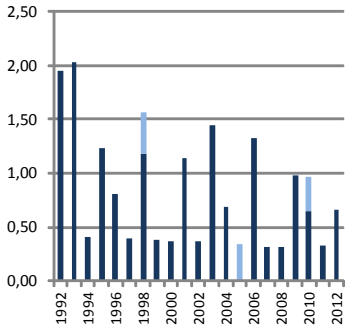

**Spina Bifida**



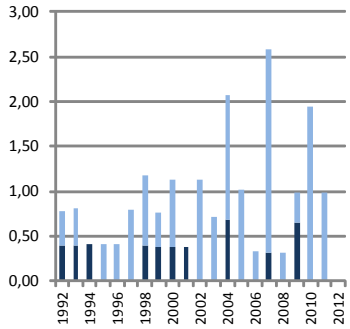

**Encephalocele**



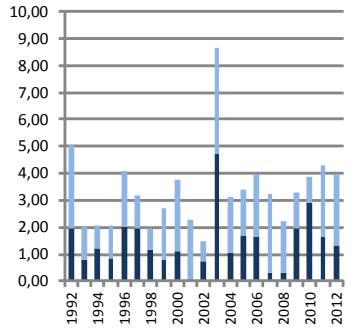

**Microcephaly**



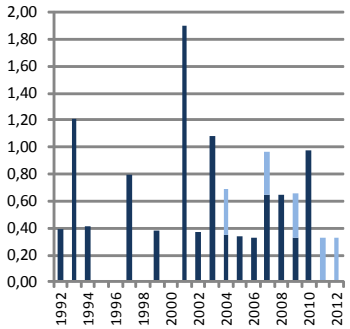

**Holoprosencephaly**



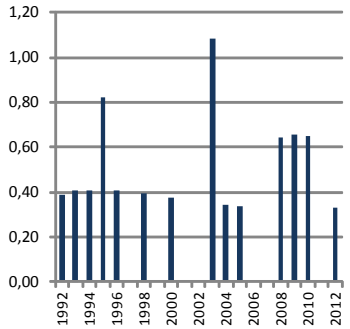

**Hydrocephaly**



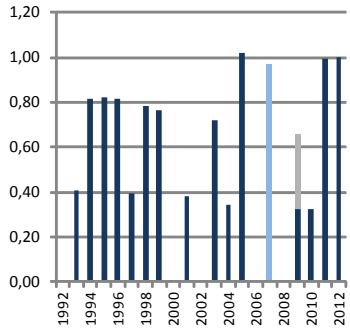

**Microphthalmos**



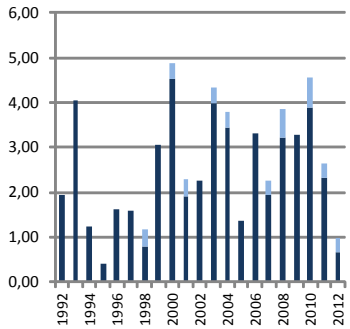

**Anotia**



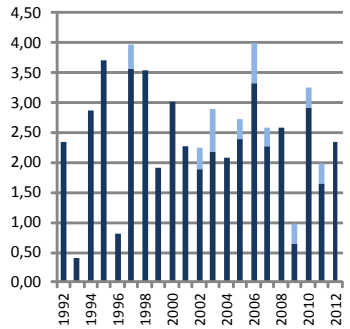

**Microtia**



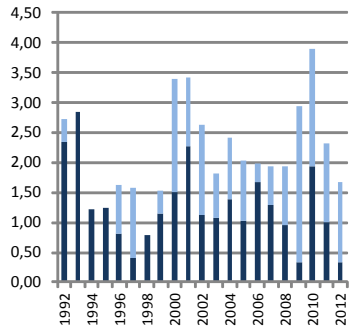

**Transposition of great vessels**




**Tetralogy of Fallot**



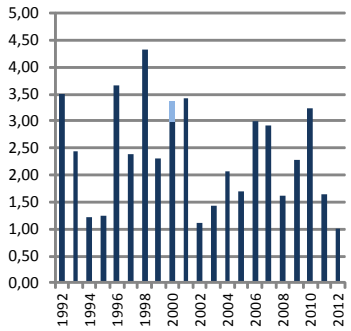

**Hypoplastic left heart syndrome**



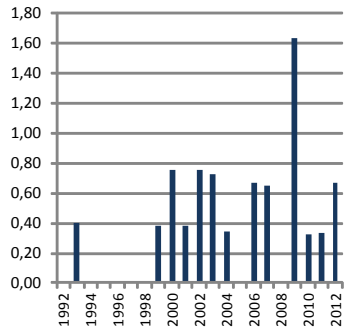
# Italy- Tuscany, Time trends 1992 – 2012

(Birth prevalence rates per 10,000)

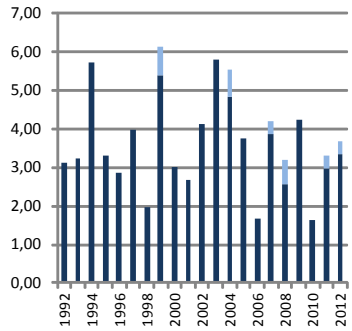
**Coarctation of aorta**



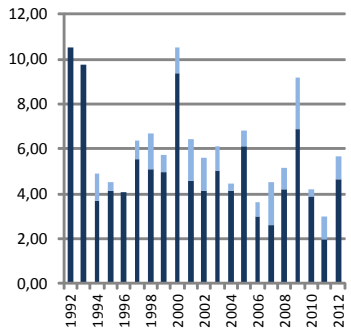

**Choanal atresia, bilateral**



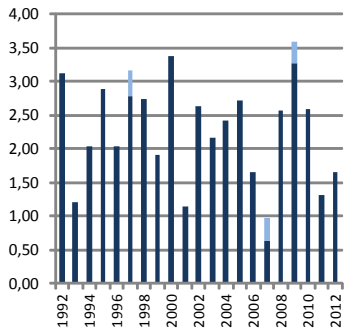

**Cleft palate without cleft lip**



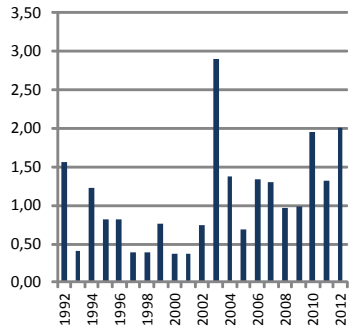

**Cleft lip with or without cleft palate**



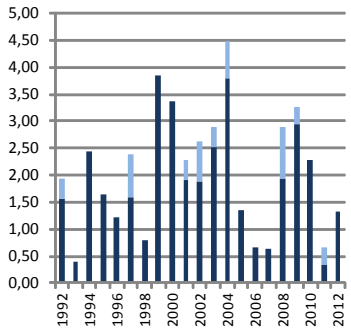

**phageal atresia/stenosis with or without f**



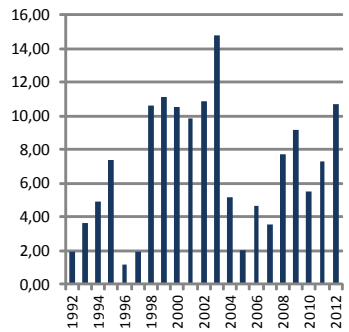

**Small intestine atresia/stenosis**



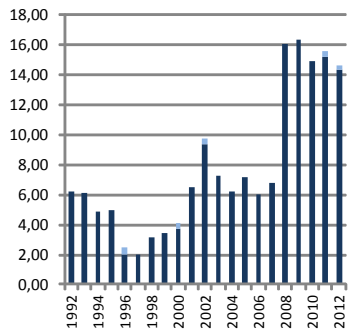

**Anorectal atresia/stenosis**



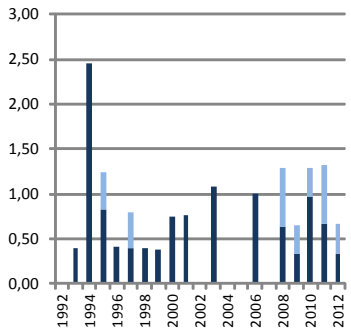

**Undescended testis**



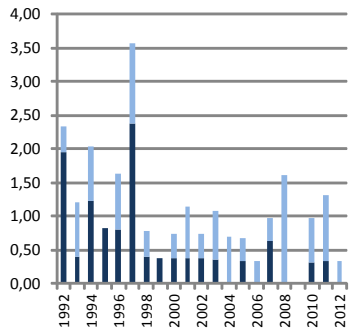

**Hypospadias**



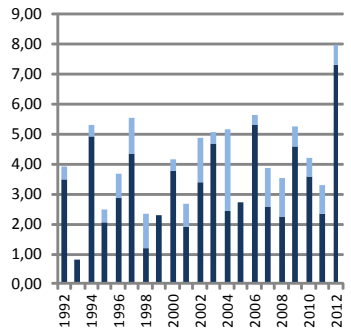

**Indeterminate sex**




**Renal agenesis**



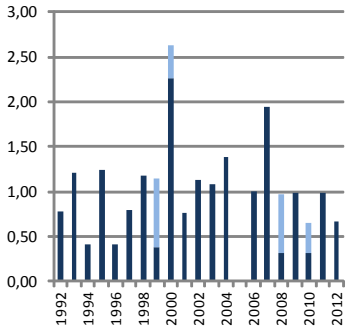

**Cystic kidney**



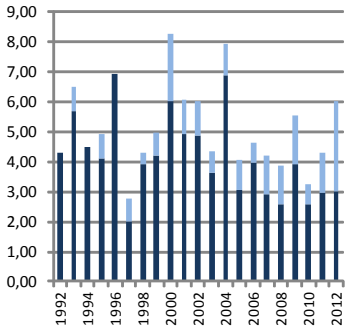
# Italy- Tuscany, Time trends 1992 – 2012

(Birth prevalence rates per 10,000)

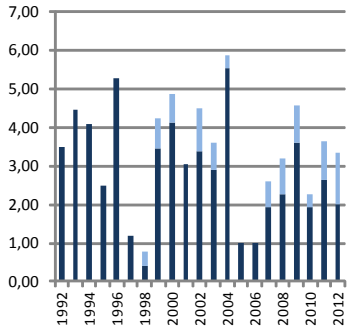
**Polydactyly, preaxial**



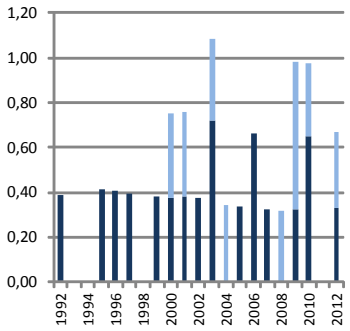

**Limb reduction defects**



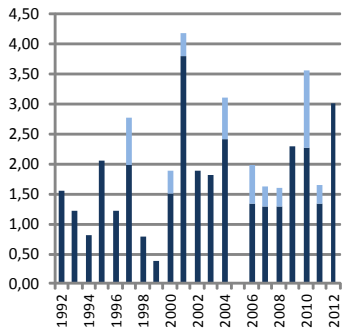

**Limb reduction defects - transverse**



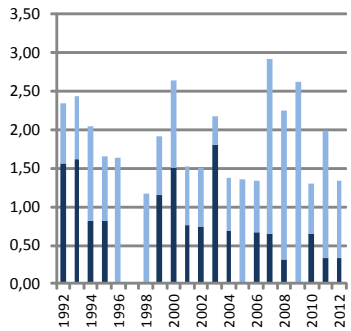

**Limb reduction defects - preaxial**



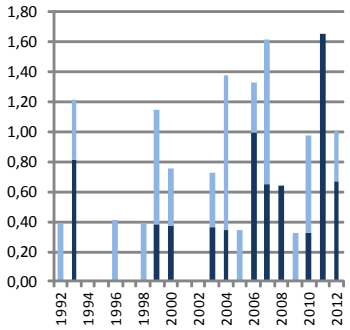

**Diaphragmatic hernia**



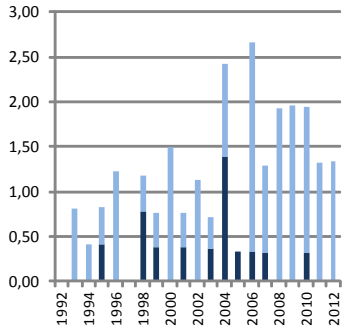

**Omphalocele**



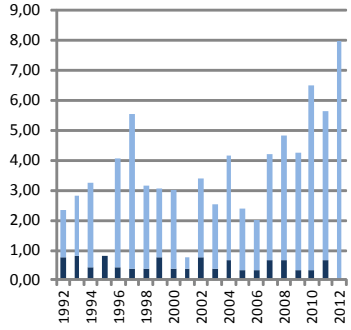

**Gastroschisis**



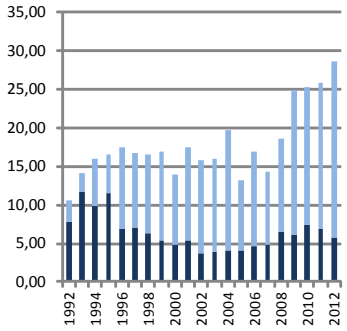

**Trisomy 13**



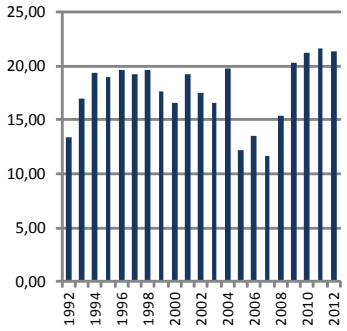

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Japan: JAOG

## Japan Association of Obstetricians and Ginaecologists

**History:**
The Programme started in 1972 and became a full member of the Clearinghouse in 1988.

**Size and coverage:**
The Programme is based on reports from 270 hospitals throughout Japan. At present approximately 100,000 births are covered, representing about 9% of all Japanese births. Stillbirths of 22 weeks or more gestation are included.

**Legislation and funding:**
The Programme is a research Programme acknowledged by the Ministry of Welfare and supported by the Japanese Association of Obstetricians and Gynecologists.

**Sources of ascertainment:**
Reports are obtained from delivery units and pediatric clinics of the participating hospitals.

**Exposure information:**
Exposure to drugs, X-ray and viral infections are available.

**Background information:**
Basic epidemiological information on all births is available from each participating hospital.

**Addresses and Staff:**
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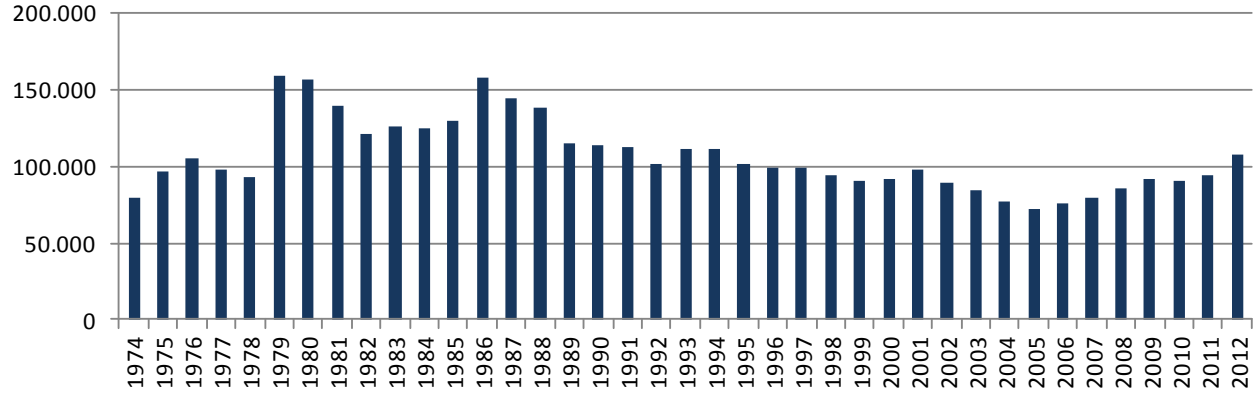
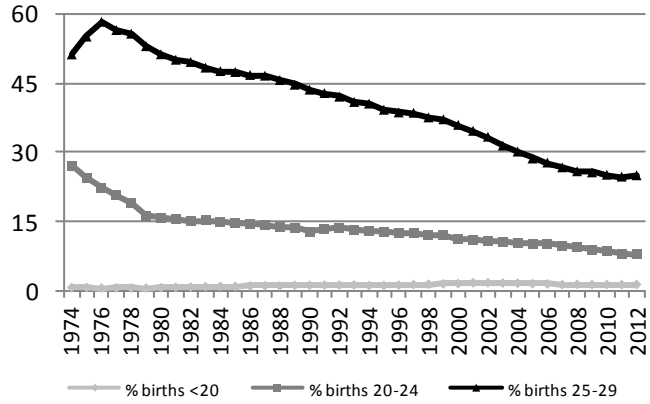
Dept. OB V GYN
3-9 Fukuura, Kanazawaku
Yokohama, 236-0004, Japan
**Phone**: 81-45-787-2689
**Fax:** 81-45-787-2689
**E-mail:** hirafu@med.yokohama-cu.ac.jp

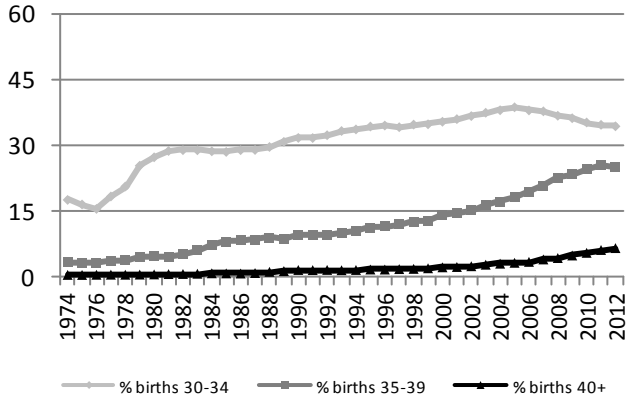
# Japan: JAOG

## Total births by year




## Percentage of births by year and maternal age



# Japan: JAOG, 2012

| | |
|---|---|
| Live births (LB) | 107,481 |
| Stillbirths (SB) | 606 |
| Total births | 108,087 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 2 | 2 | nr | 0.37 |
| Spina bifida | 50 | 6 | nr | 5.18 |
| Encephalocele | 9 | 0 | nr | 0.83 |
| Microcephaly | 11 | 2 | nr | 1.20 |
| Holoprosencephaly | 9 | 3 | nr | 1.11 |
| Hydrocephaly | 57 | 5 | nr | 5.74 |
| Anophthalmos | 3 | 1 | nr | 0.37 |
| Microphthalmos | 5 | 0 | nr | 0.46 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 16 | 6 | nr | 2.04 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 36 | 5 | nr | 3.79 |
| Tetralogy of Fallot | 75 | 0 | nr | 6.94 |
| Hypoplastic left heart syndrome | 28 | 6 | nr | 3.15 |
| Coarctation of aorta | 69 | 4 | nr | 6.75 |
| Choanal atresia, bilateral | 1 | 0 | nr | 0.09 |
| Cleft palate without cleft lip | 63 | 0 | nr | 5.83 |
| Cleft lip with or without cleft palate | 230 | 16 | nr | 22.76 |
| Oesophageal atresia/stenosis with or without fistula | 48 | 11 | nr | 5.46 |
| Small intestine atresia/stenosis | 69 | 3 | nr | 6.66 |
| Anorectal atresia/stenosis | 54 | 4 | nr | 5.37 |
| Undescended testis (36 weeks of gestation or later) | 0 | 0 | nr | 0.00 |
| Hypospadias | 61 | 1 | nr | 5.74 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 36 | 4 | nr | 3.70 |
| Cystic kidney | 43 | 9 | nr | 4.81 |
| Bladder exstrophy | 1 | 0 | nr | 0.09 |
| Polydactyly, preaxial | 72 | 2 | nr | 6.85 |
| Total Limb reduction defects (include unspecified) | 21 | 7 | nr | 2.59 |
| Transverse | 1 | 1 | nr | 0.19 |
| Preaxial | 1 | 4 | nr | 0.46 |
| Postaxial | 4 | 0 | nr | 0.37 |
| Intercalary | 5 | 1 | nr | 0.56 |
| Mixed | 9 | 1 | nr | 0.93 |
| Unspecified | 1 | 0 | nr | 0.09 |
| Diaphragmatic hernia | 55 | 4 | nr | 5.46 |
| Omphalocele | 21 | 3 | nr | 2.22 |
| Gastroschisis | 18 | 1 | nr | 1.76 |
| Unspecified Omphalocele/Gastroschisis | 4 | 1 | nr | 0.46 |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 21 | 4 | nr | 2.31 |
| Trisomy 18 | 59 | 41 | nr | 9.25 |
| Down syndrome, all ages (include age unknown) | 186 | 6 | nr | 17.76 |
| <20 | 1 | 0 | nr | 7.42 |
| 20-24 | 1 | 0 | nr | 1.19 |
| 25-29 | 14 | 0 | nr | 5.19 |
| 30-34 | 43 | 2 | nr | 12.13 |
| 35-39 | 79 | 3 | nr | 30.21 |
| 40-44 | 44 | 1 | nr | 63.54 |
| 45+ | 4 | 0 | nr | --- |
| unknown | 0 | 0 | nr | --- |

nr = data not reported or not available

# Japan: JAOG, Previous years rates 1974 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | **280,942** | **645,820** | **659,696** | **625,335** | **525,703** | **472,208** | **399,683** | **443,548** |
| Anencephaly | 8.58 | 9.55 | 8.47 | 5.96 | 2.93 | 1.63 | 1.18 | 0.74 |
| Spina bifida | 1.64 | 2.09 | 3.02 | 3.10 | 3.73 | 4.17 | 5.28 | 5.59 |
| Encephalocele | 1.14 | 1.01 | 1.20 | 1.18 | 0.99 | 0.83 | 0.78 | 0.52 |
| Microcephaly | 0.85 | 1.01 | 1.24 | 1.38 | 1.35 | 1.42 | 1.43 | 1.56 |
| Holoprosencephaly | nr | nr | nr | nr | 0.70* | 1.14 | 1.45 | 1.17 |
| Hydrocephaly | 2.56 | 3.08 | 4.64 | 6.28 | 6.81 | 7.28 | 7.58 | 7.82 |
| Anophthalmos | 0.71 | 0.77 | 0.88 | 0.54 | 0.27 | 0.19 | 0.50 | 0.27 |
| Microphthalmos | 0.39 | 0.60 | 0.65 | 0.59 | 0.55 | 0.40 | 0.50 | 0.68 |
| Unspecified Anophthalmos/Microphthalmos | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | nr | nr | nr | nr | nr | nr | 0.00* | 0.00* |
| Microtia | 0.82 | 1.25 | 1.02 | 1.17 | 1.18 | 1.33 | 1.20 | 2.12 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr | nr | nr | 0.00 | 0.00 |
| Transposition of great vessels | nr | nr | nr | nr | nr | 2.33 | 4.03 | 4.69 |
| Tetralogy of Fallot | nr | nr | nr | nr | nr | 2.77 | 5.05 | 7.03 |
| Hypoplastic left heart syndrome | nr | nr | nr | nr | nr | 1.61 | 3.53 | 4.53 |
| Coarctation of aorta | nr | nr | nr | nr | nr | 1.91 | 3.43 | 6.65 |
| Choanal atresia, bilateral | nr | nr | nr | nr | nr | nr | 0.00* | 0.02 |
| Cleft palate without cleft lip | 13.28 | 12.46 | 5.24 | 5.49 | 4.87 | 4.64 | 4.45 | 5.05 |
| Cleft lip with or without cleft palate | 15.31 | 13.19 | 13.93 | 14.68 | 15.67 | 16.88 | 20.42 | 21.67 |
| Oesophageal atresia/stenosis with or without fistula | nr | 1.20* | 1.20 | 1.70 | 2.40 | 3.60 | 4.58 | 4.98 |
| Small intestine atresia/stenosis | nr | nr | nr | nr | nr | 4.62 | 6.93 | 7.82 |
| Anorectal atresia/stenosis | 3.84 | 3.92 | 3.97 | 4.17 | 4.24 | 4.60 | 5.98 | 6.83 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr | nr | nr |
| Hypospadias | 1.46 | 2.12 | 2.40 | 2.53 | 2.87 | 3.43 | 4.23 | 5.19 |
| Epispadias | nr | nr | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr | nr | nr | nr | nr |
| Renal agenesis | nr | nr | nr | 1.23* | 1.46 | 1.78 | 2.33 | 2.91 |
| Cystic kidney | nr | nr | nr | nr | nr | 3.18 | 4.60 | 4.53 |
| Bladder exstrophy | 0.10 | 0.20* | 0.14 | 0.14 | 0.08 | 0.30 | 0.23 | 0.29 |
| Polydactyly, preaxial | nr | nr | nr | 5.89* | 6.79 | 5.93 | 6.71 | 6.74 |
| Total Limb reduction defects (include unspecified) | nr | nr | nr | nr | 3.37* | 3.22 | 3.58 | 3.81 |
| Transverse | nr | nr | nr | nr | 0.33* | 0.38 | 0.33 | 0.29 |
| Preaxial | nr | nr | nr | nr | 0.54* | 0.59 | 0.60 | 0.90 |
| Postaxial | nr | nr | nr | nr | 0.26* | 0.30 | 0.45 | 0.25 |
| Intercalary | nr | nr | nr | nr | 1.42* | 0.78 | 0.73 | 1.04 |
| Mixed | nr | nr | nr | nr | 0.57* | 0.68 | 1.00 | 0.90 |
| Unspecified | nr | nr | nr | nr | 0.26* | 0.49 | 0.48 | 0.43 |
| Diaphragmatic hernia | nr | nr | nr | 2.39* | 2.95 | 5.15 | 6.03 | 6.67 |
| Omphalocele | 0.85 | 1.22 | 1.76 | 3.26 | 2.85 | 3.43 | 3.73 | 3.86 |
| Gastroschisis | 0.96 | 1.01 | 1.02 | 1.26 | 1.48 | 2.39 | 2.65 | 2.50 |
| Unspecified Omphalocele/Gastroschisis | 0.00 | 0.00 | 0.00 | 0.21 | 0.30 | 0.28 | 0.25 | 0.23 |
| Prune belly sequence | nr | nr | nr | nr | 0.10* | 0.02 | 0.00 | 0.09 |
| Trisomy 13 | nr | nr | nr | nr | 0.61* | 1.00 | 1.68 | 2.03 |
| Trisomy 18 | nr | nr | nr | nr | 2.65* | 4.72 | 8.18 | 9.90 |
| Down syndrome, all ages (include age unknown) | nr | 4.25* | 5.24 | 5.95 | 6.60 | 8.68 | 10.88 | 13.19 |
| <20 | nr | nr | nr | nr | 4.39* | 4.56 | 5.01 | 3.65 |
| 20-24 | nr | nr | nr | nr | 2.58* | 2.90 | 4.11 | 3.60 |
| 25-29 | nr | nr | nr | nr | 4.26* | 5.19 | 5.44 | 5.74 |
| 30-34 | nr | nr | nr | nr | 5.85* | 8.11 | 9.49 | 8.07 |
| 35-39 | nr | nr | nr | nr | 15.76* | 18.95 | 21.46 | 24.34 |
| 40-44 | nr | nr | nr | nr | 60.67* | 52.33 | 50.35 | 55.20 |
| 45+ | nr | nr | nr | nr | nr | nr | nr | nr |
| unknown | --- | --- | --- | --- | --- | --- | --- | --- |

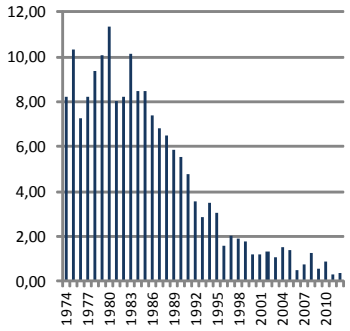
nr = data not reported or not available

* data include less than 5 years

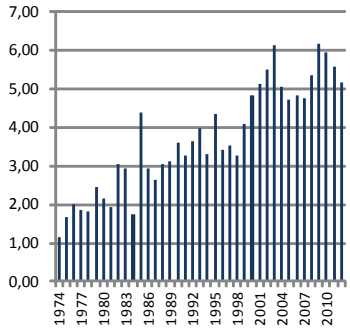
# Japan: JAOG, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

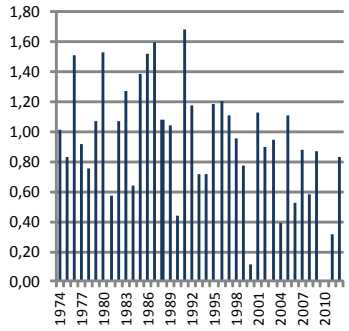
**Anencephaly**



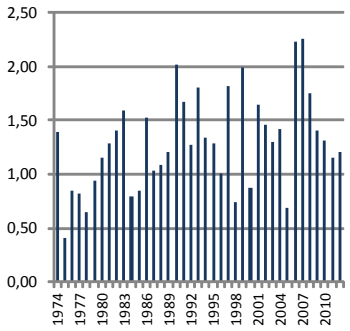

**Spina Bifida**



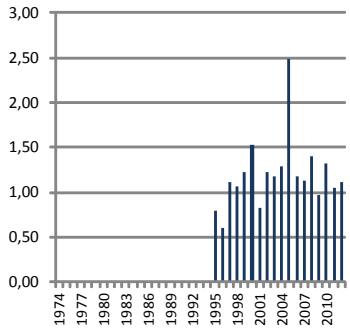

**Encephalocele**



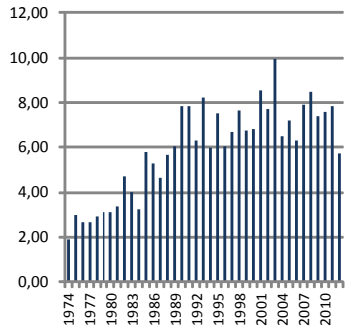

**Microcephaly**



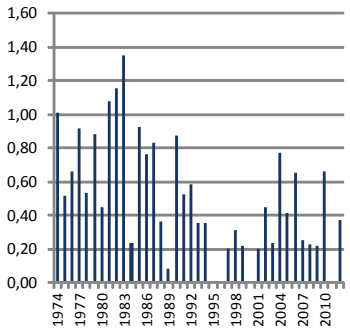

**Holoprosencephaly**



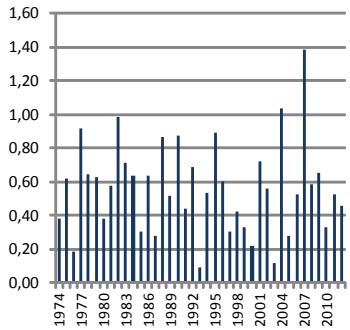

**Hydrocephaly**



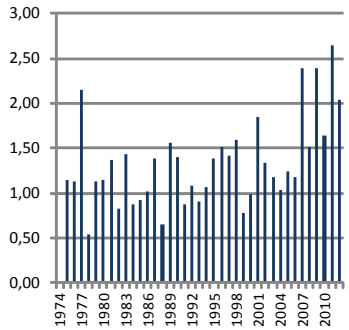

**Anophthalmos**



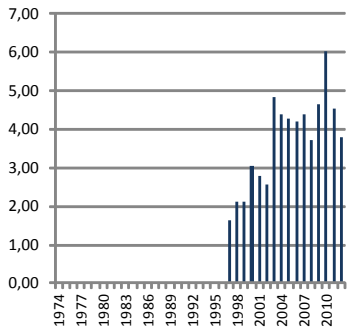

**Microphthalmos**



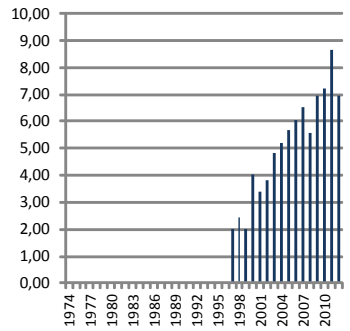

**Microtia**



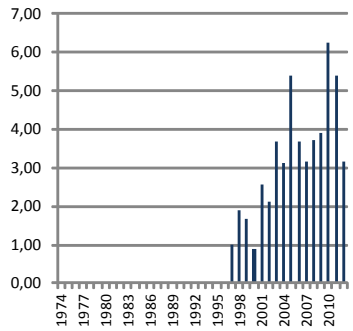

**Transposition of great vessels**




**Tetralogy of Fallot**



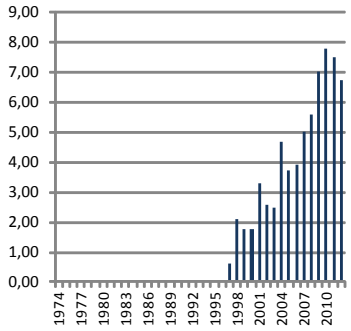

**Hypoplastic left heart syndrome**



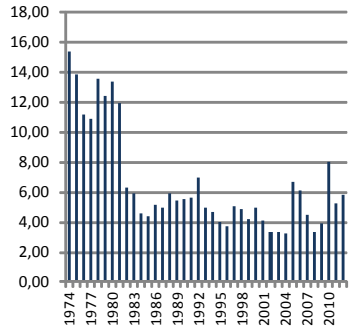
# Japan: JAOG, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

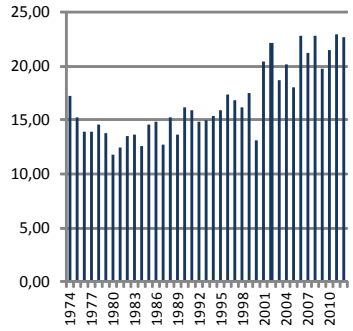
**Coarctation of aorta**



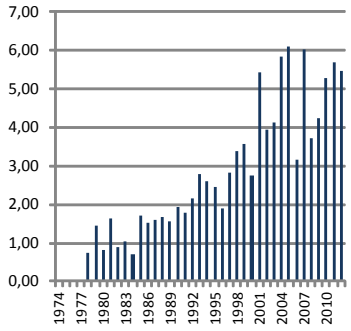

**Cleft palate without cleft lip**



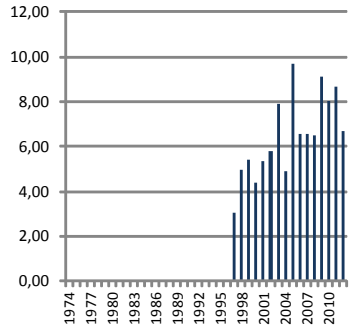

**Cleft lip with or without cleft palate**



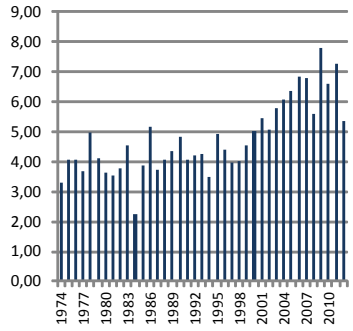

**phageal atresia/stenosis with or without f**



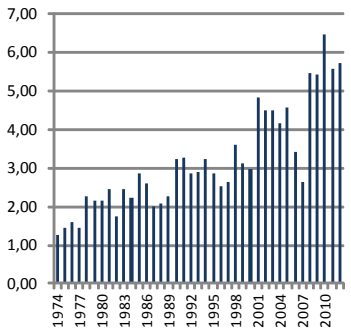

**Small intestine atresia/stenosis**



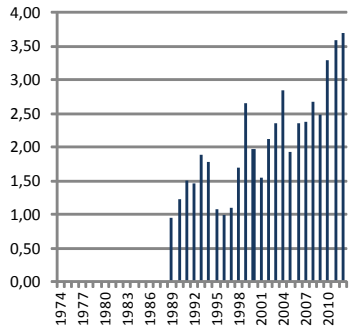

**Anorectal atresia/stenosis**



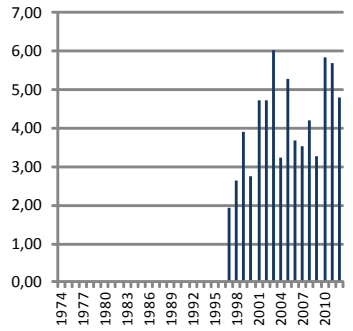

**Hypospadias**



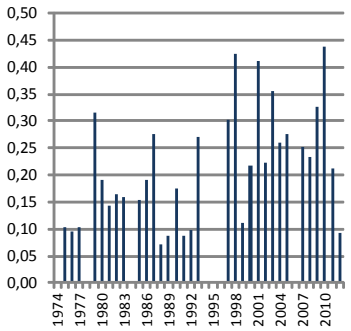

**Renal agenesis**



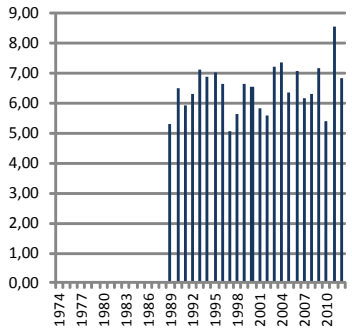

**Cystic kidney**



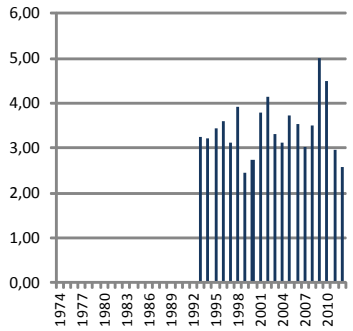

**Bladder, exstrophy**




**Polydactyly, preaxial**



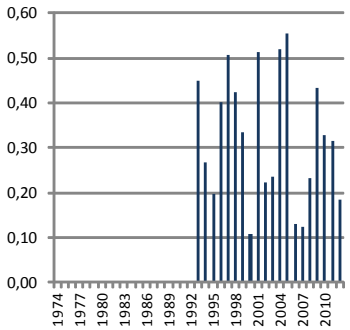

**Limb reduction defects**



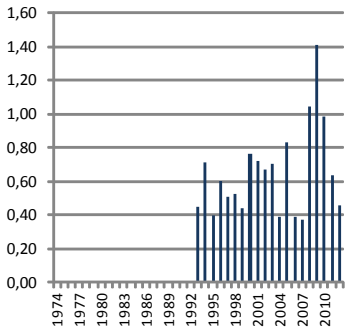
# Japan: JAOG, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

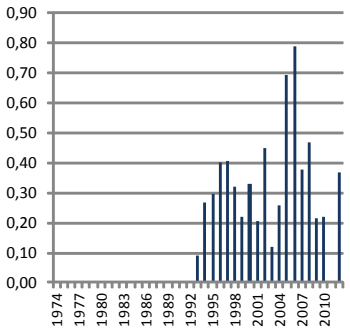
**Limb reduction defects - transverse**



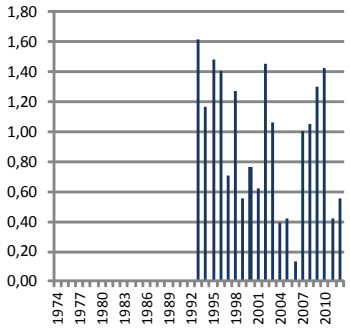

**Limb reduction defects - preaxial**



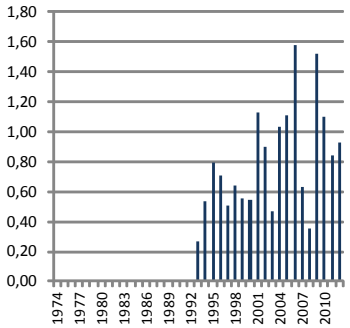

**Limb reduction defects -postaxial**



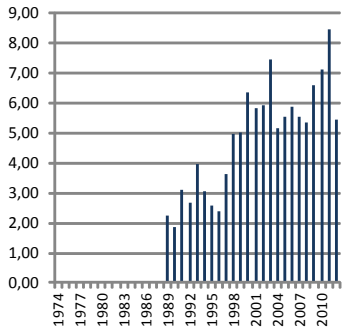

**Limb reduction defects - intercalary**



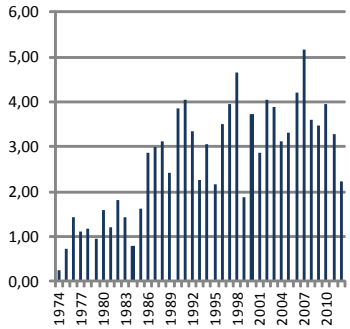

**Limb reduction defects - mixed**



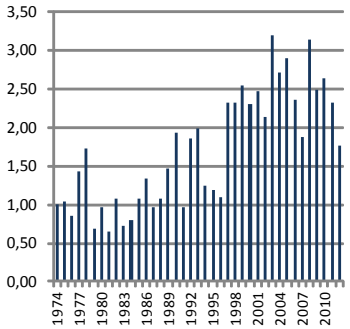

**Diaphragmatic hernia**



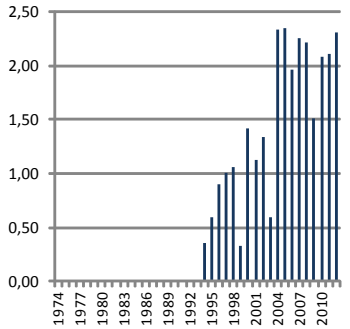

**Omphalocele**



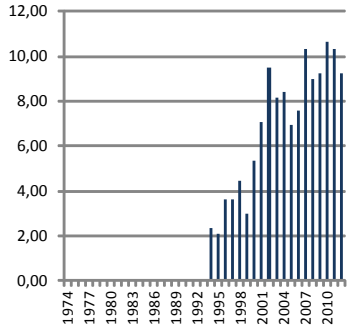

**Gastroschisis**



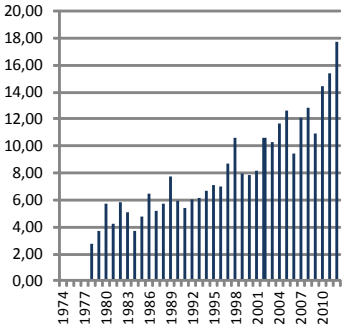

**Trisomy 13**



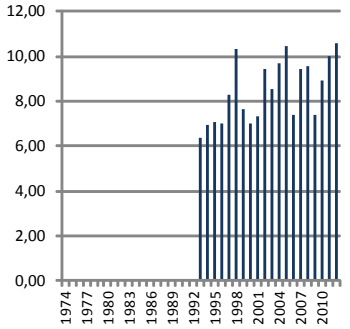

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates**

# Mexico: RYVEMCE

Mexican Registry and Epidemiological Surveillance of External Congenital Malformations

**History:**
The Programme was started in 1978. The Programme became a full member of the ICBDSR in 1980.

**Size and coverage:**
Reports are obtained from 21 hospitals in 11 cities in Mexico. Participation is voluntary. The annual number of births is approximately 62,000, about 3.5% of all births in Mexico. Stillbirths of 20 weeks or more gestation and/or at least 500g birthweight are included.

**Legislation and funding:**
The Programme is a research Programme and is funded by research grants.

**Sources of ascertainment:**
Reports are obtained from the delivery units and pediatric departments of the participating hospitals.

**Exposure information:**
The mother of each reported infant and the mother of a control infant-the next non-malformed infant born at that hospital with the same sex as the proband - are interviewed on various exposures, including drug usage and parental occupation.

**Background information:**
The total number of births in the hospitals is known.

**Addresses and Staff:**
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Mexico DF, Mexico
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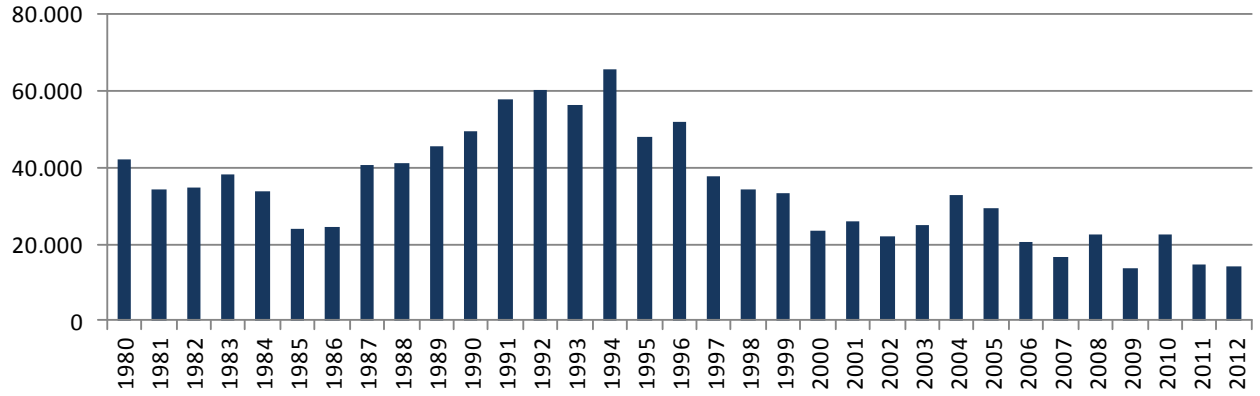
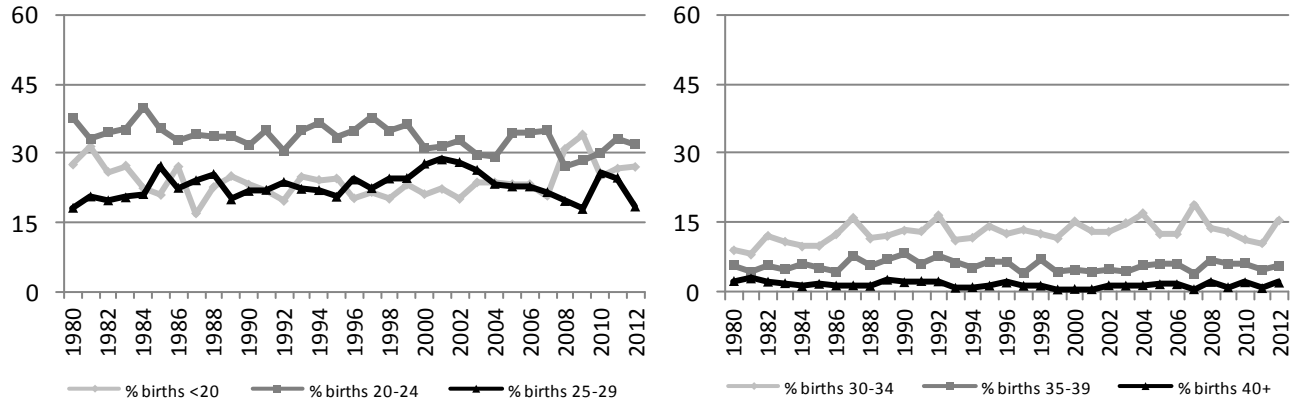
**Fax:** 52-55-56556138
**E-mail:** osvaldo@servidor.unam.mx

# Mexico: RYVEMCE

## Total births by year




## Percentage of births by year and maternal age



# Mexico: RYVEMCE, 2012

| | |
|---|---|
| Live births (LB) | 13,803 |
| Stillbirths (SB) | 204 |
| Total births | 14,007 |

Number of terminations of pregnancy (ToP) for birth defects

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 2 | | 2.14 |
| Spina bifida | 11 | 0 | | 7.85 |
| Encephalocele | 2 | 1 | | 2.14 |
| Microcephaly | 2 | 1 | | 2.14 |
| Holoprosencephaly | 3 | 0 | | 2.14 |
| Hydrocephaly | 7 | 1 | | 5.71 |
| Anophthalmos | nr | nr | | nr |
| Microphthalmos | nr | nr | | nr |
| Unspecified Anophthalmos/Microphthalmos | 3 | 0 | | 2.14 |
| Anotia | nr | nr | | nr |
| Microtia | nr | nr | | nr |
| Unspecified Anotia/Microtia | 5 | 0 | | 3.57 |
| Transposition of great vessels | 1 | 0 | | 0.71 |
| Tetralogy of Fallot | 3 | 0 | | 2.14 |
| Hypoplastic left heart syndrome | 0 | 0 | | 0.00 |
| Coarctation of aorta | 0 | 0 | | 0.00 |
| Choanal atresia, bilateral | 2 | 0 | | 1.43 |
| Cleft palate without cleft lip | 2 | 0 | | 1.43 |
| Cleft lip with or without cleft palate | 14 | 2 | | 11.42 |
| Oesophageal atresia/stenosis with or without fistula | 3 | 0 | | 2.14 |
| Small intestine atresia/stenosis | 1 | 0 | | 0.71 |
| Anorectal atresia/stenosis | 6 | 1 | | 5.00 |
| Undescended testis (36 weeks of gestation or later) | 3 | 0 | | 2.14 |
| Hypospadias | 5 | 0 | | 3.57 |
| Epispadias | 0 | 0 | | 0.00 |
| Indeterminate sex | 3 | 1 | | 2.86 |
| Renal agenesis | 0 | 0 | | 0.00 |
| Cystic kidney | 0 | 1 | | 0.71 |
| Bladder exstrophy | 0 | 0 | | 0.00 |
| Polydactyly, preaxial | 5 | 0 | | 3.57 |
| Total Limb reduction defects (include unspecified) | 3 | 1 | | 2.86 |
| Transverse | 1 | 0 | | 0.71 |
| Preaxial | 2 | 1 | | 2.14 |
| Postaxial | 0 | 0 | | 0.00 |
| Intercalary | 0 | 0 | | 0.00 |
| Mixed | 0 | 0 | | 0.00 |
| Unspecified | 0 | 0 | | 0.00 |
| Diaphragmatic hernia | 0 | 0 | | 0.00 |
| Omphalocele | 2 | 0 | | 1.43 |
| Gastroschisis | 9 | 3 | | 8.57 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | | nr |
| Prune belly sequence | 2 | 0 | | 1.43 |
| Trisomy 13 | 1 | 0 | | 0.71 |
| Trisomy 18 | 5 | 1 | | 4.28 |
| Down syndrome, all ages (include age unknown) | 23 | 0 | | 16.42 |
| <20 | 8 | 0 | | 21.16 |
| 20-24 | 6 | 0 | | 13.43 |
| 25-29 | 0 | 0 | | 0.00 |
| 30-34 | 2 | 0 | | 9.31 |
| 35-39 | 5 | 0 | | 64.65 |
| 40-44 | 1 | 0 | | 38.79 |
| 45+ | 1 | 0 | | --- |
| unknown | 0 | 0 | | --- |

nr = data not reported or not available

# Mexico: RYVEMCE, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **76,854** | **155,109** | **234,777** | **282,882** | **154,798** | **129,291** | **89,489** |
| Anencephaly | | 18.48 | 18.44 | 18.95 | 16.05 | 10.98 | 5.41 | 3.58 |
| Spina bifida | | 12.88 | 12.31 | 16.48 | 14.64 | 13.11 | 7.19 | 4.69 |
| Encephalocele | | 3.38 | 3.61 | 2.60 | 2.33 | 2.33 | 1.39 | 1.34 |
| Microcephaly | | 2.47 | 2.58 | 2.56 | 1.77 | 1.62 | 2.24 | 2.01 |
| Holoprosencephaly | | 0.13 | 0.19 | 0.43 | 0.85 | 0.84 | 1.78 | 1.79 |
| Hydrocephaly | | 5.99 | 5.35 | 4.86 | 6.01 | 6.85 | 7.27 | 5.03 |
| Anophthalmos | | 2.21 | 2.45 | 1.87 | 1.59 | 1.23 | 2.11* | 0.00* |
| Microphthalmos | | nr | nr | nr | nr | nr | nr | 0.00 |
| Unspecified Anophthalmos/Microphthalmos | | nr | nr | nr | nr | nr | 4.93* | 2.23 |
| Anotia | | nr | nr | nr | nr | nr | nr | 0.00 |
| Microtia | | nr | nr | nr | nr | nr | nr | 0.00 |
| Unspecified Anotia/Microtia | | 6.25 | 6.83 | 6.90 | 5.90 | 7.36 | 9.59 | 9.50 |
| Transposition of great vessels | | 0.13 | 0.00 | 0.13 | 0.14 | 0.32 | 0.31 | 0.78 |
| Tetralogy of Fallot | | 0.00 | 0.00 | 0.04 | 0.25 | 0.19 | 0.15 | 0.34 |
| Hypoplastic left heart syndrome | | 0.00 | 0.00 | 0.04 | 0.00 | 0.00 | 0.31 | 0.22 |
| Coarctation of aorta | | 0.13 | 0.00 | 0.04 | 0.11 | 0.00 | 0.08 | 0.67 |
| Choanal atresia, bilateral | | 0.13 | 0.26 | 0.38 | 0.57 | 0.06 | 0.23 | 0.11 |
| Cleft palate without cleft lip | | 3.51 | 3.16 | 3.62 | 3.46 | 2.71 | 3.17 | 1.90 |
| Cleft lip with or without cleft palate | | 12.10 | 13.54 | 12.39 | 12.44 | 12.98 | 15.16 | 12.96 |
| Oesophageal atresia/stenosis with or without fistula | | 1.17 | 1.22 | 2.17 | 2.09 | 2.45 | 3.17 | 2.35 |
| Small intestine atresia/stenosis | | 0.91 | 0.64 | 1.06 | 1.24 | 1.49 | 2.40 | 2.01 |
| Anorectal atresia/stenosis | | 3.77 | 4.64 | 4.43 | 4.88 | 4.46 | 5.10 | 3.35 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | nr | nr | nr | 0.00 |
| Hypospadias | | 3.51 | 4.13 | 4.51 | 5.09 | 2.91 | 4.41 | 2.68 |
| Epispadias | | nr | nr | nr | nr | nr | 0.12* | 0.11 |
| Indeterminate sex | | 1.69 | 2.00 | 2.21 | 2.58 | 1.36 | 3.02 | 3.35 |
| Renal agenesis | | 0.52 | 0.19 | 0.60 | 0.57 | 0.45 | 0.77 | 1.01 |
| Cystic kidney | | 0.26 | 0.32 | 0.47 | 0.92 | 1.42 | 1.39 | 1.01 |
| Bladder exstrophy | | 0.26 | 0.64 | 0.38 | 0.39 | 0.52 | 0.15 | 0.00 |
| Polydactyly, preaxial | | 11.71 | 12.06 | 13.63 | 12.23 | 12.02 | 12.14 | 9.72 |
| Total Limb reduction defects (include unspecified) | | 5.20 | 6.64 | 6.52 | 5.94 | 5.17 | 6.88 | 4.69 |
| Transverse | | nr | nr | nr | nr | 3.64* | 3.40 | 2.46 |
| Preaxial | | nr | nr | nr | nr | 0.81* | 1.31 | 0.45 |
| Postaxial | | nr | nr | nr | nr | 0.40* | 0.46 | 0.22 |
| Intercalary | | nr | nr | nr | nr | 0.40* | 0.46 | 0.34 |
| Mixed | | nr | nr | nr | nr | 0.61* | 0.93 | 0.89 |
| Unspecified | | nr | nr | nr | nr | 0.00* | 0.31 | 0.34 |
| Diaphragmatic hernia | | 0.52 | 0.45 | 1.06 | 1.03 | 1.03 | 1.24 | 1.01 |
| Omphalocele | | 1.95 | 1.61 | 1.36 | 1.84 | 1.49 | 2.32 | 1.79 |
| Gastroschisis | | 0.91 | 0.77 | 1.45 | 2.09 | 3.75 | 5.34 | 6.15 |
| Unspecified Omphalocele/Gastroschisis | | nr | nr | nr | nr | nr | nr | 0.00 |
| Prune belly sequence | | 1.04 | 1.35 | 1.32 | 0.67 | 0.78 | 0.54 | 0.11 |
| Trisomy 13 | | 0.52 | 0.19 | 0.34 | 0.11 | 0.13 | 0.70 | 0.56 |
| Trisomy 18 | | 1.04 | 0.52 | 0.47 | 0.28 | 0.06 | 0.62 | 0.67 |
| Down syndrome, all ages (include age unknown) | | 14.05 | 12.12 | 14.18 | 13.36 | 11.37 | 11.52 | 13.30 |
| <20 | | 8.05 | 6.27 | 11.33 | 6.76 | 6.59 | 7.46 | 0.54 |
| 20-24 | | 7.71 | 5.10 | 7.54 | 7.52 | 4.10 | 8.03 | 0.66 |
| 25-29 | | 10.21 | 5.07 | 10.12 | 12.15 | 5.91 | 6.02 | 0.42 |
| 30-34 | | 24.84 | 16.72 | 15.51 | 12.80 | 16.01 | 8.82 | 0.49 |
| 35-39 | | 52.36 | 50.44 | 38.25 | 39.85 | 54.23 | 53.91 | 2.83 |
| 40-44 | | 80.74 | 197.97 | 103.51 | 169.59 | 308.99 | 115.29 | 14.32 |
| 45+ | | 75.76 | 184.33* | 177.42* | 165.88* | 176.99* | 123.46* | nr |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

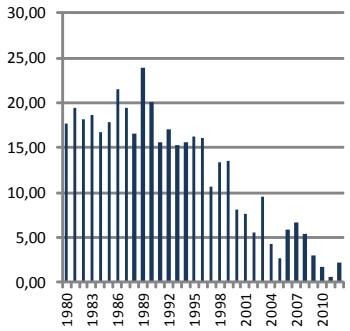
nr = data not reported or not available

* data include less than 5 years

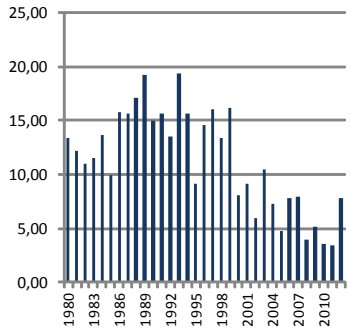
# Mexico: RYVEMCE, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

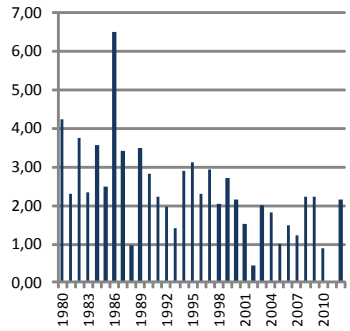
**Anencephaly**



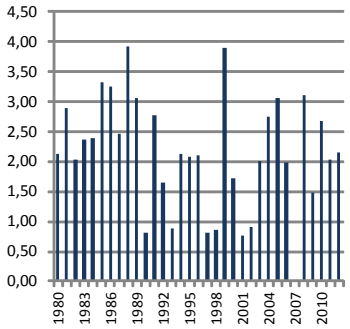

**Spina Bifida**



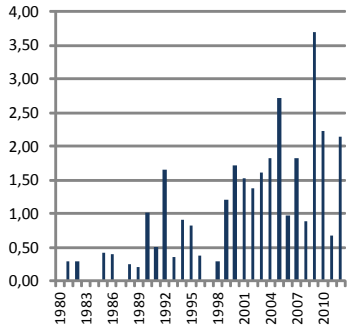

**Encephalocele**



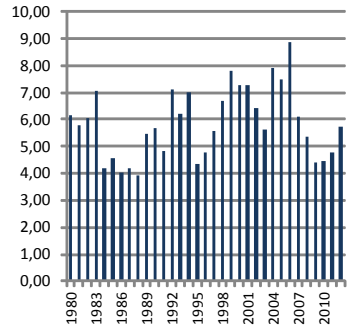

**Microcephaly**



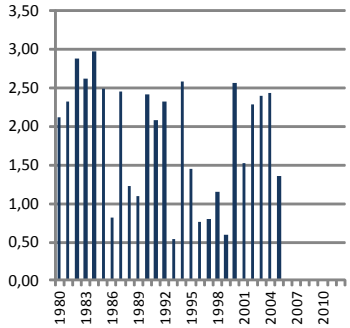

**Holoprosencephaly**



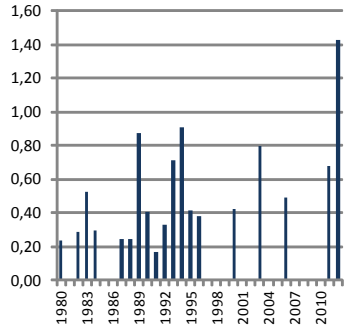

**Hydrocephaly**



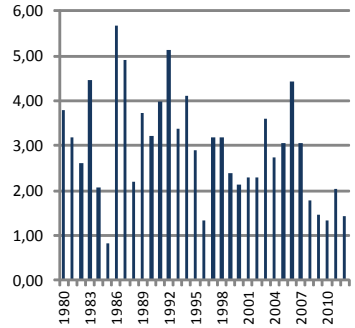

**Anophtalmos**



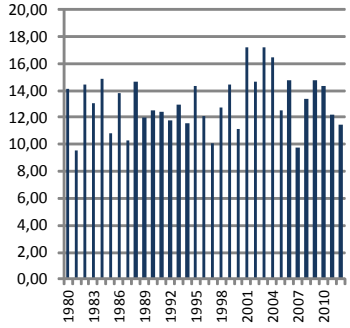

**Choanal atresia, bilateral**



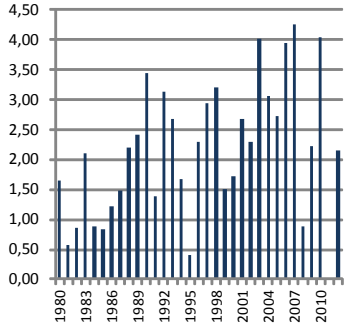

**Cleft palate without cleft lip**



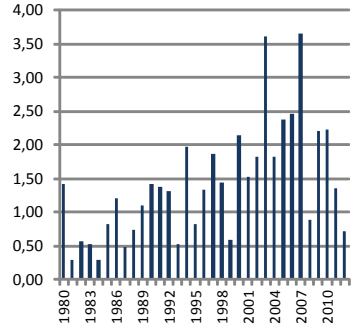

**Cleft lip with or without cleft palate**




**phageal atresia/stenosis with or without f**



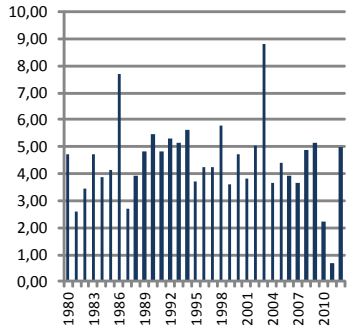

**Small intestine atresia/stenosis**



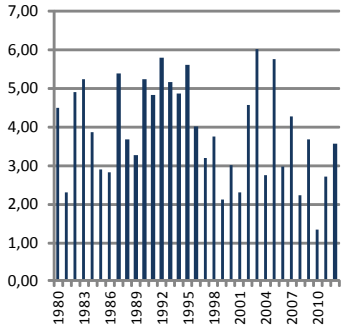
# Mexico: RYVEMCE, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

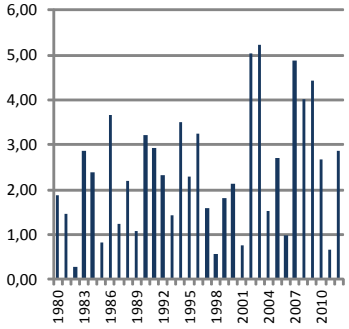
**Anorectal atresia/stenosis**



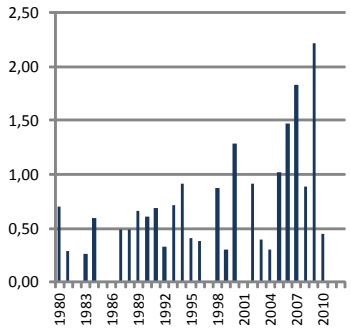

**Hypospadias**



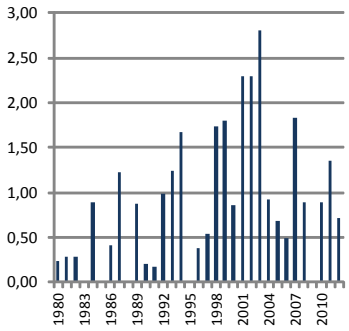

**Indeterminate sex**



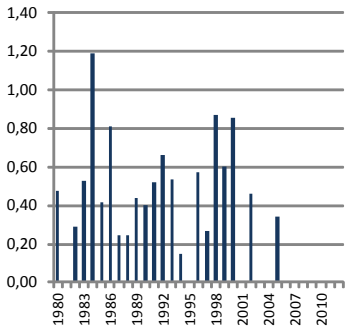

**Renal agenesis**



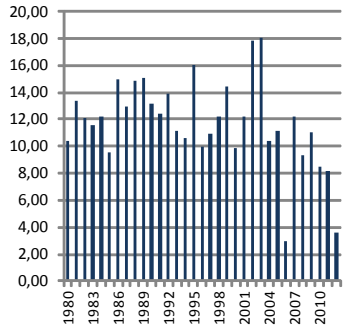

**Cystic kidney**



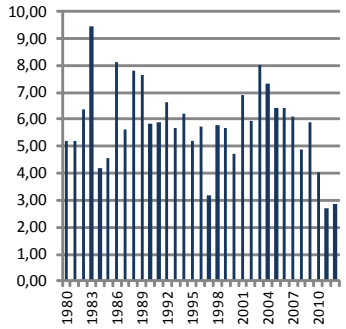

**Bladder exstrophy**



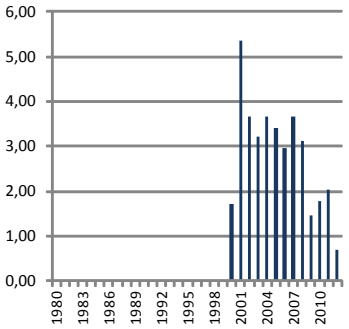

**Polydactyly, preaxial**



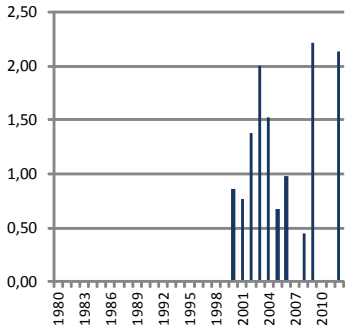

**Limb reduction defects**



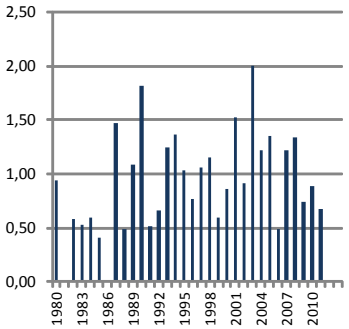

**Limb reduction defects - transverse**



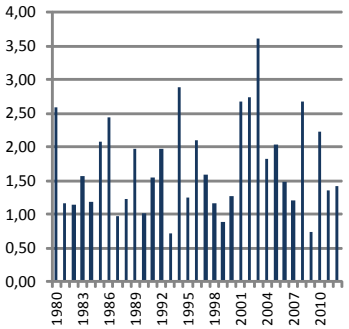

**Limb reduction defects - preaxial**




**Diaphragmatic hernia**



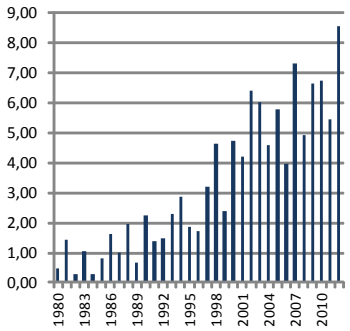

**Omphalocele**



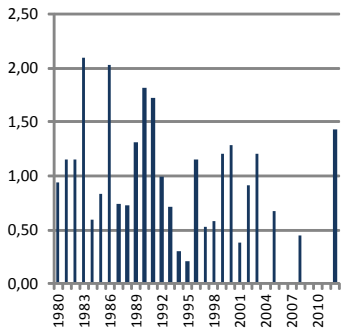
# Mexico: RYVEMCE, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

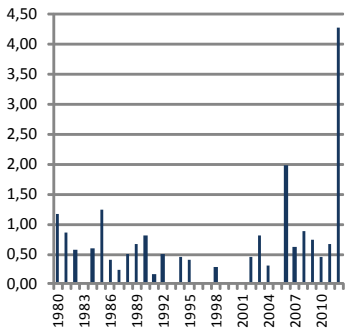
**Gastroschisis**



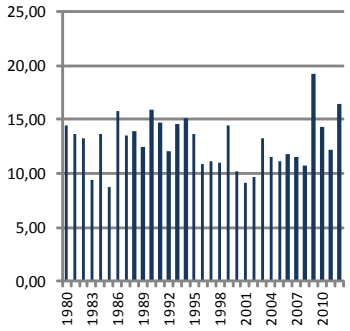

**Prune belly sequence**



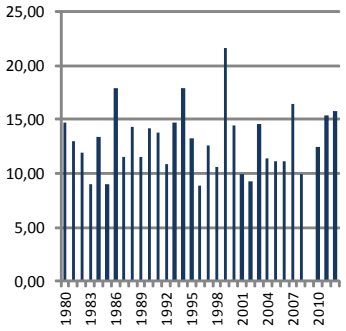

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates**

# Mexico – Monterrey, Nuevo León: BDSP

Birth Defects Surveillance Program

**History:**
Before 1999, death certificates were the only reliable data regarding birth defects. After the recognition of the significant incidence of neural tube defects (NTD) in our region, an NTD surveillance committee was created for immediate notification and registration of cases with the participation of public and private hospitals. In September 2010, a Collaborative Registry of the sub-committee for surveillance and prevention of birth defects was established with the objective of reporting birth defects in newborns and stillbirhs. We became members of the ICBDRS in 2014.

**Size and coverage:**
The number of participating hospitals has grown from 11 in 2010 to 28 in 2014. This program is population based, involving the main regional hospitals and the coverage is around 86% of all births in Nuevo Leon, Mexico (approximately 4.9 million inhabitants and 87,941 births per year). Stillbirths of 20 weeks or more gestation are being included in our records.

**Legislation and funding:**
The Records are being recognized and supported by the State Secretary of Health of Nuevo Leon.

**Sources and ascertainment:**
The prevalence of main birth defects are obtained from obstetrics and pediatric units, a software was developed for the record of cases, applying standardized questionnaire addressing social and demographic data, family history of genetic diseases and exposure to environment risk factors or teratogens. The cases are being registered according to the ICD-10.

**Addresses and Staff**:
Dr. Laura Martínez - Programme Director
Departamento de Genética y Defectos Congénitos, U.A.N.L.
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Monterrey, Nuevo León, México.
CP 64460
**Phone**: (52)-81-83294217
**Fax**: (52) 81-83294217
**E-mail**: laelmar@yahoo.com.mx

# Mexico – Monterrey, Nuevo León: BDSP, 2013

| | |
|---|---|
| Live births (LB) | 70,531 |
| Stillbirths (SB) | 461 |
| Total births | 70,992 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 10 | 2 | nr | 1.69 |
| Spina bifida | 29 | 0 | nr | 4.08 |
| Encephalocele | 0 | 0 | nr | 0.00 |
| Microcephaly | 3 | 0 | nr | 0.42 |
| Holoprosencephaly | 0 | 0 | nr | 0.00 |
| Hydrocephaly | 11 | 1 | nr | 1.69 |
| Anophthalmos | 0 | 0 | nr | 0.00 |
| Microphthalmos | 1 | 0 | nr | 0.14 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 3 | 0 | nr | 0.42 |
| Microtia | 3 | 0 | nr | 0.42 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 3 | 0 | nr | 0.42 |
| Tetralogy of Fallot | 2 | 0 | nr | 0.28 |
| Hypoplastic left heart syndrome | 3 | 0 | nr | 0.42 |
| Coarctation of aorta | 2 | 0 | nr | 0.28 |
| Choanal atresia, bilateral | 1 | 0 | nr | 0.14 |
| Cleft palate without cleft lip | 1 | 0 | nr | 0.14 |
| Cleft lip with or without cleft palate | 58 | 0 | nr | 8.17 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | nr | 1.13 |
| Small intestine atresia/stenosis | 21 | 0 | nr | 2.96 |
| Anorectal atresia/stenosis | 14 | 0 | nr | 1.97 |
| Undescended testis (36 weeks of gestation or later) | 4 | 0 | nr | 0.56 |
| Hypospadias | 7 | 0 | nr | 0.99 |
| Epispadias | 0 | 0 | nr | 0.00 |
| Indeterminate sex | 4 | 0 | nr | 0.56 |
| Renal agenesis | 2 | 0 | nr | 0.28 |
| Cystic kidney | 3 | 0 | nr | 0.42 |
| Bladder exstrophy | 0 | 0 | nr | 0.00 |
| Polydactyly, preaxial | 1 | 0 | nr | 0.14 |
| Total Limb reduction defects (include unspecified) | 4 | 0 | nr | 0.56 |
| Transverse | 2 | 0 | nr | 0.28 |
| Preaxial | nr | 0 | nr | 0.00 |
| Postaxial | nr | 0 | nr | 0.00 |
| Intercalary | nr | 0 | nr | 0.00 |
| Mixed | nr | 0 | nr | 0.00 |
| Unspecified | 2 | 0 | nr | 0.28 |
| Diaphragmatic hernia | 10 | 1 | nr | 1.55 |
| Omphalocele | 3 | 0 | nr | 0.42 |
| Gastroschisis | 11 | 0 | nr | 1.55 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 1 | 0 | nr | 0.14 |
| Trisomy 13 | 2 | 0 | nr | 0.28 |
| Trisomy 18 | 1 | 0 | nr | 0.14 |
| Down syndrome, all ages (include age unknown) | 50 | 0 | nr | 7.04 |
| <20 | 6 | 0 | nr | 5.29 |
| 20-24 | 13 | 0 | nr | 6.37 |
| 25-29 | 4 | 0 | nr | 2.35 |
| 30-34 | 9 | 0 | nr | 6.61 |
| 35-39 | 13 | 0 | nr | 20.10 |
| 40-44 | 4 | 0 | nr | 26.11 |
| 45+ | 1 | 0 | nr | 105.26 |
| unknown | 0 | 0 | nr | 0.00 |

nr = data not reported or not available

# New Zealand

New Zealand Birth Defects Registry

**History:**
The Registry (previously the New Zealand Birth Defects Monitoring Programme) began in 1975 and became a full member of the ICBDSR in 1979.

**Size and coverage:**
The Registry covers all livebirths (approximately 60,000 per year) delivered or treated in a New Zealand publicly funded hospital. Only these data are included in annual reports to the ICBDSR. Data on fetal deaths are included in the database together with additional cases derived from the national perinatal and mortality databases. In late 1995 the definition of fetal death stillbirth was changed from 28 weeks completed gestation to 20 weeks or more gestation and/or 400g birthweight. A voluntary system for the registration of birth defects in terminations of pregnancy was implemented in 2011, but other options for ascertainment are currently being explored.

**Legislation and funding:**
The NZBDR is operated by Centre for Public Health Research, Massey University, with funding from the Ministry of Health.

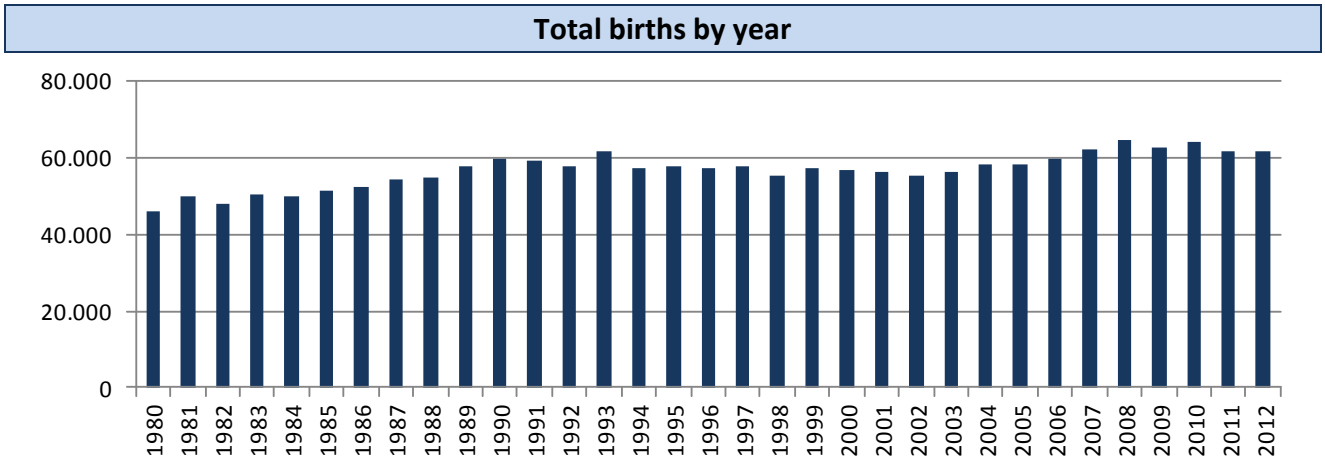
**Sources of ascertainment:**
Ascertainment is from discharge records of publicly funded hospitals, fetal death notification forms, and terminations of pregnancy.

**Exposure information:**
Limited exposure information are currently available.

**Background information:**
General epidemiological characteristics for all births are available.

**Addresses and Staff:**
Associate Professor Barry Borman
Director
New Zealand Birth Defects Registry
Centre for Public Health Research
Massey University
PO Box 756
Wellington 6140, New Zealand
**Tel:** +64-4-801-4985
**Fax:** +64-4-380-0600
**Email:** b.borman@massey.ac.nz

## New Zealand


Total births by year
80.000
60.000
40.000
20.000
0
1980
1981
1982
1983
1984
1985
1986
1987
1988
1989
1990
1991
1992
1993
1994
1995
1996
1997
1998
1999
2000
2001
2002
2003
2004
2005
2006
2007
2008
2009
2010
2011
2012

# New Zealand, 2012

| | |
|---|---|
| Live births (LB) | 61,178 |
| Stillbirths (SB) | 390 |
| Total births | 61,568 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 2 | nr | nr | 0.32 |
| Spina bifida | 4 | nr | nr | 0.65 |
| Encephalocele | 2 | nr | nr | 0.32 |
| Microcephaly | 9 | nr | nr | 1.46 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | 12 | nr | nr | 1.95 |
| Anophthalmos | 0 | nr | nr | 0.00 |
| Microphthalmos | 3 | nr | nr | 0.49 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 15 | nr | nr | 2.44 |
| Tetralogy of Fallot | 16 | nr | nr | 2.60 |
| Hypoplastic left heart syndrome | 4 | nr | nr | 0.65 |
| Coarctation of aorta | 8 | nr | nr | 1.30 |
| Choanal atresia, bilateral | 4 | nr | nr | 0.65 |
| Cleft palate without cleft lip | 28 | nr | nr | 4.55 |
| Cleft lip with or without cleft palate | 28 | nr | nr | 4.55 |
| Oesophageal atresia/stenosis with or without fistula | 7 | nr | nr | 1.14 |
| Small intestine atresia/stenosis | 6 | nr | nr | 0.97 |
| Anorectal atresia/stenosis | 10 | nr | nr | 1.62 |
| Undescended testis (36 weeks of gestation or later) | 71 | nr | nr | 11.53 |
| Hypospadias | 83 | nr | nr | 13.48 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | 1 | nr | nr | 0.16 |
| Renal agenesis | 14 | nr | nr | 2.27 |
| Cystic kidney | 18 | nr | nr | 2.92 |
| Bladder exstrophy | 0 | nr | nr | 0.00 |
| Polydactyly, preaxial | 24 | nr | nr | 3.90 |
| Total Limb reduction defects (include unspecified) | 10 | nr | nr | 1.62 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 8 | nr | nr | 1.30 |
| Omphalocele | 21 | nr | nr | 3.41 |
| Gastroschisis | nr | nr | nr | nr |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 2 | nr | nr | 0.32 |
| Trisomy 18 | 4 | nr | nr | 0.65 |
| Down syndrome, all ages (include age unknown) | 35 | nr | nr | 5.68 |
| <20 | nr | nr | nr | nr |
| 20-24 | nr | nr | nr | nr |
| 25-29 | nr | nr | nr | nr |
| 30-34 | nr | nr | nr | nr |
| 35-39 | nr | nr | nr | nr |
| 40-44 | nr | nr | nr | nr |
| 45+ | nr | nr | nr | nr |
| unknown | nr | nr | nr | nr |

nr = data not reported or not available

# New Zealand, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **95,943** | **252,560** | **286,647** | **291,957** | **283,638** | **288,243** | **316,274** |
| Anencephaly | | 6.36 | 4.12 | 1.71 | 0.55 | 0.42 | 0.45 | 0.35 |
| Spina bifida | | 11.67 | 9.38 | 5.13 | 3.84 | 2.71 | 2.12 | 2.12 |
| Encephalocele | | nr | 0.69* | 0.73* | 0.26* | 0.39 | 0.42 | 0.60 |
| Microcephaly | | nr | nr | nr | 0.70* | 3.31 | 2.84 | 2.40 |
| Holoprosencephaly | | nr | nr | nr | nr | nr | nr | 0.00 |
| Hydrocephaly | | 5.63 | 3.60 | 2.72 | 3.32 | 3.74 | 3.26 | 3.32 |
| Anophthalmos | | nr | nr | nr | 0.00* | 0.00 | 0.09* | 0.13 |
| Microphthalmos | | nr | nr | nr | 0.52* | 0.71 | 0.83* | 0.41 |
| Unspecified Anophthalmos/Microphthalmos | | nr | nr | nr | 0.00* | 0.00 | 0.10 | 0.00* |
| Anotia | | nr | nr | nr | nr | nr | nr | nr |
| Microtia | | nr | nr | nr | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | | nr | nr | nr | nr | nr | nr | 0.81* |
| Transposition of great vessels | | nr | nr | 0.55* | 5.92* | 4.79 | 4.82 | 5.15 |
| Tetralogy of Fallot | | nr | nr | nr | nr | 4.56* | 4.37 | 4.11 |
| Hypoplastic left heart syndrome | | nr | nr | 0.82 | 1.48* | 1.45 | 1.04 | 1.11 |
| Coarctation of aorta | | nr | nr | nr | nr | 2.39* | 3.23 | 2.34 |
| Choanal atresia, bilateral | | nr | nr | nr | 0.52* | 1.13 | 1.08 | 0.92 |
| Cleft palate without cleft lip | | 5.73 | 6.89 | 6.35 | 5.27 | 9.70 | 9.78 | 7.84 |
| Cleft lip with or without cleft palate | | 9.17 | 9.11 | 6.87 | 2.67 | 5.78 | 6.59 | 5.63 |
| Oesophageal atresia/stenosis with or without fistula | | 1.25 | 2.14 | 1.60 | 2.64 | 1.80 | 1.70 | 1.61* |
| Small intestine atresia/stenosis | | nr | nr | nr | 1.74* | 1.80 | 2.36* | 2.37 |
| Anorectal atresia/stenosis | | 1.98 | 2.61 | 2.09 | 3.08 | 2.33 | 2.43 | 1.96 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | nr | 69.03 | 71.47 | 46.19 |
| Hypospadias | | 10.94 | 14.41 | 12.11 | 11.83* | 26.34 | 29.00 | 22.77 |
| Epispadias | | nr | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | | nr | nr | nr | nr | 0.53 | 0.76 | 0.79 |
| Renal agenesis | | nr | 0.13* | 0.64* | nr | 3.35 | 3.09 | 2.69 |
| Cystic kidney | | nr | nr | nr | 5.05* | 6.24 | 5.20 | 5.09 |
| Bladder exstrophy | | nr | nr | nr | 0.17* | 0.48* | 0.14 | 0.16 |
| Polydactyly, preaxial | | nr | nr | nr | 4.88* | 8.52* | 10.20 | 14.58 |
| Total Limb reduction defects (include unspecified) | | 3.44 | 3.80 | 3.10 | 1.75 | 2.61 | 2.88 | 2.51* |
| Transverse | | nr | nr | nr | nr | nr | nr | nr |
| Preaxial | | nr | nr | nr | nr | nr | nr | nr |
| Postaxial | | nr | nr | nr | nr | nr | nr | nr |
| Intercalary | | nr | nr | nr | nr | nr | nr | nr |
| Mixed | | nr | nr | nr | nr | nr | nr | nr |
| Unspecified | | nr | nr | nr | nr | nr | 2.01* | nr |
| Diaphragmatic hernia | | nr | 1.52* | 1.46* | nr | 2.57* | 2.32* | 2.49* |
| Omphalocele | | 2.61 | 2.18 | 1.26 | 3.28* | nr | nr | 4.85 |
| Gastroschisis | | 0.00 | 0.36 | 0.73* | nr | nr | nr | 1.11* |
| Unspecified Omphalocele/Gastroschisis | | 0.00 | 0.24 | 0.18* | nr | nr | 5.01* | 0.95* |
| Prune belly sequence | | nr | nr | nr | nr | nr | nr | nr |
| Trisomy 13 | | nr | nr | nr | 0.35* | 0.42 | 0.59 | 0.35 |
| Trisomy 18 | | nr | nr | nr | 0.70* | 1.09 | 1.28 | 1.01 |
| Down syndrome, all ages (include age unknown) | | 8.76 | 9.23 | 9.14 | 9.33* | 11.88 | 10.96 | 9.07 |
| <20 | | 2.55 | 7.43 | 4.32* | nr | nr | nr | nr |
| 20-24 | | 6.32 | 3.86 | 1.40* | nr | nr | nr | nr |
| 25-29 | | 8.75 | 8.61 | 7.92* | nr | nr | nr | nr |
| 30-34 | | 8.75 | 9.78 | 9.29* | nr | nr | nr | nr |
| 35-39 | | 34.79 | 31.66 | 34.31* | nr | nr | nr | nr |
| 40-44 | | 65.62 | 102.54 | 452.49* | nr | nr | nr | nr |
| 45+ | | 0.00 | 215.83 | 0.00* | nr | nr | nr | nr |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

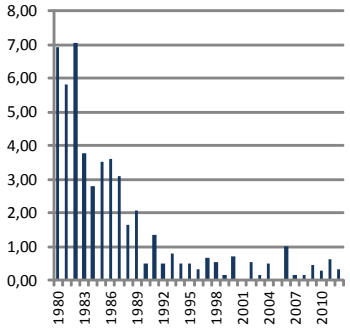
nr = data not reported or not available

* data include less than 5 years

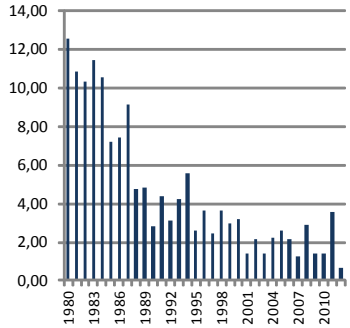
# New Zealand, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

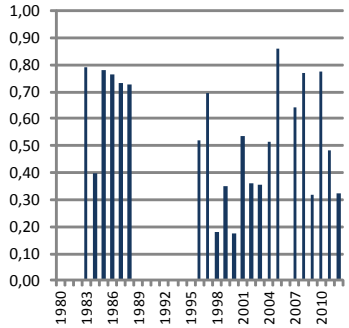
**Anencephaly**



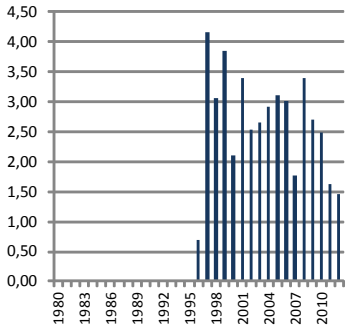

**Spina Bifida**



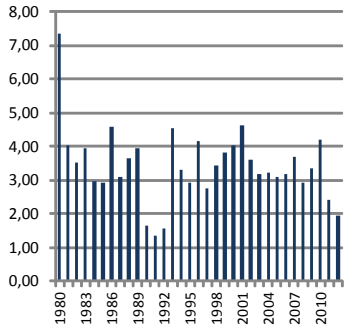

**Encephalocele**



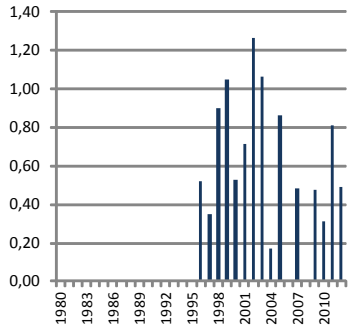

**Microcephaly**



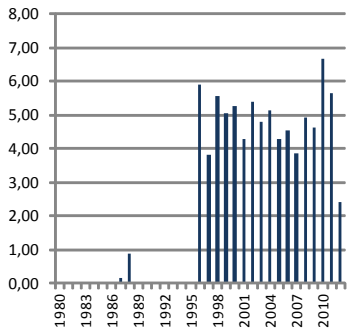

**Hydrocephaly**



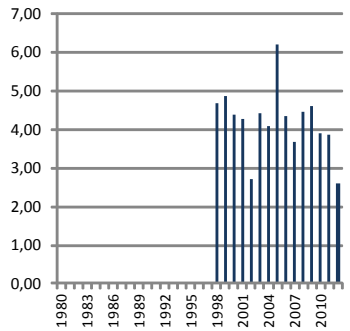

**Microphthalmos**



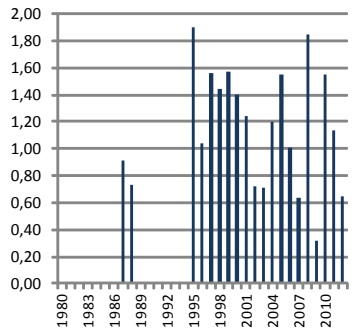

**Transposition of great vessels**



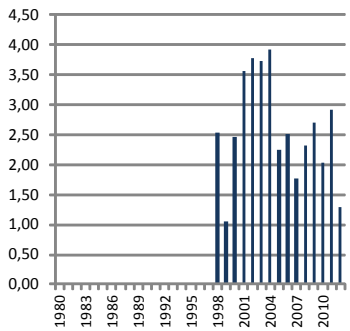

**Tetralogy of Fallot**



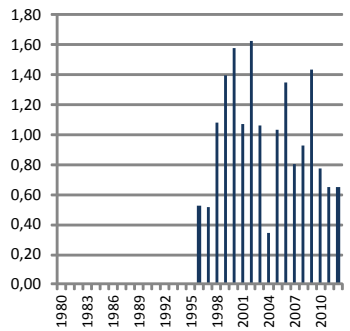

**Hypoplastic left heart syndrome**



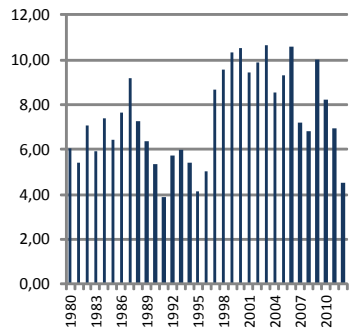

**Coarctation of aorta**




**Choanal atresia, bilateral**



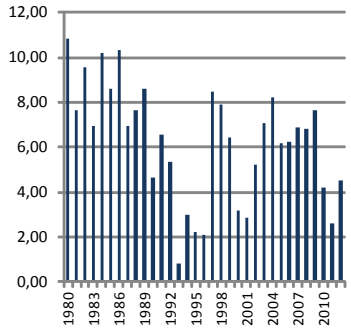

**Cleft palate without cleft lip**



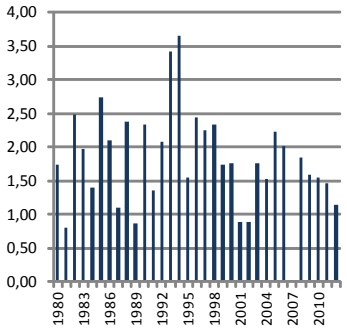
# New Zealand, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

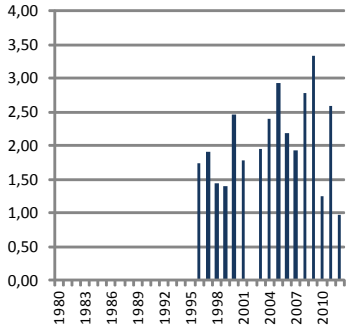
**Cleft lip with or without cleft palate**



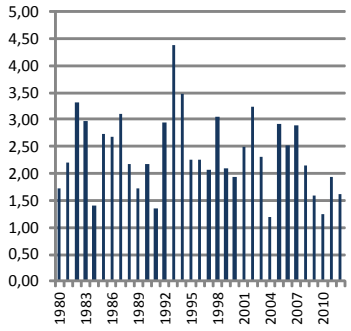

**phageal atresia/stenosis with or without f**



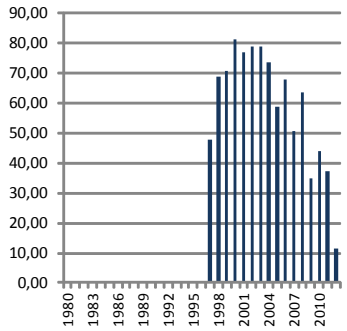

**Small intestine atresia/stenosis**



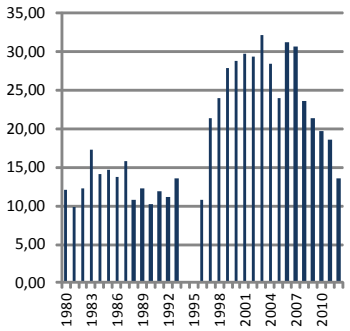

**Anorectal atresia/stenosis**



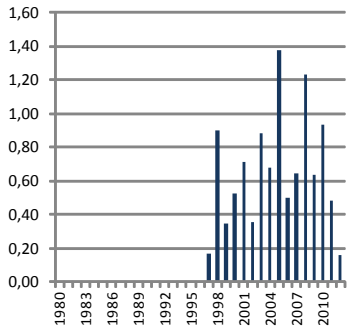

**Undescended testis**



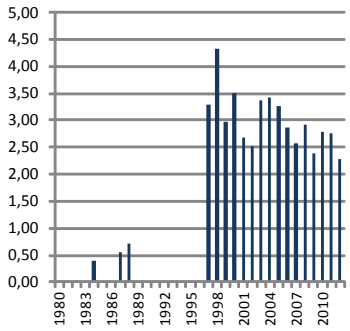

**Hypospadias**



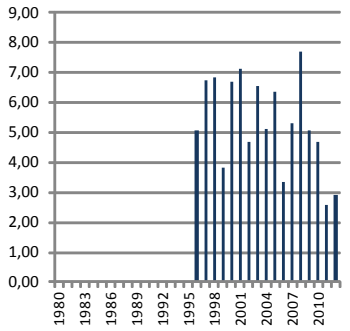

**Indeterminate sex**



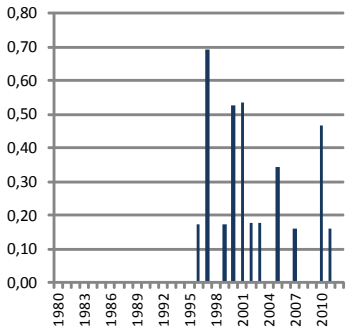

**Renal agenesis**



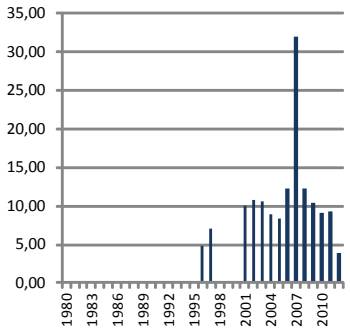

**Cystic kidney**



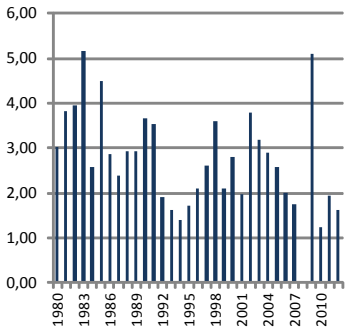

**Bladder exstrophy**




**Polydactyly, preaxial**



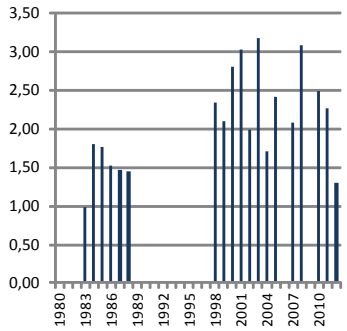

**Limb reduction defects**



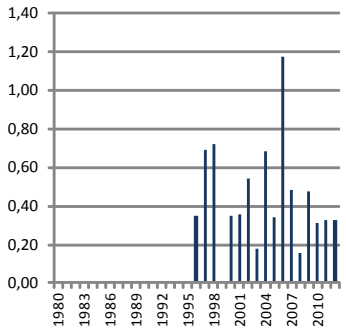
# New Zealand, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

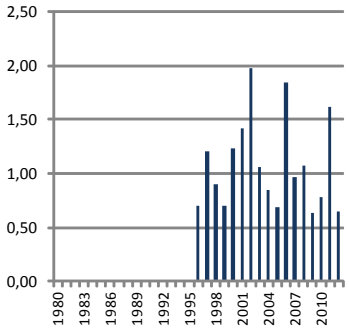
**Diaphragmatic hernia**



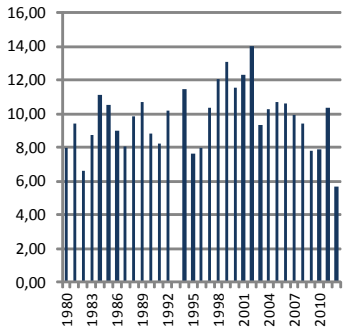

**Trisomy 13**




**Trisomy 18**




**Down Syndrome**




**L + S rates**

## Northern Netherlands

EUROCAT Registry Northern Netherlands

**History*:***
The Programme started in 1981, and became a Clearinghouse member in 1993.

**Size and coverage:**
In the beginning the Programme covered 7,500 births annually in the province of Groningen and northern Drenthe. Coverage was gradually increased to 20,000 births annually in the provinces Groningen, Friesland and Drenthe from 1989 onwards (10% of the Netherlands). Home deliveries (35% of births) are included.

**Legislation and funding:**
The Programme is funded by the Dutch Ministry of Public Health, Welfare and Sports. The registry is carried out in the Department of Genetics of the University Medical Center Groningen of the University of Groningen.

**Sources of ascertainment:**
Children and foetuses with congenital anomalies are reported on a voluntary basis by various sources: obstetricians, pediatricians, clinical geneticists, surgeons, general practitioners, midwives, well-baby clinics, pathologists and the national obstetric registry. Registry personnel is also actively involved in data collection. Children and foetuses with congenital anomalies diagnosed before or after birth are eligible for registration at the EUROCAT registry, if the mother lived in the region at the time of birth and the child has not reached the age of 10 at notification. There is no lower limit for gestational age. Spontaneous and induced abortions are included. A number of frequently occurring mild anomalies is not registered, unless they occur in combination with other congenital anomalies. Informed consent of the parents is needed.

**Exposure information:**
Since 1997 parents are asked to fill out a questionnaire including questions on occupational activities and medication use. Besides, pharmacy data are collected routinely and the actual use of the reported medications is verified with the mother.

**Background information:**
General statistics are available from the Dutch Central Bureau of Statistics (CBS).

**Addresses and Staff:**
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Department of Genetics University Medical Centre Groningen University of Groningen
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9700 RB Groningen, The Netherlands
Phone: 31-50-3617110 / 3617115
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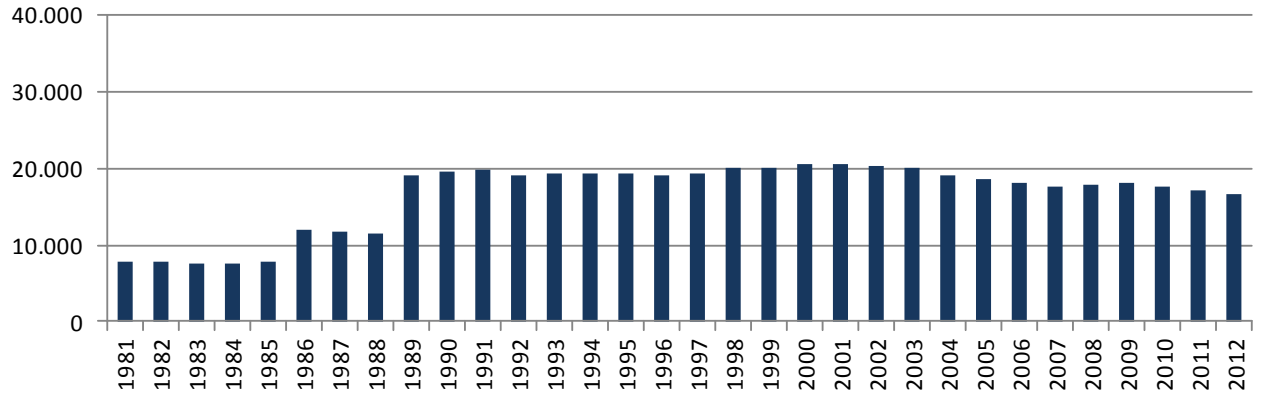
**E-mail:** m.k.bakker@umcg.nl

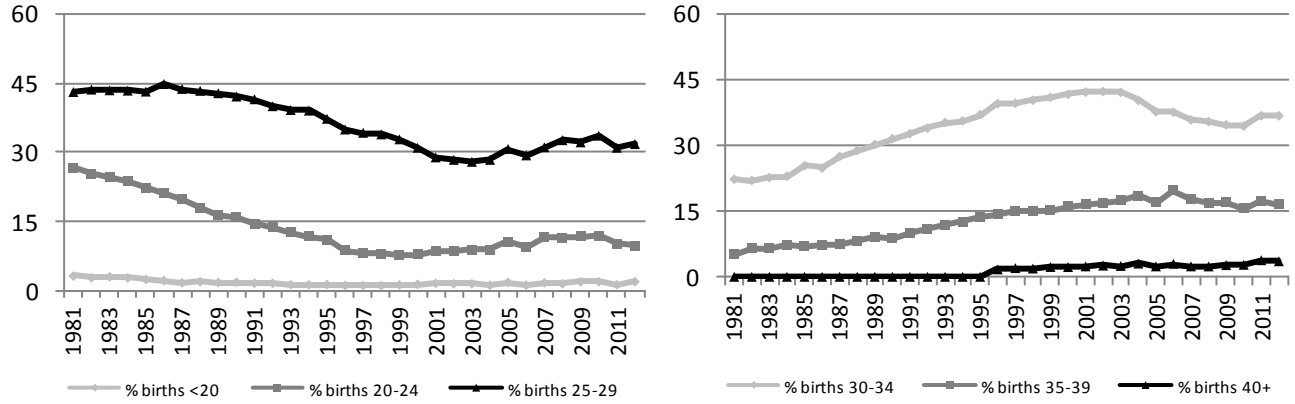
Hermien de Walle
**E-mail:** h.e.k.de.Walle@umcg.nl
**Website:** www.eurocatnederland.nl

# Northern Netherlands

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 21 | 87.5 | Cystic kidney | 12 | 29.3 |
| Spina bifida | 15 | 60.0 | Limb reduction defects | 13 | 30.2 |
| Encephalocele | 1 | 50.0 | Diaphragmatic hernia | 3 | 18.8 |
| Holoprosencephaly | 7 | 87.5 | Omphalocele | 12 | 54.5 |
| Hydrocephaly | 12 | 54.5 | Gastroschisis | 4 | 44.4 |
| Hypoplastic left heart syndrome | 14 | 58.3 | Trisomy 13 | 8 | 66.7 |
| Cleft palate without cleft lip | 1 | 5.0 | Trisomy 18 | 30 | 76.9 |
| Cleft lip with or without cleft palate | 10 | 14.5 | Down syndrome | 44 | 41.9 |
| Renal agenesis | 7 | 31.8 | | | |

Total ToPs with births defects = 245 (Ratio ToPs/Births: 4.77 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Northern Netherlands, 2012

| | |
|---|---|
| Live births (LB) | 16,512 |
| Stillbirths (SB) | 70 |
| Total births | 16,582 |
| Number of terminations of pregnancy (ToP) for birth defects | 70 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 5 | 3.02 |
| Spina bifida | 2 | 0 | 5 | 4.22 |
| Encephalocele | 0 | 0 | 0 | 0.00 |
| Microcephaly | 3 | 0 | 0 | 1.81 |
| Holoprosencephaly | 0 | 0 | 3 | 1.81 |
| Hydrocephaly | 0 | 0 | 5 | 3.02 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 0 | 0 | 0 | 0.00 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr |
| Anotia | 2 | 0 | 0 | 1.21 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 9 | 0 | 0 | 5.43 |
| Tetralogy of Fallot | 3 | 0 | 1 | 2.41 |
| Hypoplastic left heart syndrome | 4 | 1 | 4 | 5.43 |
| Coarctation of aorta | 4 | 0 | 1 | 3.02 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 1.21 |
| Cleft palate without cleft lip | 4 | 1 | 0 | 3.02 |
| Cleft lip with or without cleft palate | 16 | 0 | 4 | 12.06 |
| Oesophageal atresia/stenosis with or without fistula | 7 | 0 | 0 | 4.22 |
| Small intestine atresia/stenosis | 2 | 0 | 0 | 1.21 |
| Anorectal atresia/stenosis | 3 | 0 | 4 | 4.22 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 29 | 0 | 1 | 18.09 |
| Epispadias | 1 | 0 | 0 | 0.60 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 3 | 1 | 3 | 4.22 |
| Cystic kidney | 9 | 0 | 4 | 7.84 |
| Bladder exstrophy | 0 | 0 | 1 | 0.60 |
| Polydactyly, preaxial | 7 | 0 | 1 | 4.82 |
| Total Limb reduction defects (include unspecified) | 10 | 0 | 6 | 9.65 |
| Transverse | 5 | 0 | 6 | 6.63 |
| Preaxial | 1 | 0 | 1 | 1.21 |
| Postaxial | 3 | 0 | 0 | 1.81 |
| Intercalary | 0 | 0 | 0 | 0.00 |
| Mixed | 3 | 0 | 1 | 2.41 |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 4 | 0 | 2 | 3.62 |
| Omphalocele | 3 | 1 | 7 | 6.63 |
| Gastroschisis | 1 | 0 | 1 | 1.21 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 2 | 1.21 |
| Trisomy 18 | 2 | 1 | 10 | 7.84 |
| Down syndrome, all ages (include age unknown) | 23 | 1 | 9 | 19.90 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 0 | 0 | 0 | 0.00 |
| 25-29 | 6 | 0 | 1 | 13.30 |
| 30-34 | 7 | 1 | 0 | 13.13 |
| 35-39 | 6 | 0 | 5 | 40.18 |
| 40-44 | 4 | 0 | 3 | 123.89 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | nr | nr | nr | nr |

nr = data not reported or not available

# Northern Netherlands, Previous years rates 1981 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **7,877** | **42,560** | **82,023** | **96,178** | **100,583** | **96,266** | **88,511** |
| Anencephaly | | 3.81 | 7.52 | 2.93 | 3.12 | 2.68 | 2.08 | 3.95 |
| Spina bifida | | 6.35 | 5.64 | 8.66 | 5.61 | 4.87 | 4.88 | 4.86 |
| Encephalocele | | 0.00 | 2.11 | 0.49 | 1.25 | 0.99 | 0.73 | 0.68 |
| Microcephaly | | 3.81 | 4.70 | 3.54 | 3.95 | 4.47 | 0.93 | 2.94 |
| Holoprosencephaly | | 2.54 | 0.94 | 1.22 | 0.62 | 1.19 | 1.14 | 0.90 |
| Hydrocephaly | | 1.27 | 5.40 | 2.93 | 3.74 | 3.78 | 3.64 | 4.63 |
| Anophthalmos | | 0.00 | 0.23 | 0.37 | 0.31 | 0.00 | 0.00 | 0.23 |
| Microphthalmos | | 2.54 | 1.88 | 1.46 | 2.08 | 1.29 | 0.31 | 1.36 |
| Unspecified Anophthalmos/Microphthalmos | | nr | nr | nr | nr | 0.98* | 1.39 | nr |
| Anotia | | 2.54 | 2.35 | 0.73 | 1.04 | 0.70 | 0.21 | 1.24 |
| Microtia | | 1.27 | 0.70 | 0.98 | 0.83 | 0.80 | 0.83 | 0.34 |
| Unspecified Anotia/Microtia | | nr | nr | nr | nr | nr | nr | nr |
| Transposition of great vessels | | 0.00 | 4.23 | 3.41 | 4.78 | 4.77 | 3.74 | 5.54 |
| Tetralogy of Fallot | | 5.08 | 3.29 | 3.54 | 3.33 | 3.38 | 3.95 | 3.28 |
| Hypoplastic left heart syndrome | | 2.54 | 2.58 | 2.44 | 2.29 | 2.58 | 2.70 | 3.62 |
| Coarctation of aorta | | 6.35 | 5.87 | 5.85 | 6.34 | 4.08 | 5.09 | 3.62 |
| Choanal atresia, bilateral | | 0.00 | 1.41 | 0.73 | 1.56 | 0.80 | 1.04 | 0.56 |
| Cleft palate without cleft lip | | 8.89 | 6.34 | 6.71 | 7.59 | 7.95 | 8.31 | 6.44 |
| Cleft lip with or without cleft palate | | 15.23 | 17.39 | 14.14 | 15.80 | 14.12 | 14.23 | 13.22 |
| Oesophageal atresia/stenosis with or without fistula | | 1.27 | 2.82 | 2.68 | 3.12 | 3.88 | 3.95 | 1.47 |
| Small intestine atresia/stenosis | | 3.81 | 2.58 | 2.56 | 3.02 | 2.19 | 1.45 | 2.03 |
| Anorectal atresia/stenosis | | 1.27 | 2.82 | 3.66 | 2.91 | 3.88 | 3.53 | 5.42 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | nr | nr | nr | nr |
| Hypospadias | | 19.04 | 13.63 | 9.88 | 10.71 | 16.70 | 21.30 | 22.48 |
| Epispadias | | 0.00 | 0.23 | 0.73 | 0.52 | 0.60 | 0.42 | 0.79 |
| Indeterminate sex | | 0.00 | 0.23 | 0.24 | 0.10 | 0.50 | 0.52 | 0.79 |
| Renal agenesis | | 3.81 | 3.99 | 4.27 | 4.78 | 4.87 | 4.57 | 5.20 |
| Cystic kidney | | 2.54 | 2.11 | 6.34 | 4.68 | 3.48 | 5.92 | 7.23 |
| Bladder exstrophy | | 0.00 | 0.23 | 0.24 | 0.10 | 0.20 | 0.42 | 0.68 |
| Polydactyly, preaxial | | 0.00 | 2.82 | 1.58 | 2.08 | 2.49 | 0.62 | 1.69 |
| Total Limb reduction defects (include unspecified) | | 8.89 | 7.28 | 5.49 | 7.17 | 5.57 | 6.65 | 6.33 |
| Transverse | | 5.08 | 4.46 | 2.68 | 4.05 | 3.38 | 4.57 | 4.63 |
| Preaxial | | 1.27 | 1.41 | 0.61 | 1.04 | 0.60 | 1.14 | 1.13 |
| Postaxial | | 2.54 | 0.47 | 1.22 | 1.66 | 0.80 | 0.00 | 1.92 |
| Intercalary | | 1.27 | 0.00 | 0.00 | 0.31 | 0.20 | 0.21 | 0.23 |
| Mixed | | 1.27 | 0.23 | 0.24 | 0.42 | 0.30 | 0.52 | 2.03 |
| Unspecified | | nr | nr | nr | nr | nr | nr | nr |
| Diaphragmatic hernia | | 2.54 | 2.35 | 2.68 | 2.60 | 2.98 | 2.70 | 2.49 |
| Omphalocele | | 2.54 | 1.41 | 2.93 | 3.22 | 1.89 | 1.97 | 2.60 |
| Gastroschisis | | 1.27 | 1.17 | 0.49 | 0.42 | 1.19 | 0.93 | 1.81 |
| Unspecified Omphalocele/Gastroschisis | | nr | nr | nr | nr | nr | nr | nr |
| Prune belly sequence | | 0.00 | 0.23 | 0.49 | 0.42 | 0.50 | 0.10 | 0.11 |
| Trisomy 13 | | 0.00 | 0.94 | 1.22 | 1.35 | 0.89 | 1.25 | 2.60 |
| Trisomy 18 | | 3.81 | 1.88 | 2.56 | 2.08 | 3.38 | 5.82 | 7.46 |
| Down syndrome, all ages (include age unknown) | | 10.16 | 13.16 | 14.14 | 14.66 | 15.31 | 16.72 | 18.53 |
| <20 | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| 20-24 | | 9.58 | 8.12 | 8.15 | 8.14 | 2.47 | 6.79 | 5.00 |
| 25-29 | | 5.90 | 8.05 | 11.19 | 4.65 | 10.23 | 9.37 | 7.75 |
| 30-34 | | 11.42 | 14.88 | 11.18 | 15.20 | 9.47 | 10.65 | 15.64 |
| 35-39 | | 49.38 | 44.20 | 38.51 | 36.14 | 37.60 | 33.09 | 41.44 |
| 40-44 | | nr | nr | nr | 160.77* | 91.00 | 120.63 | 83.72 |
| 45+ | | nr | nr | nr | 0.00* | 149.25 | 121.95 | 857.14 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

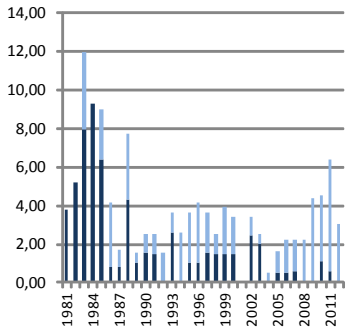
nr = data not reported or not available

* data include less than 5 years

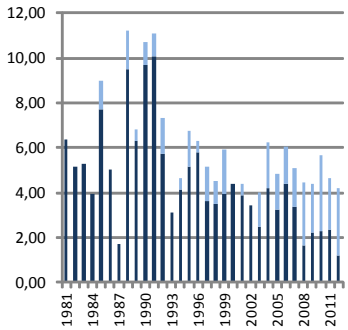
# Northern Netherlands, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

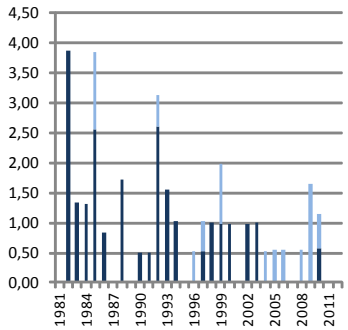
### Anencephaly



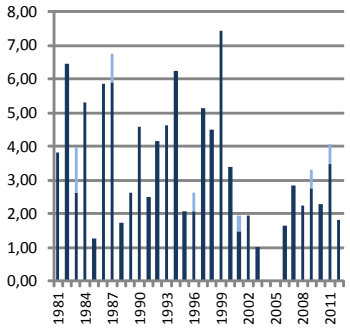

### Spina Bifida



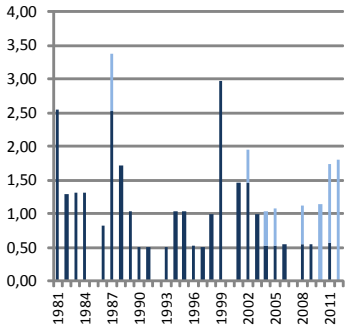

### Encephalocele



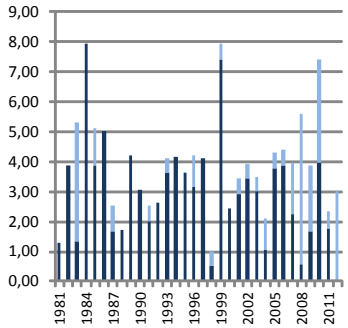

### Microcephaly



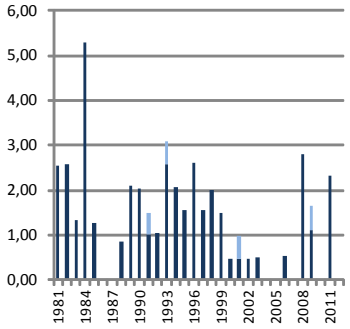

### Holoprosencephaly



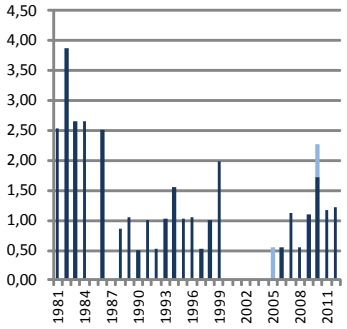

### Hydrocephaly



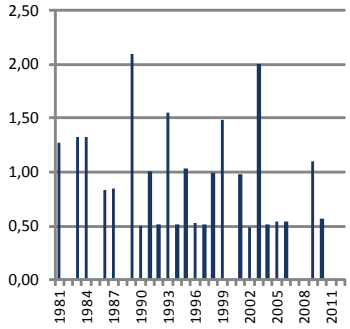

### Microphthalmos



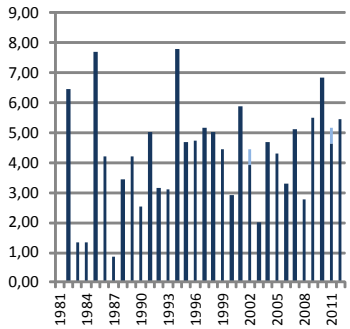

### Anotia



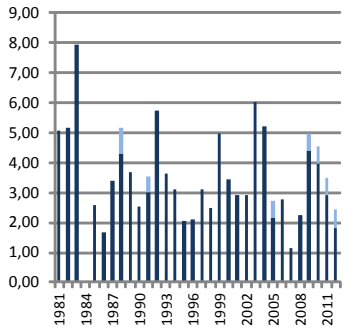

### Microtia



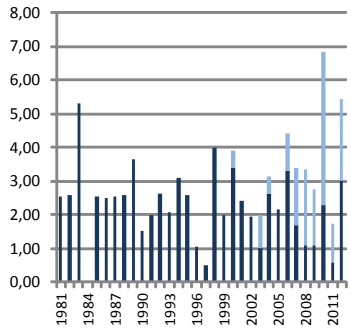

### Transposition of great vessels




### Tetralogy of Fallot



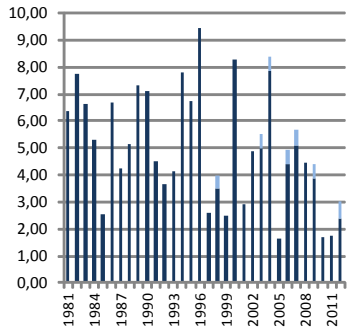

### Hypoplastic left heart syndrome



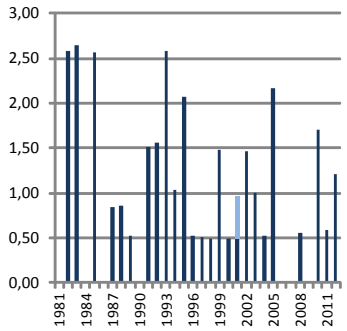
# Northern Netherlands, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

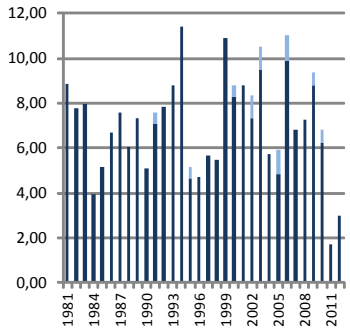
**Coarctation of aorta**



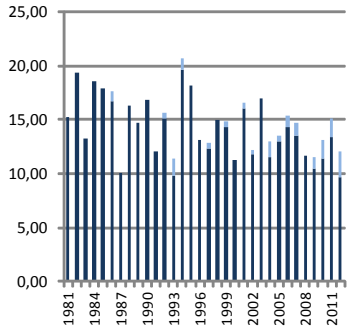

**Choanal atresia, bilateral**



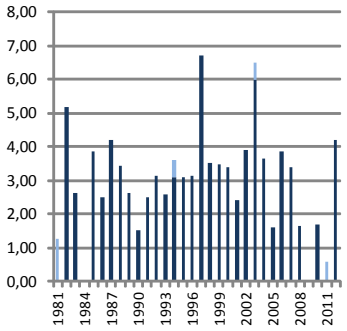

**Cleft palate without cleft lip**



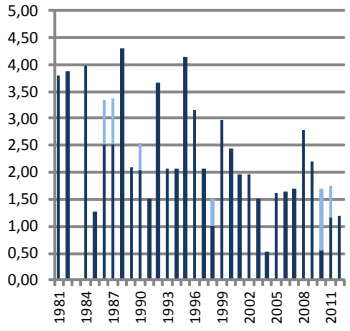

**Cleft lip with or without cleft palate**



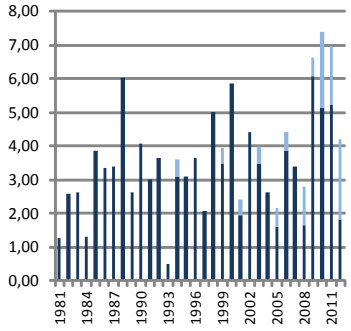

**phageal atresia/stenosis with or without f**



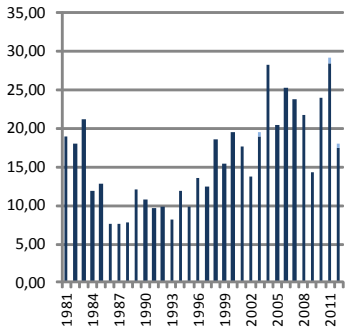

**Small intestine atresia/stenosis**



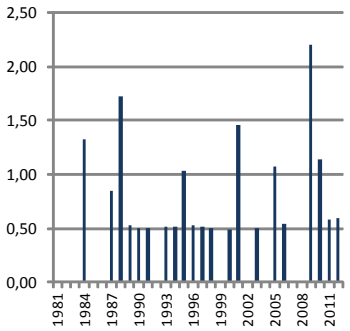

**Anorectal atresia/stenosis**



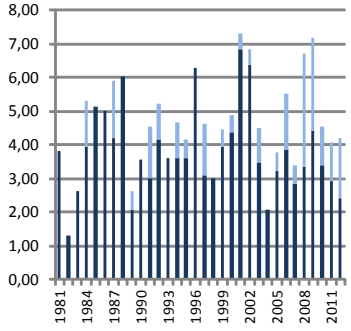

**Hypospadias**



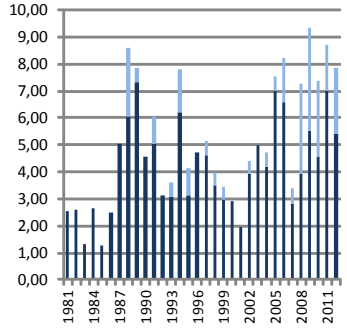

**Epispadias**



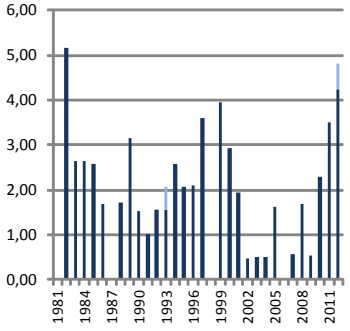

**Renal agenesis**




**Cystic kidney**



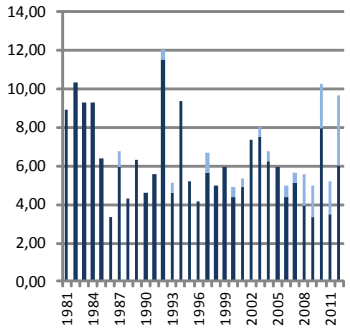

**Polydactyly, preaxial**



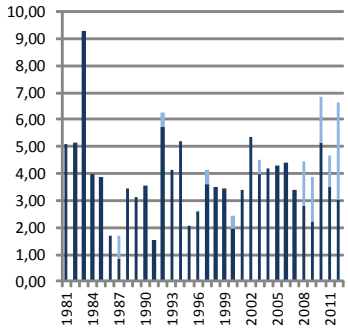
# Northern Netherlands, Time trends 1981 – 2012

(Birth prevalence rates per 10,000)

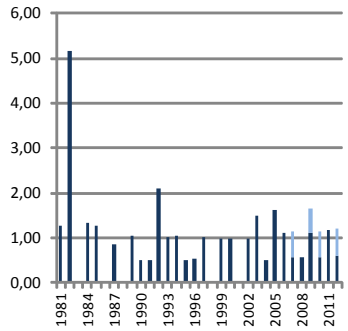
**Limb reduction defects**



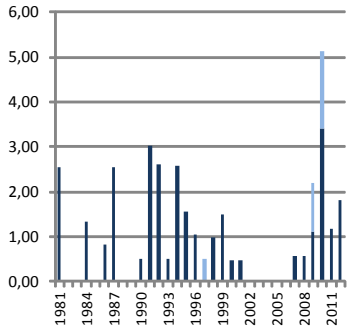

**Limb reduction defects - transverse**



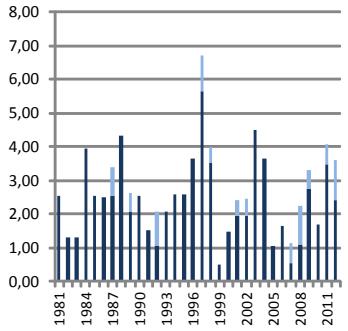

**Limb reduction defects - preaxial**



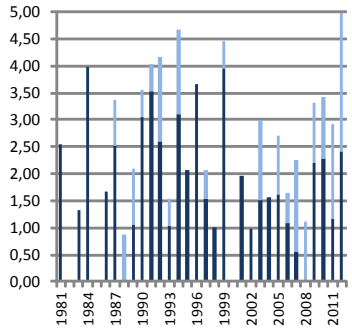

**Limb reduction defects - postaxial**



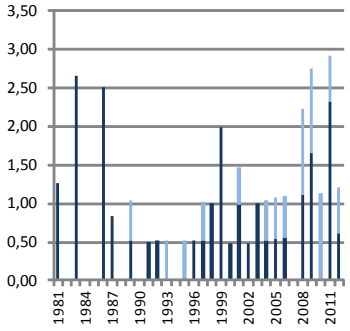

**Diaphragmatic hernia**



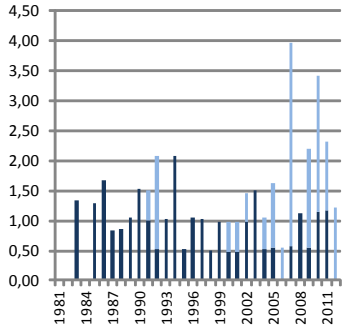

**Omphalocele**



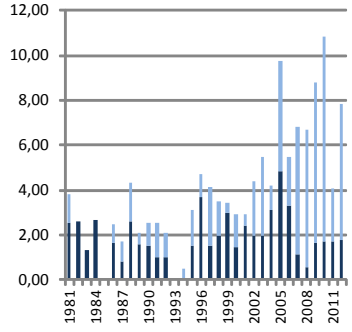

**Gastroschisis**



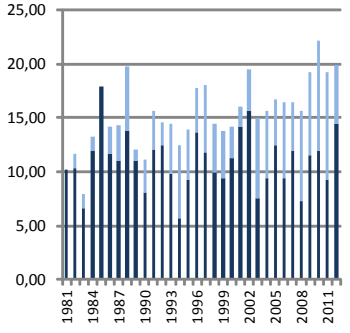

**Trisomy 13**



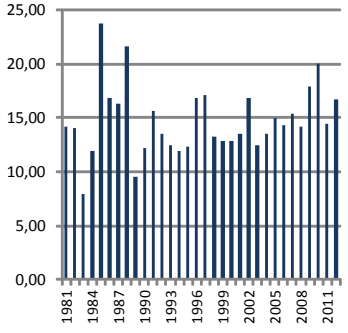

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Slovak Republic

Teratologic Information Centre, Slovak Medical University in Bratislava

**History:**
All "Reports on Birth Defects" from neonatal clinics in Slovakia receives and processes the National Health Information Centre of SR (NHIC). The obligation to report all categories of congenital malformations results from valid legislative standards. Reporting of congenital malformations began in 1964.The Programme of Slovak Teratological Information Center (STIC) was established in 2003 year and consists in cooperation of the Slovak Medical University, NHIC and the Centers of Medical Genetics or neonatal clinics. Work on research projects with the issue of congenital malformation and collaboration with Departmens of Clinical Genetics in Slovakia started from 1995 year, under the responsibility of Dr. Elena Szabova, PhD.

**Size and coverage:**
The registry covers all births in about 55. 000-60. 000 births per year, received from NHIC, by the Reports of birth defects". The detailed information about cases of CM are collected in the Centers of Medical Genetics, or under the running research projects at the Slovak Medical University.

**Legislation and funding:**
Reporting is compulsory. Analysis of data is supported only by grant projects.

**Sources of ascertainment:**
Reports are received from NHIC, delivery units, neonatal, pediatric clinics, or departments of clinical genetics.

**Exposure Information:**
Detailed information on maternal and paternal occupation, drug use, etc. are collected by interviews only according to the running research projects.

**Background information:**
Some background information is available from the general population statistics.

**Addresses and Staff:**
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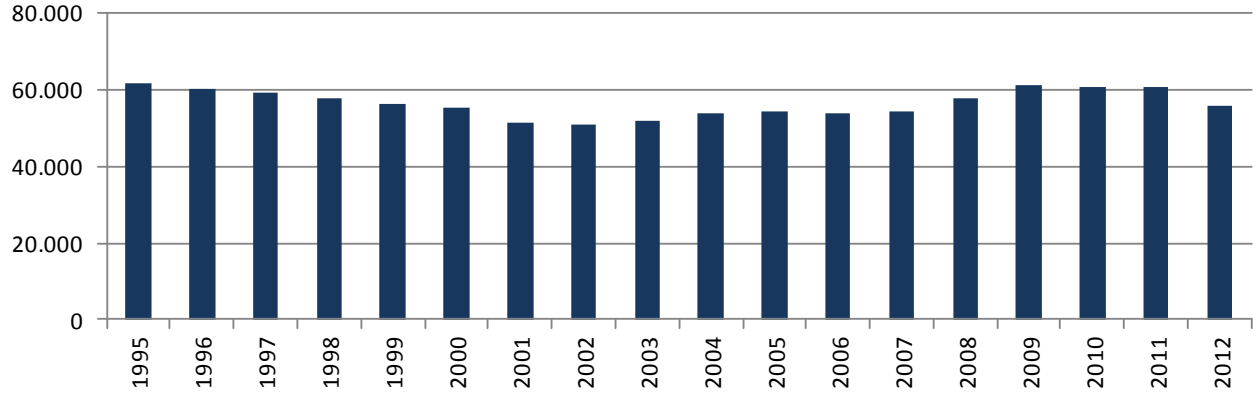
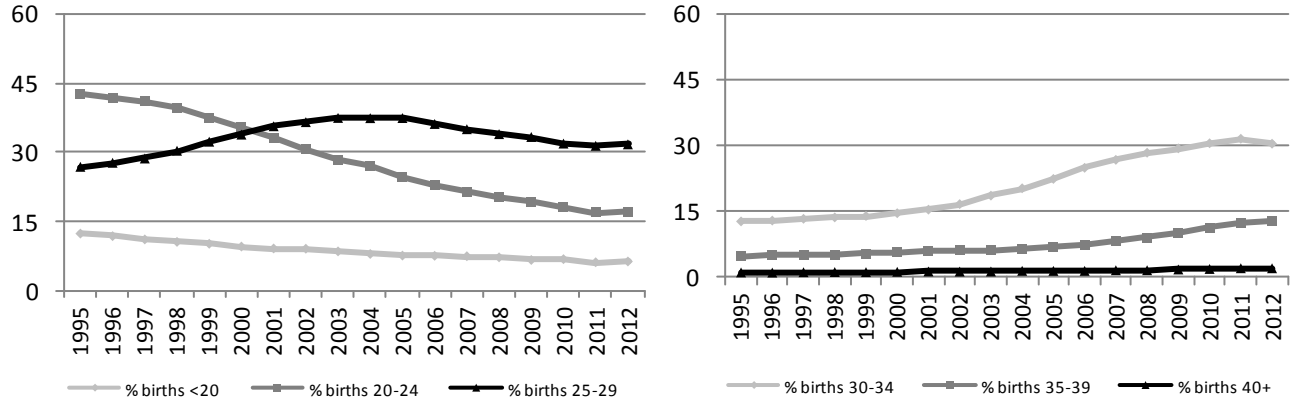
Programme Director
Slovak Teratologic Information Centre
Slovak Medical University
Limbova 12
833 03 Bratislava, Slovak Republic
**Phone:** 00421 2 59370324
**E-mail:** elena.szabova@szu.sk

# Slovak Republic

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 14 | 70.0 | Cystic kidney | 1 | 3.6 |
| Spina bifida | 4 | 10.3 | Limb reduction defects | 0 | 0.0 |
| Encephalocele | 5 | 50.0 | Diaphragmatic hernia | 0 | 0.0 |
| Holoprosencephaly | 2 | 14.3 | Omphalocele | 2 | 13.3 |
| Hydrocephaly | 9 | 15.0 | Gastroschisis | 1 | 6.3 |
| Hypoplastic left heart syndrome | 0 | 0.0 | Trisomy 13 | 0 | 0.0 |
| Cleft palate without cleft lip | 4 | 4.1 | Trisomy 18 | 6 | 46.2 |
| Cleft lip with or without cleft palate | 1 | 0.6 | Down syndrome | 22 | 15.0 |
| Renal agenesis | 1 | 0.9 | | | |

Total ToPs with births defects = 119 (Ratio ToPs/Births: 0.67 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# Slovak Republic, 2012

| | |
|---|---|
| Live births (LB) | 55,535 |
| Stillbirths (SB) | 180 |
| Total births | 55,715 |
| Number of terminations of pregnancy (ToP) for birth defects | 0 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 3 | 0 | 0 | 0.54 |
| Spina bifida | 10 | 0 | 0 | 1.79 |
| Encephalocele | 1 | 0 | 0 | 0.18 |
| Microcephaly | 7 | 0 | 0 | 1.26 |
| Holoprosencephaly | 6 | 0 | 0 | 1.08 |
| Hydrocephaly | 27 | 0 | 0 | 4.85 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 1 | 0 | 0 | 0.18 |
| Unspecified Anophthalmos/Microphthalmos | 3 | 0 | 0 | 0.54 |
| Anotia | 2 | 0 | 0 | 0.36 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 9 | 0 | 0 | 1.62 |
| Tetralogy of Fallot | 11 | 0 | 0 | 1.97 |
| Hypoplastic left heart syndrome | 11 | 0 | 0 | 1.97 |
| Coarctation of aorta | 8 | 0 | 0 | 1.44 |
| Choanal atresia, bilateral | 1 | 0 | 0 | 0.18 |
| Cleft palate without cleft lip | 39 | 0 | 0 | 7.00 |
| Cleft lip with or without cleft palate | 53 | 0 | 0 | 9.51 |
| Oesophageal atresia/stenosis with or without fistula | 10 | 0 | 0 | 1.79 |
| Small intestine atresia/stenosis | 13 | 0 | 0 | 2.33 |
| Anorectal atresia/stenosis | 13 | 0 | 0 | 2.33 |
| Undescended testis (36 weeks of gestation or later) | 144 | 0 | 0 | 25.85 |
| Hypospadias | 176 | 0 | 0 | 31.59 |
| Epispadias | 2 | 0 | 0 | 0.36 |
| Indeterminate sex | 1 | 0 | 0 | 0.18 |
| Renal agenesis | 36 | 0 | 0 | 6.46 |
| Cystic kidney | 12 | 0 | 0 | 2.15 |
| Bladder exstrophy | 1 | 0 | 0 | 0.18 |
| Polydactyly, preaxial | 23 | 0 | 0 | 4.13 |
| Total Limb reduction defects (include unspecified) | 20 | 0 | 0 | 3.59 |
| Transverse | nr | 0 | 0 | nr |
| Preaxial | nr | 0 | 0 | nr |
| Postaxial | nr | 0 | 0 | nr |
| Intercalary | nr | 0 | 0 | nr |
| Mixed | nr | 0 | 0 | nr |
| Unspecified | nr | 0 | 0 | nr |
| Diaphragmatic hernia | 9 | 0 | 0 | 1.62 |
| Omphalocele | 7 | 0 | 0 | 1.26 |
| Gastroschisis | 2 | 0 | 0 | 0.36 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 1 | 0 | 0 | 0.18 |
| Trisomy 13 | 3 | 0 | 0 | 0.54 |
| Trisomy 18 | 0 | 0 | 0 | 0.00 |
| Down syndrome, all ages (include age unknown) | 44 | 0 | 0 | 7.90 |
| <20 | 3 | 0 | 0 | 8.66 |
| 20-24 | 4 | 0 | 0 | 4.24 |
| 25-29 | 7 | 0 | 0 | 3.98 |
| 30-34 | 12 | 0 | 0 | 7.12 |
| 35-39 | 12 | 0 | 0 | 16.89 |
| 40-44 | 6 | 0 | 0 | 57.92 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

# Slovak Republic, Previous years rates 1995 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996* | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | **122,031** | **280,410** | **265,670** | **295,264** |
| Anencephaly | | | | | 0.82 | 0.68 | 0.72 | 1.08 |
| Spina bifida | | | | | 3.77 | 3.46 | 2.75 | 2.24 |
| Encephalocele | | | | | 1.64 | 1.21 | 0.90 | 0.68 |
| Microcephaly | | | | | 1.31 | 1.36 | 0.94 | 0.95 |
| Holoprosencephaly | | | | | 0.00 | 0.29 | 0.38 | 0.37 |
| Hydrocephaly | | | | | 5.74 | 4.96 | 4.52 | 2.61 |
| Anophthalmos | | | | | 0.00 | 0.07 | 0.08 | 0.00 |
| Microphthalmos | | | | | 0.25 | 0.14 | 0.38 | 0.10 |
| Unspecified Anophthalmos/Microphthalmos | | | | | 0.00 | 0.00 | 0.00* | 0.00* |
| Anotia | | | | | 0.08 | 0.18 | 0.04 | 0.20 |
| Microtia | | | | | 0.41 | 0.32 | 0.23 | 0.10 |
| Unspecified Anotia/Microtia | | | | | 0.16 | 0.21 | 0.68 | 0.72* |
| Transposition of great vessels | | | | | 0.82 | 0.89 | 1.39 | 1.52 |
| Tetralogy of Fallot | | | | | 1.15 | 1.00 | 1.58 | 1.59 |
| Hypoplastic left heart syndrome | | | | | 0.82 | 1.75 | 2.22 | 1.80 |
| Coarctation of aorta | | | | | 0.41 | 0.53 | 0.79 | 0.81 |
| Choanal atresia, bilateral | | | | | 0.16 | 0.25 | 0.11 | 0.17 |
| Cleft palate without cleft lip | | | | | 5.65 | 5.17 | 5.76 | 4.54 |
| Cleft lip with or without cleft palate | | | | | 9.42 | 10.77 | 9.41 | 8.50 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 0.98 | 1.18 | 1.66 | 1.08 |
| Small intestine atresia/stenosis | | | | | 1.39 | 1.39 | 2.45 | 1.32 |
| Anorectal atresia/stenosis | | | | | 0.82 | 2.57 | 2.71 | 2.61 |
| Undescended testis (36 weeks of gestation or later) | | | | | 5.16 | 7.70 | 8.13 | 12.60 |
| Hypospadias | | | | | 23.52 | 23.07 | 22.51 | 16.02 |
| Epispadias | | | | | 0.08 | 0.21 | 0.19 | 0.20 |
| Indeterminate sex | | | | | 0.41 | 0.57 | 0.19 | 0.24 |
| Renal agenesis | | | | | 1.48 | 4.03 | 5.95 | 5.93 |
| Cystic kidney | | | | | 0.57 | 1.11 | 1.77 | 1.02 |
| Bladder exstrophy | | | | | 0.00 | 0.25 | 0.11 | 0.07 |
| Polydactyly, preaxial | | | | | 1.56 | 2.21 | 3.27 | 2.84 |
| Total Limb reduction defects (include unspecified) | | | | | 4.43 | 3.03 | 4.10 | 2.71 |
| Transverse | | | | | nr | nr | nr | nr |
| Preaxial | | | | | nr | nr | nr | nr |
| Postaxial | | | | | nr | nr | nr | nr |
| Intercalary | | | | | nr | nr | nr | nr |
| Mixed | | | | | nr | nr | nr | nr |
| Unspecified | | | | | 0.08 | 0.00 | 0.04 | 0.00* |
| Diaphragmatic hernia | | | | | 0.98 | 1.39 | 1.69 | 1.69 |
| Omphalocele | | | | | 0.33 | 0.68 | 0.68 | 0.58 |
| Gastroschisis | | | | | 0.57 | 1.00 | 1.17 | 0.91 |
| Unspecified Omphalocele/Gastroschisis | | | | | 0.00 | 0.00 | 0.00* | 0.11* |
| Prune belly sequence | | | | | 0.00 | 0.04 | 0.23 | 0.03 |
| Trisomy 13 | | | | | 0.16 | 0.32 | 0.38 | 0.20 |
| Trisomy 18 | | | | | 0.08 | 0.32 | 0.64 | 0.78 |
| Down syndrome, all ages (include age unknown) | | | | | 9.18 | 9.59 | 10.01 | 8.64 |
| <20 | | | | | 8.18 | 5.67 | 2.81 | 3.77* |
| 20-24 | | | | | 7.57 | 5.14 | 3.96 | 3.60* |
| 25-29 | | | | | 6.02 | 6.58 | 6.93 | 4.42* |
| 30-34 | | | | | 12.87 | 12.43 | 10.80 | 9.35 |
| 35-39 | | | | | 27.30 | 40.77 | 37.53 | 23.15* |
| 40-44 | | | | | 43.29 | 90.36 | 109.02 | 61.46* |
| 45+ | | | | | 0.00 | 241.94 | 283.69 | 188.68* |
| unknown | | | | | --- | --- | --- | --- |

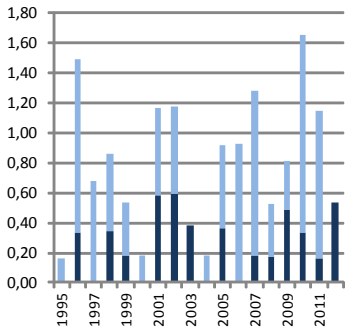
nr = data not reported or not available

* data include less than 5 years

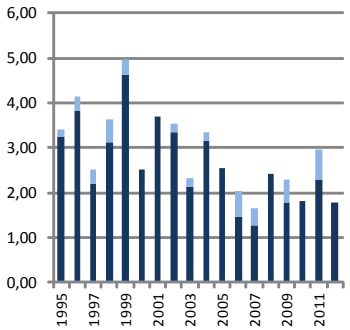
# Slovak Republic, Time trends 1995 – 2012

(Birth prevalence rates per 10,000)

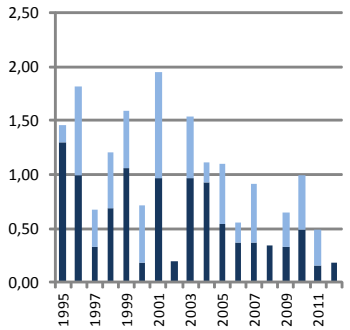
### Anencephaly



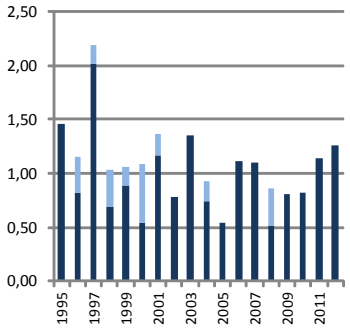

### Spina Bifida



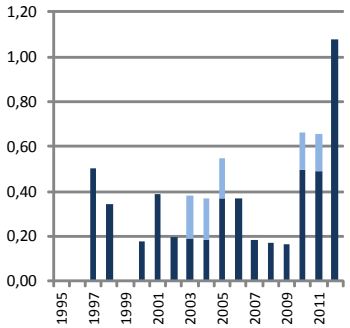

### Encephalocele



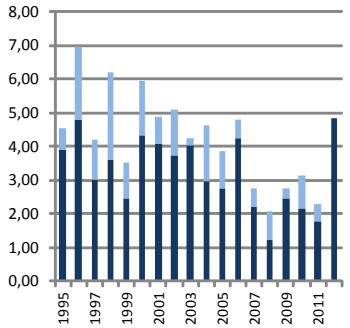

### Microcephaly



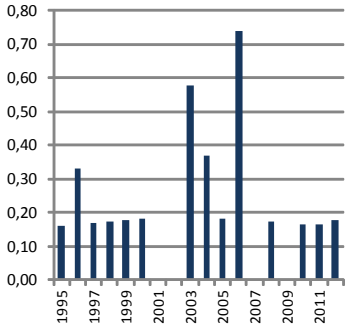

### Holoprosencephaly



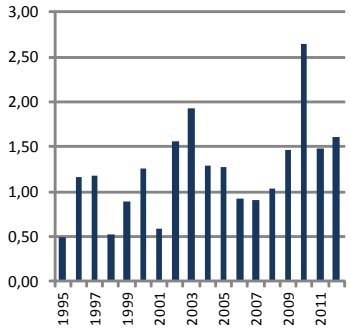

### Hydrocephaly



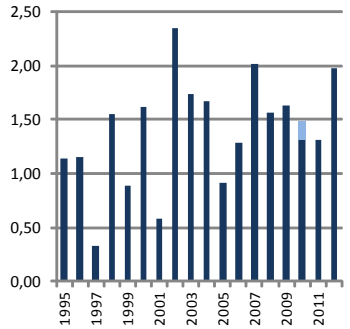

### Microphthalmos



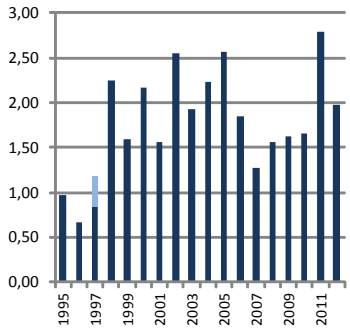

### Transposition of great vessels



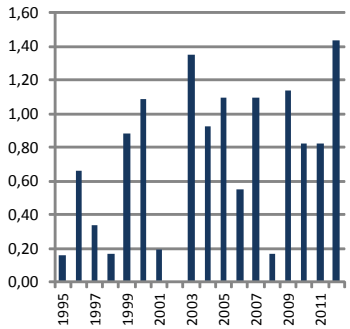

### Tetralogy of Fallot



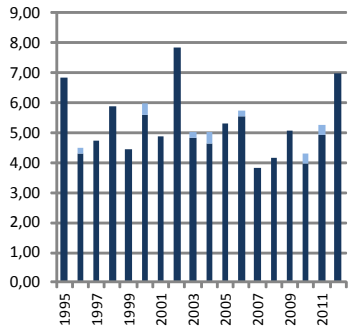

### Hypoplastic left heart syndrome




### Coarctation of aorta



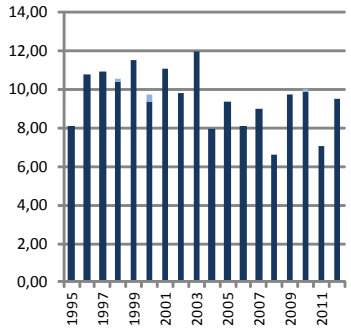

### Cleft palate without cleft lip



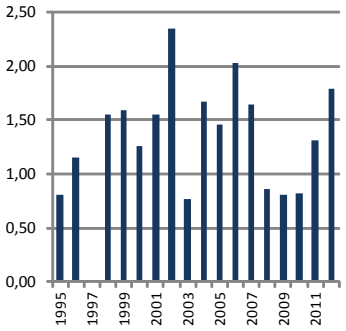
# Slovak Republic, Time trends 1995 – 2012

(Birth prevalence rates per 10,000)

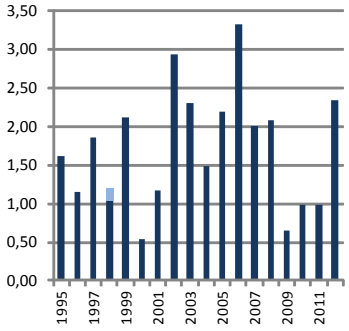
**Cleft lip with or without cleft palate**



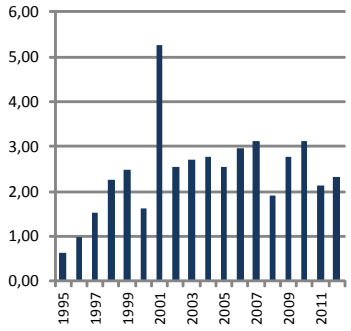

**phageal atresia/stenosis with or without f**



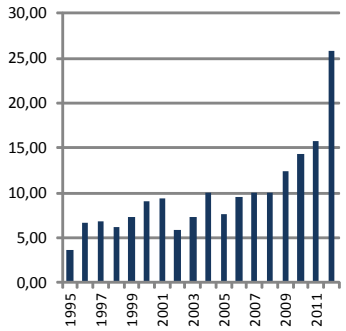

**Small intestine atresia/stenosis**



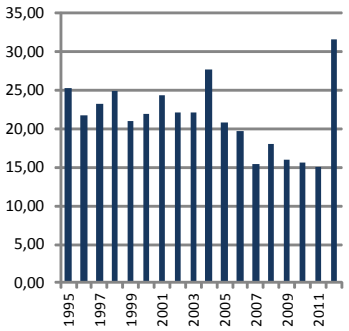

**Anorectal atresia/stenosis**



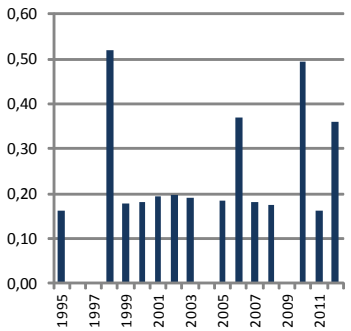

**Undescended testis**



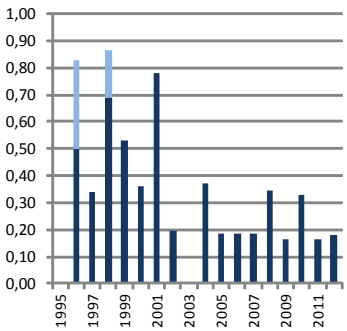

**Hypospadias**



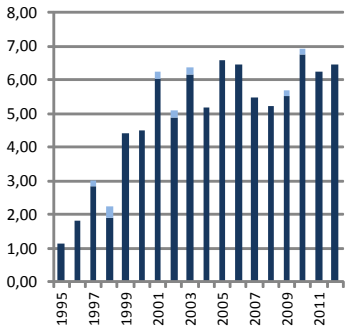

**Epispadias**



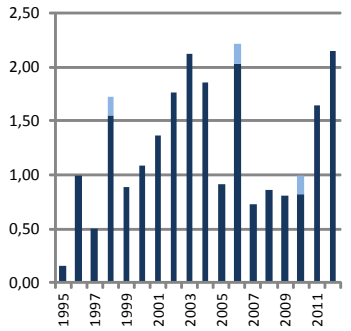

**Indeterminate sex**



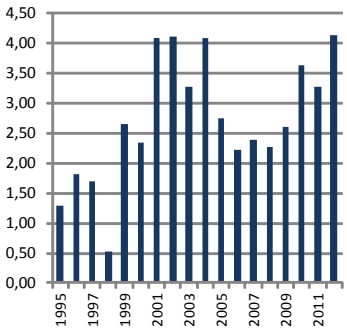

**Renal agenesis**



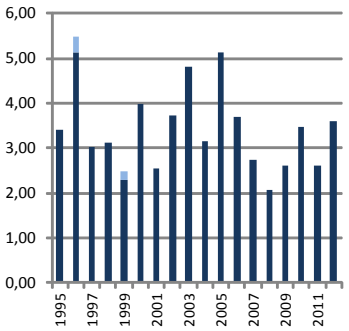

**Cystic kidney**




**Polydactyly, preaxial**



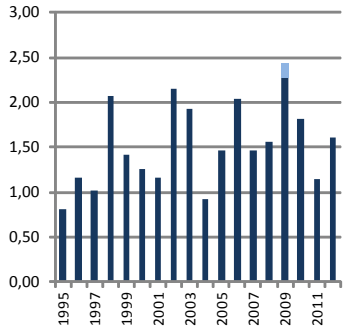

**Limb reduction defects**



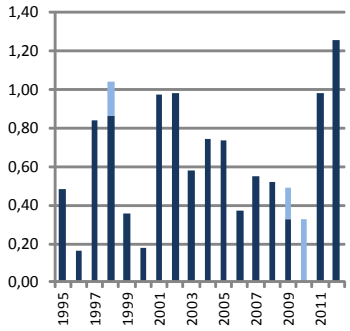
# Slovak Republic, Time trends 1995 – 2012

(Birth prevalence rates per 10,000)

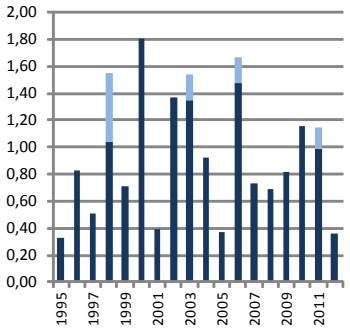
**Diaphragmatic hernia**



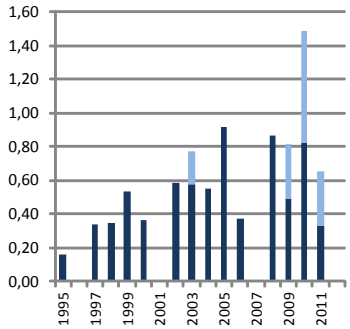

**Omphalocele**



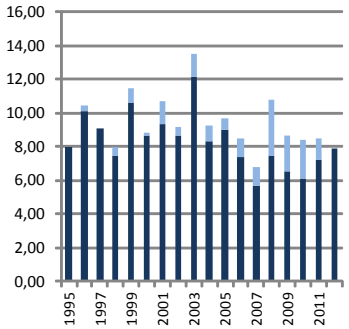

**Gastroschisis**



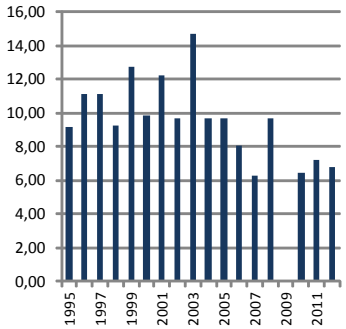

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# South America: ECLAMC

Latin American Collaborative Study of Congenital Malformations

**History:**
The Programme started in 1967 and has grown in size and coverage. The Programme became a full member of the International Clearinghouse in 1977.

**Size and coverage:**
The number of participating hospitals has grown from 20 in 1977 to 70 at the present time, distributed over most South Americans countries. The annual number of births covered is at present approximately 150,000, less than 1% of all births. Stillbirths of at least 500g birthweight have been included since 1978.

**Legislation and funding:**
The Programme is a research Programme with voluntary participation of hospitals and funded by research grants provided from several sources, mainly the national research councils of Argentina and Brazil.

**Sources of ascertainment:**
Reporting is made by collaborating pediatricians at the delivery units of participating hospitals.

**Exposure information:**
The mother of each reported infant and the mother of a control infant - the next nonmalformed infant born at that hospital with the same sex as the proband - are interviewed on various exposures, including drug usage and parental occupation.

**Background information:**
Background information is obtained partly from summarising tables of births in each participating hospitals, partly from the matched control newborns.

**Addresses and Staff:**
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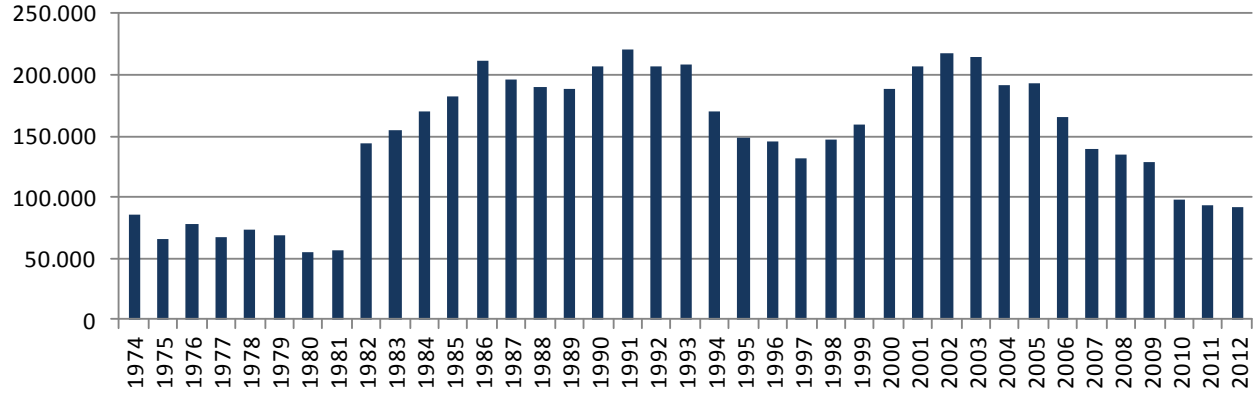
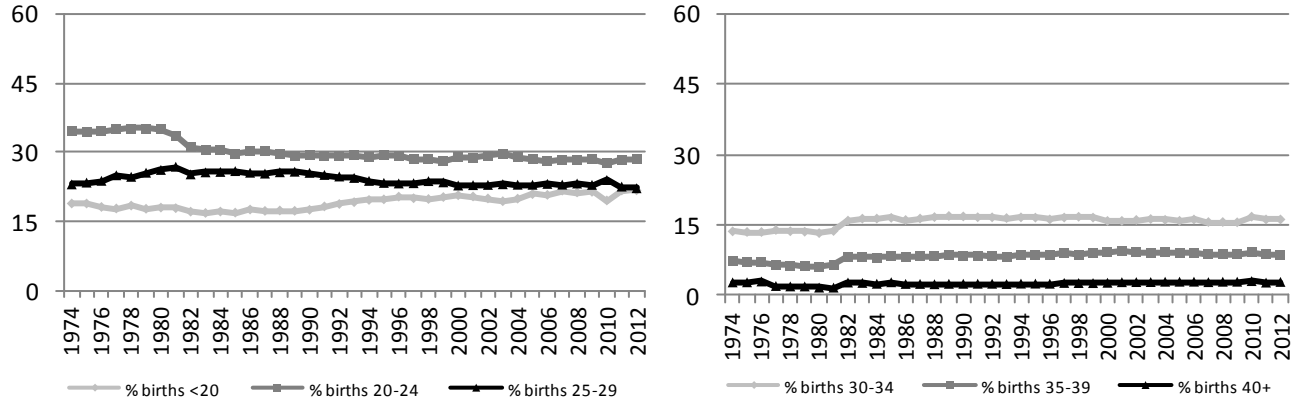
20010-970 Rio de Janeiro, Brazil
**Phone:** 55-21-25528952
**Fax:** 55-21-22604282(5521)
**E-mail:** castilla@centroin.com.br

# South America: ECLAMC

## Total births by year




## Percentage of births by year and maternal age



# South America: ECLAMC, 2012

| | |
|---|---|
| Live births (LB) | 91,221 |
| Stillbirths (SB) | 1,206 |
| Total births | 92,427 |
| Number of terminations of pregnancy (ToP) for birth defects | not permitted |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 27 | 16 | nr | 4.65 |
| Spina bifida | 70 | 4 | nr | 8.01 |
| Encephalocele | 22 | 2 | nr | 2.60 |
| Microcephaly | 35 | 4 | nr | 4.22 |
| Holoprosencephaly | 6 | 1 | nr | 0.76 |
| Hydrocephaly | 132 | 12 | nr | 15.58 |
| Anophthalmos | 5 | 0 | nr | 0.54 |
| Microphthalmos | 10 | 3 | nr | 1.41 |
| Unspecified Anophthalmos/Microphthalmos | 2 | 1 | nr | 0.32 |
| Anotia | 1 | 0 | nr | 0.11 |
| Microtia | 47 | 3 | nr | 5.41 |
| Unspecified Anotia/Microtia | 2 | 0 | nr | 0.22 |
| Transposition of great vessels | 6 | 0 | nr | 0.65 |
| Tetralogy of Fallot | 15 | 0 | nr | 1.62 |
| Hypoplastic left heart syndrome | 7 | 1 | nr | 0.87 |
| Coarctation of aorta | 3 | 0 | nr | 0.32 |
| Choanal atresia, bilateral | 5 | 0 | nr | 0.54 |
| Cleft palate without cleft lip | 35 | 1 | nr | 3.89 |
| Cleft lip with or without cleft palate | 101 | 12 | nr | 12.23 |
| Oesophageal atresia/stenosis with or without fistula | 28 | 1 | nr | 3.14 |
| Small intestine atresia/stenosis | 40 | 4 | nr | 4.76 |
| Anorectal atresia/stenosis | 47 | 3 | nr | 5.41 |
| Undescended testis (36 weeks of gestation or later) | 101 | 3 | nr | 11.25 |
| Hypospadias | 66 | 0 | nr | 7.14 |
| Epispadias | 2 | 0 | nr | 0.22 |
| Indeterminate sex | 17 | 6 | nr | 2.49 |
| Renal agenesis | 27 | 1 | nr | 3.03 |
| Cystic kidney | 37 | 2 | nr | 4.22 |
| Bladder exstrophy | 1 | 0 | nr | 0.11 |
| Polydactyly, preaxial | 24 | 0 | nr | 2.60 |
| Total Limb reduction defects (include unspecified) | 36 | 3 | nr | 4.22 |
| Transverse | 3 | 1 | nr | 0.43 |
| Preaxial | 11 | 1 | nr | 1.30 |
| Postaxial | 0 | 0 | nr | 0.00 |
| Intercalary | 3 | 0 | nr | 0.32 |
| Mixed | 2 | 0 | nr | 0.22 |
| Unspecified | 17 | 1 | nr | 1.95 |
| Diaphragmatic hernia | 32 | 0 | nr | 3.46 |
| Omphalocele | 34 | 4 | nr | 4.11 |
| Gastroschisis | 87 | 12 | nr | 10.71 |
| Unspecified Omphalocele/Gastroschisis | 13 | 0 | nr | 1.41 |
| Prune belly sequence | 2 | 0 | nr | 0.22 |
| Trisomy 13 | 6 | 1 | nr | 0.76 |
| Trisomy 18 | 9 | 0 | nr | 0.97 |
| Down syndrome, all ages (include age unknown) | 164 | 1 | nr | 17.85 |
| <20 | 12 | 0 | nr | 6.21 |
| 20-24 | 16 | 0 | nr | 6.31 |
| 25-29 | 23 | 1 | nr | 12.17 |
| 30-34 | 16 | 0 | nr | 11.11 |
| 35-39 | 41 | 0 | nr | 53.17 |
| 40-44 | 47 | 0 | nr | 202.76 |
| 45+ | 9 | 0 | nr | 378.15 |
| unknown | 0 | 0 | nr | --- |

nr = data not reported or not available

# South America: ECLAMC, Previous years rates 1974 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | **228,167** | **321,052** | **861,574** | **1,001,967** | **878,906** | **832,415** | **981,311** | **592,191** |
| Anencephaly | 2.76 | 4.95 | 6.28 | 6.58 | 7.84 | 7.03 | 6.08 | 5.79 |
| Spina bifida | 5.48 | 6.73 | 6.29 | 7.44 | 8.25 | 10.56 | 9.54 | 8.46 |
| Encephalocele | 1.40 | 1.68 | 1.73 | 1.87 | 2.14 | 2.86 | 2.77 | 3.28 |
| Microcephaly | 2.28 | 2.59 | 2.63 | 2.61 | 2.75 | 3.72 | 3.91 | 4.83 |
| Holoprosencephaly | 0.18 | 0.59 | 0.51 | 0.27 | 0.61 | 1.36 | 1.60 | 0.78 |
| Hydrocephaly | 2.24 | 3.89 | 4.53 | 6.01 | 9.88 | 11.66 | 12.52 | 17.21 |
| Anophthalmos | 0.18 | 0.28 | 0.38 | 0.36 | 0.39 | 0.44 | 1.48 | 1.18 |
| Microphthalmos | 1.23 | 1.18 | 0.97 | 1.27 | 1.58 | 1.62 | 1.56 | 1.71 |
| Unspecified Anophthalmos/Microphthalmos | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | nr | nr | nr | nr | nr | 0.34 | 0.40 | 0.24 |
| Microtia | nr | nr | nr | nr | nr | 3.90 | 5.83 | 5.67 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr | nr | 0.11 | 0.09 | 0.39 |
| Transposition of great vessels | 0.13 | 0.09 | 0.57 | 0.57 | 0.74* | 1.57 | 1.18 | 0.66 |
| Tetralogy of Fallot | 0.00 | 0.16 | 0.53 | 0.79 | 1.46 | 1.65 | 1.57 | 1.64 |
| Hypoplastic left heart syndrome | 0.00 | 0.00 | 0.01 | 0.17 | 0.50 | 1.07 | 1.20 | 0.95 |
| Coarctation of aorta | 0.13 | 0.09 | 0.19 | 0.54 | 0.74 | 1.01 | 0.62 | 0.52 |
| Choanal atresia, bilateral | 0.00 | 0.00 | 0.14 | 0.23 | 0.24 | 0.14 | 0.21 | 0.32 |
| Cleft palate without cleft lip | 2.76 | 3.33 | 3.49 | 3.41 | 3.81 | 4.50 | 4.91 | 4.46 |
| Cleft lip with or without cleft palate | 11.04 | 10.96 | 10.40 | 10.46 | 11.54 | 12.61 | 14.01 | 11.72 |
| Oesophageal atresia/stenosis with or without fistula | 1.84 | 2.15 | 2.39 | 2.90 | 2.90 | 3.53 | 3.64 | 4.10 |
| Small intestine atresia/stenosis | 0.13 | 1.12 | 1.56 | 1.52 | 1.90 | 2.61 | 3.12 | 2.94 |
| Anorectal atresia/stenosis | 2.32 | 3.36 | 3.69 | 3.97 | 4.78 | 5.18 | 5.60 | 5.62 |
| Undescended testis (36 weeks of gestation or later) | 1.36 | 2.09 | 4.02 | 4.61 | 4.95 | 5.99 | 7.63 | 9.79 |
| Hypospadias | 3.90 | 3.58 | 4.87 | 3.66 | 4.86 | 5.20 | 4.64 | 9.19 |
| Epispadias | 0.18 | 0.06 | 0.37 | 0.35 | 0.22 | 0.23 | 0.18 | 0.12 |
| Indeterminate sex | 0.92 | 1.40 | 2.25 | 1.66 | 1.92 | 2.05 | 2.39 | 2.85 |
| Renal agenesis | 0.44 | 0.44 | 0.71 | 1.11 | 1.92 | 2.39 | 2.56 | 2.97 |
| Cystic kidney | 0.57 | 0.59 | 1.17 | 1.80 | 2.47 | 4.37 | 3.78 | 5.29 |
| Bladder exstrophy | 0.04 | 0.19 | 0.26 | 0.29 | 0.32 | 0.35 | 0.29 | 0.20 |
| Polydactyly, preaxial | 2.89 | 2.49 | 2.44 | 2.64 | 2.74 | 3.21 | 4.22 | 3.14 |
| Total Limb reduction defects (include unspecified) | 3.64 | 4.80 | 5.26 | 4.90 | 5.72 | 6.48 | 7.44 | 7.60 |
| Transverse | 1.84 | 2.59 | 2.62 | 2.56 | 2.88 | 3.22 | 3.38 | 1.86 |
| Preaxial | 0.53 | 0.84 | 1.09 | 0.92 | 1.29 | 1.61 | 1.35 | 1.10 |
| Postaxial | 0.26 | 0.34 | 0.41 | 0.28 | 0.49 | 0.43 | 0.44 | 0.35 |
| Intercalary | 0.53 | 0.44 | 0.45 | 0.47 | 0.40 | 0.60 | 0.62 | 0.74 |
| Mixed | 0.39 | 0.44 | 0.59 | 0.54 | 0.51 | 0.50 | 1.46 | 1.49 |
| Unspecified | 0.09 | 0.16 | 0.09 | 0.13 | 0.16 | 0.11 | 0.19 | 2.06 |
| Diaphragmatic hernia | 0.75 | 0.93 | 1.35 | 1.99 | 2.58 | 3.74 | 3.72 | 3.88 |
| Omphalocele | 1.23 | 1.34 | 2.19 | 2.26 | 2.73 | 3.20 | 3.82 | 4.54 |
| Gastroschisis | 0.04 | 0.22 | 0.53 | 0.70 | 1.75 | 2.94 | 4.12 | 9.59 |
| Unspecified Omphalocele/Gastroschisis | 0.35 | 0.34 | 0.43 | 0.34 | 0.75 | 1.26 | 1.12 | 0.61 |
| Prune belly sequence | 0.00 | 0.03 | 0.70 | 0.72 | 0.96 | 1.14 | 0.87 | 0.62 |
| Trisomy 13 | 0.18 | 0.19 | 0.55 | 0.42 | 0.66 | 0.94 | 0.76 | 0.57 |
| Trisomy 18 | 0.22 | 0.25 | 0.93 | 0.93 | 1.23 | 2.05 | 1.83 | 1.32 |
| Down syndrome, all ages (include age unknown) | 13.63 | 15.48 | 14.53 | 15.80 | 16.35 | 18.84 | 19.02 | 17.70 |
| <20 | 4.82 | 9.94 | 6.23 | 7.08 | 7.29 | 7.98 | 7.83 | 8.43 |
| 20-24 | 7.80 | 6.92 | 6.40 | 7.37 | 8.35 | 9.24 | 9.21 | 8.40 |
| 25-29 | 7.87 | 8.41 | 7.67 | 7.39 | 8.86 | 10.07 | 8.91 | 10.12 |
| 30-34 | 11.83 | 17.42 | 14.04 | 17.04 | 15.04 | 17.58 | 16.62 | 16.66 |
| 35-39 | 49.82 | 56.79 | 42.10 | 48.65 | 45.92 | 54.88 | 54.95 | 52.77 |
| 40-44 | 132.97 | 207.77 | 147.69 | 142.88 | 167.15 | 168.16 | 179.76 | 140.05 |
| 45+ | 240.30 | 405.06 | 240.47 | 312.15 | 266.38 | 371.45 | 358.82 | 263.79 |
| unknown | --- | --- | --- | --- | --- | --- | --- | --- |

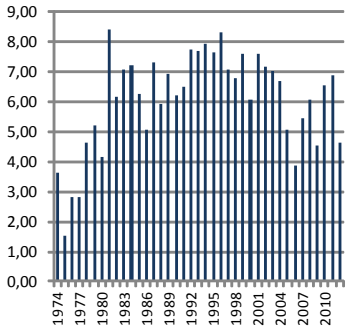
nr = data not reported or not available

* data include less than 5 years

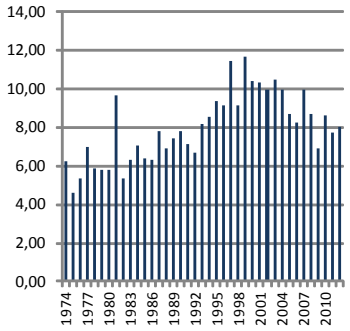
# South America: ECLAMC, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

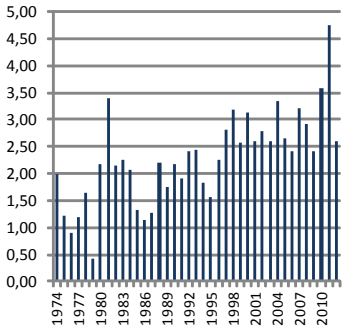
**Anencephaly**



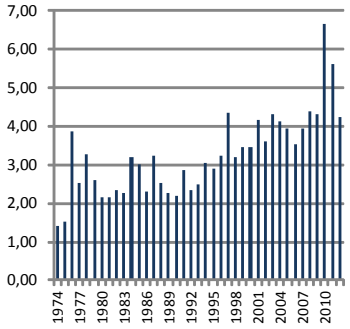

**Spina Bifida**



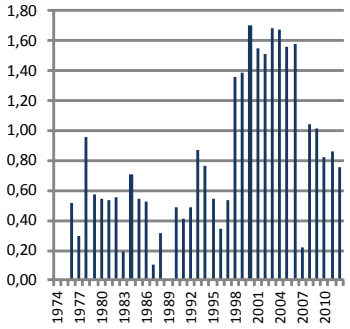

**Encephalocele**



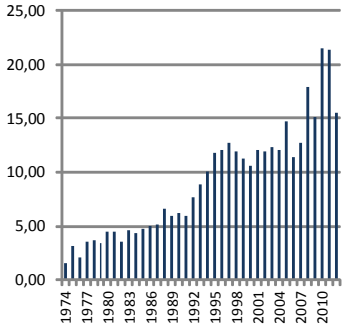

**Microcephaly**



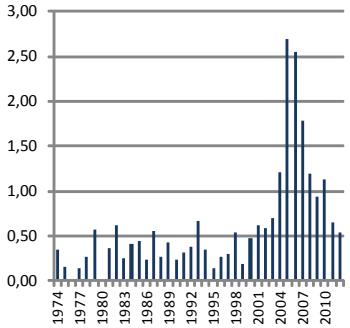

**Holoprosencephaly**



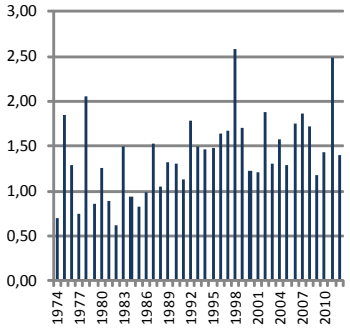

**Hydrocephaly**



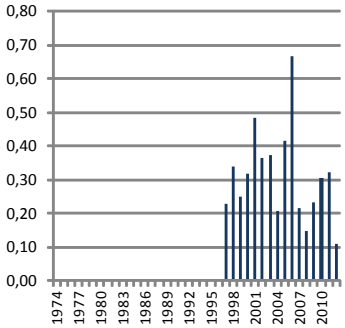

**Anophthalmos**



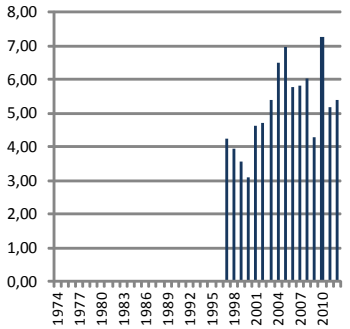

**Microphthalmos**



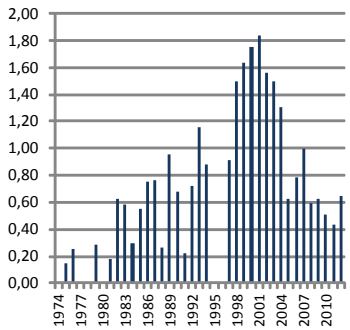

**Anotia**



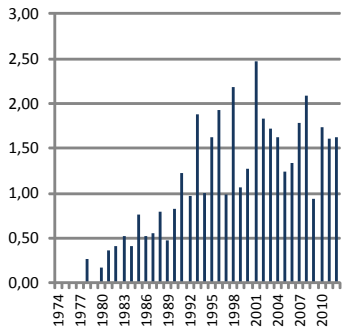

**Microtia**




**Transposition of great vessels**



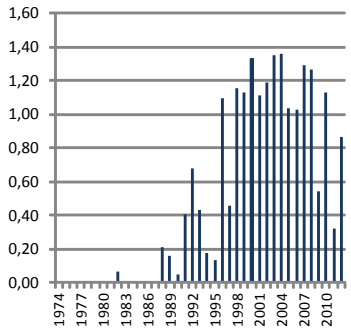

**Tetralogy of Fallot**



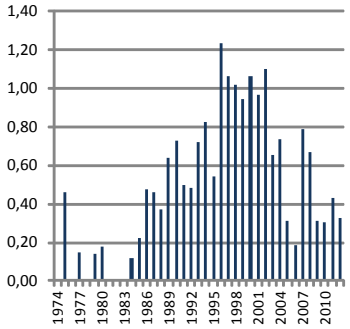
# South America: ECLAMC, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

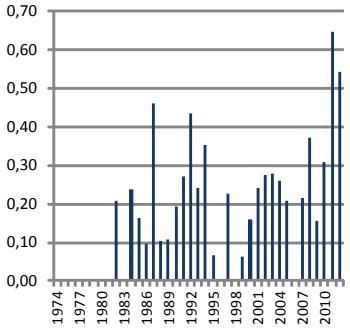
**Hypoplastic left heart syndrome**



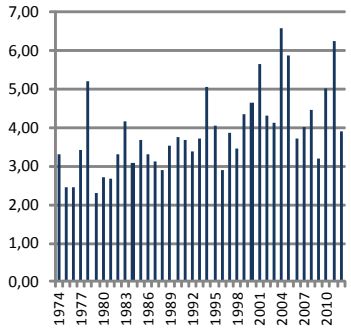

**Coarctation of aorta**



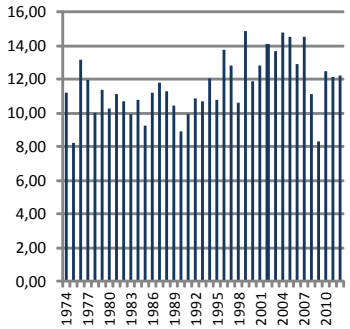

**Choanal atresia, bilateral**



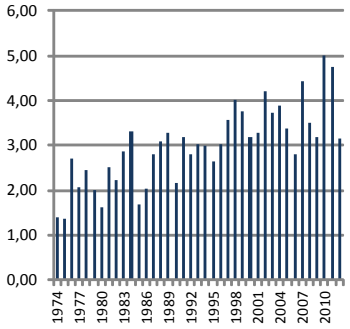

**Cleft palate without cleft lip**



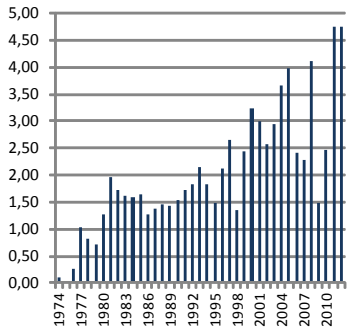

**Cleft lip with or without cleft palate**



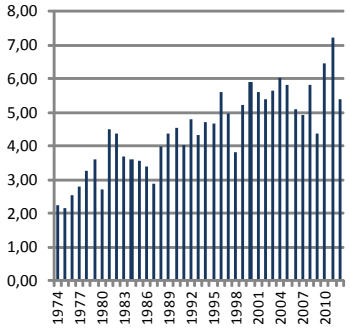

**phageal atresia/stenosis with or without f**



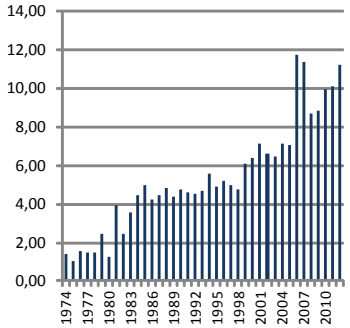

**Small intestine atresia/stenosis**



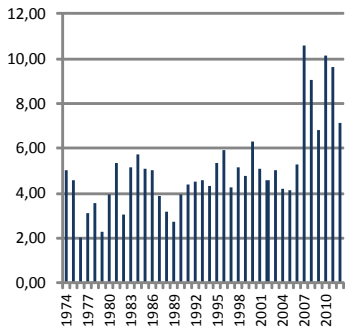

**Anorectal atresia/stenosis**



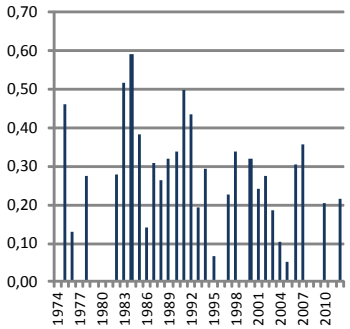

**Undescended testis**



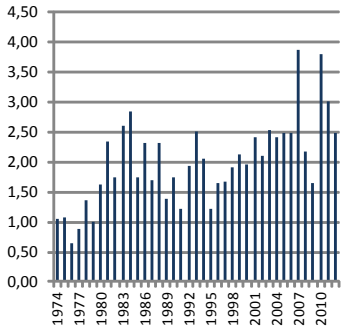

**Hypospadias**




**Epispadias**



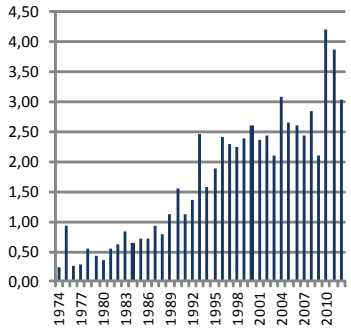

**Indeterminate sex**



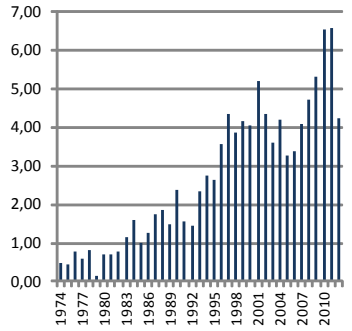
# South America: ECLAMC, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

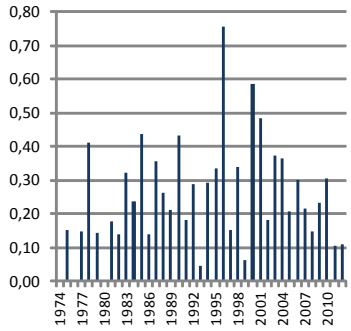
**Renal agenesis**



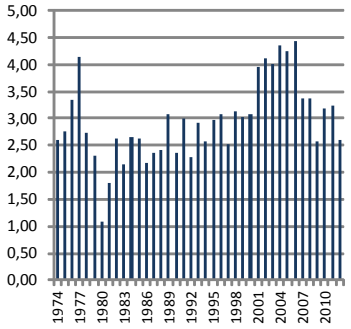

**Cystic kidney**



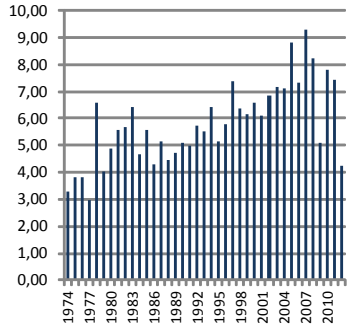

**Bladder, exstrophy**



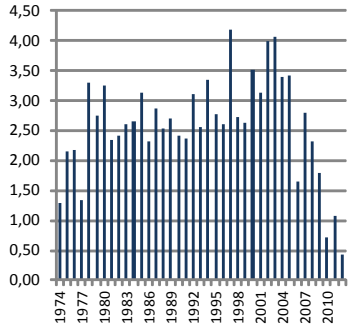

**Polydactyly, preaxial**



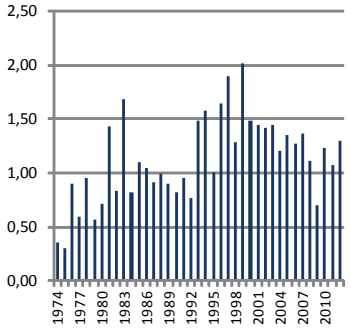

**Limb reduction defects**



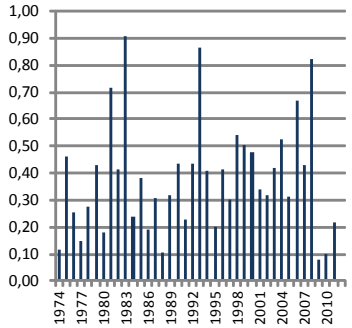

**Limb reduction defects - transverse**



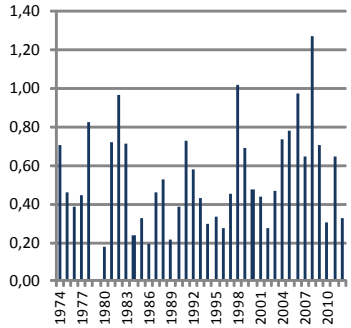

**Limb reduction defects - preaxial**



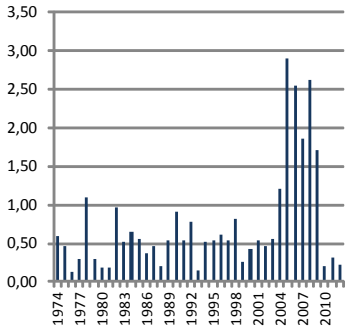

**Limb reduction defects -postaxial**



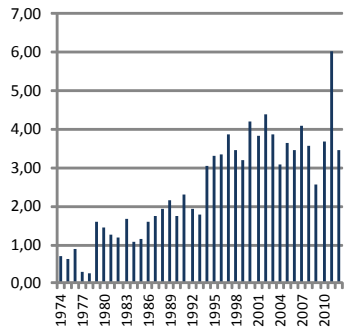

**Limb reduction defects - intercalary**



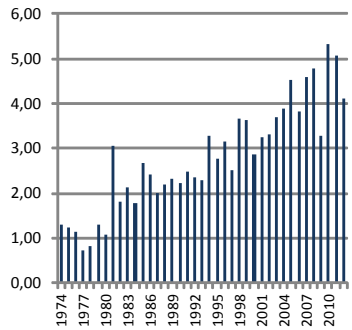

**Limb reduction defects - mixed**




**Diaphragmatic hernia**



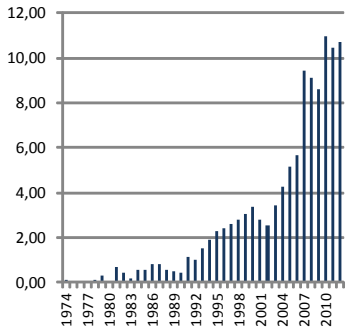

**Omphalocele**



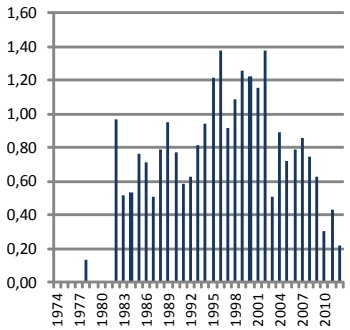
# South America: ECLAMC, Time trends 1974 – 2012

(Birth prevalence rates per 10,000)

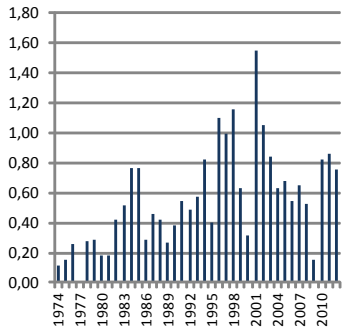
**Gastroschisis**



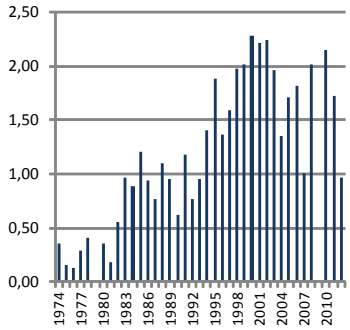

**Prune belly sequence**



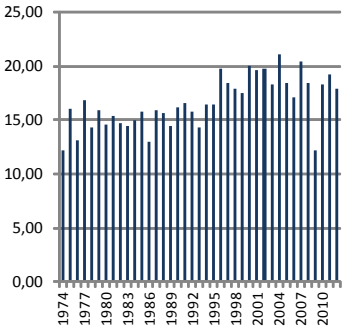

**Trisomy 13**



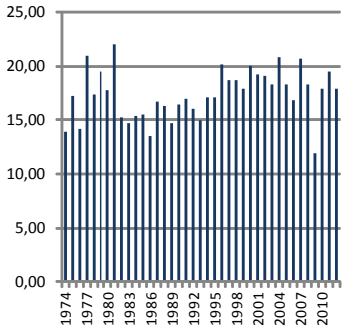

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates**

# Spain: ECEMC

## Spanish Collaborative Study of Congenital Malformations

**History:**

The programme was created in 1976 by Prof. Dr. María Luisa Martínez-Frías, as a hospital-based case-control study and surveillance system. ECEMC joined ICBDSR in 1979. It is also a member of EUROCAT, contributing with data since 1980. In January 2002, the ECEMC Programme started its activities into the CIAC (Research Center on Congenital Anomalies), of the Instituto de Salud Carlos III (ISCIII), now dependent from the Ministry of Economy and Competitiveness, of Spain. In 2006 the ECEMC was recognized as a group of excellence in research and was selected to be integrated into the CIBERER (Centre for Biomedical Research on Rare Diseases). ECEMC also operates two Teratology Information Services (TIS) since 1991, one for the general population and another one for physicians. ECEMC and the two TIS are directed by Prof. Martínez-Frías. In June 2012, ECEMC's Clinical Network was formally constituted, although it has operated since 1976. Much activity is developed to provide continued medical education, as well as to inform the population about preventive measures regarding birth defects.

**Size and coverage:**

Data are obtained from about 70 hospitals distributed all over Spain. The annual number of births is about 90,000, representing near 20% of all Spanish births. Stillbirths of at least 24 weeks or 500 g. have been included since 1980. Data on terminations of pregnancy due to the presence of congenital anomalies, which can be legally performed under defined circumstances, can be gathered on a routine basis only in some participating hospitals.

**Legislation and funding:**

ECEMC is a research programme with voluntary participation of hospitals (but mandatory subjugation to the operating rules expressed in the Operating Manual, for those participating), and is financed mainly by the Spanish Administration and, partially, by non-governmental organisations. ECEMC has the approval of the Research Ethics Committee of ISCIII, and is declared to the Spanish Agency of Data Protection.

**Sources of ascertainment:**

The detection period comprises the first 3 days of life, including major and/or minor/mild defects. For some selected cases a longer follow-up can be performed. Controls are defined as the next non-malformed infant born at the same hospital and with the same sex as the malformed infant. The information comes from delivery units and paediatric departments of the participating hospitals. Mothers are interviewed directly by the participating physicians, during those first 3 days after infant's delivery, to fill in the ECEMC standard protocols, which include more than 310 data for each child whether case or control. The information for each case and its control is gathered by the same physician after the written informed consent of parents. In many instances, photographs, imaging studies, high-resolution bands karyotypes and molecular analyses when needed (which are performed at the central group of the ECEMC), and other complementary studies are available. Biological samples are also stored in the ECEMC registry for those cases and controls for which the collaborating physicians send them, also with the informed consent of the parents. A multidisciplinary

approach is applied to analyse and characterize each subject included in the registry. For research, the interaction with other groups, even from other related fields, is always considered.

**Exposure information:**
The mother of each reported infant (case or control) is interviewed within the first three days after delivery to obtain data on several exposures (parental occupation, maternal acute or chronic diseases, drug usage, illicit drugs, alcohol and tobacco maternal consumption, exposure to other chemical or physical factors), apart from the other data gathered (family history, obstetrical and demographic data, paternal exposures among others). All the data are gathered in ECEMC's specific forms. It is important to note that when the paediatricians detect the cases and select the control children, they are blinded to the different maternal and family data that they are going to collect.

**Background information:**
Total number of births by sex and number of twin pairs in each participating hospital are gathered. Other background information is obtained from the control material.

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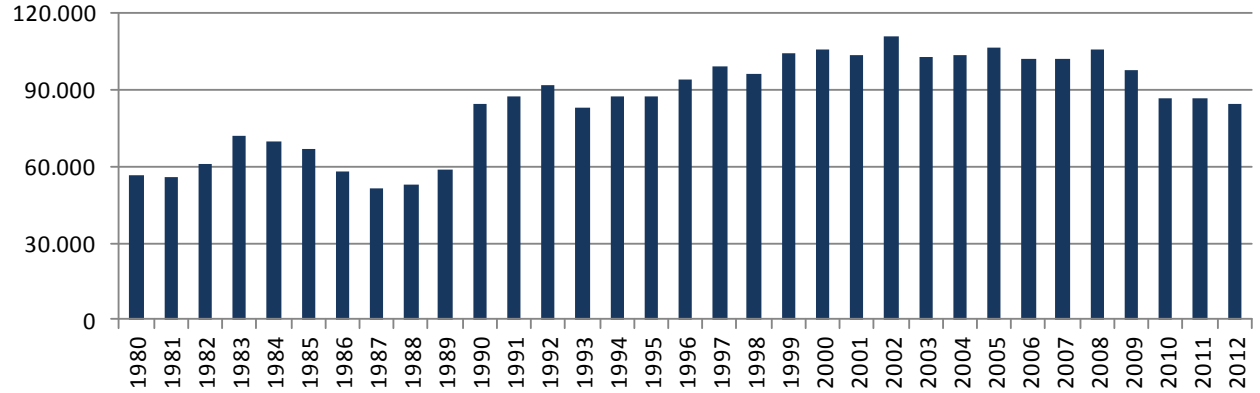
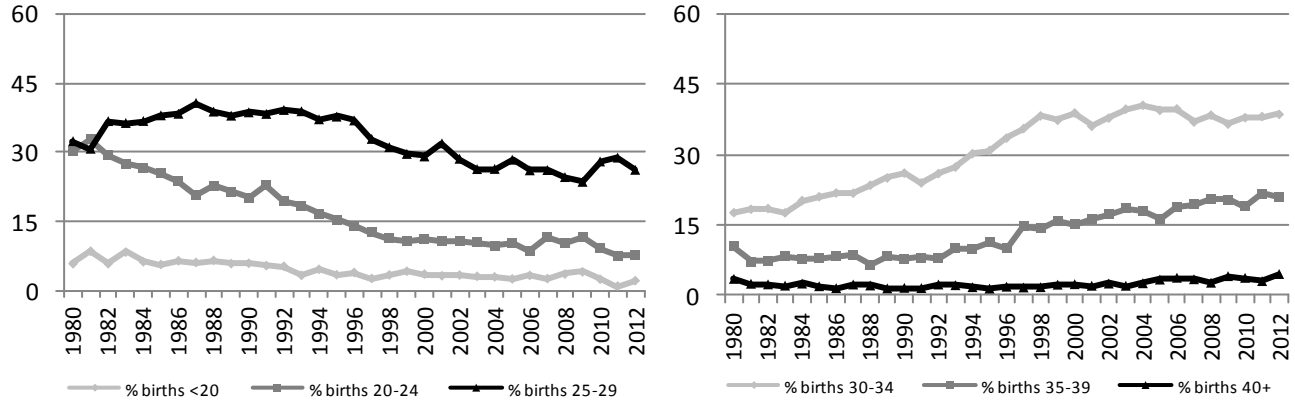
# Spain: ECEMC

## Total births by year




## Percentage of births by year and maternal age



# Spain: ECEMC, 2012

| | |
|---|---|
| Live births (LB) | 83,963 |
| Stillbirths (SB) | 272 |
| Total births | 84,235 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | nr | 0.12 |
| Spina bifida | 2 | 0 | nr | 0.24 |
| Encephalocele | 1 | 0 | nr | 0.12 |
| Microcephaly | 9 | 0 | nr | 1.07 |
| Holoprosencephaly | 0 | 0 | nr | 0.00 |
| Hydrocephaly | 10 | 0 | nr | 1.19 |
| Anophthalmos | 0 | 0 | nr | 0.00 |
| Microphthalmos | 6 | 0 | nr | 0.71 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 8 | 0 | nr | 0.95 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 6 | 0 | nr | 0.71 |
| Tetralogy of Fallot | 5 | 0 | nr | 0.59 |
| Hypoplastic left heart syndrome | 3 | 0 | nr | 0.36 |
| Coarctation of aorta | 0 | 0 | nr | 0.00 |
| Choanal atresia, bilateral | 1 | 0 | nr | 0.12 |
| Cleft palate without cleft lip | 25 | 0 | nr | 2.97 |
| Cleft lip with or without cleft palate | 23 | 1 | nr | 2.85 |
| Oesophageal atresia/stenosis with or without fistula | 13 | 0 | nr | 1.54 |
| Small intestine atresia/stenosis | 3 | 0 | nr | 0.36 |
| Anorectal atresia/stenosis | 6 | 0 | nr | 0.71 |
| Undescended testis (36 weeks of gestation or later) | 14 | 0 | nr | 1.66 |
| Hypospadias | 10 | 0 | nr | 1.19 |
| Epispadias | 1 | 0 | nr | 0.12 |
| Indeterminate sex | 1 | 0 | nr | 0.12 |
| Renal agenesis | 2 | 0 | nr | 0.24 |
| Cystic kidney | 16 | 0 | nr | 1.90 |
| Bladder exstrophy | 0 | 0 | nr | 0.00 |
| Polydactyly, preaxial | 15 | 0 | nr | 1.78 |
| Total Limb reduction defects (include unspecified) | 31 | 0 | nr | 3.68 |
| Transverse | 9 | 0 | nr | 1.07 |
| Preaxial | 2 | 0 | nr | 0.24 |
| Postaxial | 0 | 0 | nr | 0.00 |
| Intercalary | 3 | 0 | nr | 0.36 |
| Mixed | 9 | 0 | nr | 1.07 |
| Unspecified | 8 | 0 | nr | 0.95 |
| Diaphragmatic hernia | 4 | 0 | nr | 0.47 |
| Omphalocele | 5 | 0 | nr | 0.59 |
| Gastroschisis | 5 | 0 | nr | 0.59 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 2 | 0 | nr | 0.24 |
| Trisomy 13 | 1 | 0 | nr | 0.12 |
| Trisomy 18 | 2 | 1 | nr | 0.36 |
| Down syndrome, all ages (include age unknown) | 52 | 1 | nr | 6.29 |
| <20 | 0 | 0 | nr | 0.00 |
| 20-24 | 2 | 0 | nr | 3.11 |
| 25-29 | 6 | 0 | nr | 2.73 |
| 30-34 | 8 | 0 | nr | 2.46 |
| 35-39 | 20 | 0 | nr | 11.30 |
| 40-44 | 13 | 0 | nr | 34.91 |
| 45+ | 1 | 0 | nr | 91.74 |
| unknown | 2 | 1 | nr | --- |

nr = data not reported or a routine basis by all the participating hospitals

# Spain: ECEMC, Previous years rates 1980 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981* | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | **113,012** | **327,623** | **334,870** | **443,187** | **509,008** | **526,739** | **480,647** |
| Anencephaly | | 5.31 | 4.03 | 1.94 | 0.77 | 0.37 | 0.15 | 0.29 |
| Spina bifida | | 3.89 | 4.88 | 4.42 | 2.84 | 1.91 | 1.08 | 0.67 |
| Encephalocele | | 1.50 | 0.89 | 0.78 | 0.65 | 0.24 | 0.21 | 0.17 |
| Microcephaly | | 2.12 | 2.23 | 2.09 | 2.14 | 1.55 | 1.06 | 0.87 |
| Holoprosencephaly | | 0.35 | 0.43 | 0.63 | 0.52 | 0.43 | 0.25 | 0.37 |
| Hydrocephaly | | 3.19 | 2.23 | 2.96 | 2.98 | 2.12 | 1.88 | 1.83 |
| Anophthalmos | | 0.71 | 0.64 | 0.30 | 0.27 | 0.14 | 0.17 | 0.19 |
| Microphthalmos | | 1.50 | 2.11 | 1.76 | 1.62 | 1.20 | 0.95 | 1.06 |
| Unspecified Anophthalmos/Microphthalmos | | 0.00 | 0.00 | 0.00 | 0.00 | 0.02 | 0.00 | 0.29 |
| Anotia | | 0.00 | 0.03 | 0.03 | 0.23 | 0.10 | 0.09 | 0.06 |
| Microtia | | 2.21 | 2.05 | 1.49 | 1.31 | 1.45 | 1.52 | 1.75 |
| Unspecified Anotia/Microtia | | 0.00 | 0.00 | 0.00 | 0.00 | 0.02 | 0.00 | 0.27 |
| Transposition of great vessels | | 0.62 | 0.67 | 1.25 | 1.49 | 1.24 | 1.22 | 0.67 |
| Tetralogy of Fallot | | 0.18 | 0.24 | 0.66 | 1.13 | 1.12 | 0.93 | 0.67 |
| Hypoplastic left heart syndrome | | 0.27 | 0.37 | 0.75 | 0.70 | 0.43 | 0.30 | 0.08 |
| Coarctation of aorta | | 0.71 | 0.24 | 0.69 | 0.79 | 0.88 | 0.74 | 0.67 |
| Choanal atresia, bilateral | | 0.00 | 0.15 | 0.51 | 0.18 | 0.18 | 0.17 | 0.21 |
| Cleft palate without cleft lip | | 4.96 | 4.91 | 5.05 | 4.31 | 3.79 | 4.10 | 3.08 |
| Cleft lip with or without cleft palate | | 5.93 | 5.68 | 5.76 | 5.26 | 3.75 | 3.85 | 3.06 |
| Oesophageal atresia/stenosis with or without fistula | | 1.68 | 2.41 | 1.85 | 2.14 | 1.57 | 2.13 | 1.21 |
| Small intestine atresia/stenosis | | 0.53 | 0.52 | 0.57 | 0.52 | 0.33 | 0.63 | 0.52 |
| Anorectal atresia/stenosis | | 2.48 | 2.66 | 1.97 | 2.08 | 2.20 | 1.92 | 1.46 |
| Undescended testis (36 weeks of gestation or later) | | 1.24 | 2.14 | 2.69 | 2.59 | 3.10 | 2.11 | 2.33 |
| Hypospadias | | 2.65 | 2.66 | 2.21 | 1.69 | 1.93 | 1.97 | 1.64 |
| Epispadias | | 0.44 | 0.15 | 0.30 | 0.05 | 0.12 | 0.06 | 0.10 |
| Indeterminate sex | | 0.53 | 1.16 | 1.08 | 0.65 | 0.67 | 0.44 | 0.44 |
| Renal agenesis | | 0.62 | 0.73 | 0.87 | 0.59 | 0.28 | 0.08 | 0.15 |
| Cystic kidney | | 1.50 | 1.13 | 1.70 | 1.67 | 1.69 | 1.50 | 1.48 |
| Bladder exstrophy | | 0.27 | 0.27 | 0.27 | 0.25 | 0.31 | 0.15 | 0.12 |
| Polydactyly, preaxial | | 2.57 | 2.38 | 2.96 | 2.91 | 2.65 | 2.15 | 1.93 |
| Total Limb reduction defects (include unspecified) | | 7.52 | 6.84 | 7.23 | 6.54 | 5.21 | 4.46 | 3.95 |
| Transverse | | 2.83 | 3.11 | 3.08 | 2.19 | 2.22 | 1.71 | 1.69 |
| Preaxial | | 1.15 | 1.16 | 0.93 | 0.90 | 0.67 | 0.57 | 0.52 |
| Postaxial | | 0.27 | 0.09 | 0.15 | 0.25 | 0.18 | 0.11 | 0.10 |
| Intercalary | | 0.53 | 0.46 | 0.33 | 0.59 | 0.16 | 0.30 | 0.15 |
| Mixed | | 1.50 | 0.92 | 1.28 | 1.08 | 1.04 | 0.85 | 0.75 |
| Unspecified | | 1.24 | 1.10 | 1.46 | 1.53 | 0.90 | 0.49 | 0.71 |
| Diaphragmatic hernia | | 2.48 | 2.56 | 2.21 | 2.08 | 1.12 | 0.66 | 0.98 |
| Omphalocele | | 2.12 | 1.50 | 1.49 | 1.02 | 0.61 | 0.55 | 0.56 |
| Gastroschisis | | 0.80 | 0.40 | 0.48 | 0.36 | 0.41 | 0.46 | 0.60 |
| Unspecified Omphalocele/Gastroschisis | | 0.27 | 0.37 | 0.33 | 0.09 | 0.06 | 0.02 | 0.02 |
| Prune belly sequence | | 0.44 | 0.58 | 0.66 | 0.41 | 0.20 | 0.21 | 0.27 |
| Trisomy 13 | | 0.27 | 0.37 | 0.48 | 0.45 | 0.47 | 0.36 | 0.23 |
| Trisomy 18 | | 0.44 | 1.34 | 0.90 | 0.81 | 0.67 | 0.61 | 0.58 |
| Down syndrome, all ages (include age unknown) | | 14.60 | 15.02 | 13.92 | 11.76 | 9.80 | 7.35 | 6.80 |
| <20 | | 8.72 | 7.08 | 10.76 | 3.34 | 1.18 | 3.83 | 6.12 |
| 20-24 | | 8.44 | 5.86 | 5.27 | 5.69 | 4.72 | 5.20 | 3.30 |
| 25-29 | | 5.33 | 7.23 | 8.17 | 6.55 | 6.11 | 3.85 | 2.55 |
| 30-34 | | 12.31 | 11.74 | 14.28 | 12.88 | 9.03 | 6.55 | 4.77 |
| 35-39 | | 40.80 | 48.10 | 39.94 | 32.58 | 17.72 | 11.65 | 12.48 |
| 40-44 | | 117.61 | 189.43 | 129.80 | 51.00 | 52.03 | 31.95 | 33.45 |
| 45+ | | 163.27 | 246.91 | 137.93 | 265.49 | 2666.67 | 42.55 | 112.57 |
| unknown | | --- | --- | --- | --- | --- | --- | --- |

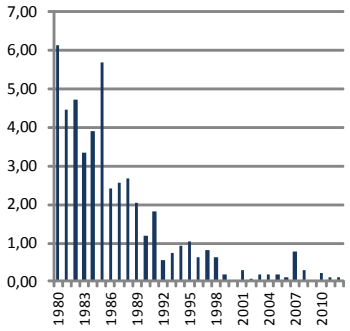
nr = data not reported or not available

* data include less than 5 years

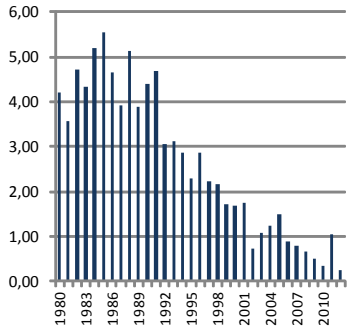
# Spain: ECEMC, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

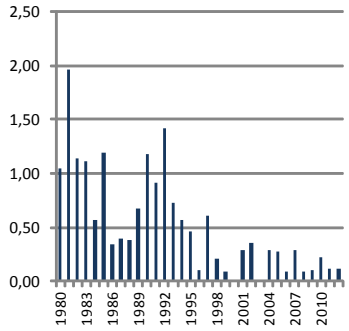
**Anencephaly**



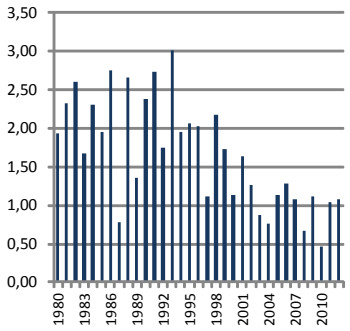

**Spina Bifida**



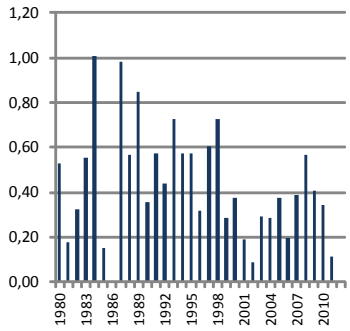

**Encephalocele**



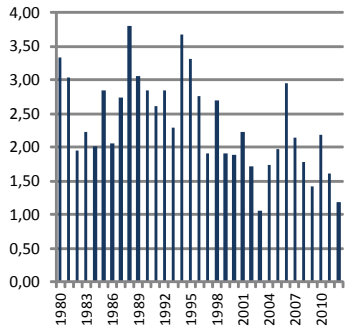

**Microcephaly**



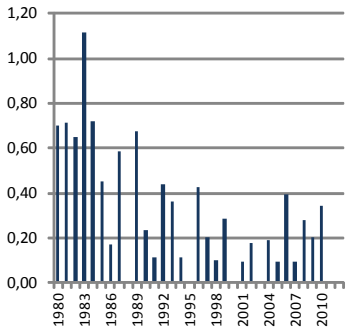

**Holoprosencephaly**



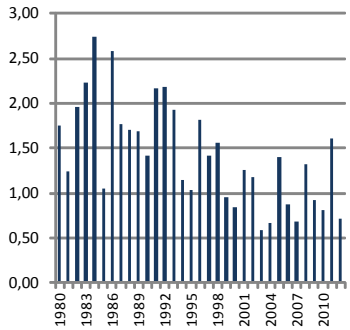

**Hydrocephaly**



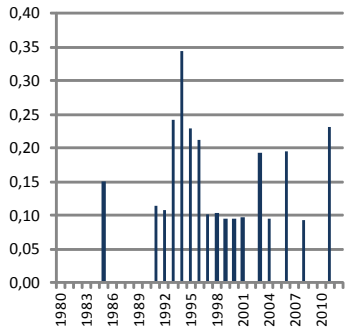

**Anophthalmos**



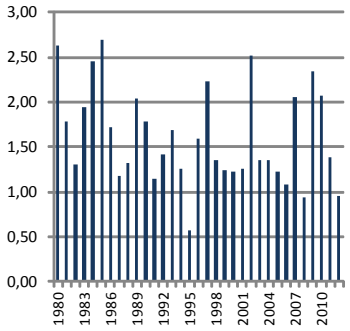

**Microphthalmos**



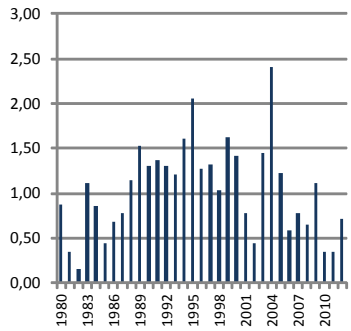

**Anotia**



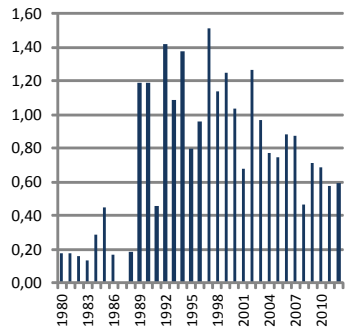

**Microtia**




**Transposition of great vessels**



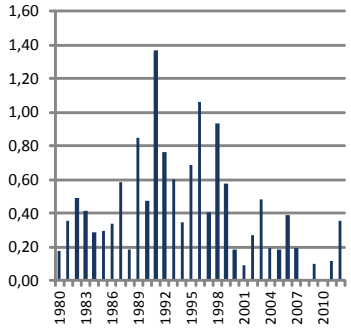

**Tetralogy of Fallot**



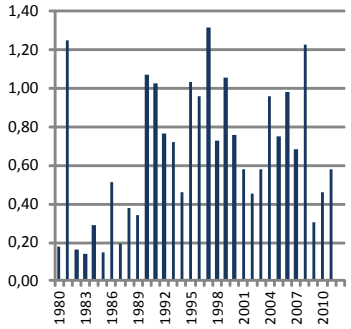
# Spain: ECEMC, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

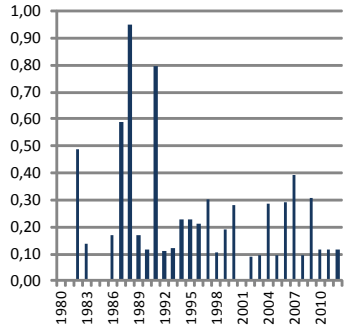
**Hypoplastic left heart syndrome**



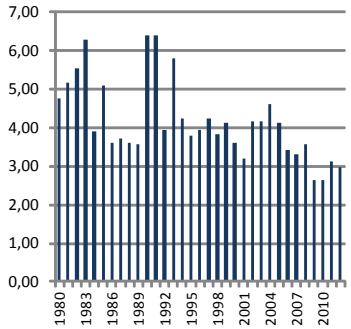

**Coarctation of aorta**



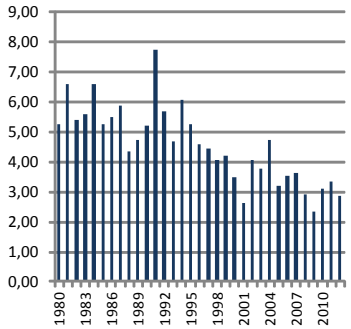

**Choanal atresia, bilateral**



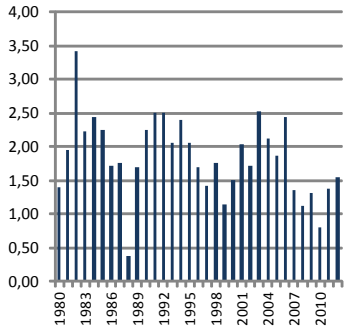

**Cleft palate without cleft lip**



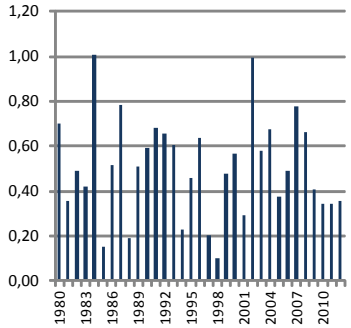

**Cleft lip with or without cleft palate**



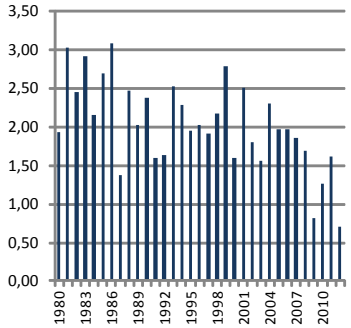

**phageal atresia/stenosis with or without f**



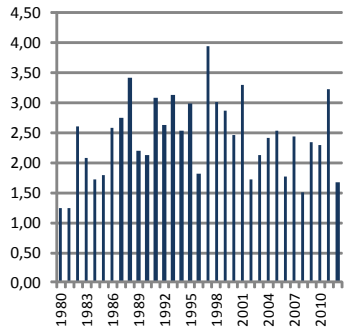

**Small intestine atresia/stenosis**



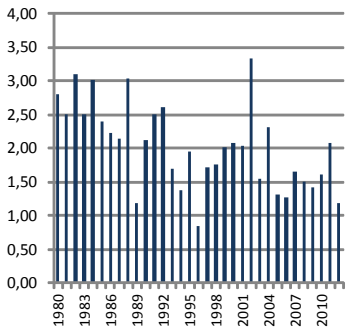

**Anorectal atresia/stenosis**



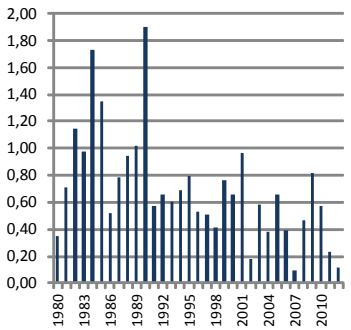

**Undescended testis**



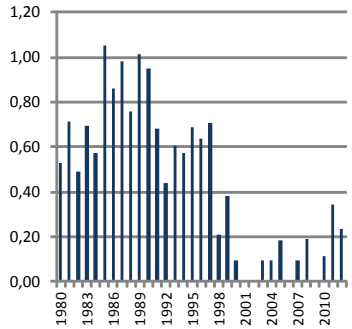

**Hypospadias**




**Indeterminate sex**



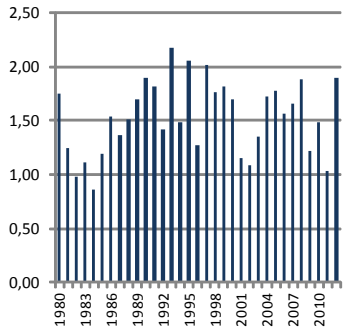

**Renal agenesis**



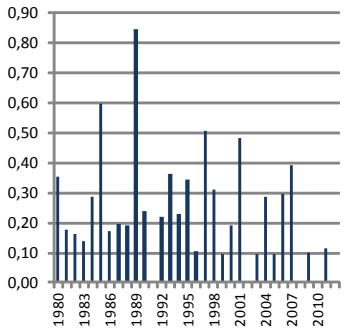
# Spain: ECEMC, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

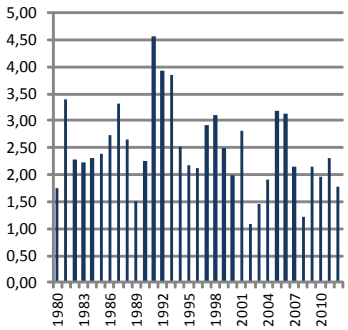
**Cystic kidney**



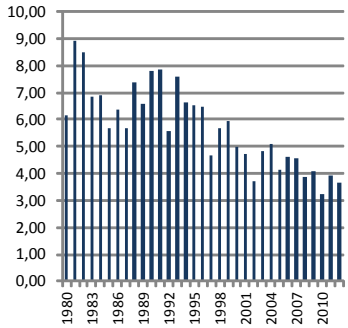

**Bladder exstrophy**



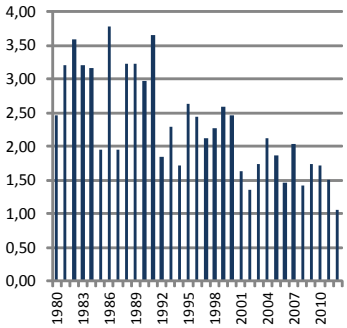

**Polydactyly, preaxial**



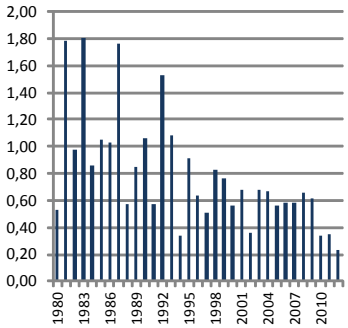

**Limb reduction defects**



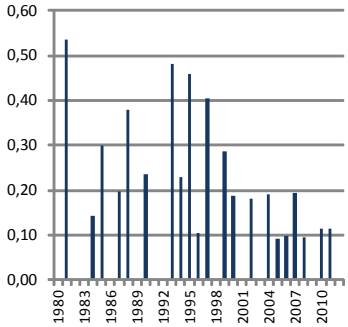

**Limb reduction defects - transverse**



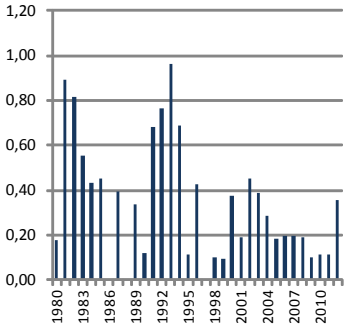

**Limb reduction defects - preaxial**



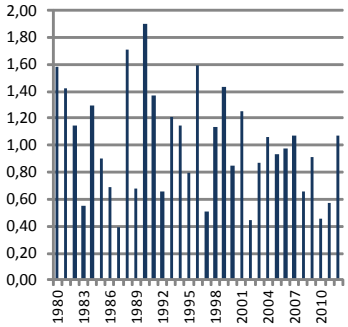

**Limb reduction defects - postaxial**



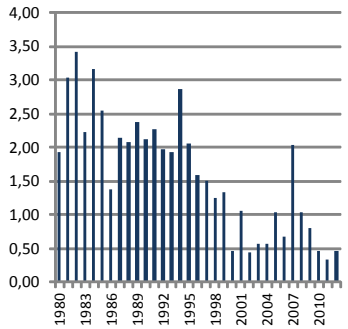

**Limb reduction defects - intercalary**



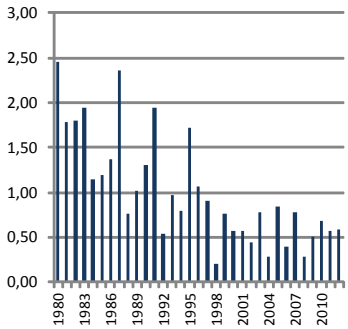

**Limb reduction defects - mixed**



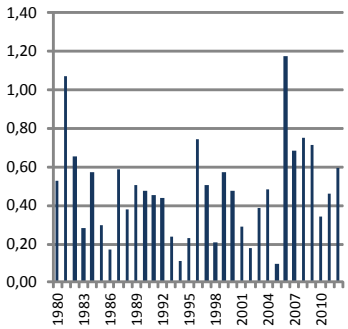

**Diaphragmatic hernia**




**Omphalocele**



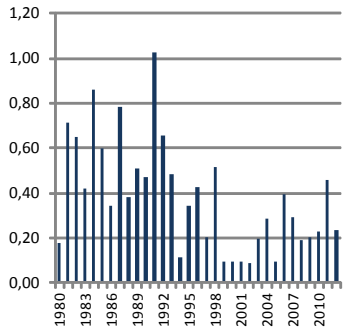

**Gastroschisis**



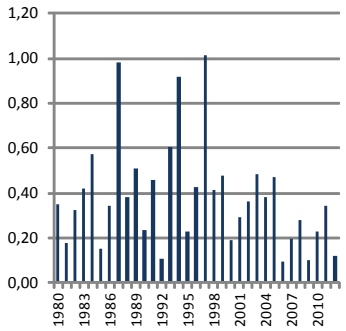
# Spain: ECEMC, Time trends 1980 – 2012

(Birth prevalence rates per 10,000)

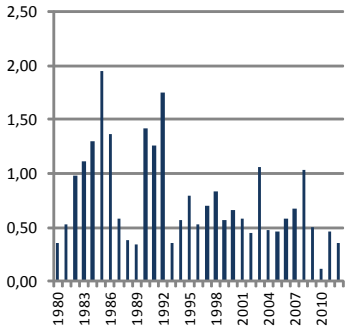
**Prune belly sequence**



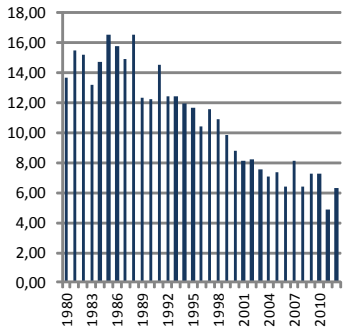

**Trisomy 13**



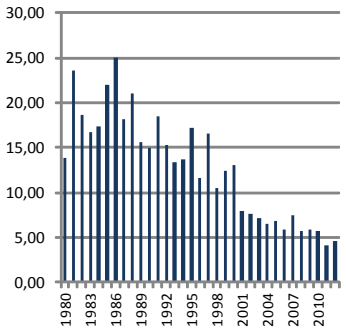

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates**

# Ukraine: OMNI-Net

## Ukraine Birth Defects Program

**History:**
Population based birth defects surveillance began in 2000 in the framework of the Ukrainian-American Birth Defects Program (UABDP) funded by the United States Agency for International Development (USAID). The program became an associate member of ICBDSR in 2001. In 2005 the USAID component was completed and the program was assumed by OMNI-Net, a not-forprofit international organization incorporated in Ukraine, and is continued as OMNI-Net Ukraine Birth Defects Program. OMNI-Net represents five resource OMNI-Centers all of which provide care for children with birth defects, promote prevention programs, participate in parental organizations and engage in collaborative programs with national and international partners.

Program objectives include universal folic acid flour fortification, methods to reduce alcohol impact on child development in collaboration with partners and promoting international partnerships.

**Legislation and funding:**
OMNI-Net personnel are financed from regional budgets. The legislation and rules by the Ministry of Health mandates the reporting of birth defects. BD data is reported by Oblast Vital Statistics Centrum who aggregates, formats and forwards the data to the Ministry of Health.

**Population Coverage:**
BD surveillance annually covers about 30000 births in two oblasts (provinces) of Northwestern Ukraine – Rivne and Volyn, representing approximately 6% of births in Ukraine. The population is relatively homogeneous and stable (data is pooled from these two oblasts). The northern counties (rayons) of both oblasts are contaminated from Chornobyl disaster.

**Sources of ascertainment**:
Relevant hospital admission/discharge summaries are systematically reviewed. Qualified Registry specialists also routinely review all medical records of regional pediatric cardiology centres and obtain ascertainment of diagnostic details. Data from specialty clinics, laboratories (including cytogenetic one) and other services are explored. Our cytogenetic laboratories are the only ones in the region and they provide us with study reports. Pregnancy, obstetrics, delivery, neonatal and pediatrics records are reviewed. The information is substantial regarding service providers located in regional centres, but limited regarding service providers in rural environments.

**Maximum Age at Diagnosis:**
Up to 1 year of age.

**Exposure information:**
Routine information collection is limited except when ad hoc circumstances are noted. An expansion of exposure data collection is in progress.

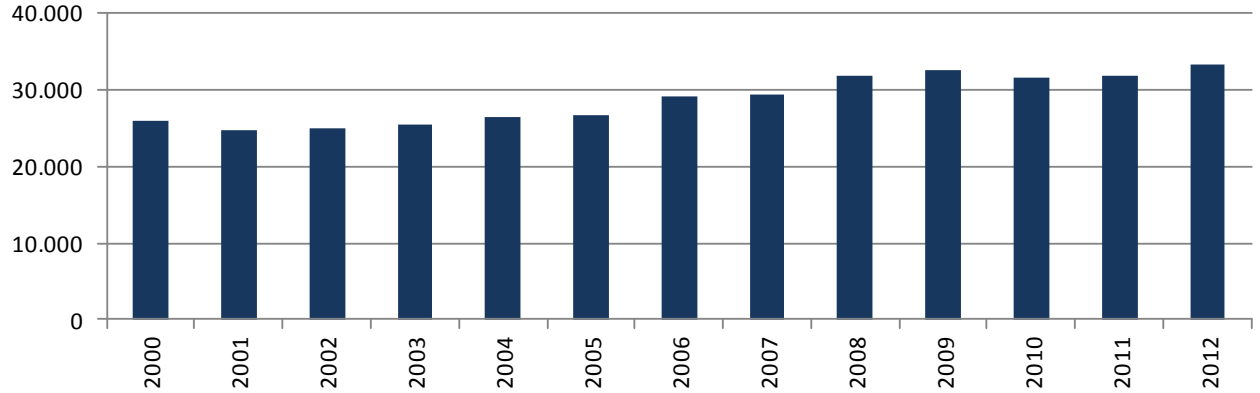
**Prenatal diagnosis information:**
The information is substantial regarding service providers located in regional centers, but limited regarding service providers in rural environment.

**Background information.**
Data regarding ionizing radiation pollution in contaminated rayons is available by special agreements. Data from a population based neonatal registry is also available by special agreements.

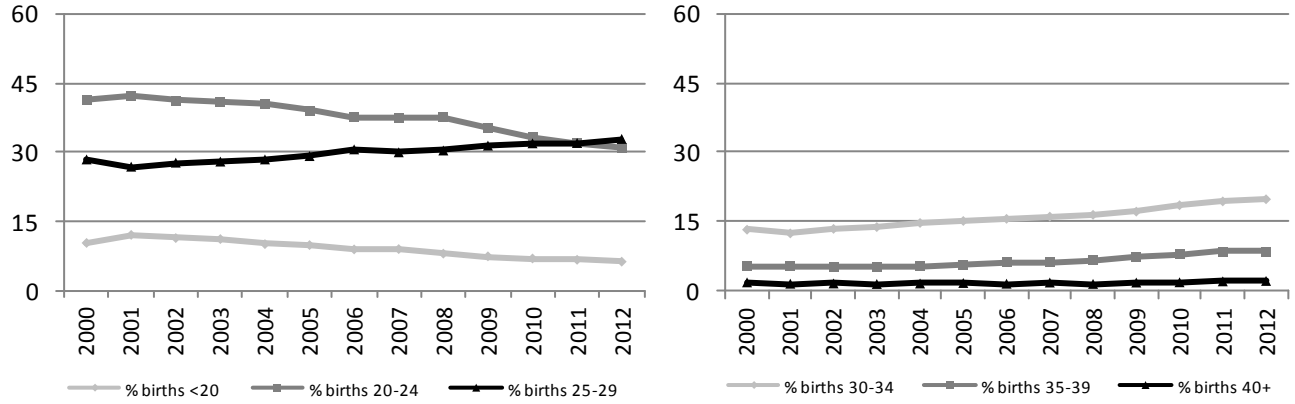
**Addresses and Staff:**
Program Director: Dr. Wladimir Wertelecki Medical Coordinator: Dr. Lyubov Yevtushok "OMNI-Net for Children", 36, 16 Lypnya Str., Room 709, Rivne, Ukraine 33028
**Phone/Fax:** 38 036 262 3447
**E-mail:** werteleckiomni@gmail.com
yevtushokl@gmail.com
rivneomni2@gmail.com
**Website:** http://www.ibis-birthdefects.org/

# Ukraine: OMNI-Net

## Total births by year




## Percentage of births by year and maternal age



# Ukraine: OMNI-Net, 2012

| | |
|---|---|
| Live births (LB) | 33,263 |
| Stillbirths (SB) | 179 |
| Total births | 33,442 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP([1]) | Total rate |
| Anencephaly | 0 | 6 | 18 | 7.18 |
| Spina bifida | 14 | 0 | 15 | 8.67 |
| Encephalocele | 1 | 0 | 10 | 3.29 |
| Microcephaly | 17 | 1 | nr | 5.38 |
| Holoprosencephaly([2]) | 2 | 0 | nr | 0.60 |
| Hydrocephaly | 11 | 2 | nr | 3.89 |
| Anophthalmos([2]) | 0 | 0 | nr | 0.00 |
| Microphthalmos | 0 | 1 | nr | 0.30 |
| Unspecified Anophthalmos/Microphthalmos([2]) | 0 | 0 | nr | 0.00 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 7 | 0 | nr | 2.09 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 17 | 1 | nr | 5.38 |
| Tetralogy of Fallot | 3 | 1 | nr | 1.20 |
| Hypoplastic left heart syndrome | 8 | 1 | nr | 2.69 |
| Coarctation of aorta | 10 | 0 | nr | 2.99 |
| Choanal atresia, bilateral | 0 | 0 | nr | 0.00 |
| Cleft palate without cleft lip | 20 | 0 | nr | 5.98 |
| Cleft lip with or without cleft palate | 25 | 3 | nr | 8.37 |
| Oesophageal atresia/stenosis with or without fistula | 9 | 0 | nr | 2.69 |
| Small intestine atresia/stenosis | 3 | 0 | nr | 0.90 |
| Anorectal atresia/stenosis | 5 | 0 | nr | 1.50 |
| Undescended testis (36 weeks of gestation or later) | 61 | 0 | nr | 18.24 |
| Hypospadias([3]) | 13 | 0 | nr | 3.89 |
| Epispadias | 0 | 0 | nr | 0.00 |
| Indeterminate sex | 0 | 0 | nr | 0.00 |
| Renal agenesis | 3 | 1 | nr | 1.20 |
| Cystic kidney | 14 | 2 | nr | 4.78 |
| Bladder exstrophy | 0 | 0 | nr | 0.00 |
| Polydactyly, preaxial | 8 | 0 | nr | 2.39 |
| Total Limb reduction defects (include unspecified) | 9 | 0 | nr | 2.69 |
| Transverse | 4 | 0 | nr | 1.20 |
| Preaxial | 1 | 0 | nr | 0.30 |
| Postaxial | 1 | 0 | nr | 0.30 |
| Intercalary | 2 | 0 | nr | 0.60 |
| Mixed | 0 | 0 | nr | 0.00 |
| Unspecified | 0 | 0 | nr | 0.00 |
| Diaphragmatic hernia | 4 | 1 | nr | 1.50 |
| Omphalocele | 2 | 2 | nr | 1.20 |
| Gastroschisis | 5 | 0 | nr | 1.50 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13([4]) | 0 | 1 | nr | 0.30 |
| Trisomy 18([5]) | 4 | 1 | nr | 1.50 |
| Down syndrome, all ages (include age unknown)([2,6]) | 48 | 0 | nr | 14.35 |
| <20 | 0 | 0 | nr | 0.00 |
| 20-24 | 5 | 0 | nr | 4.84 |
| 25-29 | 11 | 0 | nr | 10.01 |
| 30-34 | 9 | 0 | nr | 13.65 |
| 35-39 | 14 | 0 | nr | 50.25 |
| 40-44 | 8 | 0 | nr | 130.51 |
| 45+ | 1 | 0 | nr | 200.00 |
| unknown | 0 | 0 | nr | --- |

nr = data not reported or not available
(1) Number of terminations of pregnancy (ToP) for birth defects is not reported, except for NTD;
(2) Clinical diagnosis only; with photodocumentation or measurements documented;
(3) Includes penile, scrotal, and perineal hypospadias only;
(4) One ToPs with Trisomy 13 confirmed by amniocentesis
(5) Two ToPs with Trisomy 18 confirmed by amniocentesis
(6) Nine ToPs with Down Syndrome confirmed by amniocentesis

# Ukraine: OMNI-Net, Previous years rates 2000 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001* | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | **50,771** | **132,575** | **157,170** |
| Anencephaly | | | | | | 8.47 | 8.75 | 7.00 |
| Spina bifida | | | | | | 9.85 | 11.54 | 10.37 |
| Encephalocele | | | | | | 2.36 | 2.04 | 1.78 |
| Microcephaly | | | | | | 2.36 | 3.17 | 5.85 |
| Holoprosencephaly | | | | | | 0.00 | 0.75 | 1.53 |
| Hydrocephaly | | | | | | 6.30 | 5.58 | 5.28 |
| Anophthalmos | | | | | | 0.20 | 0.00 | 0.25 |
| Microphthalmos | | | | | | 1.77 | 0.83 | 1.34 |
| Unspecified Anophthalmos/Microphthalmos | | | | | | 0.00 | 0.00 | 0.06 |
| Anotia | | | | | | 0.20 | 0.45 | 0.25 |
| Microtia | | | | | | 1.38 | 1.96 | 2.23 |
| Unspecified Anotia/Microtia | | | | | | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | | | | | 3.94 | 3.47 | 4.20 |
| Tetralogy of Fallot | | | | | | 1.38 | 2.26 | 2.67 |
| Hypoplastic left heart syndrome | | | | | | 0.59 | 1.36 | 2.04 |
| Coarctation of aorta | | | | | | 0.79 | 1.43 | 1.72 |
| Choanal atresia, bilateral | | | | | | 0.00 | 0.00 | 0.06 |
| Cleft palate without cleft lip | | | | | | 2.95 | 4.98 | 7.19 |
| Cleft lip with or without cleft palate | | | | | | 8.67 | 9.13 | 7.83 |
| Oesophageal atresia/stenosis with or without fistula | | | | | | 1.77 | 1.96 | 2.23 |
| Small intestine atresia/stenosis | | | | | | 1.38 | 1.58 | 1.53 |
| Anorectal atresia/stenosis | | | | | | 2.36 | 2.41 | 2.16 |
| Undescended testis (36 weeks of gestation or later) | | | | | | 36.64 | 41.71 | 31.56 |
| Hypospadias | | | | | | 3.55 | 3.32 | 2.80 |
| Epispadias | | | | | | 0.59 | 0.15 | 0.06 |
| Indeterminate sex | | | | | | 0.59 | 0.45 | 0.25 |
| Renal agenesis | | | | | | 0.79 | 0.75 | 0.89 |
| Cystic kidney | | | | | | 0.98 | 2.87 | 4.77 |
| Bladder exstrophy | | | | | | 0.79 | 0.75 | 0.38 |
| Polydactyly, preaxial | | | | | | 2.76 | 3.62 | 4.14 |
| Total Limb reduction defects (include unspecified) | | | | | | 4.33 | 3.02 | 4.84 |
| Transverse | | | | | | 2.56 | 1.66 | 3.05 |
| Preaxial | | | | | | 0.39 | 0.45 | 0.51 |
| Postaxial | | | | | | 0.59 | 0.15 | 0.25 |
| Intercalary | | | | | | 0.39 | 0.30 | 0.25 |
| Mixed | | | | | | 0.20 | 0.23 | 0.45 |
| Unspecified | | | | | | 0.20 | 0.23 | 0.00 |
| Diaphragmatic hernia | | | | | | 2.17 | 1.66 | 2.93 |
| Omphalocele | | | | | | 0.98 | 1.36 | 1.97 |
| Gastroschisis | | | | | | 0.59 | 1.66 | 1.34 |
| Unspecified Omphalocele/Gastroschisis | | | | | | 0.00 | 0.00 | 0.00 |
| Prune belly sequence | | | | | | 0.00 | 0.00 | 0.00 |
| Trisomy 13 | | | | | | 0.20 | 0.38 | 0.13 |
| Trisomy 18 | | | | | | 0.59 | 0.23 | 0.45 |
| Down syndrome, all ages (include age unknown) | | | | | | 10.83 | 13.50 | 14.38 |
| <20 | | | | | | 8.86 | 10.38 | 6.77 |
| 20-24 | | | | | | 5.66 | 7.59 | 7.08 |
| 25-29 | | | | | | 7.84 | 9.95 | 9.81 |
| 30-34 | | | | | | 15.49 | 15.15 | 17.54 |
| 35-39 | | | | | | 15.10 | 43.28 | 35.27 |
| 40-44 | | | | | | 136.61 | 112.23 | 160.89 |
| 45+ | | | | | | 638.30 | 666.67 | 259.74 |
| unknown | | | | | | --- | --- | --- |

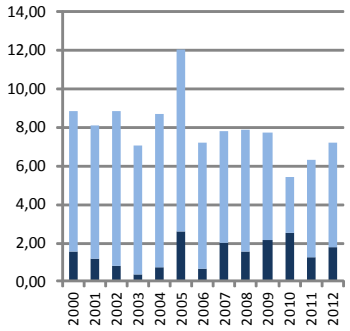
nr = data not reported or not available

* data include less than 5 years

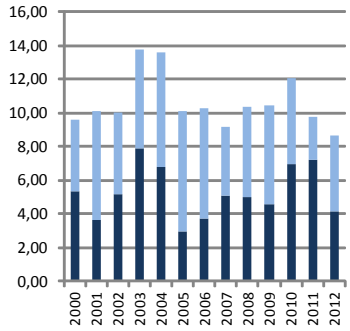
# Ukraine: OMNI-Net, Time trends 2000 – 2012

(Birth prevalence rates per 10,000)

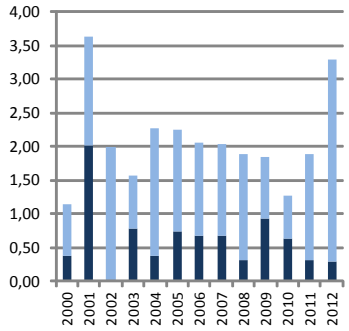
**Anencephaly**



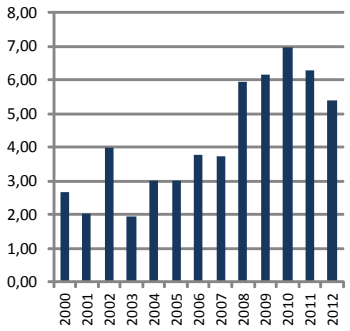

**Spina Bifida**



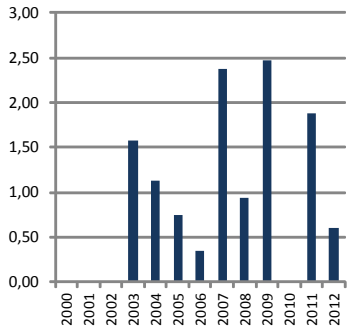

**Encephalocele**



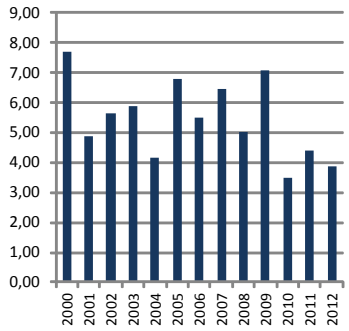

**Microcephaly**



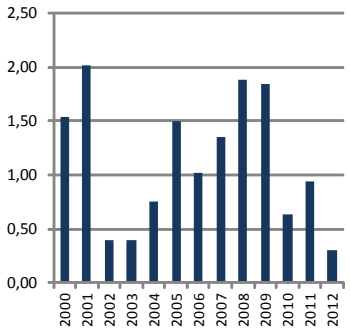

**Holoprosencephaly**



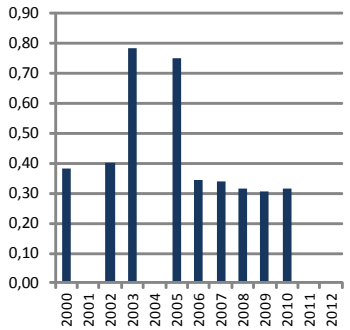

**Hydrocephaly**



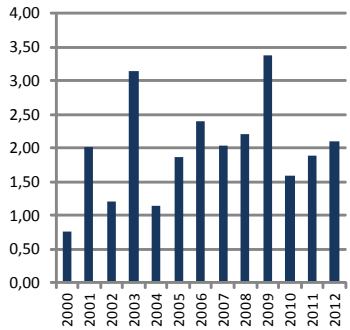

**Microphthalmos**



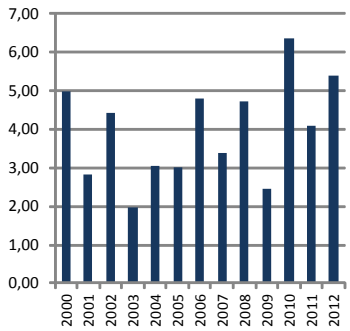

**Anotia**



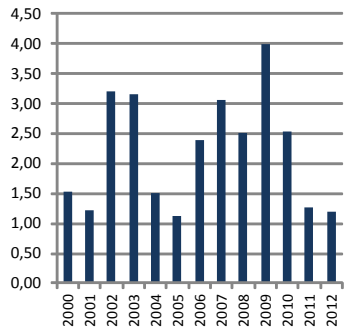

**Microtia**



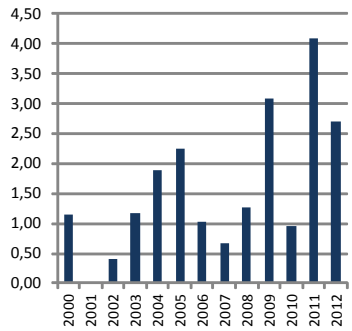

**Transposition of great vessels**




**Tetralogy of Fallot**



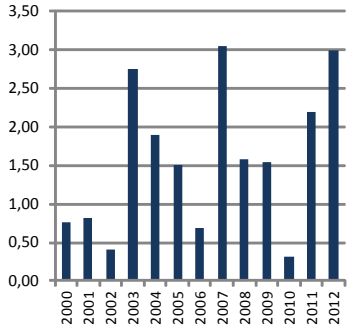

**Hypoplastic left heart syndrome**



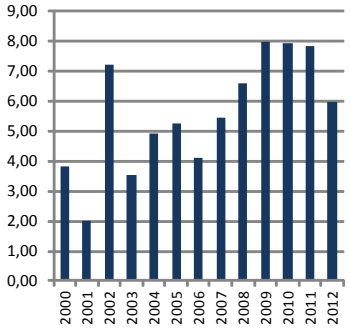
# Ukraine: OMNI-Net, Time trends 2000 – 2012

(Birth prevalence rates per 10,000)

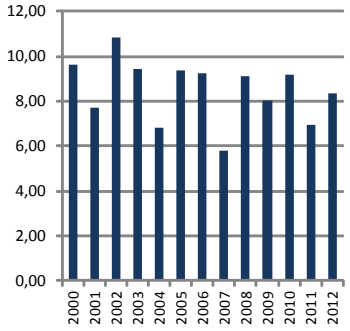
**Coarctation of aorta**



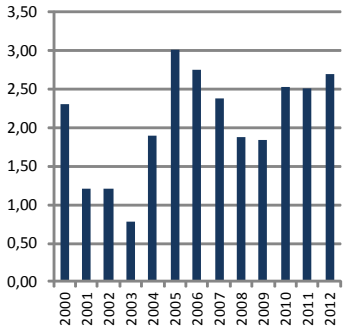

**Cleft palate without cleft lip**



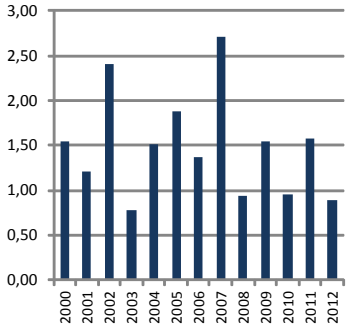

**Cleft lip with or without cleft palate**



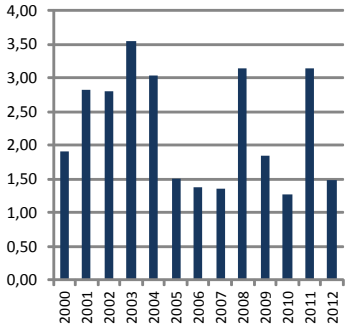

**phageal atresia/stenosis with or without f**



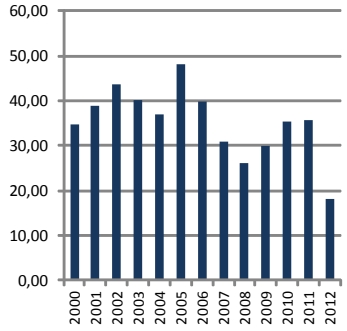

**Small intestine atresia/stenosis**



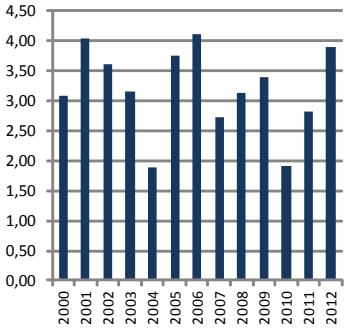

**Anorectal atresia/stenosis**



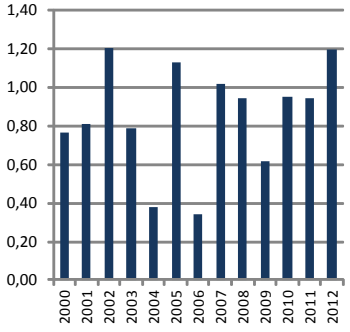

**Undescended testis**



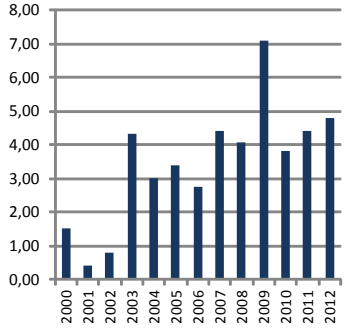

**Hypospadias**



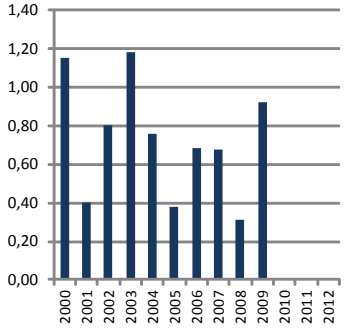

**Renal agenesis**



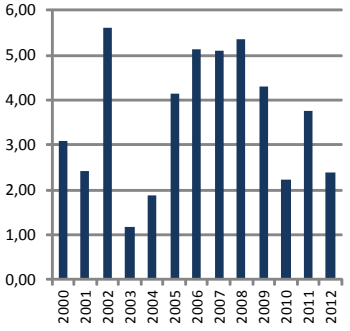

**Cystic kidney**




**Bladder exstrophy**



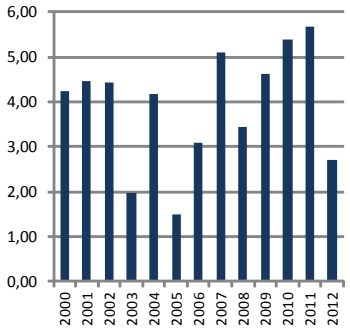

**Polydactyly, preaxial**



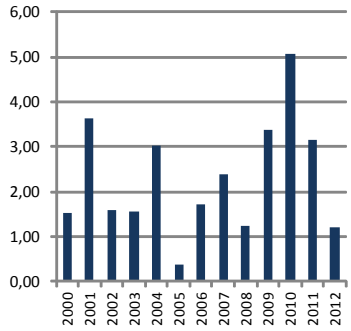
# Ukraine: OMNI-Net, Time trends 2000 – 2012

(Birth prevalence rates per 10,000)

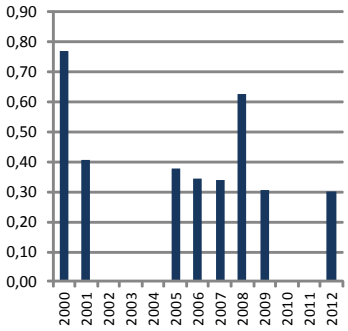
**Limb reduction defects**



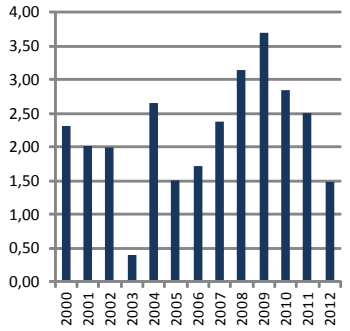

**Limb reduction defects - transverse**



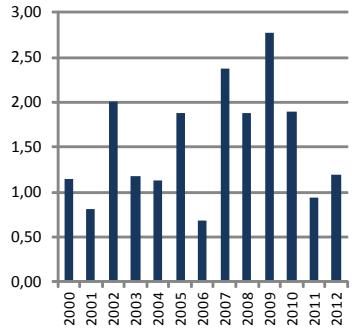

**Limb reduction defects - postaxialis**



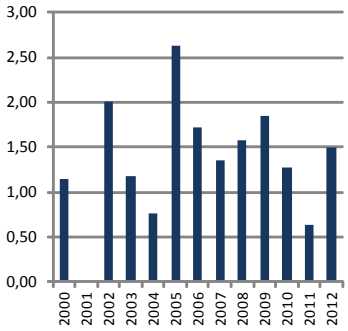

**Diaphragmatic hernia**



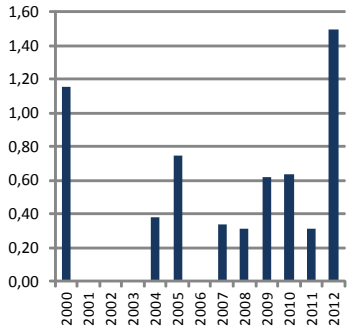

**Omphalocele**



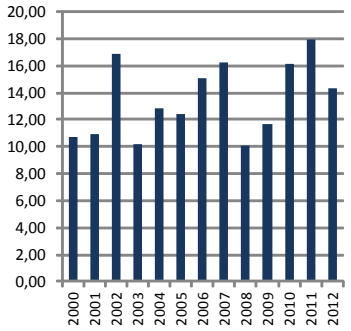

**Gastroschisis**



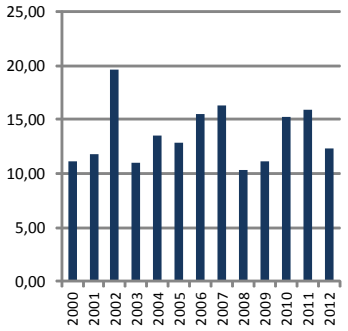

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# United Kingdom-Wales: CARIS

## Congenital Anomaly Register and Information System for Wales

**History:**
CARIS aims to describe the pattern of congenital anomalies in Wales and provide:

- a description of anomalies and rates
- an assessment of antenatal detection and interventions
- information for health care planning
- identification of clusters and causes

Start of data collection: 1.1.1998. ICBDSR member: 2004. EUROCAT member: 1998. Funding: Public Health Wales. Base: Singleton Hospital, Swansea.

**Population Coverage:**
All pregnancies of mothers normally resident in Wales. This includes spontaneous fetal losses and terminations of pregnancy. Annual live birth rate of 35,000.

**Sources of Ascertainment:**
Voluntary reporting Mutiple source reporting including inpatient data Clinical obstetric and paediatric champion in each delivery unit Data coordinator in each delivery unit Data exchange with bordering registers in England

**Termination of Pregnancy:**
Legal up to 24 weeks gestation in any pregnancy but no upper age limit for cases of major anomaly.

**Stillbirth Definition and Early Fetal Deaths:**
Stillbirth = fetal death at or after 24 weeks gestation. No lower limit for inclusion of spontaneous fetal losses.

**Exposure Data Availability:**
Maternal drugs, folic acid dosage and timing, maternal and paternal diseases and occupations.

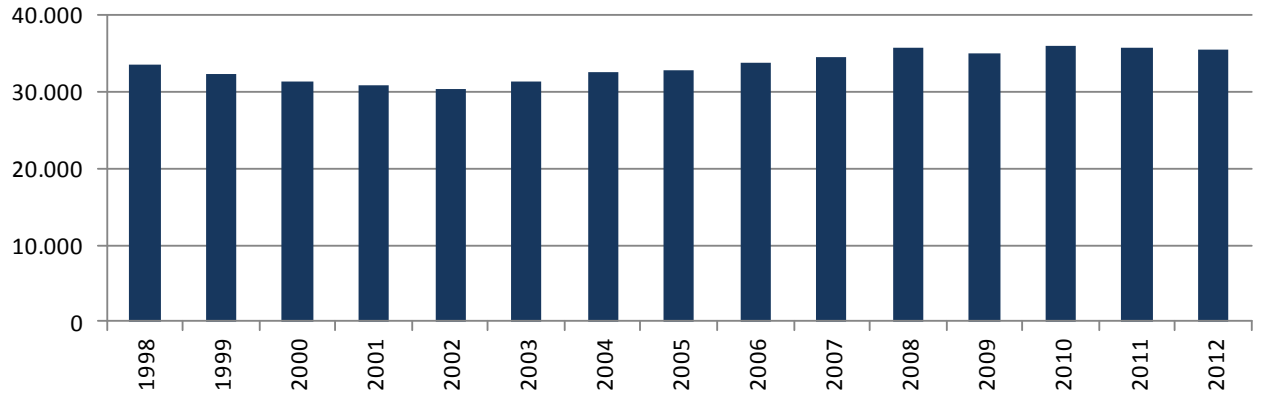
**Denominators and Controls Information:**
Data obtained from Office for National Statistics.

**Address and Staff:**
Margery Morgan, Programme Director
Congenital Anomaly Register and Information Service for Wales (CARIS)
Public Health Wales
Singleton Hospital
Sketty Lane
Swansea, Wales, UK, SA2 8QA
**Phone:** 44-1792-285241
**Fax:** 44-1792-285241

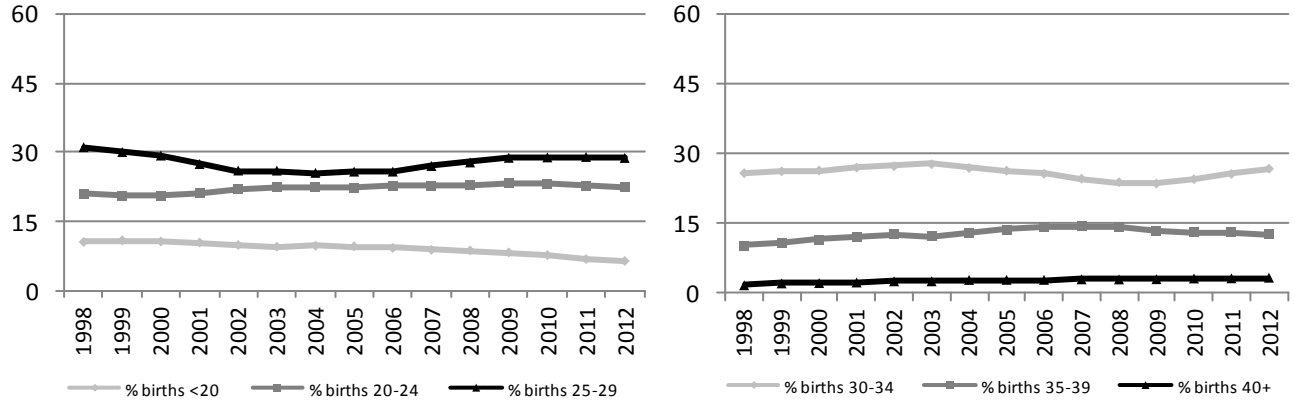
Relevant Contact Person: David Tucker
**E-mail:** david.tucker2@wales.nhs.uk

# United Kingdom-Wales: CARIS

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 51 | 98.1 | Cystic kidney | 22 | 20.4 |
| Spina bifida | 50 | 61.0 | Limb reduction defects | 19 | 31.7 |
| Encephalocele | 14 | 82.4 | Diaphragmatic hernia | 13 | 32.5 |
| Holoprosencephaly | 10 | 83.3 | Omphalocele | 17 | 43.6 |
| Hydrocephaly | 29 | 37.2 | Gastroschisis | 4 | 8.5 |
| Hypoplastic left heart syndrome | 14 | 45.2 | Trisomy 13 | 16 | 84.2 |
| Cleft palate without cleft lip | 8 | 8.6 | Trisomy 18 | 46 | 80.7 |
| Cleft lip with or without cleft palate | 14 | 13.3 | Down syndrome | 113 | 47.7 |
| Renal agenesis | 12 | 85.7 | | | |

Total ToPs with births defects = 508 (Ratio ToPs/Births: 4.73 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# United Kingdom-Wales: CARIS, 2012

| | |
|---|---|
| Live births (LB) | 35,238 |
| Stillbirths (SB) | 181 |
| Total births | 35,419 |
| Number of terminations of pregnancy (ToP) for birth defects | 157 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 0 | 14 | 4.24 |
| Spina bifida | 9 | 1 | 12 | 6.21 |
| Encephalocele | 0 | 0 | 2 | 0.56 |
| Microcephaly | 11 | 0 | 0 | 3.11 |
| Holoprosencephaly | 0 | 0 | 8 | 2.26 |
| Hydrocephaly | 17 | 2 | 12 | 8.75 |
| Anophthalmos | 1 | 0 | 0 | 0.28 |
| Microphthalmos | 1 | 0 | 0 | 0.28 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.28 |
| Microtia | 1 | 0 | 0 | 0.28 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 11 | 0 | 2 | 3.67 |
| Tetralogy of Fallot | 12 | 0 | 0 | 3.39 |
| Hypoplastic left heart syndrome | 10 | 0 | 5 | 4.24 |
| Coarctation of aorta | 16 | 0 | 0 | 4.52 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.56 |
| Cleft palate without cleft lip | 27 | 0 | 4 | 8.75 |
| Cleft lip with or without cleft palate | 31 | 0 | 8 | 11.01 |
| Oesophageal atresia/stenosis with or without fistula | 13 | 1 | 1 | 4.24 |
| Small intestine atresia/stenosis | 4 | 0 | 0 | 1.13 |
| Anorectal atresia/stenosis | 12 | 0 | 5 | 4.80 |
| Undescended testis (36 weeks of gestation or later) | 52 | 0 | 0 | 14.68 |
| Hypospadias | 95 | 0 | 0 | 26.82 |
| Epispadias | 2 | 0 | 0 | 0.56 |
| Indeterminate sex | 1 | 0 | 0 | 0.28 |
| Renal agenesis | 0 | 1 | 5 | 1.69 |
| Cystic kidney | 41 | 0 | 14 | 15.53 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 0 | 0 | 1 | 0.28 |
| Total Limb reduction defects (include unspecified) | 12 | 0 | 7 | 5.36 |
| Transverse | 6 | 0 | 5 | 3.11 |
| Preaxial | 3 | 0 | 2 | 1.41 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 2 | 0 | 0 | 0.56 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 1 | 0 | 0 | 0.28 |
| Diaphragmatic hernia | 11 | 0 | 2 | 3.67 |
| Omphalocele | 7 | 1 | 4 | 3.39 |
| Gastroschisis | 10 | 0 | 0 | 2.82 |
| Unspecified Omphalocele/Gastroschisis | 2 | 0 | 5 | 1.98 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | 5 | 1.69 |
| Trisomy 18 | 2 | 3 | 11 | 4.52 |
| Down syndrome, all ages (include age unknown) | 39 | 2 | 28 | 19.48 |
| <20 | 3 | 0 | 0 | 13.33 |
| 20-24 | 3 | 0 | 4 | 8.82 |
| 25-29 | 7 | 0 | 3 | 9.83 |
| 30-34 | 5 | 0 | 4 | 9.53 |
| 35-39 | 11 | 1 | 8 | 44.76 |
| 40-44 | 8 | 1 | 9 | 167.13 |
| 45+ | 2 | 0 | 0 | 289.86 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

# United Kingdom-Wales: CARIS, Previous years rates 1998 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001* | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | **128,106** | **160,736** | **177,409** |
| Anencephaly | | | | | | 7.34 | 6.41 | 5.07 |
| Spina bifida | | | | | | 8.66 | 7.28 | 7.21 |
| Encephalocele | | | | | | 2.34 | 2.12 | 2.03 |
| Microcephaly | | | | | | 7.26 | 4.98 | 4.34 |
| Holoprosencephaly | | | | | | 1.33 | 1.68 | 1.01 |
| Hydrocephaly | | | | | | 9.76 | 9.21 | 8.17 |
| Anophthalmos | | | | | | 0.62 | 0.25 | 0.00 |
| Microphthalmos | | | | | | 2.34 | 1.62 | 1.24 |
| Unspecified Anophthalmos/Microphthalmos | | | | | | 0.00 | 0.00 | 0.00 |
| Anotia | | | | | | 0.39 | 0.25 | 0.85 |
| Microtia | | | | | | 0.47 | 0.81 | 0.68 |
| Unspecified Anotia/Microtia | | | | | | 0.00 | 0.00 | 0.06 |
| Transposition of great vessels | | | | | | 5.46 | 4.23 | 4.57 |
| Tetralogy of Fallot | | | | | | 3.20 | 3.05 | 4.73 |
| Hypoplastic left heart syndrome | | | | | | 3.51 | 3.36 | 2.76 |
| Coarctation of aorta | | | | | | 6.09 | 6.28 | 5.02 |
| Choanal atresia, bilateral | | | | | | 0.16 | 0.25 | 0.34 |
| Cleft palate without cleft lip | | | | | | 9.76 | 9.83 | 8.40 |
| Cleft lip with or without cleft palate | | | | | | 10.23 | 10.51 | 11.44 |
| Oesophageal atresia/stenosis with or without fistula | | | | | | 3.28 | 3.36 | 2.93 |
| Small intestine atresia/stenosis | | | | | | 2.19 | 1.62 | 1.69 |
| Anorectal atresia/stenosis | | | | | | 5.23 | 3.17 | 4.17 |
| Undescended testis (36 weeks of gestation or later) | | | | | | 24.75 | 13.25 | 27.51 |
| Hypospadias | | | | | | 31.54 | 32.10 | 31.28 |
| Epispadias | | | | | | 0.62 | 0.25 | 0.23 |
| Indeterminate sex | | | | | | 0.31 | 0.87 | 0.51 |
| Renal agenesis | | | | | | 2.89 | 1.80 | 1.63 |
| Cystic kidney | | | | | | 10.30 | 9.89 | 8.23 |
| Bladder exstrophy | | | | | | 0.39 | 0.12 | 0.62 |
| Polydactyly, preaxial | | | | | | 0.94 | 1.24 | 0.73 |
| Total Limb reduction defects (include unspecified) | | | | | | 10.93 | 9.27 | 7.72 |
| Transverse | | | | | | 5.00 | 4.67 | 4.23 |
| Preaxial | | | | | | 1.72 | 1.31 | 1.30 |
| Postaxial | | | | | | 0.78 | 0.25 | 0.17 |
| Intercalary | | | | | | 1.41 | 1.93 | 0.90 |
| Mixed | | | | | | 0.86 | 1.00 | 0.62 |
| Unspecified | | | | | | 1.33 | 0.87 | 0.39 |
| Diaphragmatic hernia | | | | | | 3.75 | 3.80 | 3.83 |
| Omphalocele | | | | | | 3.43 | 4.29 | 4.06 |
| Gastroschisis | | | | | | 4.84 | 7.15 | 5.07 |
| Unspecified Omphalocele/Gastroschisis | | | | | | 0.47 | 0.56 | 0.28 |
| Prune belly sequence | | | | | | 0.23 | 0.06 | 0.11 |
| Trisomy 13 | | | | | | 2.81 | 1.80 | 2.09 |
| Trisomy 18 | | | | | | 4.06 | 5.72 | 6.26 |
| Down syndrome, all ages (include age unknown) | | | | | | 20.14 | 21.53 | 22.72 |
| <20 | | | | | | 10.35 | 5.88 | 9.87 |
| 20-24 | | | | | | 8.28 | 6.97 | 10.82 |
| 25-29 | | | | | | 11.66 | 10.13 | 8.78 |
| 30-34 | | | | | | 18.44 | 15.15 | 20.60 |
| 35-39 | | | | | | 54.57 | 59.08 | 54.70 |
| 40-44 | | | | | | 150.06 | 172.63 | 148.12 |
| 45+ | | | | | | 256.41 | 454.55 | 261.19 |
| unknown | | | | | | --- | --- | --- |

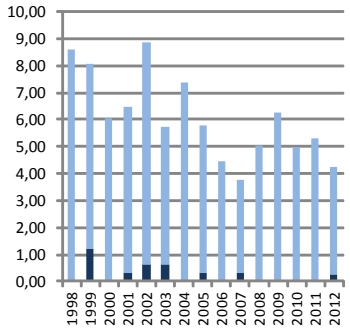
nr = data not reported or not available

* data include less than 5 years

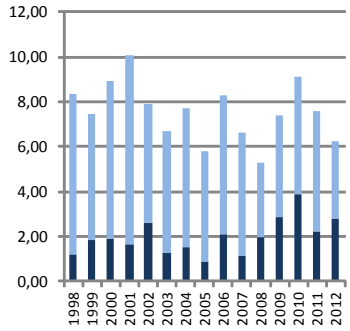
# United Kingdom-Wales: CARIS, Time trends 1998 – 2012

(Birth prevalence rates per 10,000)

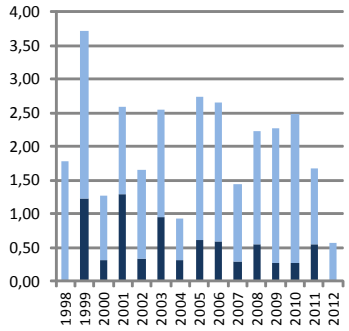
**Anencephaly**



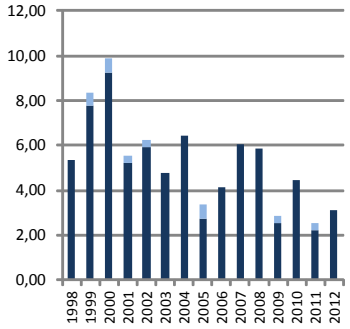

**Spina Bifida**



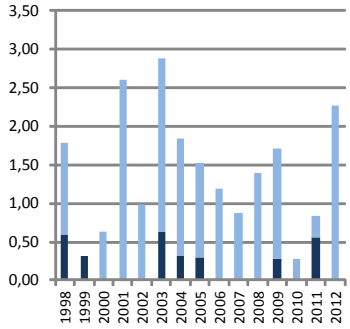

**Encephalocele**



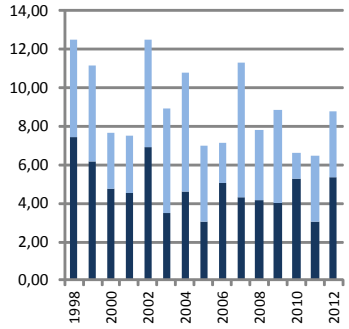

**Microcephaly**



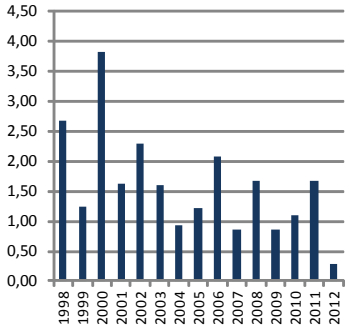

**Holoprosencephaly**



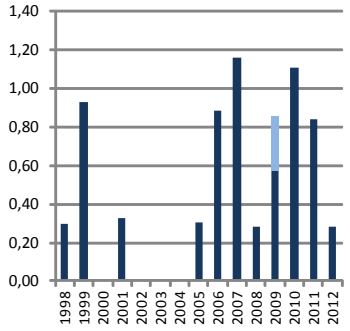

**Hydrocephaly**



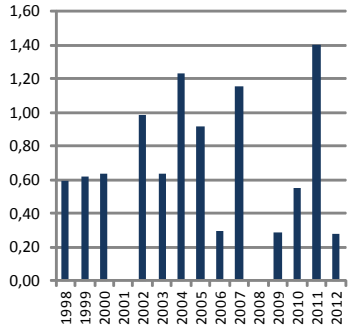

**Microphthalmos**



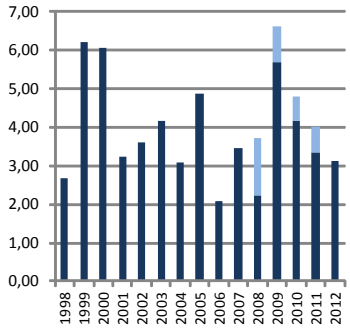

**Anotia**



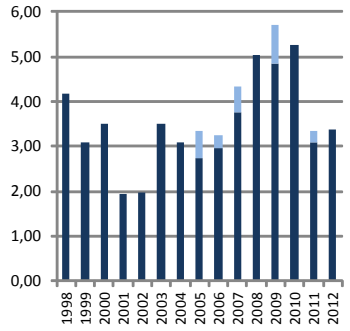

**Microtia**



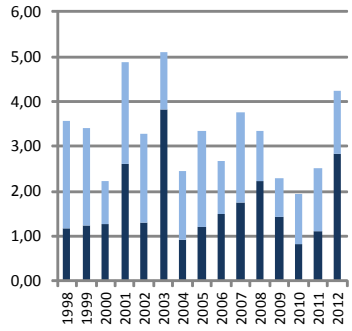

**Transposition of great vessels**




**Tetralogy of Fallot**



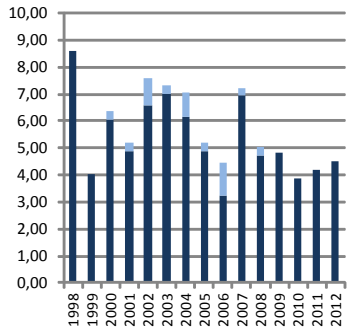

**Hypoplastic left heart syndrome**



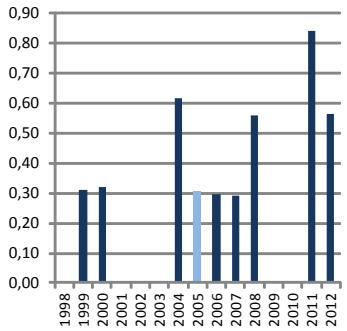
# United Kingdom-Wales: CARIS, Time trends 1998 – 2012

(Birth prevalence rates per 10,000)

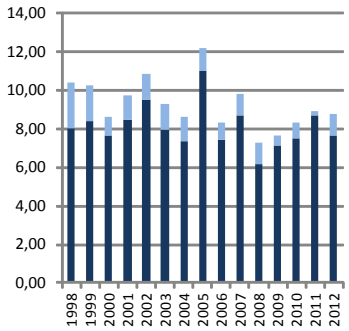
**Coarctation of aorta**



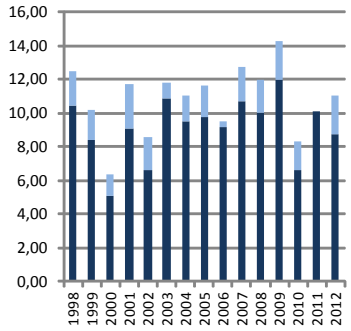

**Choanal atresia, bilateral**



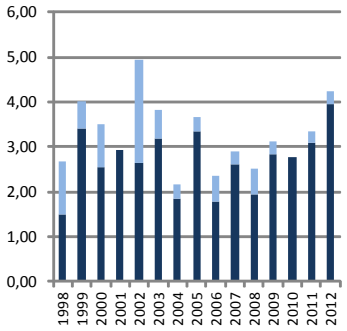

**Cleft palate without cleft lip**



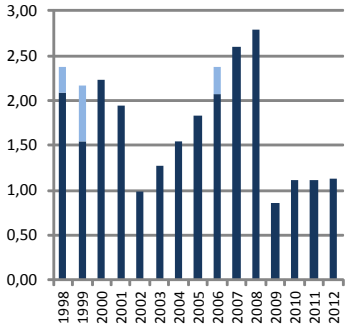

**Cleft lip with or without cleft palate**



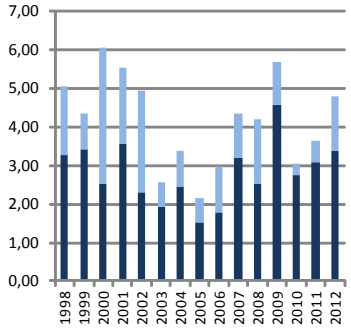

**phageal atresia/stenosis with or without f**



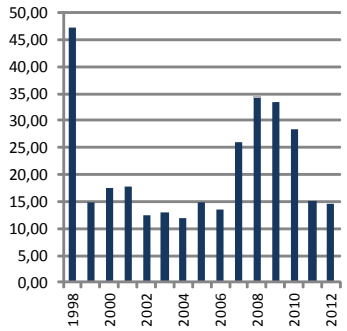

**Small intestine atresia/stenosis**



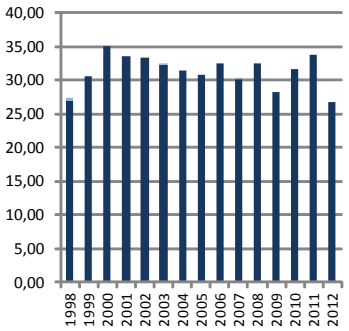

**Anorectal atresia/stenosis**



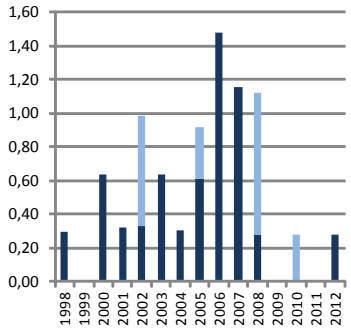

**Undescended testis**



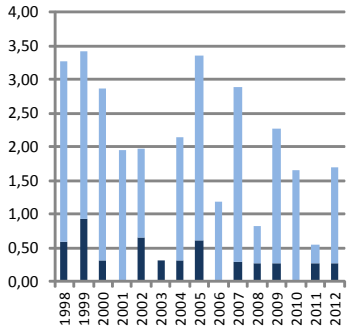

**Hypospadias**



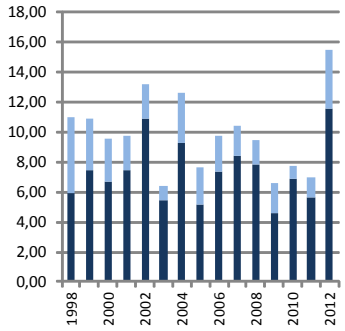

**Indeterminate sex**




**Renal agenesis**



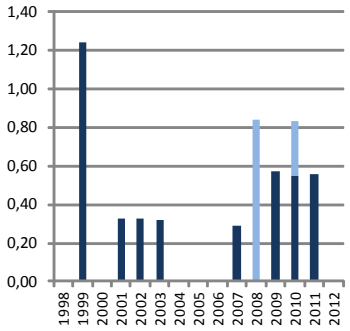

**Cystic kidney**



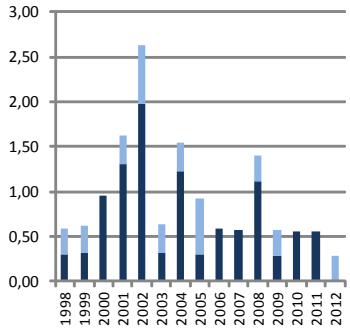
# United Kingdom-Wales: CARIS, Time trends 1998 – 2012

(Birth prevalence rates per 10,000)

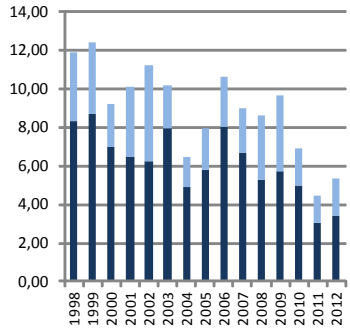
**Bladder exstrophy**



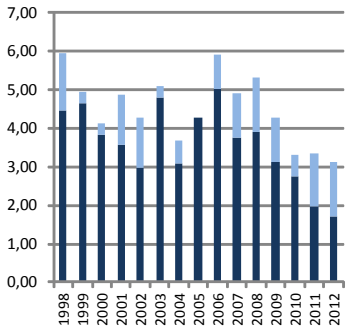

**Polydactyly, preaxial**



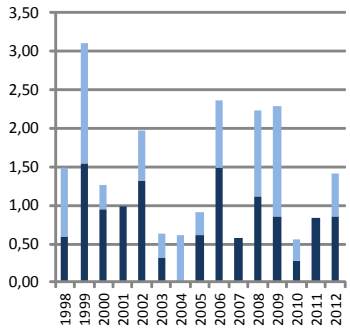

**Limb reduction defects**



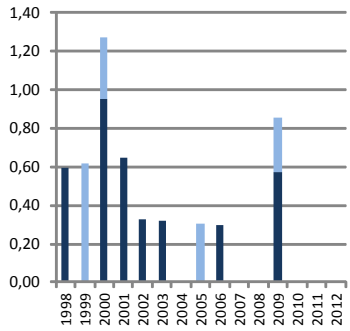

**Limb reduction defects -transverse**



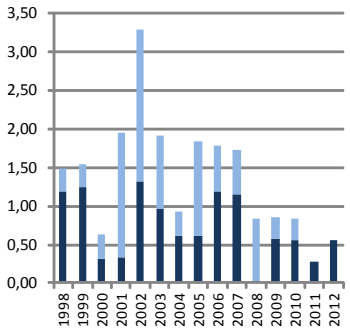

**Limb reduction defects - preaxial**



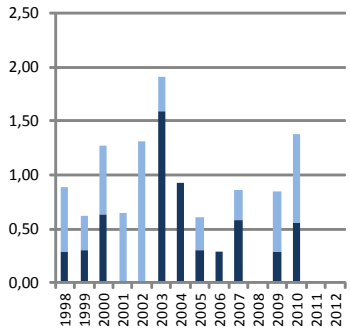

**Limb reduction defects - postaxial**



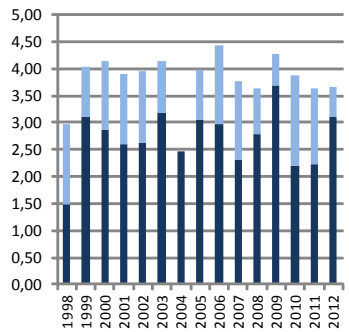

**Limb reduction defects - intercalary**



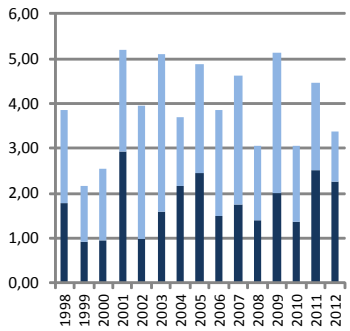

**Limb reduction defects - mixed**



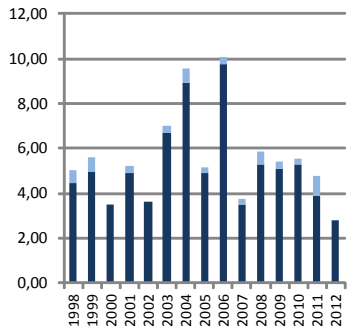

**Diaphragmatic hernia**



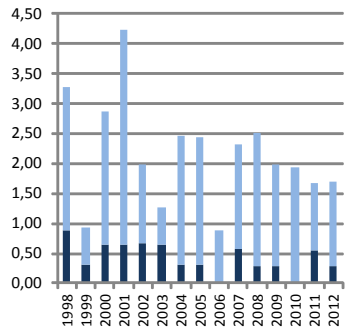

**Omphalocele**




**Gastroschisis**



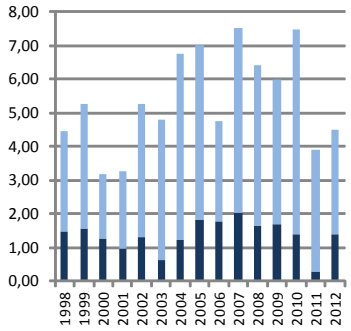

**Trisomy 13**



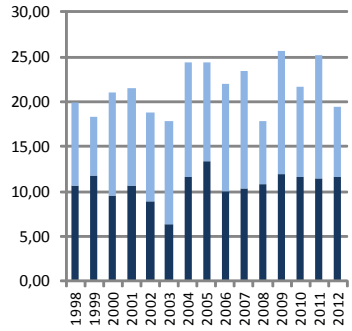
# United Kingdom-Wales: CARIS, Time trends 1998 – 2012

(Birth prevalence rates per 10,000)

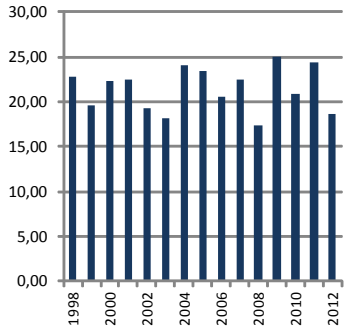
**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# USA-Atlanta: MACDP

## Metropolitan Atlanta Congenital Defects Program

**History:**
The Program started in 1967 and was a founding member of the ICBDSR.

**Size and coverage:**
Between 1967 and 2011, the Program covered all births within a five-county area in metropolitan Atlanta, Georgia. The annual number of births in this area is approximately 50,000. Beginning in 2012, the area covered by the Program was reduced to 3 counties in metropolitan Atlanta, with approximately 35,000 live births. Stillbirths of at least 20 weeks gestation and elective terminations at any gestational age are included

**Legislation and funding:**
In 1994 the Georgia Department of Human Resources (now the Georgia Department of Public Health) added birth defects to the list of legally reportable conditions in Georgia. In 1997 the GDHR requested the staff of MACDP to act with them in the collection of public health surveillance data related to birth defects and stillbirths. The Program is funded by the Centers for Disease Control and Prevention

**Sources of ascertainment:**
Multiple sources, such as delivery units, pediatric departments, neonatal intensive care units, laboratories, prenatal diagnostic centers, tertiary care centers, and vital records are used to ascertain malformed infants born in the defined area with a follow-up to age six years.

**Exposure information:**
Exposure information is obtained through interviews with mothers of reported malformed infants and often with mothers of infants without defects who participate in specific research projects.

**Background information:**
Number of live births and demographic information for the included counties are obtained from Georgia vital records.

**Addresses and Staff:**
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Centers for Disease Control and Prevention
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**Fax:** 404.498.3040
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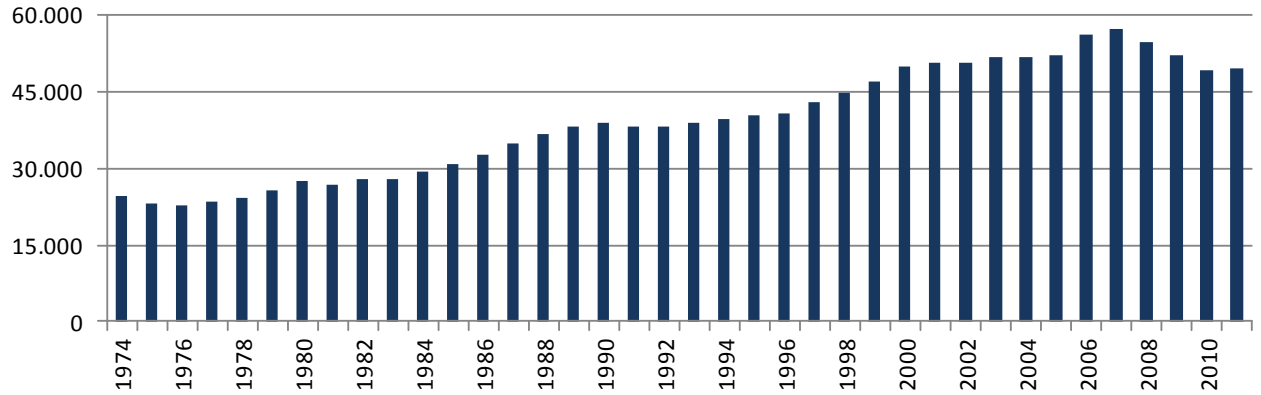
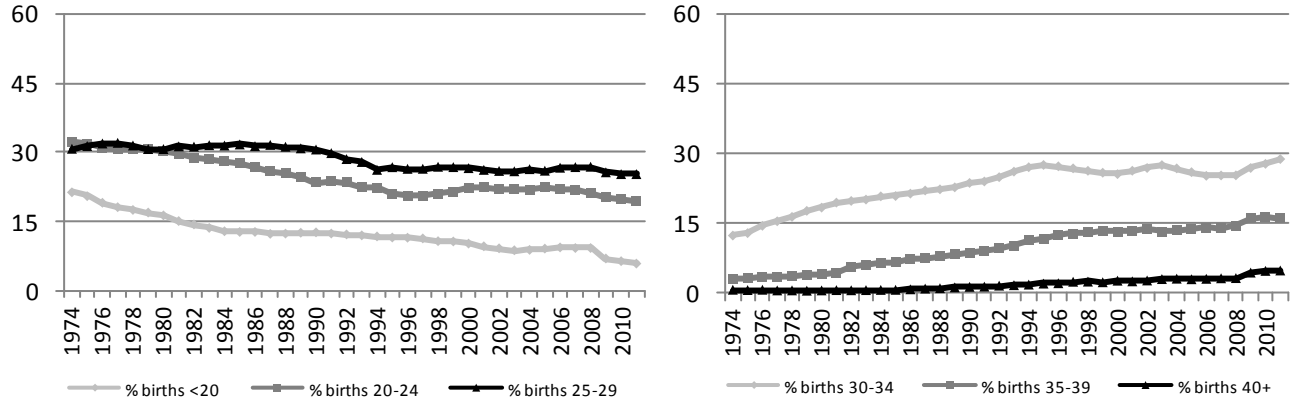
**Fax:** 404.498.3040
**E-mail:** JCragan@cdc.gov

# USA-Atlanta: MACDP

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2009-2011)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 38 | 67.9 | Cystic kidney | 9 | 16.4 |
| Spina bifida | 23 | 29.5 | Limb reduction defects | 14 | 17.3 |
| Encephalocele | 8 | 40.0 | Diaphragmatic hernia | 8 | 16.0 |
| Holoprosencephaly | 9 | 29.0 | Omphalocele | 27 | 43.5 |
| Hydrocephaly | 16 | 13.6 | Gastroschisis | 7 | 12.5 |
| Hypoplastic left heart syndrome | 13 | 28.9 | Trisomy 13 | 20 | 50.0 |
| Cleft palate without cleft lip | 5 | 6.3 | Trisomy 18 | 59 | 55.1 |
| Cleft lip with or without cleft palate | 23 | 16.8 | Down syndrome | 93 | 28.6 |
| Renal agenesis | 14 | 14.1 | | | |

Total ToPs with births defects = 260 (Ratio ToPs/Births: 1.72 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# USA-Atlanta: MACDP, 2011

| | |
|---|---|
| Live births (LB) | 49,220 |
| Stillbirths (SB) | 479 |
| Total births | 49,699 |
| Number of terminations of pregnancy (ToP) for birth defects | 140 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly(*) | 3 | 4 | 10 | 3.42 |
| Spina bifida | 11 | 4 | 9 | 4.83 |
| Encephalocele(*) | 4 | 0 | 2 | 1.21 |
| Microcephaly | nr | nr | nr | nr |
| Holoprosencephaly | 8 | 3 | 5 | 3.22 |
| Hydrocephaly | nr | nr | nr | nr |
| Anophthalmos | 1 | 0 | 1 | 0.40 |
| Microphthalmos | 8 | 0 | 0 | 1.61 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr |
| Anotia | 3 | 0 | 1 | 0.80 |
| Microtia | 10 | 0 | 0 | 2.01 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 13 | 0 | 1 | 2.82 |
| Tetralogy of Fallot | 20 | 3 | 3 | 5.23 |
| Hypoplastic left heart syndrome | 9 | 0 | 7 | 3.22 |
| Coarctation of aorta | 31 | 1 | 5 | 7.44 |
| Choanal atresia, bilateral(**) | 7 | 0 | 0 | 1.41 |
| Cleft palate without cleft lip | 15 | 2 | 1 | 3.62 |
| Cleft lip with or without cleft palate | 32 | 1 | 6 | 7.85 |
| Oesophageal atresia/stenosis with or without fistula | 12 | 0 | 0 | 2.41 |
| Small intestine atresia/stenosis | 22 | 0 | 0 | 4.43 |
| Anorectal atresia/stenosis | 24 | 2 | 0 | 5.23 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 200 | 0 | 1 | 40.44 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis(***) | 39 | 1 | 6 | 9.26 |
| Cystic kidney | nr | nr | nr | nr |
| Bladder exstrophy | 0 | 0 | 2 | 0.40 |
| Polydactyly, preaxial | nr | nr | nr | nr |
| Total Limb reduction defects (include unspecified) | 20 | 3 | 5 | 5.63 |
| Transverse | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 7 | 2 | 3 | 2.41 |
| Omphalocele | 6 | 4 | 13 | 4.63 |
| Gastroschisis | 18 | 2 | 2 | 4.43 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 5 | 6 | 5 | 3.22 |
| Trisomy 18 | 7 | 6 | 19 | 6.44 |
| Down syndrome, all ages (include age unknown) | 72 | 4 | 26 | 20.52 |
| <20 | 3 | 0 | 0 | 10.44 |
| 20-24 | 5 | 0 | 1 | 6.22 |
| 25-29 | 13 | 0 | 1 | 11.17 |
| 30-34 | 14 | 2 | 4 | 14.00 |
| 35-39 | 20 | 1 | 12 | 41.38 |
| 40-44 | 14 | 1 | 8 | 105.60 |
| 45+ | 3 | 0 | 0 | 149.25 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

Pregnancies diagnosed prenatally with unknown outocome that did not deliver in a hospital are assumed to have been electively terminated

(*) Cases with both anencephaly and encephalocele are included only in the category for anencephaly

(**) Information on laterality for choanal atresia (unilateral vs bilateral) was not available. All cases are included

(***) Renal agenesis includes bilateral renal dysplasia and hypoplasia.

# USA-Atlanta: MACDP, Previous years rates 1974 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | **70,480** | **127,639** | **148,828** | **186,757** | **198,299** | **235,616** | **262,250** | **263,574** |
| Anencephaly | 4.97 | 5.33 | 3.70 | 3.11 | 3.53 | 3.40 | 2.02 | 3.34 |
| Spina bifida | 7.66 | 6.58 | 6.85 | 5.03 | 4.64 | 3.95 | 4.12 | 4.63 |
| Encephalocele | 1.56 | 2.59 | 2.22 | 1.61 | 1.51 | 1.53 | 1.18 | 1.40 |
| Microcephaly | 4.82 | 6.19 | 5.64 | 5.46 | 6.05 | 7.89 | 5.95 | 5.10* |
| Holoprosencephaly | 0.28 | 0.71 | 0.74 | 1.55 | 1.41 | 0.72 | 1.14 | 2.10* |
| Hydrocephaly | 8.51 | 11.67 | 8.06 | 5.94 | 5.90 | 8.15 | 6.67 | 9.21* |
| Anophthalmos | 0.28 | 0.78 | 0.54 | 0.59 | 0.71 | 0.30 | 0.46 | 0.23 |
| Microphthalmos | 3.69 | 4.23 | 3.83 | 2.62 | 3.28 | 2.46 | 1.79 | 1.10 |
| Unspecified Anophthalmos/Microphthalmos | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00* |
| Anotia | 0.43 | 0.00 | 0.20 | 0.16 | 0.20 | 0.21 | 0.19 | 0.34 |
| Microtia | 1.99 | 0.86 | 1.41 | 1.66 | 1.41 | 1.32 | 1.22 | 1.40 |
| Unspecified Anotia/Microtia | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.00* |
| Transposition of great vessels | 4.11 | 5.17 | 5.71 | 4.93 | 5.60 | 5.35 | 4.69 | 4.29 |
| Tetralogy of Fallot | 2.41 | 3.60 | 3.83 | 4.18 | 4.14 | 3.78 | 4.39 | 4.74 |
| Hypoplastic left heart syndrome | 2.41 | 2.51 | 2.49 | 3.05 | 2.57 | 3.06 | 2.06 | 2.39 |
| Coarctation of aorta | 3.69 | 4.31 | 3.90 | 5.30 | 4.14 | 5.60 | 5.41 | 5.46 |
| Choanal atresia, bilateral | 0.43 | 0.24 | 0.27 | 0.32 | 0.45 | 0.34 | 0.42 | 0.61 |
| Cleft palate without cleft lip | 8.09 | 5.48 | 4.84 | 5.25 | 5.09 | 6.45 | 5.07 | 4.82 |
| Cleft lip with or without cleft palate | 12.49 | 10.89 | 10.48 | 9.48 | 9.48 | 8.45 | 9.27 | 9.11 |
| Oesophageal atresia/stenosis with or without fistula | 2.70 | 2.66 | 2.28 | 1.93 | 2.32 | 2.33 | 1.79 | 2.35 |
| Small intestine atresia/stenosis | 1.70 | 1.41 | 1.48 | 1.98 | 1.66 | 1.95 | 1.91 | 2.80* |
| Anorectal atresia/stenosis | 4.97 | 4.15 | 3.90 | 3.91 | 3.33 | 3.82 | 2.82 | 4.34* |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | 16.34 | 15.44 | 7.48* |
| Hypospadias | 1.28 | 0.94 | 3.56 | 4.50 | 5.35 | 9.17 | 7.13 | 17.60 |
| Epispadias | 0.85 | 1.02 | 0.87 | 0.64 | 0.50 | 0.30 | 0.65 | 0.33* |
| Indeterminate sex | 1.28 | 3.06 | 1.14 | 1.12 | 1.21 | 1.49 | 1.53 | 1.64* |
| Renal agenesis | 1.56 | 2.19 | 1.68 | 1.29 | 1.56 | 0.85 | 0.99 | 4.10 |
| Cystic kidney | 2.84 | 2.04 | 3.90 | 3.91 | 5.70 | 6.24 | 6.14 | 6.87* |
| Bladder exstrophy | 0.57 | 0.47 | 0.07 | 0.21 | 0.30 | 0.13 | 0.11 | 0.23* |
| Polydactyly, preaxial | 1.28 | 2.35 | 1.88 | 3.27 | 3.08 | 2.29 | 2.25 | 2.25* |
| Total Limb reduction defects (include unspecified) | 6.53 | 5.17 | 4.57 | 4.39 | 6.25 | 6.32 | 4.08 | 4.89 |
| Transverse | 4.40 | 2.98 | 3.36 | 2.62 | 4.14 | 3.23 | 2.21 | 2.80* |
| Preaxial | 1.14 | 1.02 | 0.47 | 0.80 | 1.01 | 1.36 | 0.69 | 0.61* |
| Postaxial | 0.14 | 0.31 | 0.13 | 0.32 | 0.30 | 0.25 | 0.31 | 0.18* |
| Intercalary | 0.28 | 0.55 | 0.20 | 0.32 | 0.25 | 0.21 | 0.19 | 0.49* |
| Mixed | 0.00 | 0.16 | 0.34 | 0.21 | 0.35 | 0.98 | 0.53 | 0.67* |
| Unspecified | 0.57 | 0.16 | 0.07 | 0.11 | 0.20 | 0.30 | 0.11 | 0.24* |
| Diaphragmatic hernia | 3.26 | 1.96 | 2.35 | 3.00 | 2.12 | 2.42 | 2.94 | 2.96 |
| Omphalocele | 4.26 | 3.60 | 3.49 | 2.46 | 2.47 | 2.55 | 1.72 | 3.30 |
| Gastroschisis | 0.85 | 2.04 | 2.08 | 2.78 | 2.22 | 2.25 | 3.36 | 4.40 |
| Unspecified Omphalocele/Gastroschisis | 0.00 | 0.00 | 0.00 | 0.05 | 0.00 | 0.00 | 0.00 | 0.00* |
| Prune belly sequence | 0.57 | 0.86 | 0.47 | 0.32 | 0.20 | 0.51 | 0.38 | 0.49 |
| Trisomy 13 | 1.14 | 1.02 | 1.48 | 1.39 | 1.46 | 1.95 | 1.87 | 1.93 |
| Trisomy 18 | 0.57 | 1.18 | 1.95 | 2.14 | 2.98 | 4.71 | 4.35 | 5.46 |
| Down syndrome, all ages (include age unknown) | 9.51 | 10.03 | 9.94 | 10.98 | 15.63 | 17.15 | 17.12 | 19.27 |
| <20 | nr | 10.62* | 5.62 | 7.36 | 7.76 | 7.76 | 8.07 | 9.56 |
| 20-24 | nr | 6.78* | 7.47 | 7.85 | 7.84 | 7.90 | 6.76 | 7.02 |
| 25-29 | nr | 11.33* | 6.42 | 6.97 | 8.75 | 7.53 | 7.15 | 9.50 |
| 30-34 | nr | 17.63 | 14.71 | 13.33 | 13.12 | 13.50 | 14.30 | 14.79 |
| 35-39 | nr | 31.52* | 17.91 | 23.38 | 39.40 | 48.35 | 41.21 | 41.40 |
| 40-44 | nr | 106.38* | 67.34 | 49.02 | 116.89 | 108.46 | 127.35 | 98.85 |
| 45+ | nr | 0.00* | 0.00 | 0.00 | 413.22 | 241.94 | 103.36 | 144.17 |
| unknown | --- | --- | --- | --- | --- | --- | --- | --- |

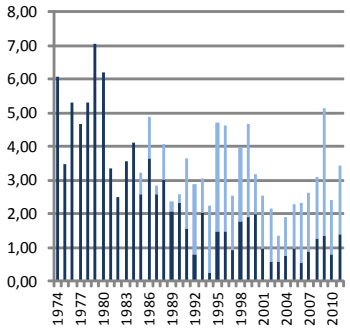
nr = data not reported or not available

* data include less than 5 years

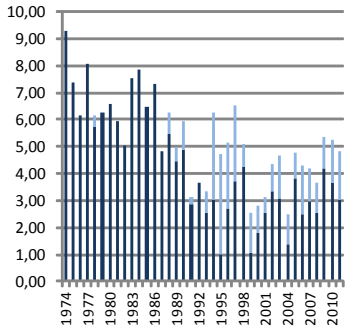
# USA-Atlanta: MACDP, Time trends 1974 – 2011

(Birth prevalence rates per 10,000)

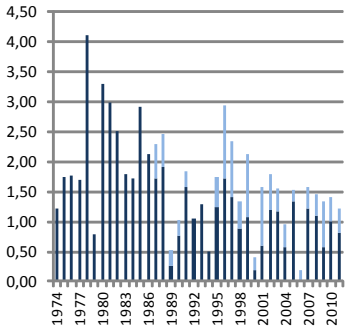
**Anencephaly**



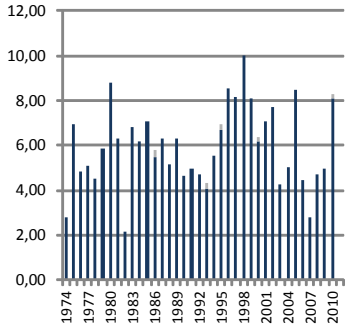

**Spina Bifida**



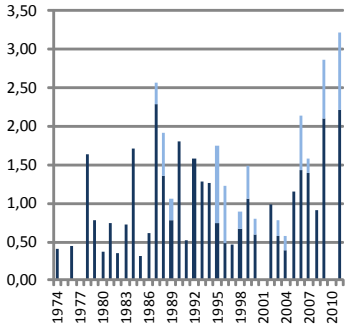

**Encephalocele**



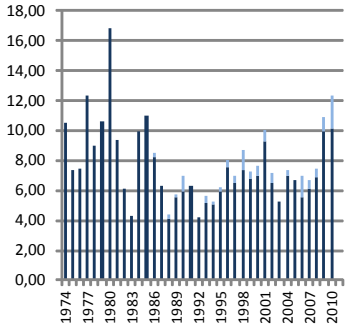

**Microcephaly**



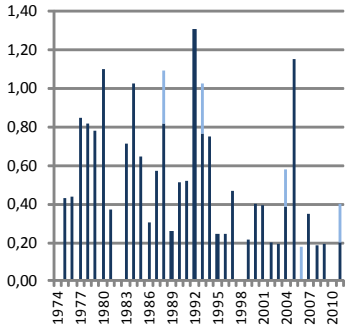

**Holoprosencephaly**



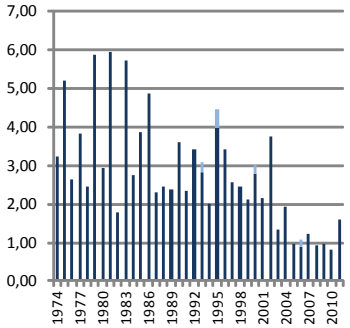

**Hydrocephaly**



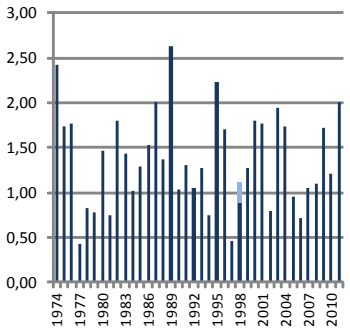

**Anophthalmos**



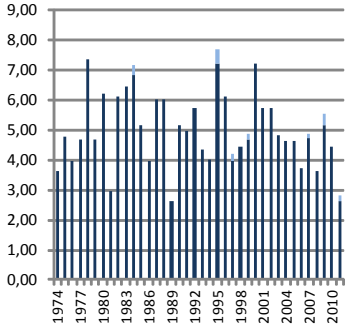

**Microphthalmos**



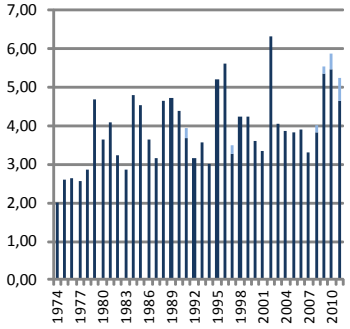

**Microtia**



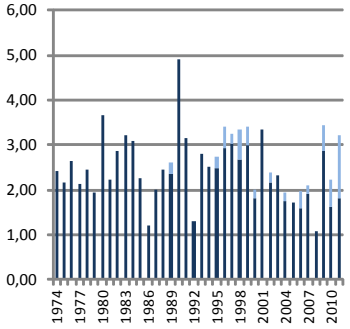

**Transposition of great vessels**




**Tetralogy of Fallot**



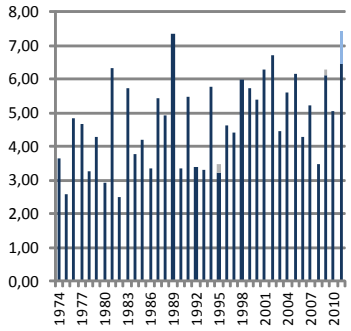

**Hypoplastic left heart syndrome**



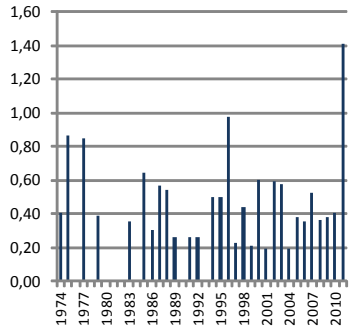
# USA-Atlanta: MACDP, Time trends 1974 – 2011

(Birth prevalence rates per 10,000)

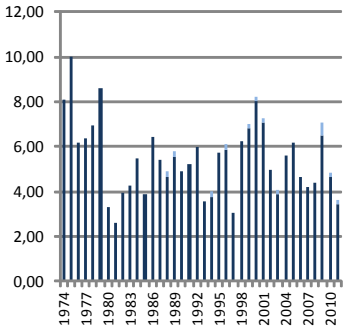
**Coarctation of aorta**



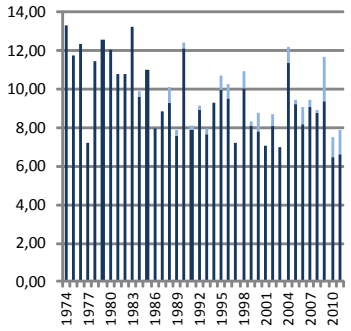

**Choanal atresia, bilateral**



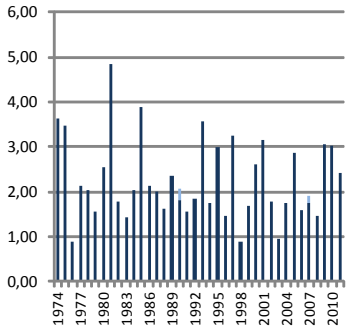

**Cleft palate without cleft lip**



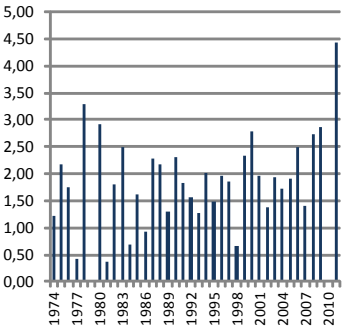

**Cleft lip with or without cleft palate**



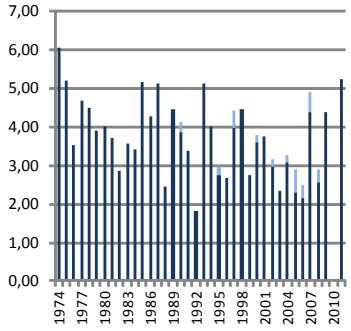

**phageal atresia/stenosis with or without f**



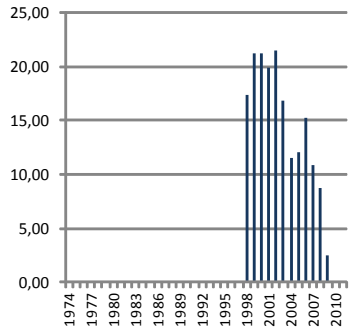

**Small intestine atresia/stenosis**



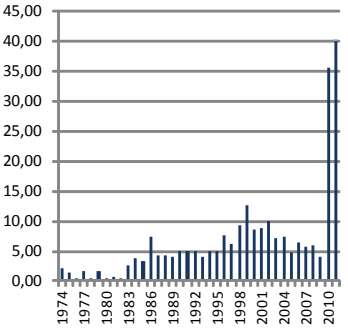

**Anorectal atresia/stenosis**



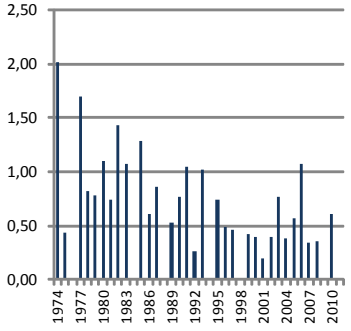

**Undescended testis**



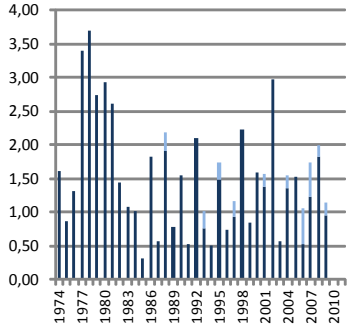

**Hypospadias**



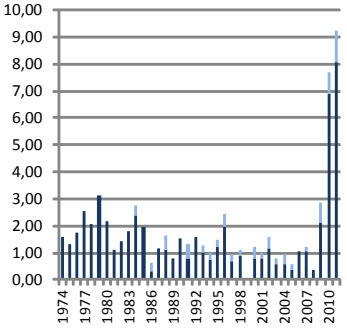

**Epispadias**




**Indeterminate sex**



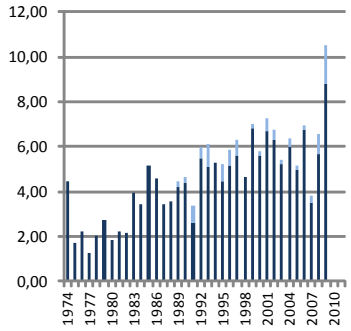

**Renal agenesis**



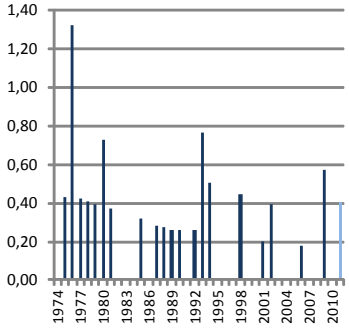
# USA-Atlanta: MACDP, Time trends 1974 – 2011

(Birth prevalence rates per 10,000)

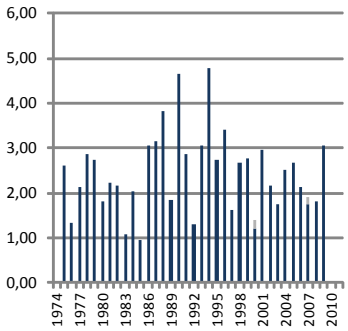
**Cystic kidney**



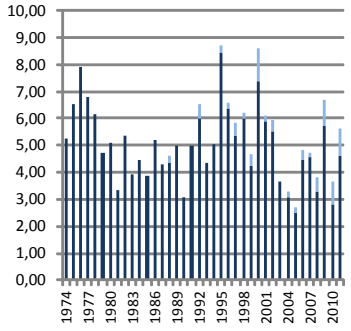

**Bladder exstrophy**



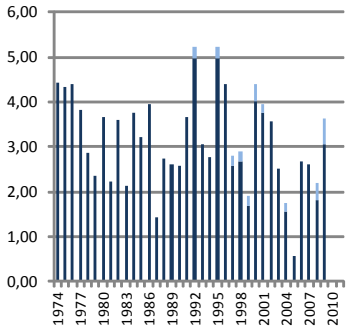

**Polydactyly, preaxial**



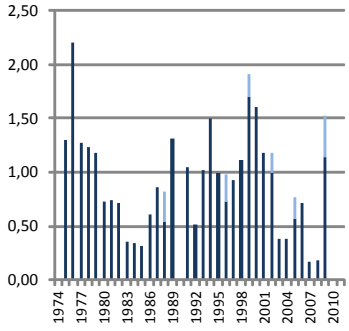

**Limb reduction defects**



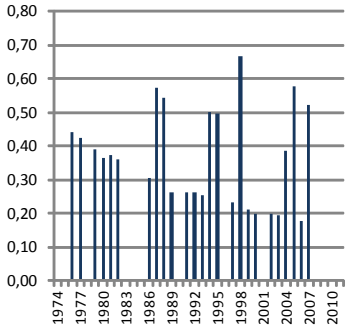

**Limb reduction defects - transverse**



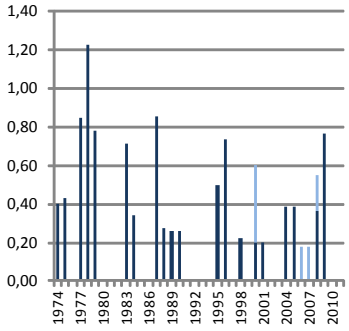

**Limb reduction defects - preaxial**



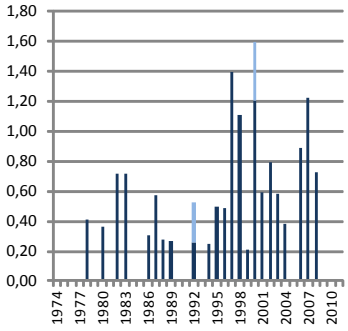

**Limb reduction defects - postaxial**



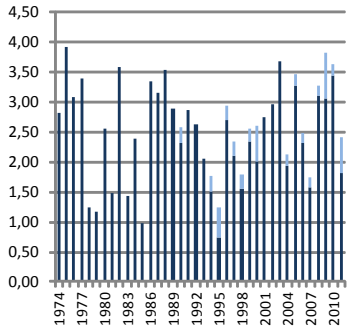

**Limb reduction defects - intercalary**



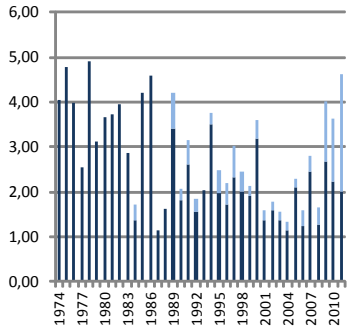

**Limb reduction defects - mixed**



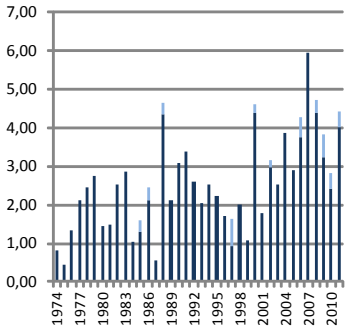

**Diaphragmatic hernia**




**Omphalocele**



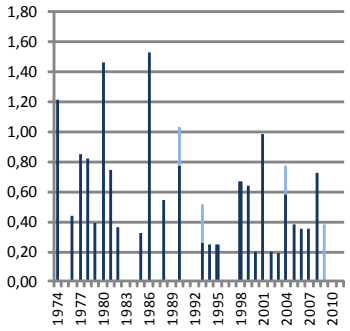

**Gastroschisis**



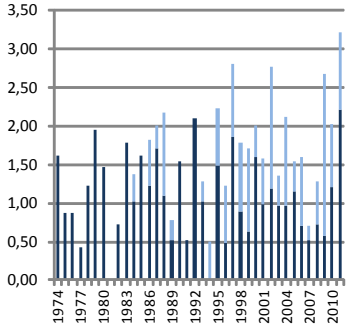
# USA-Atlanta: MACDP, Time trends 1974 – 2011

(Birth prevalence rates per 10,000)

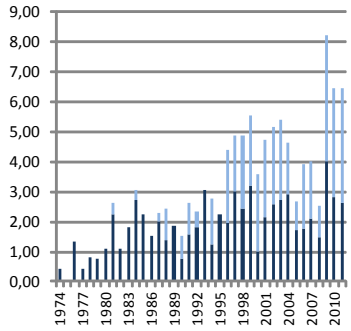
**Prune belly sequence**



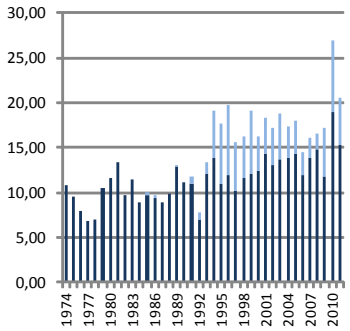

**Trisomy 13**



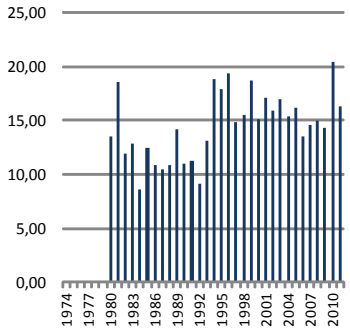

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

## USA-Arkansas: ARHMS

Arkansas Reproductive Health Monitoring System

**History:**
The Program began as the state's official birth defect surveillance system in 1985. It became a member of the ICBDSR in 2013.

**Size and coverage:**
Between 1985 and 1992, ARHMS covered births within 3-7 counties in central Arkansas. The number of counties/births covered during this period varied each year as funding was available. Since 1993, ARHMS has monitored births statewide. The annual number of births in the state has increased since 1993 with the most recent years seeing approximately 40,000 births annually. Stillbirths of at least 20 weeks gestation, early fetal losses and elective terminations at any gestational age are included.

**Legislation and funding:**
In 1985 the Arkansas State Legislators and Governor established ARHMS. Funding for the program is appropriated on an annual basis by the State Legislators.

**Sources of ascertainment:**
Hospital disease indices and customized diagnosis reports from specific hospitals serve as the initial case finding documents. Following medical record review by ARHMS staff, information on eligible subjects are ascertained into ARHMS. Information from all delivering hospitals, prenatal diagnostic centers, and tertiary care centers are used to ascertain malformed infants born in the defined area with a follow-up of initial diagnosis to age two years.

**Exposure information:**
Demographic and clinical information is obtained through medical record review by ARHMS staff. Exposure information is obtained for specific research studies by protocol established by each study.

**Background information:**
Number of live births for the state are obtained from the Center for Health Statistics at the Arkansas Department of Health.

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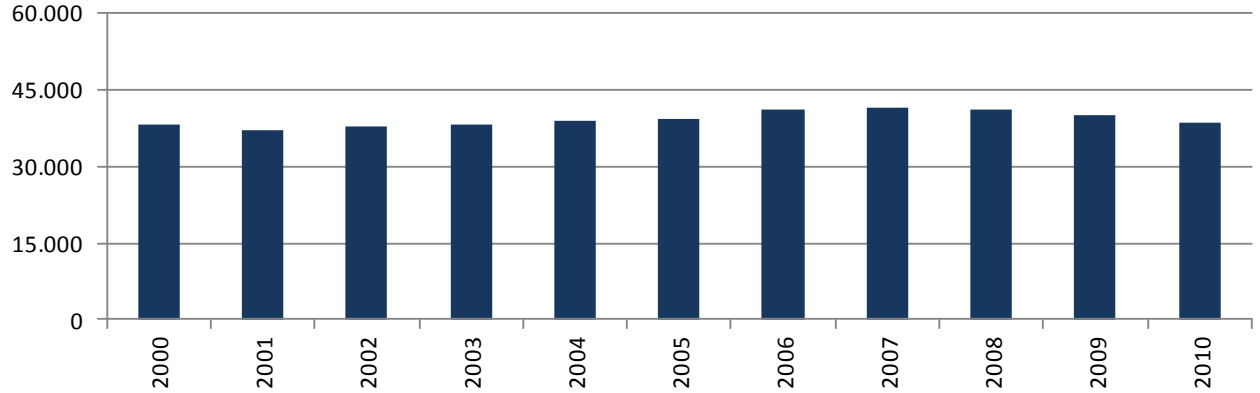
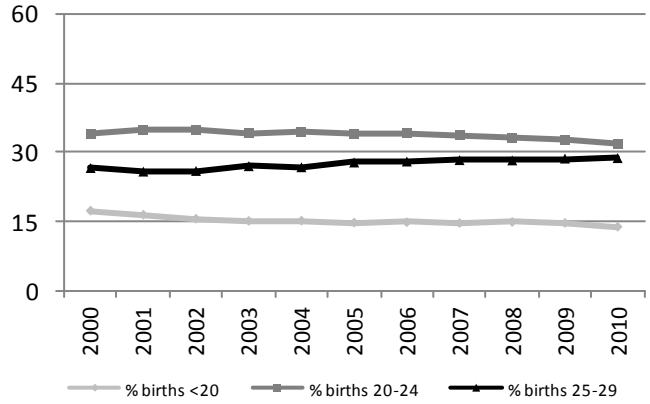
**Email**: hobbscharlotte@uams.edu

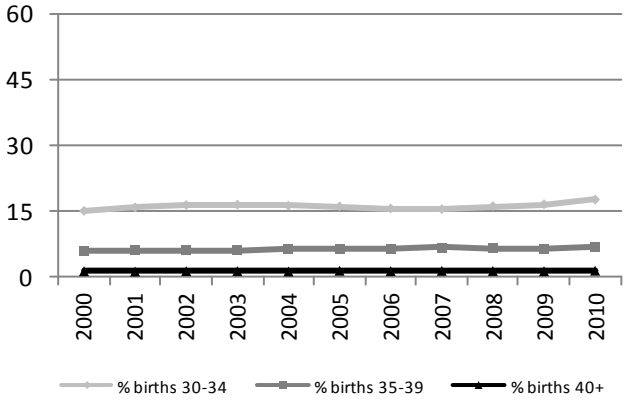
# USA-Arkansas: ARHMS

## Total births by year




## Percentage of births by year and maternal age



# USA-Arkansas: ARHMS, 2010

| | |
|---|---|
| Live births (LB) | 38,217 |
| Stillbirths (SB) | 283 |
| Total births | 38,500 |
| Number of terminations of pregnancy (ToP) for birth defects | 9 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 4 | 4 | 3 | 2.86 |
| Spina bifida | 13 | 0 | 2 | 3.90 |
| Encephalocele | 2 | 2 | 0 | 1.04 |
| Microcephaly | 17 | 1 | 0 | 4.68 |
| Holoprosencephaly | 7 | 2 | 0 | 2.34 |
| Hydrocephaly | 18 | 2 | 1 | 5.45 |
| Anophthalmos | 2 | 0 | 0 | 0.52 |
| Microphthalmos | 4 | 0 | 0 | 1.04 |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | nr | nr |
| Anotia | 0 | 0 | 1 | 0.26 |
| Microtia | 9 | 0 | 0 | 2.34 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 13 | 0 | 0 | 3.38 |
| Tetralogy of Fallot | 15 | 0 | 0 | 3.90 |
| Hypoplastic left heart syndrome | 11 | 0 | 0 | 2.86 |
| Coarctation of aorta | 36 | 0 | 0 | 9.35 |
| Choanal atresia, bilateral | 1 | 0 | 0 | 0.26 |
| Cleft palate without cleft lip | 24 | 0 | 0 | 6.23 |
| Cleft lip with or without cleft palate | 42 | 0 | 1 | 11.17 |
| Oesophageal atresia/stenosis with or without fistula | 15 | 1 | 0 | 4.16 |
| Small intestine atresia/stenosis | 9 | 0 | 0 | 2.34 |
| Anorectal atresia/stenosis | 13 | 0 | 0 | 3.38 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 197 | 0 | 0 | 51.17 |
| Epispadias | 3 | 0 | 0 | 0.78 |
| Indeterminate sex | 7 | 0 | 0 | 1.82 |
| Renal agenesis | 7 | 0 | 3 | 2.60 |
| Cystic kidney | 4 | 0 | 0 | 1.04 |
| Bladder exstrophy | 2 | 0 | 0 | 0.52 |
| Polydactyly, preaxial | 16 | 0 | 0 | 4.16 |
| Total Limb reduction defects (include unspecified) | 30 | 2 | 0 | 8.31 |
| Transverse | 17 | 0 | 0 | 4.42 |
| Preaxial | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr |
| Intercalary | 1 | 0 | 0 | 0.26 |
| Mixed | nr | nr | nr | nr |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | 13 | 0 | 0 | 3.38 |
| Omphalocele | 7 | 2 | 0 | 2.34 |
| Gastroschisis | 20 | 2 | 1 | 5.97 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 5 | 1 | 0 | 1.56 |
| Trisomy 18 | 10 | 7 | 0 | 4.42 |
| Down syndrome, all ages (include age unknown) | 55 | 4 | 0 | 15.32 |
| <20 | 4 | 0 | 0 | 7.62 |
| 20-24 | 9 | 1 | 0 | 8.25 |
| 25-29 | 11 | 1 | 0 | 10.93 |
| 30-34 | 13 | 0 | 0 | 19.28 |
| 35-39 | 14 | 1 | 0 | 57.74 |
| 40-44 | 4 | 1 | 0 | 101.21 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available

# USA-Arkansas: ARHMS, Previous years rates 2000 – 2010

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001* | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | **75,334** | **195,357** | |
| Anencephaly | | | | | | 4.65 | 3.28 | |
| Spina bifida | | | | | | 4.51 | 4.04 | |
| Encephalocele | | | | | | 1.06 | 0.97 | |
| Microcephaly | | | | | | 3.45 | 2.20 | |
| Holoprosencephaly | | | | | | 1.73 | 1.13 | |
| Hydrocephaly | | | | | | 9.42 | 7.27 | |
| Anophthalmos | | | | | | 0.13 | 0.46 | |
| Microphthalmos | | | | | | 1.73 | 1.89 | |
| Unspecified Anophthalmos/Microphthalmos | | | | | | nr | nr | |
| Anotia | | | | | | 0.27 | 0.15 | |
| Microtia | | | | | | 1.19 | 1.69 | |
| Unspecified Anotia/Microtia | | | | | | nr | nr | |
| Transposition of great vessels | | | | | | 3.98 | 4.76 | |
| Tetralogy of Fallot | | | | | | 3.72 | 4.10 | |
| Hypoplastic left heart syndrome | | | | | | 3.45 | 2.51 | |
| Coarctation of aorta | | | | | | 5.84 | 5.32 | |
| Choanal atresia, bilateral | | | | | | 0.00 | 0.26 | |
| Cleft palate without cleft lip | | | | | | 5.84 | 6.50 | |
| Cleft lip with or without cleft palate | | | | | | 8.89 | 11.98 | |
| Oesophageal atresia/stenosis with or without fistula | | | | | | 3.05 | 2.25 | |
| Small intestine atresia/stenosis | | | | | | 4.65 | 3.69 | |
| Anorectal atresia/stenosis | | | | | | 5.58 | 4.56 | |
| Undescended testis (36 weeks of gestation or later) | | | | | | nr | nr | |
| Hypospadias | | | | | | 33.32 | 43.87 | |
| Epispadias | | | | | | 0.40 | 0.82 | |
| Indeterminate sex | | | | | | 1.59 | 2.00 | |
| Renal agenesis | | | | | | 4.12 | 2.76 | |
| Cystic kidney | | | | | | 1.86 | 1.28 | |
| Bladder exstrophy | | | | | | 0.27 | 0.36 | |
| Polydactyly, preaxial | | | | | | 0.66 | 3.12 | |
| Total Limb reduction defects (include unspecified) | | | | | | 5.31 | 6.35 | |
| Transverse | | | | | | 2.26 | 2.97 | |
| Preaxial | | | | | | nr | nr | |
| Postaxial | | | | | | nr | nr | |
| Intercalary | | | | | | 0.40 | 0.46 | |
| Mixed | | | | | | nr | nr | |
| Unspecified | | | | | | nr | nr | |
| Diaphragmatic hernia | | | | | | 3.32 | 2.87 | |
| Omphalocele | | | | | | 3.19 | 3.28 | |
| Gastroschisis | | | | | | 3.85 | 5.99 | |
| Unspecified Omphalocele/Gastroschisis | | | | | | nr | nr | |
| Prune belly sequence | | | | | | 0.53 | 0.36 | |
| Trisomy 13 | | | | | | 1.06 | 1.23 | |
| Trisomy 18 | | | | | | 1.86 | 2.35 | |
| Down syndrome, all ages (include age unknown) | | | | | | 11.42 | 11.67 | |
| <20 | | | | | | 10.25 | 7.50 | |
| 20-24 | | | | | | 5.79 | 7.03 | |
| 25-29 | | | | | | 4.06 | 8.14 | |
| 30-34 | | | | | | 7.74 | 10.80 | |
| 35-39 | | | | | | 49.14 | 37.85 | |
| 40-44 | | | | | | 203.16 | 145.05 | |
| 45+ | | | | | | 377.36 | 0.00 | |
| unknown | | | | | | --- | --- | |

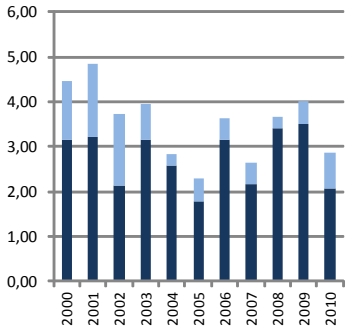
nr = data not reported or not available

* data include less than 5 years

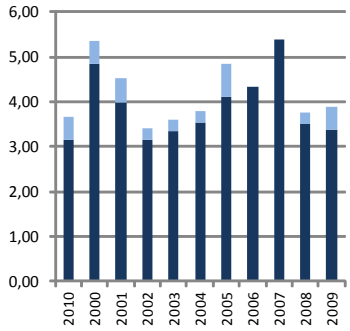
# USA-Arkansas: ARHMS, Time trends 2000 – 2010

(Birth prevalence rates per 10,000)

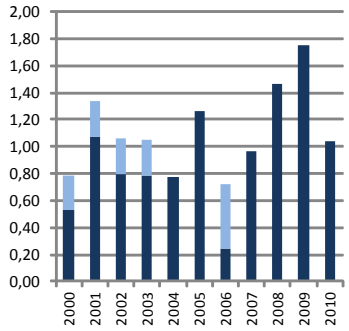
**Anencephaly**



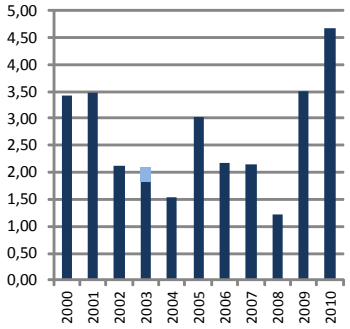

**Spina Bifida**



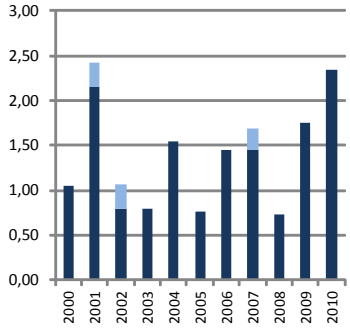

**Encephalocele**



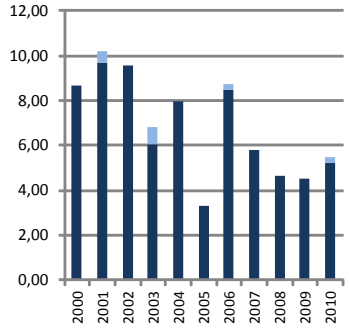

**Microcephaly**



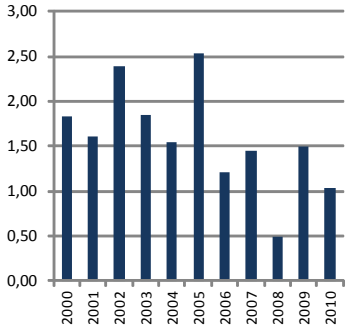

**Holoprosencephaly**



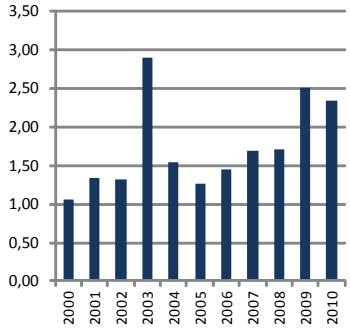

**Hydrocephaly**



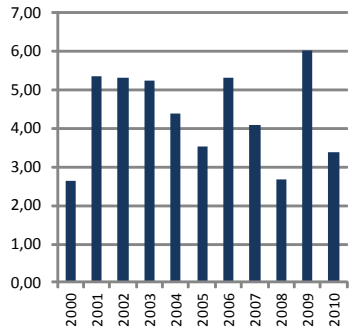

**Microphthalmos**



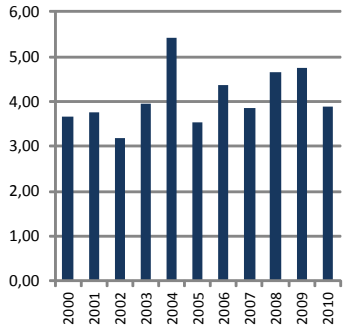

**Microtia**



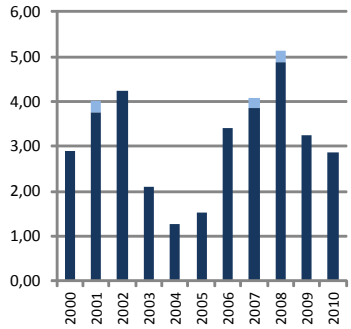

**Transposition of great vessels**



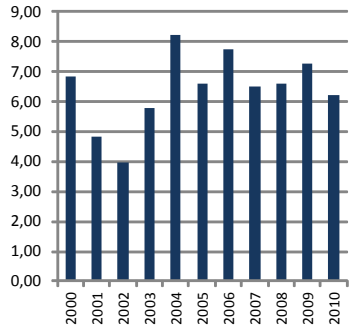

**Tetralogy of Fallot**




**Hypoplastic left heart syndrome**



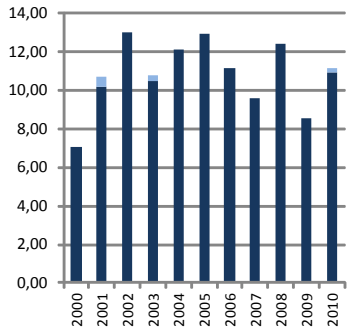

**Cleft palate without cleft lip**



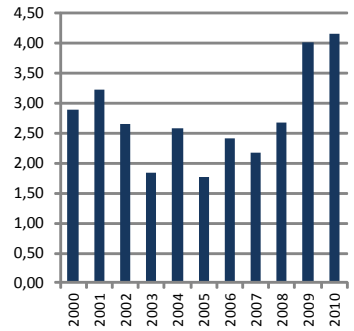
# USA-Arkansas: ARHMS, Time trends 2000 – 2010

(Birth prevalence rates per 10,000)

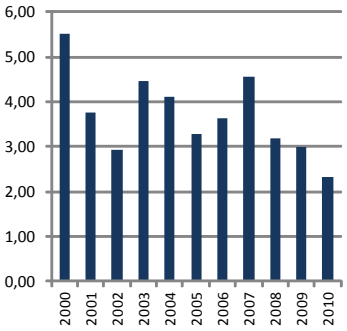
**Cleft lip with or without cleft palate**



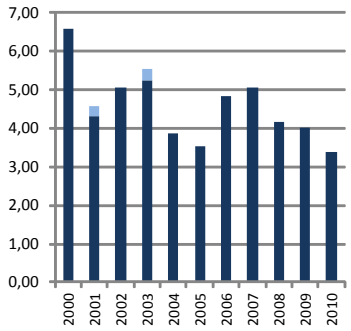

**phageal atresia/stenosis with or without f**



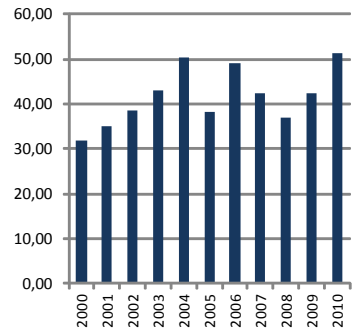

**Small intestine atresia/stenosis**



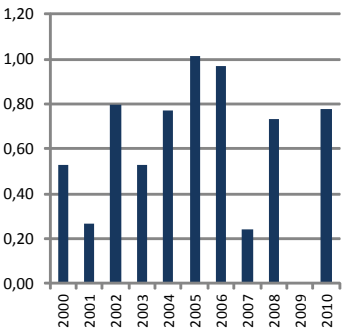

**Anorectal atresia/stenosis**



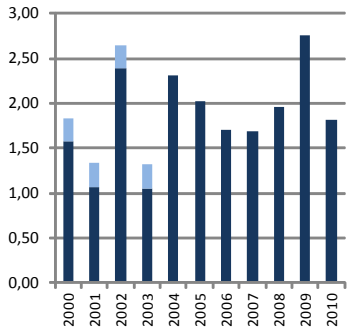

**Hypospadias**



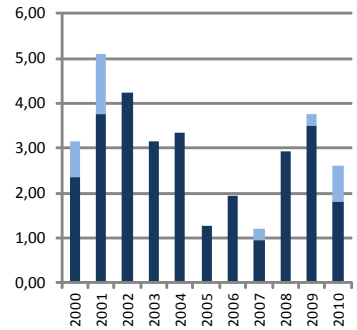

**Epispadias**



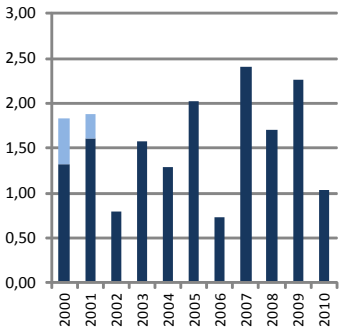

**Indeterminate sex**



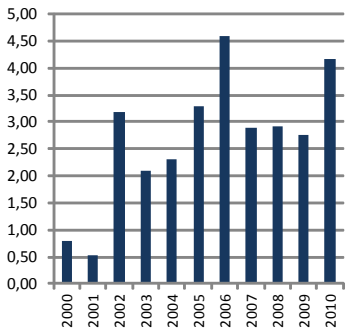

**Renal agenesis**



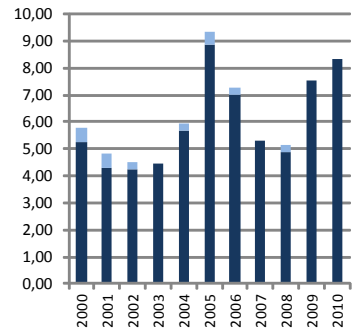

**Cystic kidney**



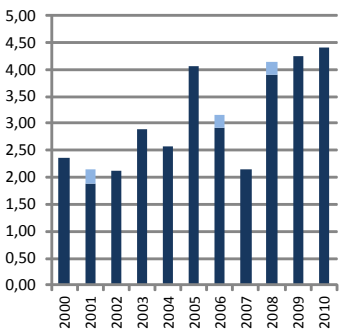

**Polydactyly, preaxial**




**Limb reduction defects**



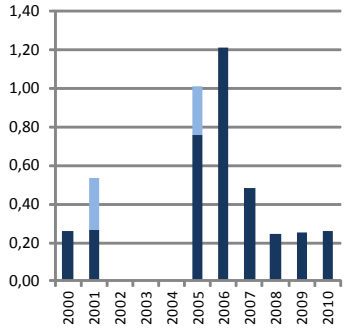

**Limb reduction defects - Transverse**



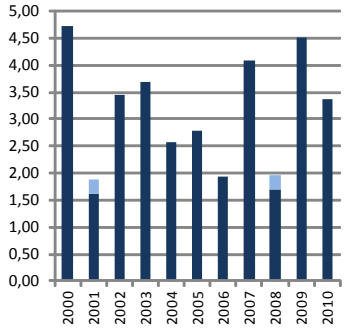
# USA-Arkansas: ARHMS, Time trends 2000 – 2010

(Birth prevalence rates per 10,000)

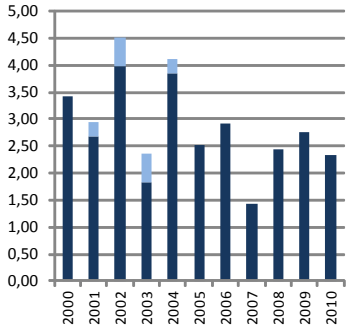
**Limb reduction defects - intercalary**



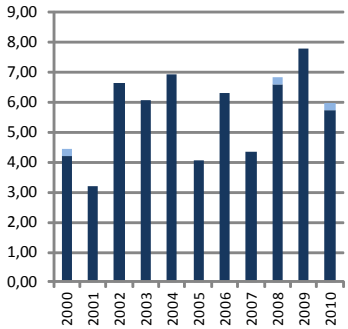

**Diaphragmatic hernia**



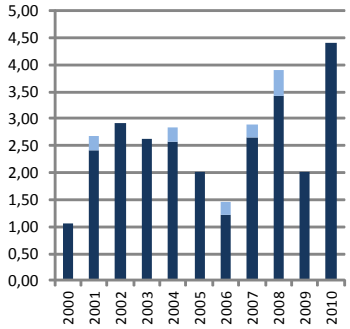

**Omphalocele**



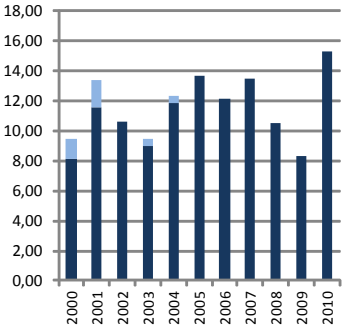

**Gastroschisis**



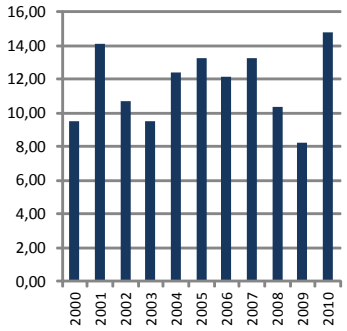

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# USA-Texas: BDES

## Texas Birth Defects Epidemiology and Surveillance Branch

**History:**
BDES was established after an unusual cluster of anencephaly cases that occurred in Brownsville, Texas in 1991. Epidemiologic investigations revealeda higher than expected rate of neural tube defectsamong children born to Hispanic mothers living in South Texas. In recognition that epidemiologic resources are routinely needed to investigate birth defects clusters, the Texas State Legislature passed the Texas Birth Defects Act in 1993, which authorized the establishment of BDES. Since 1994, BDES has maintained the Texas Birth Defects Registry, an active population-based birth defects surveillance system, which has been statewide since 1999. Through multiple sources of information, the Registry monitors all births in Texas and identifies cases of birth defects. Children identified through the Registry are referred to appropriate medical and community services. In 1996, the CDC-funded Texas Center for Birth Defects Research and Prevention was established under the auspices of BDES . The Programme is a full member of the ICBDSR.

**Size and coverage:**
The Programme covers all deliveries to mothers residing in Texas (approximately 380,000 annually). Stillbirths and terminations of any gestational age are included. Cases diagnosed up to age one are included (up to any age for fetal alcohol syndrome). As of 2006, there were over 100,000 birth defect cases in the Registry.

**Legislation and funding:**
Birth defects surveillance was mandated by the Texas Birth Defects Act in 1993, and is codified in the Texas Health and Safety Code Chapter 87. About one-half of funding for the birth defects registry is from state general revenue with the remainder from federal block grants.

**Sources of ascertainment:**
Birth hospitals, birthing centres, lay midwives, hospitals where affected children are treated.

**Exposure information:**
Limited information on maternal illnesses and conditions, limited information on maternal exposures such as medications.

**Background information:**
Basic demographics, reproductive history, gestational age, delivery information.

**Addresses and Staff:**
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Texas Birth Defects Epidemiology and Surveillance
PO Box 149347
Austin, Texas 78714-9347, USA
**Phone:** 512-776-7232
**Fax:** 512-776-7330
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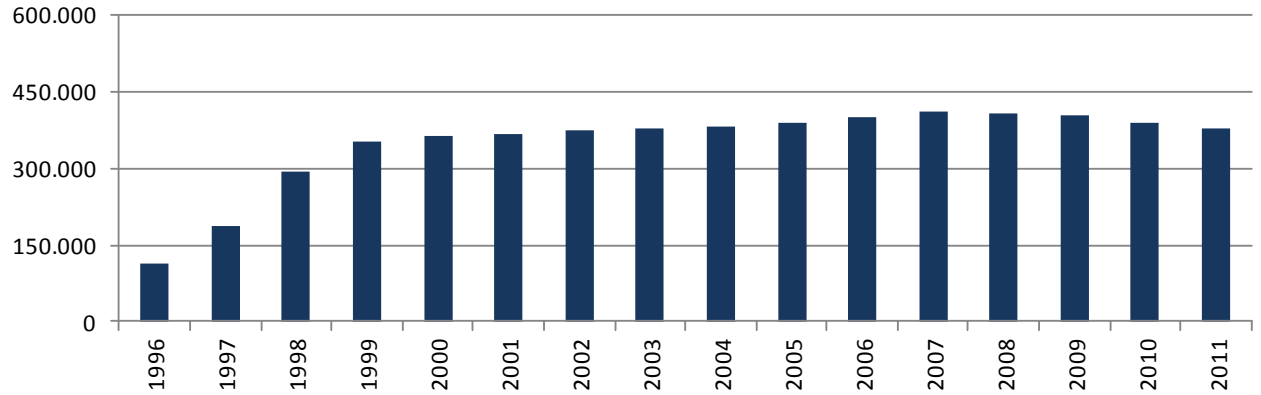
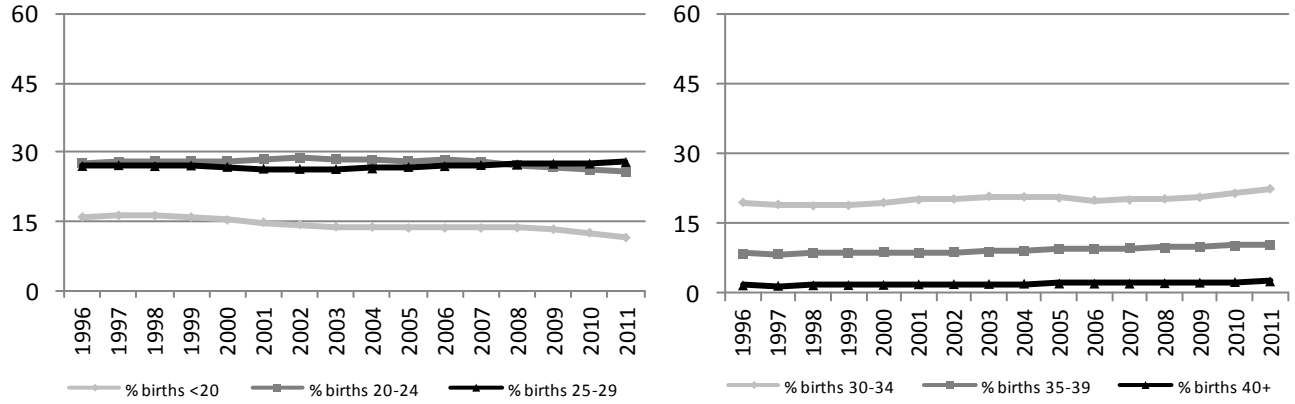
Lisa K. Marengo, MS
Texas Birth Defects Epidemiology and Surveillance
Epidemiologist
PO Box 149347
Austin, Texas 78714-9347, USA
**Phone:** 512-776-6657
**Fax:** 512-776-7330
**E-mail:** Lisa.Marengo@dshs.state.tx.us
**Website:**
http://www.dshs.state.tx.us/birthdefects/

## USA-Texas: BDES

### Total births by year




### Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2009-2011)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 110 | 32.7 | Cystic kidney | 8 | 1.1 |
| Spina bifida | 20 | 4.2 | Limb reduction defects | 23 | 3.1 |
| Encephalocele | 13 | 10.6 | Diaphragmatic hernia | 3 | 0.9 |
| Holoprosencephaly | 8 | 6.7 | Omphalocele | 19 | 7.9 |
| Hydrocephaly | 9 | 1.0 | Gastroschisis | 9 | 1.3 |
| Hypoplastic left heart syndrome | 3 | 1.1 | Trisomy 13 | 25 | 16.1 |
| Cleft palate without cleft lip | 2 | 0.3 | Trisomy 18 | 69 | 21.1 |
| Cleft lip with or without cleft palate | 39 | 3.2 | Down syndrome | 40 | 2.4 |
| Renal agenesis | 19 | 8.3 | | | |

Total ToPs with births defects = 376 (Ratio ToPs/Births: 0.32 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# USA-Texas: BDES, 2011

| | |
|---|---|
| Live births (LB) | 377,336 |
| Stillbirths (SB) | 2,087 |
| Total births | 379,423 |
| Number of terminations of pregnancy (ToP) for birth defects | 167 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 33 | 30 | 38 | 2.66 |
| Spina bifida | 153 | 7 | 4 | 4.32 |
| Encephalocele | 36 | 1 | 5 | 1.11 |
| Microcephaly | 540 | 1 | 0 | 14.26 |
| Holoprosencephaly | 37 | 4 | 4 | 1.19 |
| Hydrocephaly | 293 | 4 | 2 | 7.88 |
| Anophthalmos | 17 | 0 | 2 | 0.50 |
| Microphthalmos | 108 | 3 | 1 | 2.95 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 10 | 1 | 0 | 0.29 |
| Microtia | 126 | 0 | 0 | 3.32 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 189 | 2 | 2 | 5.09 |
| Tetralogy of Fallot | 162 | 1 | 0 | 4.30 |
| Hypoplastic left heart syndrome | 82 | 3 | 0 | 2.24 |
| Coarctation of aorta | 211 | 3 | 0 | 5.64 |
| Choanal atresia, bilateral | 53 | 0 | 0 | 1.40 |
| Cleft palate without cleft lip | 190 | 11 | 0 | 5.30 |
| Cleft lip with or without cleft palate | 418 | 20 | 12 | 11.86 |
| Oesophageal atresia/stenosis with or without fistula | 87 | 4 | 0 | 2.40 |
| Small intestine atresia/stenosis | 67 | 0 | 0 | 1.77 |
| Anorectal atresia/stenosis | 176 | 15 | 5 | 5.17 |
| Undescended testis (36 weeks of gestation or later) | 560 | 2 | 0 | 14.81 |
| Hypospadias | 659 | 2 | 0 | 17.42 |
| Epispadias | 36 | 0 | 0 | 0.95 |
| Indeterminate sex | 11 | 27 | 8 | 1.21 |
| Renal agenesis | 57 | 8 | 5 | 1.84 |
| Cystic kidney | 231 | 3 | 1 | 6.19 |
| Bladder exstrophy | 6 | 0 | 0 | 0.16 |
| Polydactyly, preaxial | 159 | 1 | 0 | 4.22 |
| Total Limb reduction defects (include unspecified) | 220 | 17 | 8 | 6.46 |
| Transverse | 100 | 11 | 4 | 3.03 |
| Preaxial | 62 | 2 | 0 | 1.69 |
| Postaxial | 7 | 0 | 0 | 0.18 |
| Intercalary | 6 | 0 | 1 | 0.18 |
| Mixed | 33 | 2 | 3 | 1.00 |
| Unspecified | 12 | 2 | 0 | 0.37 |
| Diaphragmatic hernia | 100 | 1 | 0 | 2.66 |
| Omphalocele | 48 | 12 | 9 | 1.82 |
| Gastroschisis | 202 | 13 | 2 | 5.72 |
| Unspecified Omphalocele/Gastroschisis | 13 | 7 | 3 | 0.61 |
| Prune belly sequence | 6 | 1 | 0 | 0.18 |
| Trisomy 13 | 28 | 7 | 8 | 1.13 |
| Trisomy 18 | 60 | 25 | 24 | 2.87 |
| Down syndrome, all ages (include age unknown) | 519 | 25 | 13 | 14.68 |
| <20 | 24 | 2 | 0 | 6.00 |
| 20-24 | 73 | 1 | 1 | 7.73 |
| 25-29 | 73 | 4 | 0 | 7.30 |
| 30-34 | 98 | 6 | 4 | 12.74 |
| 35-39 | 150 | 6 | 5 | 41.21 |
| 40-44 | 87 | 4 | 3 | 107.40 |
| 45+ | 14 | 2 | 0 | 318.73 |
| unknown | 0 | 0 | 0 | --- |

nr = data not reported or not available
Only definite diagnosed cases are reported

# USA-Texas: BDES, Previous years rates 1996 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996* | 1997-2001 | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | **114,765** | **1,565,369** | **1,927,544** | **1,988,714** |
| Anencephaly | | | | | 4.01 | 3.03 | 2.39 | 2.71 |
| Spina bifida | | | | | 5.75 | 3.99 | 3.55 | 3.91 |
| Encephalocele | | | | | 1.48 | 1.00 | 0.80 | 1.03 |
| Microcephaly | | | | | 5.49 | 6.44 | 8.87 | 12.88 |
| Holoprosencephaly | | | | | 1.57 | 1.30 | 1.20 | 1.10 |
| Hydrocephaly | | | | | 6.36 | 6.96 | 6.06 | 7.20 |
| Anophthalmos | | | | | 0.26 | 0.35 | 0.35 | 0.34 |
| Microphthalmos | | | | | 1.92 | 2.51 | 2.68 | 2.90 |
| Unspecified Anophthalmos/Microphthalmos | | | | | 0.00 | 0.00 | 0.00 | 0.00 |
| Anotia | | | | | 0.35 | 0.22 | 0.31 | 0.25 |
| Microtia | | | | | 2.53 | 2.49 | 2.73 | 3.31 |
| Unspecified Anotia/Microtia | | | | | 0.00 | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | | | | 4.79 | 4.94 | 4.81 | 5.30 |
| Tetralogy of Fallot | | | | | 2.70 | 3.12 | 3.58 | 3.88 |
| Hypoplastic left heart syndrome | | | | | 2.27 | 2.05 | 2.12 | 2.19 |
| Coarctation of aorta | | | | | 5.23 | 4.43 | 5.21 | 5.20 |
| Choanal atresia, bilateral | | | | | 0.96 | 1.25 | 1.13 | 1.29 |
| Cleft palate without cleft lip | | | | | 5.58 | 5.85 | 5.30 | 5.89 |
| Cleft lip with or without cleft palate | | | | | 10.19 | 10.86 | 10.76 | 10.43 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 2.09 | 2.12 | 2.02 | 2.13 |
| Small intestine atresia/stenosis | | | | | 1.66 | 1.75 | 1.69 | 2.00 |
| Anorectal atresia/stenosis | | | | | 4.01 | 4.50 | 5.06 | 4.86 |
| Undescended testis (36 weeks of gestation or later) | | | | | 5.66 | 8.37 | 10.32 | 13.74 |
| Hypospadias | | | | | 15.77 | 18.19 | 15.82 | 16.62 |
| Epispadias | | | | | 0.70 | 0.68 | 0.71 | 0.96 |
| Indeterminate sex | | | | | 1.48 | 1.47 | 0.79 | 0.94 |
| Renal agenesis | | | | | 1.66 | 2.11 | 1.89 | 2.00 |
| Cystic kidney | | | | | 4.71 | 4.39 | 5.18 | 6.03 |
| Bladder exstrophy | | | | | 0.17 | 0.20 | 0.22 | 0.18 |
| Polydactyly, preaxial | | | | | 2.18 | 2.97 | 3.51 | 4.00 |
| Total Limb reduction defects (include unspecified) | | | | | 5.49 | 5.51 | 5.30 | 6.01 |
| Transverse | | | | | 2.35 | 2.66 | 2.81 | 3.03 |
| Preaxial | | | | | 1.39 | 1.12 | 1.10 | 1.37 |
| Postaxial | | | | | 0.35 | 0.24 | 0.23 | 0.21 |
| Intercalary | | | | | 0.17 | 0.10 | 0.13 | 0.21 |
| Mixed | | | | | 1.13 | 1.20 | 0.81 | 0.95 |
| Unspecified | | | | | 0.09 | 0.20 | 0.21 | 0.26 |
| Diaphragmatic hernia | | | | | 2.61 | 2.63 | 2.72 | 2.91 |
| Omphalocele | | | | | 1.83 | 2.33 | 2.03 | 2.00 |
| Gastroschisis | | | | | 3.22 | 3.90 | 4.66 | 6.14 |
| Unspecified Omphalocele/Gastroschisis | | | | | 0.96 | 0.63 | 0.60 | 0.60 |
| Prune belly sequence | | | | | 0.44 | 0.26 | 0.31 | 0.26 |
| Trisomy 13 | | | | | 0.96 | 1.24 | 1.08 | 1.26 |
| Trisomy 18 | | | | | 3.14 | 2.27 | 2.41 | 2.75 |
| Down syndrome, all ages (include age unknown) | | | | | 11.94 | 12.54 | 12.79 | 14.04 |
| <20 | | | | | 6.57 | 7.34 | 7.50 | 6.96 |
| 20-24 | | | | | 5.35 | 7.01 | 6.39 | 7.27 |
| 25-29 | | | | | 6.14 | 7.38 | 7.07 | 7.75 |
| 30-34 | | | | | 12.56 | 12.20 | 12.50 | 13.05 |
| 35-39 | | | | | 38.39 | 36.15 | 36.11 | 37.79 |
| 40-44 | | | | | 138.01 | 118.50 | 112.97 | 114.60 |
| 45+ | | | | | 0.00 | 167.60 | 187.01 | 208.50 |
| unknown | | | | | --- | --- | --- | --- |

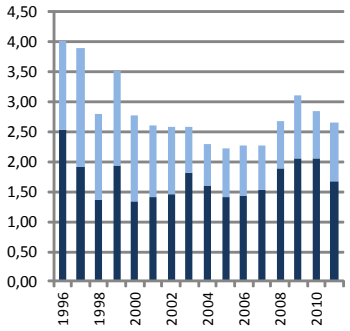
nr = data not reported or not available

* data include less than 5 years

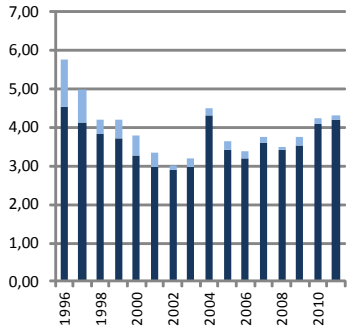
# USA-Texas: BDES, Time trends 1996 – 2011

(Birth prevalence rates per 10,000)

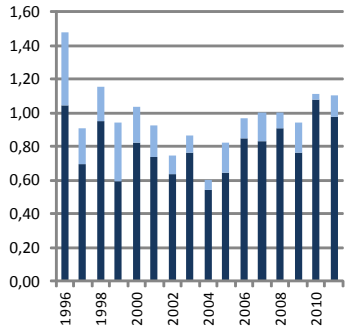
**Anencephaly**



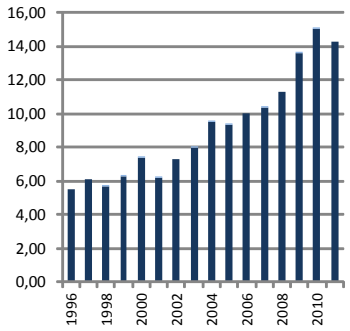

**Spina Bifida**



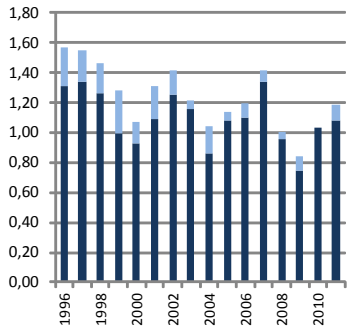

**Encephalocele**



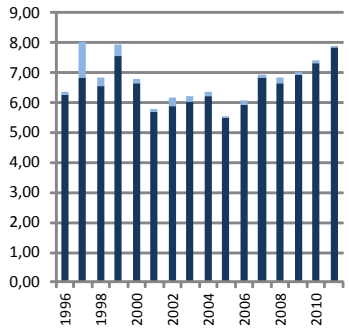

**Microcephaly**



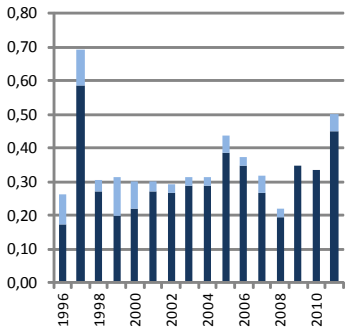

**Holoprosencephaly**



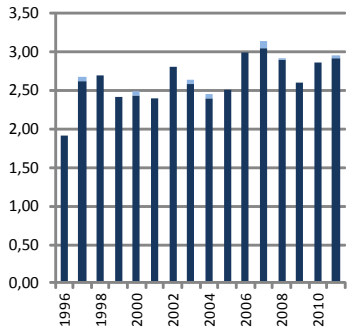

**Hydrocephaly**



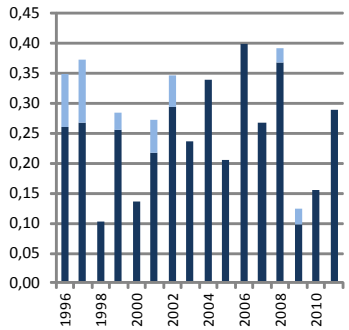

**Anophthalmos**



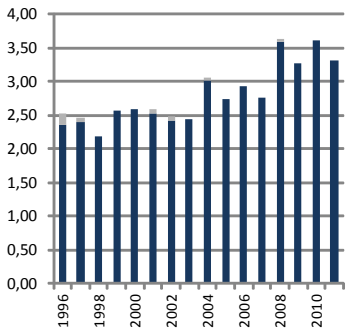

**Microphthalmos**



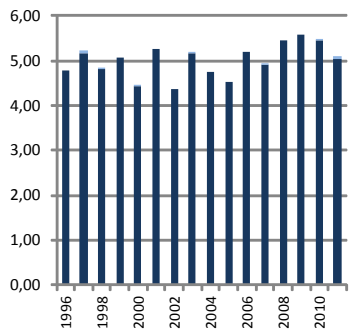

**Anotia**



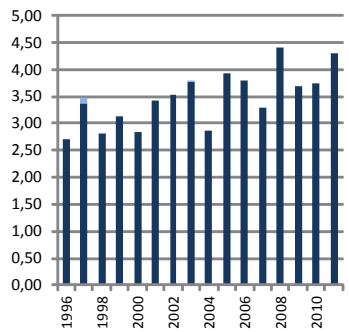

**Microtia**




**Transposition of great vessels**



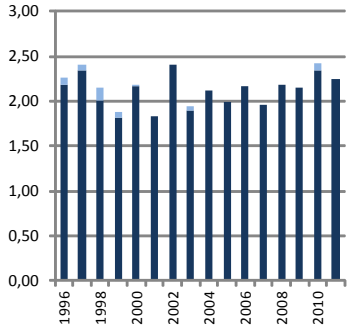

**Tetralogy of Fallot**



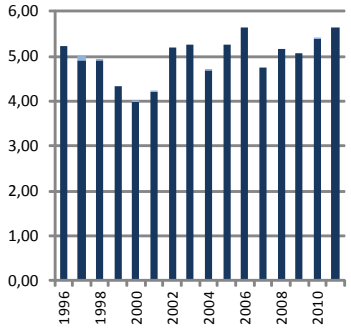
# USA-Texas: BDES, Time trends 1996 – 2011

(Birth prevalence rates per 10,000)

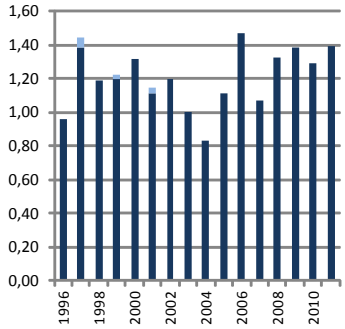
**Hypoplastic left heart syndrome**



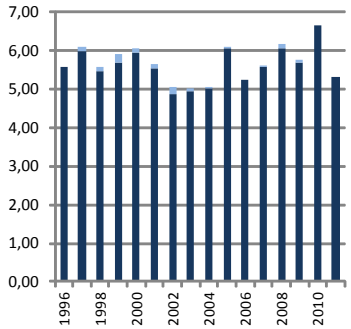

**Coarctation of aorta**



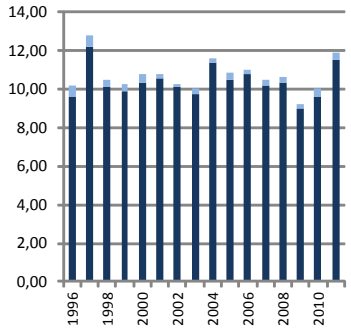

**Choanal atresia, bilateral**



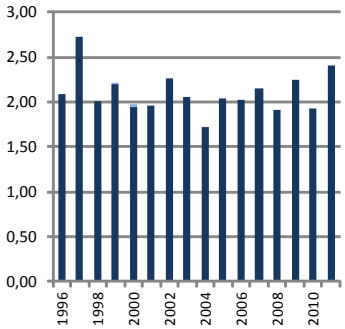

**Cleft palate without cleft lip**



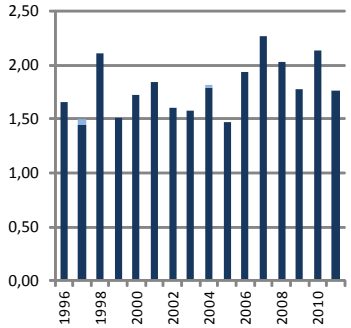

**Cleft lip with or without cleft palate**



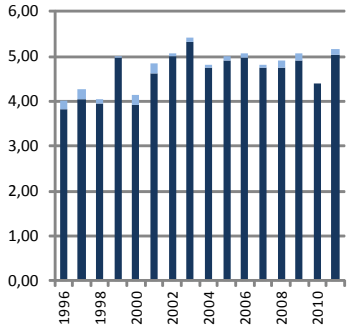

**phageal atresia/stenosis with or without f**



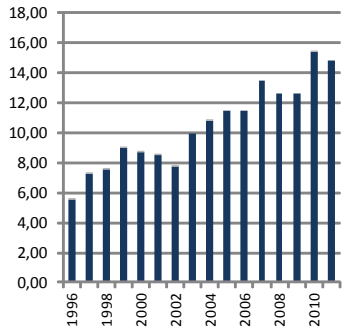

**Small intestine atresia/stenosis**



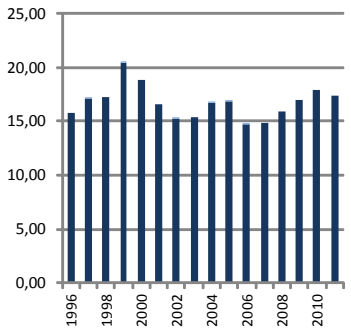

**Anorectal atresia/stenosis**



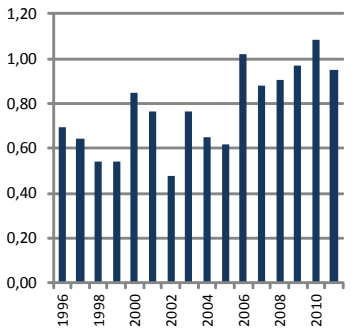

**Undescended testis**



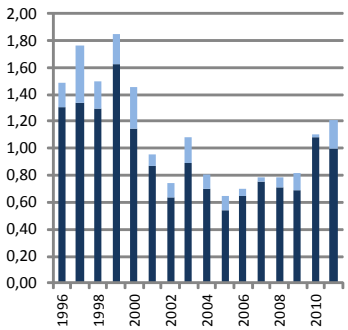

**Hypospadias**




**Epispadias**



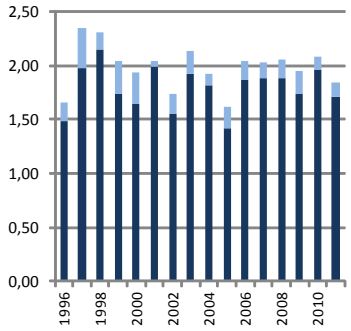

**Indeterminate sex**



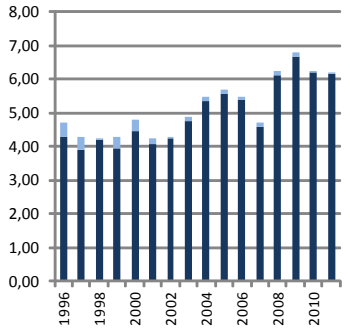
# USA-Texas: BDES, Time trends 1996 – 2011

(Birth prevalence rates per 10,000)

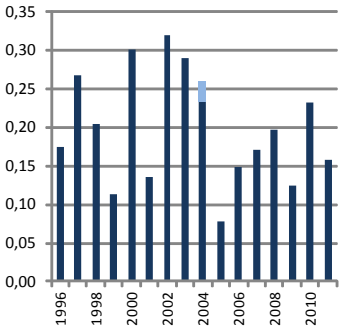
**Renal agenesis**



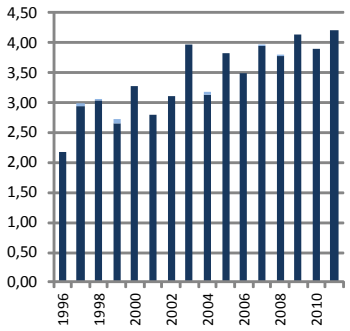

**Cystic kidney**



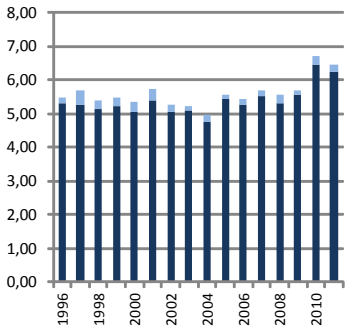

**Bladder exstrophy**



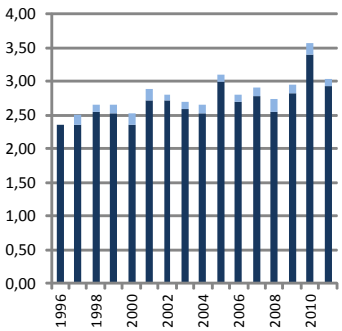

**Polydactyly, preaxial**



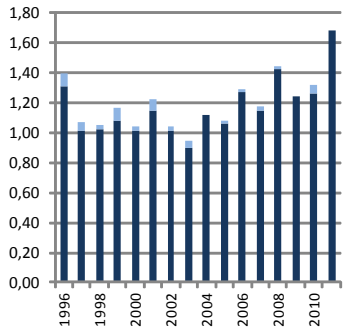

**Limb reduction defects**



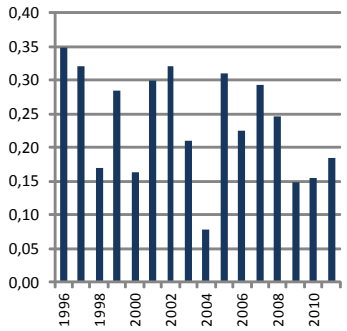

**Limb reduction defects - transverse**



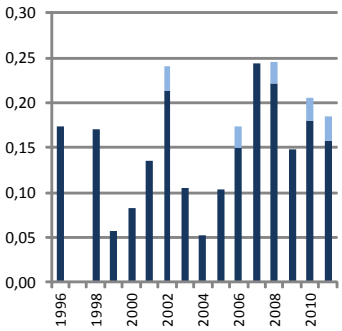

**Limb reduction defects - preaxial**



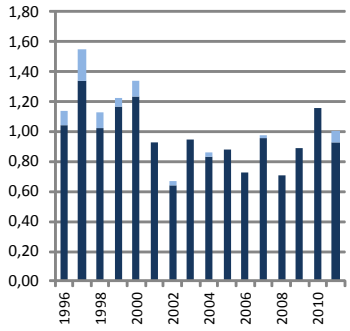

**Limb reduction defects - postaxial**



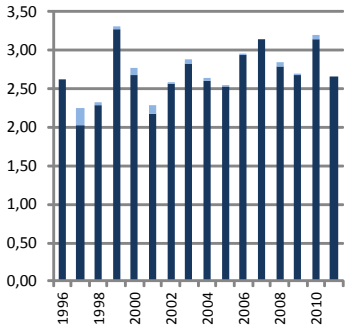

**Limb reduction defects - intercalary**



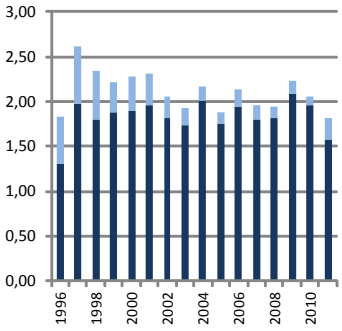

**Limb reduction defects - mixed**




**Diaphragmatic hernia**



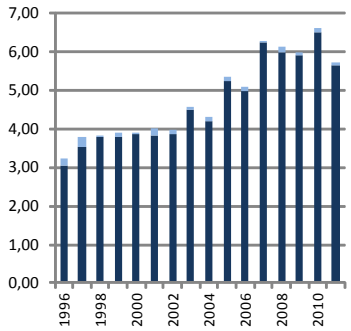

**Omphalocele**



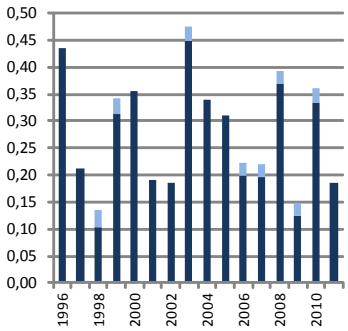
# USA-Texas: BDES, Time trends 1996 – 2011

(Birth prevalence rates per 10,000)

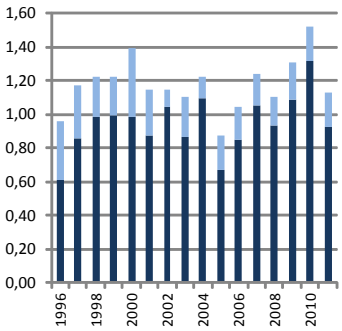
**Gastroschisis**



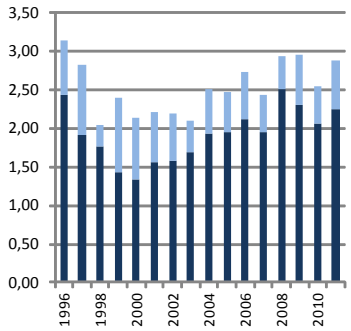

**Prune belly sequence**



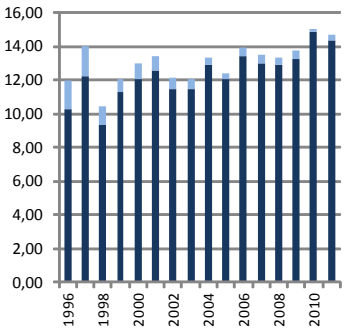

**Trisomy 13**



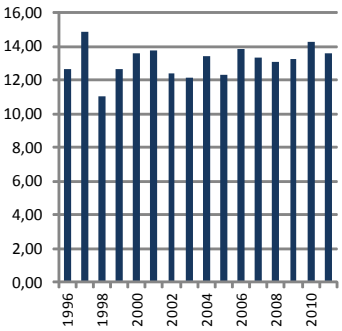

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# USA-Utah: UBDN

Utah Birth Defects Network

**History:**
The Utah Birth Defect Network (UBDN) began in 1994 monitoring neural tube defects, expanding its identification of major malformations through 1999 when all major structural birth defects were identified. The program is a full member of the ICBDSR.

**Size and coverage:**
The UBDN is a statewide population-based surveillance system covering over 50,000 births annually.

**Legislation and funding:**
In 1999 an Administrative Rule was enacted under the Utah Health Code Statute which mandates all delivery hospitals and laboratories to report any pregnancy or infant diagnosed with a birth defect. This administrative rule also covers those health care providers and other agencies that voluntarily report a birth defect case to the UBDN. The UBDN surveillance staff is funded by both state and federal funds. The UBDN has many research projects funded from federal sources (e.g., Centers for Disease Control and Prevention).

**Sources of ascertainment:**
Multiple sources (n=128), such as delivery units, paediatric departments, laboratories, prenatal diagnostic centers, hospital discharge data, other specialties, and champions are used to ascertained malformed infants born in Utah. These sources include reports that are generated by the facilities, case reports submitted by individual care providers, as well as reports actively obtained by UBDN staff reviewing records or log books.

**Exposure information:**
Basic risk factors including medications taken during pregnancy, infections, chronic conditions are all recorded based on medical records abstraction.

**Background information:**
Detailed background information including demographics, reproductive history, gestational age, prenatal diagnostics, and family history are all collected from the medical record. The number of births and basic demographic data are obtained from vital statistics.

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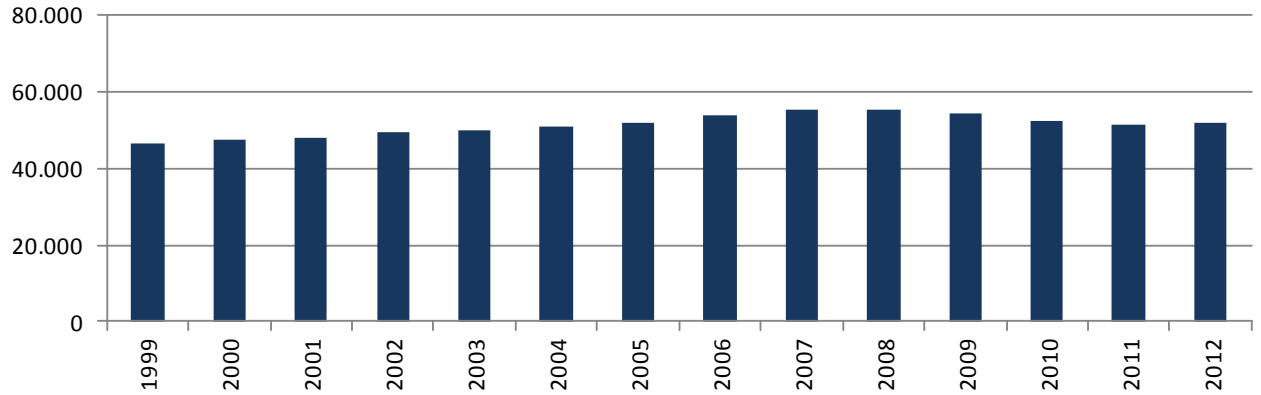
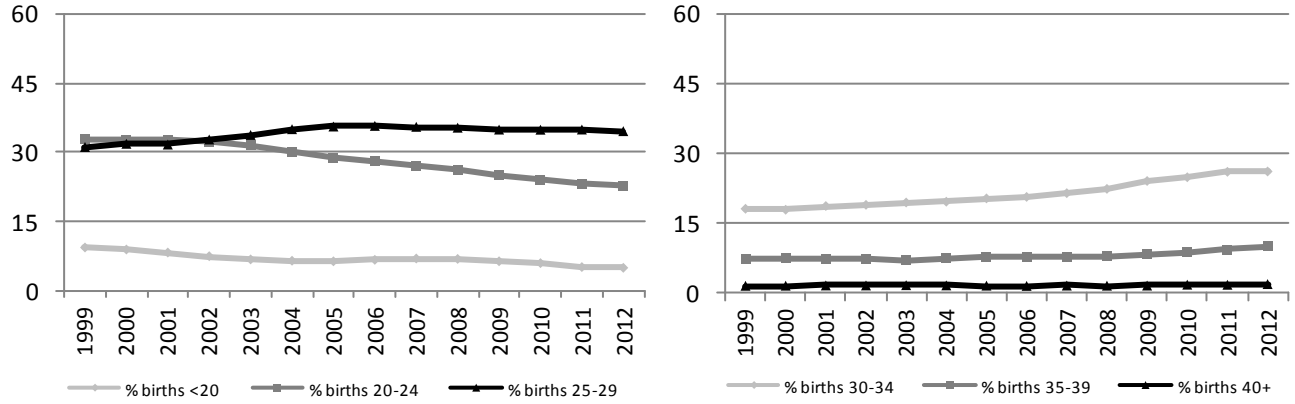
**Website:**
http://www.health.utah.gov/birthdefect/

# USA-Utah: UBDN

## Total births by year




## Percentage of births by year and maternal age




**Terminations of pregnancy (ToPs) in selected malformations (2010-2012)**
**(Total cases: isolated + multiples + syndromes)**

| Birth defects | ToPs | % of ToPs (*) | Birth defects | ToPs | % of ToPs (*) |
|---|---|---|---|---|---|
| Anencephaly | 18 | 52.9 | Cystic kidney | 7 | 8.4 |
| Spina bifida | 6 | 13.6 | Limb reduction defects | 5 | 5.9 |
| Encephalocele | 2 | 22.2 | Diaphragmatic hernia | 2 | 4.3 |
| Holoprosencephaly | 5 | 33.3 | Omphalocele | 11 | 22.0 |
| Hydrocephaly | 0 | 0.0 | Gastroschisis | 1 | 1.9 |
| Hypoplastic left heart syndrome | 7 | 15.6 | Trisomy 13 | 9 | 36.0 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 13 | 23.2 |
| Cleft lip with or without cleft palate | 9 | 4.1 | Down syndrome | 17 | 7.8 |
| Renal agenesis | 12 | 19.4 | | | |

Total ToPs with births defects = 102 (Ratio ToPs/Births: 0.66 per 1.000)
(*) % of ToPs = ToPs/(ToPs+Births)

# USA-Utah: UBDN, 2012

| | |
|---|---|
| Live births (LB) | 51,439 |
| Stillbirths (SB) | 301 |
| Total births | 51,740 |
| Number of terminations of pregnancy (ToP) for birth defects | 31 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 4 | 0 | 1 | 0.97 |
| Spina bifida | 15 | 1 | 2 | 3.48 |
| Encephalocele | 1 | 0 | 1 | 0.39 |
| Microcephaly | 11 | 0 | 0 | 2.13 |
| Holoprosencephaly | 2 | 1 | 2 | 0.97 |
| Hydrocephaly | 3 | 0 | 0 | 0.58 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 7 | 1 | 0 | 1.55 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 1 | 0 | 0.19 |
| Microtia | 13 | 0 | 1 | 2.71 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 10 | 1 | 0 | 2.13 |
| Tetralogy of Fallot | 13 | 0 | 1 | 2.71 |
| Hypoplastic left heart syndrome | 12 | 0 | 3 | 2.90 |
| Coarctation of aorta | 32 | 3 | 0 | 6.76 |
| Choanal atresia, bilateral | 10 | 0 | 0 | 1.93 |
| Cleft palate without cleft lip | 26 | 1 | 0 | 5.22 |
| Cleft lip with or without cleft palate | 69 | 3 | 3 | 14.50 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | 0 | 1.55 |
| Small intestine atresia/stenosis | 7 | 0 | 0 | 1.35 |
| Anorectal atresia/stenosis | 17 | 0 | 1 | 3.48 |
| Undescended testis (36 weeks of gestation or later) | 0 | 0 | 0 | 0.00 |
| Hypospadias | 53 | 0 | 1 | 10.44 |
| Epispadias | 1 | 0 | 0 | 0.19 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 14 | 2 | 5 | 4.06 |
| Cystic kidney | 19 | 0 | 3 | 4.25 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 0 | 0 | 0 | 0.00 |
| Total Limb reduction defects (include unspecified) | 14 | 1 | 2 | 3.29 |
| Transverse | 3 | 0 | 0 | 0.58 |
| Preaxial | 6 | 1 | 0 | 1.35 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 0 | 0 | 1 | 0.19 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 3 | 0 | 1 | 0.77 |
| Diaphragmatic hernia | 15 | 1 | 1 | 3.29 |
| Omphalocele | 9 | 5 | 6 | 3.87 |
| Gastroschisis | 16 | 0 | 0 | 3.09 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 1 | 1 | 1 | 0.58 |
| Trisomy 13 | 6 | 1 | 3 | 1.93 |
| Trisomy 18 | 9 | 5 | 6 | 3.87 |
| Down syndrome, all ages (include age unknown) | 62 | 2 | 3 | 12.95 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 11 | 1 | 0 | 10.23 |
| 25-29 | 9 | 0 | 0 | 5.04 |
| 30-34 | 15 | 0 | 1 | 11.86 |
| 35-39 | 20 | 1 | 2 | 44.56 |
| 40-44 | 5 | 0 | 0 | 55.13 |
| 45+ | 2 | 0 | 0 | 363.64 |
| unknown | --- | --- | --- | --- |

nr = data not reported or not available

There are several birth defects that appear to have decreased in prevalence in 2012. These include anencephaly, encephalocele, hydrocephalus, anopthalmia, coarctation, esophogeal atresia/TEF, transverse limb deficiency, postaxial limb reduction defects, all limb deficiencies, and gastroschisis. The prevalence of these defects is much lower than the previous seven years. The reason(s) for this decrease in counts (and therefore, prevalence) is not known but should be viewed as potentially incomplete or under-reported.

# USA-Utah: UBDN, Previous years rates 1999 – 2011

Birth prevalence rates: (LB+SB+TOP) * 10,000

| Birth Defects | 1974-1976 | 1977-1981 | 1982-1986 | 1987-1991 | 1992-1996 | 1997-2001* | 2002-2006 | 2007-2011 |
|---|---|---|---|---|---|---|---|---|
| **Total births** | | | | | | **142,188** | **255,878** | **268,952** |
| Anencephaly | | | | | | 2.18 | 2.31 | 2.68 |
| Spina bifida | | | | | | 3.31 | 4.38 | 3.50 |
| Encephalocele | | | | | | 0.91 | 0.86 | 0.82 |
| Microcephaly | | | | | | 2.88 | 6.06 | 3.42 |
| Holoprosencephaly | | | | | | 1.27 | 1.60 | 1.75 |
| Hydrocephaly | | | | | | 3.94 | 4.53 | 2.19 |
| Anophthalmos | | | | | | 0.14 | 0.35 | 0.52 |
| Microphthalmos | | | | | | 1.55 | 1.45 | 1.49 |
| Unspecified Anophthalmos/Microphthalmos | | | | | | 0.00 | 0.00 | 0.00 |
| Anotia | | | | | | 0.21 | 0.16 | 0.11 |
| Microtia | | | | | | 1.76 | 2.85 | 3.79 |
| Unspecified Anotia/Microtia | | | | | | 0.00 | 0.00 | 0.00 |
| Transposition of great vessels | | | | | | 5.42 | 4.77 | 3.01 |
| Tetralogy of Fallot | | | | | | 4.99 | 3.79 | 3.09 |
| Hypoplastic left heart syndrome | | | | | | 3.73 | 3.44 | 3.42 |
| Coarctation of aorta | | | | | | 7.60 | 8.64 | 9.56 |
| Choanal atresia, bilateral | | | | | | 0.07 | 0.31 | 0.89 |
| Cleft palate without cleft lip | | | | | | 7.45 | 7.27 | 5.47 |
| Cleft lip with or without cleft palate | | | | | | 14.56 | 13.25 | 13.61 |
| Oesophageal atresia/stenosis with or without fistula | | | | | | 2.74 | 2.46 | 2.75 |
| Small intestine atresia/stenosis | | | | | | 1.27 | 1.41 | 1.41 |
| Anorectal atresia/stenosis | | | | | | 3.09 | 3.52 | 3.23 |
| Undescended testis (36 weeks of gestation or later) | | | | | | nr | nr | nr |
| Hypospadias | | | | | | 4.29 | 6.96 | 9.52 |
| Epispadias | | | | | | 0.28 | 0.12 | 0.11 |
| Indeterminate sex | | | | | | nr | nr | nr |
| Renal agenesis | | | | | | 3.59 | 3.67 | 3.27 |
| Cystic kidney | | | | | | 5.20 | 5.39 | 4.80 |
| Bladder exstrophy | | | | | | 0.28 | 0.12 | 0.26 |
| Polydactyly, preaxial | | | | | | nr | nr | nr |
| Total Limb reduction defects (include unspecified) | | | | | | 5.84 | 6.41 | 6.43 |
| Transverse | | | | | | 3.31 | 3.20 | 3.31 |
| Preaxial | | | | | | 1.48 | 1.56 | 1.45 |
| Postaxial | | | | | | 0.07 | 0.12 | 0.33 |
| Intercalary | | | | | | 0.07 | 0.12 | 0.26 |
| Mixed | | | | | | 0.70 | 1.02 | 0.52 |
| Unspecified | | | | | | 0.14 | 0.16 | 0.37 |
| Diaphragmatic hernia | | | | | | 3.38 | 3.36 | 3.27 |
| Omphalocele | | | | | | 2.60 | 2.74 | 2.94 |
| Gastroschisis | | | | | | 4.22 | 5.20 | 4.61 |
| Unspecified Omphalocele/Gastroschisis | | | | | | 0.00 | 0.00 | 0.00 |
| Prune belly sequence | | | | | | 0.14 | 0.16 | 0.56 |
| Trisomy 13 | | | | | | 1.48 | 1.91 | 1.75 |
| Trisomy 18 | | | | | | 3.38 | 3.56 | 3.38 |
| Down syndrome, all ages (include age unknown) | | | | | | 15.12 | 15.32 | 14.54 |
| <20 | | | | | | 8.03 | 11.73 | 6.62 |
| 20-24 | | | | | | 8.18 | 9.23 | 6.70 |
| 25-29 | | | | | | 8.04 | 9.05 | 7.97 |
| 30-34 | | | | | | 12.37 | 14.03 | 13.71 |
| 35-39 | | | | | | 60.45 | 46.57 | 41.87 |
| 40-44 | | | | | | 143.21 | 142.97 | 173.61 |
| 45+ | | | | | | 413.79 | 406.09 | 326.09 |
| unknown | | | | | | --- | --- | --- |

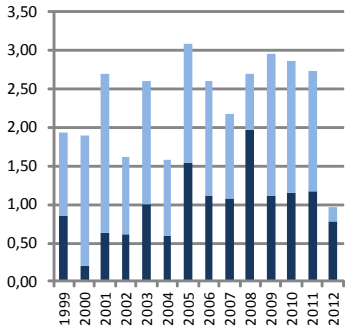
nr = data not reported or not available

* data include less than 5 years

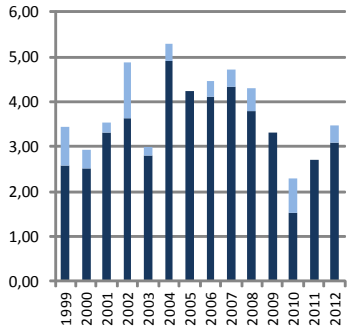
# USA-Utah: UBDN, Time trends 1999 – 2012

(Birth prevalence rates per 10,000)

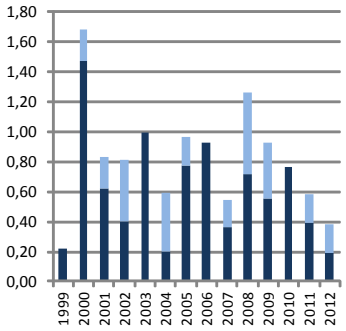
**Anencephaly**



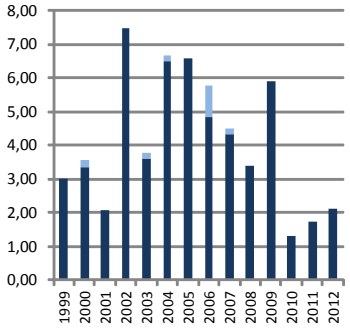

**Spina Bifida**



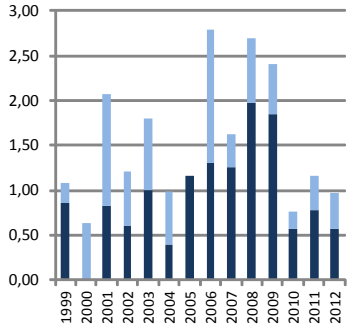

**Encephalocele**



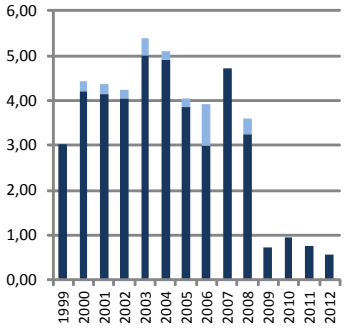

**Microcephaly**



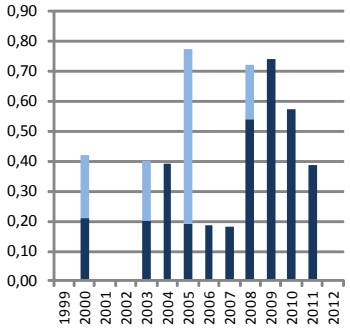

**Holoprosencephaly**



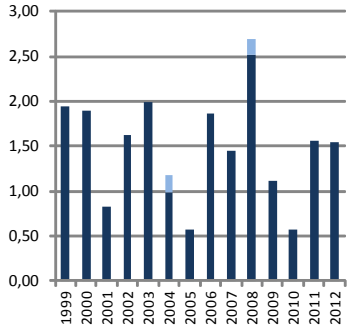

**Hydrocephaly**



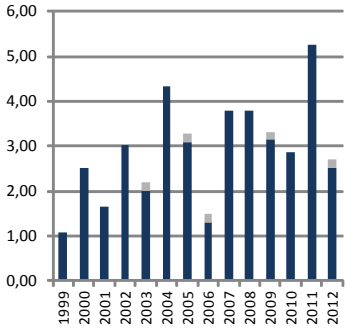

**Anophthalmos**



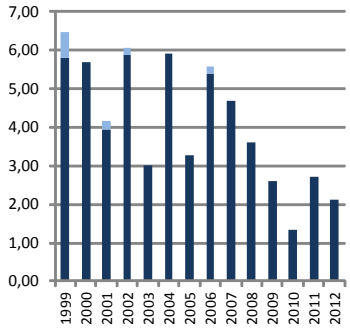

**Microphthalmos**



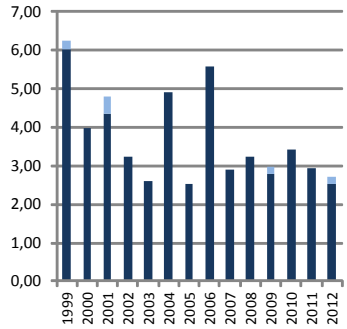

**Microtia**



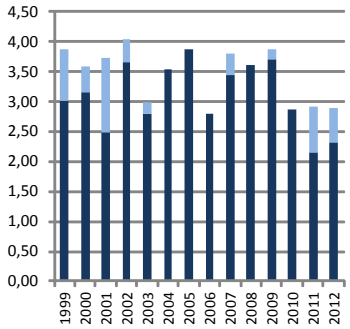

**Transposition of great vessels**




**Tetralogy of Fallot**



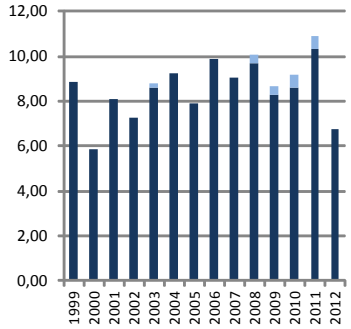

**Hypoplastic left heart syndrome**



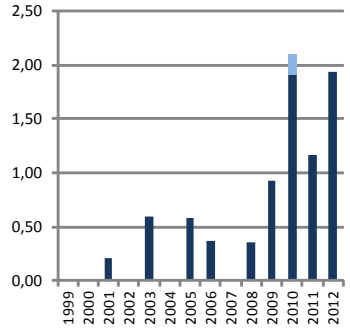
# USA-Utah: UBDN, Time trends 1999 – 2012

(Birth prevalence rates per 10,000)

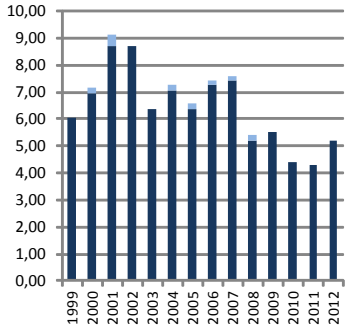
**Coarctation of aorta**



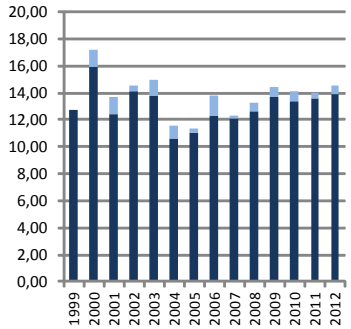

**Choanal atresia, bilateral**



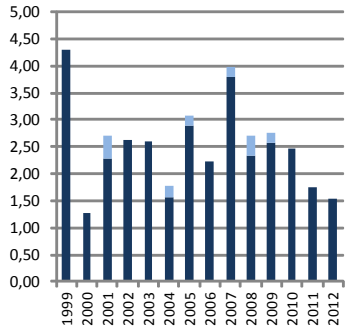

**Cleft palate without cleft lip**



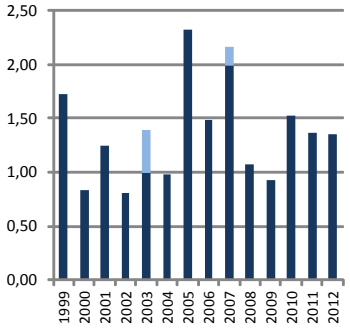

**Cleft lip with or without cleft palate**



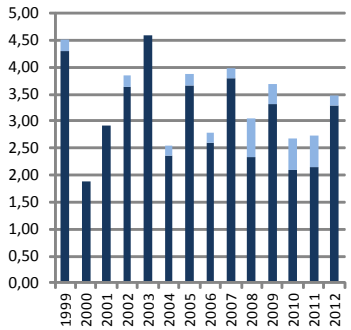

**phageal atresia/stenosis with or without f**



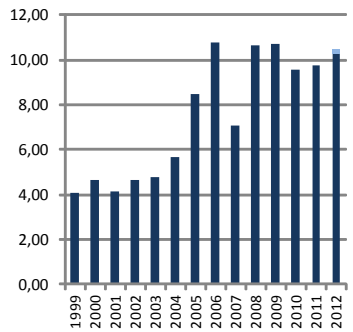

**Small intestine atresia/stenosis**



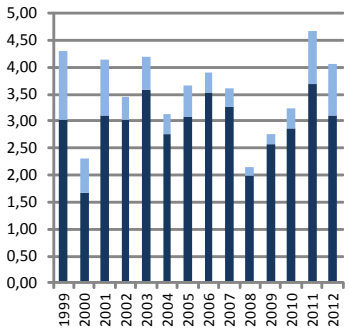

**Anorectal atresia/stenosis**



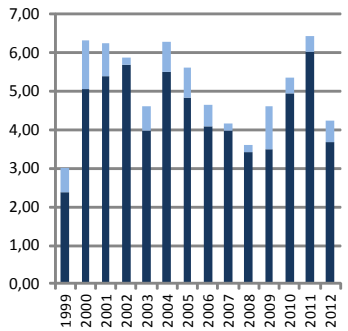

**Hypospadias**



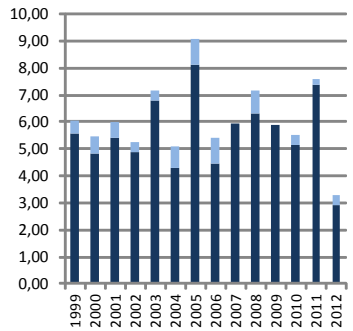

**Renal agenesis**



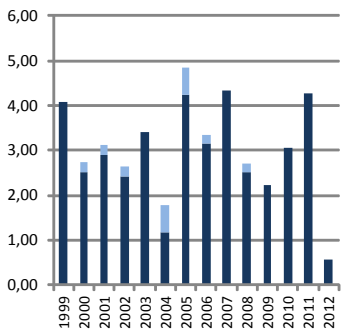

**Cystic kidney**




**Limb reduction defects**



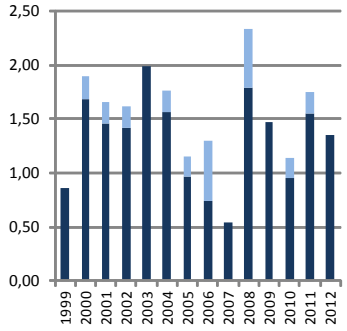

**Limb reduction defects - transverse**



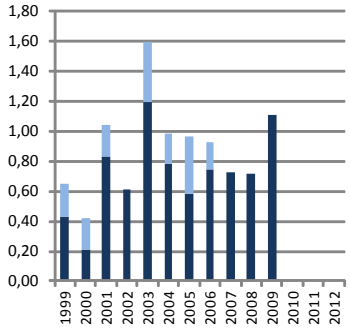
# USA-Utah: UBDN, Time trends 1999 – 2012

(Birth prevalence rates per 10,000)

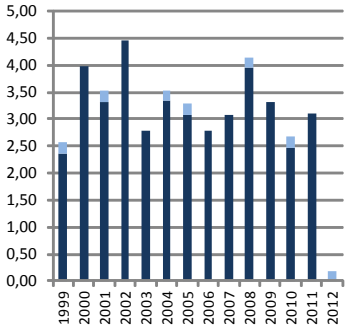
**Limb reduction defects - preaxial**



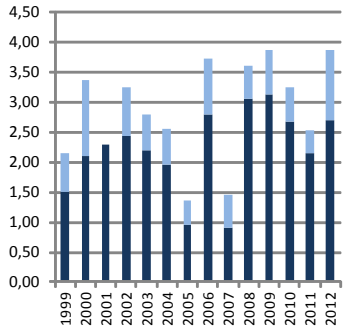

**Limb reduction defects - mixed**



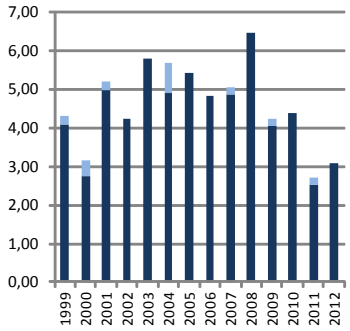

**Diaphragmatic hernia**



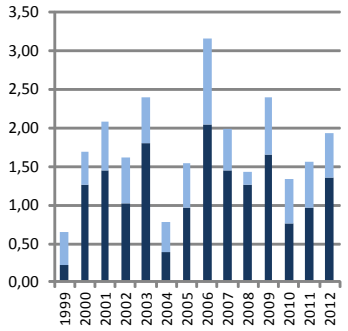

**Omphalocele**



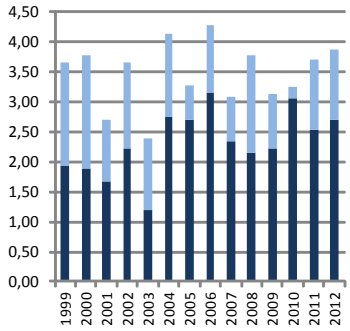

**Gastroschisis**



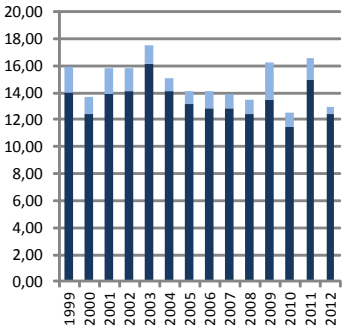

**Trisomy 13**



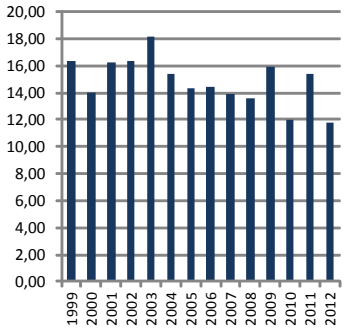

**Trisomy 18**




**Down Syndrome**




**Down Syndrome standardized total rate**




**L + S rates** **ToP rates**

# Monitoring Systems, not contributing with Annual Data: description of the registry

# Chile-Maule: RRMC-SSM

Regional Register Congenital Malformational Maule Health Service

**History:**
The register started in 2001 defined by order of Director Maule Health Service and assessed for South America.ECLAMC (Latin American Collaborative Study of Congenital Malformations) RRMC-SSM became a member of ICBDSR in 2003.

**Size and coverage:**
RRMC-SSM is located in a Region in the center of Chile, in Talca Maule Region. Maule Region is situated between 34º 41' & 36º 33' S and 70º 20' & 72º 44' W. The surface is 30.535 kms2 (4 % of Chile). 930,306 habitants. 37,4% rurality. Cellulosa producer and agricultural products. The number of participating are 13 public hospitals from 2001 and since 2004 will included the unique private maternity of the region. There are around 13.500 births annually (2002).
The information about livebirths and stillbirths are collected from 13 maternity hospitals in the region for pediatricians and midwives. Stillbirths of at least 500g birthweight have been included since 2001.

**Legislation and funding:**
The registry is based on the information of births and notification of congenital malformation ECLAMC from 2001 and funded by the Maule Health Service.

**Sources of ascertainment:**
Reporting is made by collaborating pediatricians and midwives at the delivery units of participating hospitals.

**Exposure information:**
Detailed information on various risk factor exposures, maternal and paternal occupation, diseases and other information available.

**Background information:**
Epidemiological information on all births is available from participating hospitals and statisticals units.

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Rosa Gajardo Abarza
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## Colombia-Bogota: BCMSP

Bogota Congenital Malformations Surveillance Program

**History:**
The Bogota Congenital Malformations Surveillance Program was initiated by the Institute of Human Genetics of the Pontificia Universidad Javeriana in the year 2001 and was developed based on the Latin American Collaborative Study of Congenital Malformations (ECLAMC). In 2006 the health authorities of the city of Bogotá (District Health Secretary of Bogotá) joined the program and since then have become a key ally for its adequate functioning.

**Size and Coverage:**
The program is hospital based register. In 2001 surveillance began in one hospital of Bogotá D.C. and coverage has been expanded up to a total of 56 hospitals in 2012. In the past year approximately 104,700 births were monitored.

**Legislation and funding:**
The program is based on the Latin American Collaborative Study of Congenital Malformations, ECLAMC, and is financed by the health authorities of the city of Bogota (District Health Secretary of Bogotá) together with the Pontificia Universidad Javeriana. In 2007 the Ministry of Social Protection issued a decree which enforced the implementation of birth defects surveillance systems.

**Sources of ascertainment:**
There are two modalities for surveillance: monitor and case-control. The first one depends on the staff of each hospital (nurses, gynecologists, neonatologists), and the latter is held by physicians who are previously trained to actively search for congenital anomalies through a systematic physical exam. Both modalities include a format that obliges health care providers to realize a textual and thorough description of the anomalies according to the ECLAMC manual.

**Exposure Information:**
The format that is filled out by physicians that participate in the case-control modality includes many variables such as immunizations, acute diseases during pregnancy, chronic diseases, physical factors (x-rays, surgery, radiotherapy etc.), drugs, smoking, recreational drugs, alcohol, level of education of parents and place where they lived during the periconceptional period.

**Background information:**
Epidemiological information may be accessed at www.anomaliascongenitas.org

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# Cuba: RECUMAC

## Cuban Register of Congenital malformation

**History:**
The program started in 1985 and has grown in size and coverage. The registry became a member of ICBDSR in 2003.

**Size and coverage:**
Reports are obtained from hospitals distributed all over Cuba. The number of partecipating hospitals has grown in 1986 to 60 at the present time. The annual number of birth is approximately 121,000 representing almost 96% of all births.

**Legislation and funding:**
RIt is a research programme with voluntary participation of hospitals. The registry is associated with the National Centre of Medical Genetics, and is financed by Health Public Ministry of Cuba.

**Sources of ascertainment:**
Reports are obtained from delivery units paediatric departments of the participating hospitals. Mothers are also interviewed directly to gather information and fill in the RECUMAC standard protocols.

**Exposure information:**
The mother of each reported infant and the mother of a control infant, the next non malformed infant born at the hospital with the same sex as the proband are interviewed on various exposures, including drug usage and parental occupation.

**Background information:**
Total number of birth by sex and number of twin pairs in each participating hospital are known. Other background information is obtained partly from summarizing tables of births in each participating hospital, partly from the control material.

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# Finland

## The Finnish Register of Congenital Malformations 19.2.2012

**History:**
The registry was established in 1963 and regular monitoring started in 1977. It was a founding member of the ICBDSR. In 1998 the registry became an associate member of EUROCAT. The data content and the data collection practices of the registry have been revised in 1985, 1993 and 2005.

**Size and coverage:**
The registry is national and population based. All births in Finland are covered, at present approximately 60,000 annually. Stillbirths of 22 weeks / 500 grams or more are registered. Information on congenital anomalies is principally collected up to the age of 1 year, but later information is also included. Elective terminations of pregnancy for fetal anomalies and spontaneous abortions with congenital anomalies have been included since 1987.

**Legislation and funding:**
Reporting is compulsory. The registry is regulated by the act and statute on the national health care registers with personal data. The registry is run and financed by THL, National Institute for Health and Welfare (under the Ministry of Social Affairs and Health).

**Sources and ascertainment:**
Reports are obtained from delivery units, neonatal, paediatric and pathology departments, death certificates and cytogenetic laboratories. Case information is also received from the national Medical Birth Register, the Care Register for Health Care (including Information on Outpatient Services in Specialised Health Care), the Register on Induced Abortions and the Register of Visual Impairment, all maintained by THL, from the National Supervisory Authority for Welfare and Health (Valvira) as well as from the Cause of Death Statistics, maintained by Statistics Finland. The diagnoses of the cases with congenital anomalies received from these other sources are confirmed from the hospitals.

**Exposure information:**
Until 1986, extensive exposure information was obtained from maternity health centres and by personal interviews for cases with selected congenital anomalies and their controls. In 1987–1992 only parental occupation was reported. Exposure information, like maternal occupation, medication, X-rays and diseases, etc., has been obtained since 1993. Some exposure information on all births is also available in the Medical Birth Register since 1987.

**Background information:**
Epidemiological background data are available on all births in the Medical Birth Register and in the Statistics Finland.

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**Phone:** +358 29 52 47376
**Fax:** +358 29 53 47459
**E-mail:** annukka.ritvanen(at)thl.fi
**Website:** http://www.thl.fi or http://www.thl.fi/statistics/congenitalmalformations

# India: BDRI

## Birth Defects Registry of India

**History**:
BDRI is a part of Fetal Care Research Association a not for profit organisation that is dedicated to Preventive Curative and Supportive care of Birth Defects. With a population of 1.21 billion India is second only to China in population. Every year, India adds more people than any other nation in the world, and in fact the individual population of some of its states is equal to the total population of many countries. Founded in 2001, BDRI started with a few chennai hospitals and reported 15000 births. Initially BDRI encouraged each district to have a nodal leader which in turn would collect data from participant hospitals and submit it to the Central Registry. The data was sent as hard copy files by post. But in time it was found that there was more reception to the idea of individual reporting and therefore we now have around 750 hospitals reporting data from all over India across 28 states and three union territories. The Registry now has the facilty of online reporting which has made it user friendly. BDRI has so far analysed almost 10 lakh births . As a result of these studies important conclusions have been made on birth defects in general and neural tube defects in particular. In return to the member hospitals who contribute data, BDRI shares its study in the form of quarterly meetings and quarterly newsletters, thereby helping in evolving strategies on handling birth defects.Out of a total of birth of 25 million a year BDRI represents only an annual birth of 2 lakhs a year as it is a voluntary hospital based passive Registry. Statistical Report is published annually.

**Legislation and funding**:
The funding is by Fetal Care Research Foundation and we do not have any external funding. But however as a fallout of this program the Government collaborated with us for Project on NTD.

**Sources of ascertainment:**
All our contributing hospitals are Obstetrics hospitals and the idea of Paediatricians and neonatologists contributing is just picking up.

**Exposure information**:
We do not have any exposure information.

**Background information**:
BDRI is a hospital based passive registry. The inclusion criteria is for both major and minor anomalies diagnosed in the antenatal period up to children of one year of age.The exclusion criteria is for Functional problems without any obvious structural anomaly; e.g. murmur with no
structural abnormalities in the heart & Hydrops due to Rh iso immunisation or unknown etiology, IUGR due to placental causes & Preterm births.

**Addresses and Staff:**
Dr Prof S.Suresh From 2001 (inception) till date.
Director, Birth Defects Registry Of India
Managing Director, MediScan Systems
197, Dr Natesan Road
Mylapore, Chennai – 4 India
**E-mail:** mediscan@gmail.com
**Website:** www.mediscansystems.org.in

## Israel: IBDSP

Israel Birth Defects Surveillance Program

**History :**
the Programme started in one hospital in 1966 and was a founding member of Clearinghouse.

**Size and coverage:**
Reports are now obtained from five hospitals located in all regions of the country, with more than 40,000 births per year (about 25% of all annual births in Israel). Stillbirths of 20 weeks gestation or more and 500g or more are included. The registry of termination of pregnancy began in 1995.

**Legislation and funding :**
The Programme is a research and surveillance one supported by the Directors of the Departments of Neonatology and by research grants without any governmental support.

**Sources of ascertainment :**
Reporting is voluntary. Reports are obtained from Delivery units and Departments of Neonatology in the participating hospitals. The five included hospitals are: Rabin Medical Center, Beilinson and Schneider Hospitals, Petah Tikva ( Prof L.Sirota , Prof N. Linder ); Kaplan Hospital, Rehovot (Prof E. Shinwell ); Lis Medical Center, Tel-Aviv (Prof Dohlberg ). These hospitals are affiliated to Sackler School of Medicine, Tel-Aviv University. Soroka Medical Center, Beer-Sheva ( Prof E. Zmora, Dr D. Landau ) affiliated to Ben-Gurion University of Negev; Bnai-Zion Medical Center, Haifa ( Prof. D. Bader, Dr M Grun) affiliated to the Technion University, Haifa.

**Exposure information :**
Completeness is obtained by interviews of mothers of all malformed infants. All the other women with normal newborns complete a similar form at birth.

**Background information:**
Epidemiological information on all births occurring in the participating hospitals is available.

**Addresses and Staff:**
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# Italy-Campania: BDRCam

## Birth Defects Registry of Campania

**History**:
The Registry started in 1991 and became a full member of the ICBDSR in 1996.

**Size and coverage:**
The Registry is based on reporting from hospitals distributed in Campania, a region in southern Italy. Naples is the main city. Initially 38 hospitals reported and the annual number of births was 38.000. Until 2001 the registry is hospital-based covering approximately 50.000 annual births. Actually beginning from 2002, the registry is population based covering approximately 100% of all births. Stillbirths and induced abortions are included. In 2002 is started officially a link with birth regional registry (CEDAP). After 2004 started a new link with Hospital discharge schedules registry (SDO). The last link allows to enclose the data after pediatric hospital discharge in the first year of life and to complete the birth data on baby with birth defects. Thus the birh defects ratio is about 5% and not 2%. Unfortunately, the data obtained from SDO registry allows to analyse only minimum data set (birh date, number of birth defects, mother's place of residence. No informations on exposure is possible.

**Legislation and funding:**
The Registry is a surveillance Programme supported by grants from Regional Health Authorities. Participation was voluntary up to 1995. From 1996 participation is mandatory.

**Sources of ascertainment:**
Reports are obtained from delivery units and pediatric clinics at the participating hospitals. For selected malformations multiple sources are used with follow-up to one year using specific records from pediatric specialities departments dealing with malformed infants.

**Exposure information:**
For each malformed infant reported, information is given on certain exposures, including maternal drug usage and parental occupation. Beginning from 2002 informations on controls are available but only partially on induced abortions.

**Background information:**
Always from 2002 background information is given on certain exposures, including maternal drug usage and parental occupation. Informations on controls are available.

**Addresses and Staff:**
Gioacchino Scarano, Registro Campano Difetti Congeniti (BDRCam) Osservatorio Epidemiologico Regione Campania and Medical Genetics Division Azienda Ospedaliera "G. Rummo", Via dell'Angelo 1
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## Italy-Emilia Romagna: IMER

Emilia Romagna Registry of Congenital Malformations

**History**:
The registry was started in 1978 in a few hospitals and has increased in size to now include 45 delivery units. The Programme became an associate member of the Clearinghouse in 1985.

**Size and coverage:**
The Programme is based on approximately 90% of all births in the Emilia-Romagna region, or approximately 25,000 annual births (4% of all births in Italy). Stillbirths of 28 weeks or more gestation are included.

**Legislation and funding:**
The Programme is recognised and financed by the health authorities, the National Research Council, and the Regional Health Council. Hospital participation is voluntary.

**Sources of ascertainment:**
Reporting is made by neonatologists and pediatricians during the first week of the infant's life. Selected malformations are followed up.

**Exposure information:**
Detailed exposure information is obtained by interviews of the mothers of malformed infants. For each malformed infant, a control is chosen (the baby born before or after the malformed case in the same hospital) and its mother is interviewed in a similar way.

**Background information:**
Some general demographic information is known for all births in the area. For each participating hospital, the number of livebirths and stillbirths are known.

**Addresses and Staff:**
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# Italy-North East

## North-East Italy Registry of Congenital Malformations

**History:**
The Registry was established in 1981 to include the Veneto, Friuli Venezia Giulia and Trentino Alto Adige regions. The Registry became member of Eurocat in 1985, and member of Clearinghouse in 1997.

**Size and coverage:**
Reports are obtained from 60 participating hospitals, with a total of approximately 57,000 annual births; the actual coverage is estimated at 73%.

**Legislation and funding:**
Reporting is voluntary. The Programme is partly run by privately funded research organisations and partly by Regional Health Authorities.

**Sources of ascertainment:**
Reports are obtained on specific forms from delivery units, induced abortion units, pediatric, cardiology, ophthalmology and pathology departments, regional induced abortion database and cytogenetic laboratories. 32 selected malformations are recorded within 7 days from birth (within 3 years of age for cardiovascular and ophthalmological anomalies only). In induced abortions all fetal anomalies are recorded. Two control infants are
selected for each malformed one.

**Exposure information:**
Detailed information on various exposures, including maternal or paternal occupation, diseases and drug use is obtained by interview of the mothers at the birth of the malformed nfants and controls. Only selected malformations are colected.

**Background information:**
Some epidemiological background data of all births are available. For each participating hospital the number of livebirths and stillbirths by sex and number of twin pairs are known.

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# Malta: MCAR

## Malta Congenital Anomalies Register

**History:**
The register started in 1985 as a research project of the University of Malta. It started as a hospital based register collecting data regarding congenital anomalies diagnosed in babies born at the main general hospital. It became a member of EUROCAT in 1986. Funding for the research project was stopped in 1995 and in 1997 the Department of Health Information assumed the functions of data collection increasing coverage to all hospitals on the islands making it a population based register. The Register was accepted as an associate member of the Clearinghouse in 2000.

**Size and coverage:**
The registry is population based and now covers 4,000 births per year.

**Legislation and funding:**
The registry is run and funded by the state Department of Health Information and Research. Reporting is not statutory.

**Sources of ascertainment:**
The registry employs active data collection from multiple sources including delivery and obstetric wards, doctors' reporting, cardiac lab records, genetics clinic records, National Mortality Register, National Obstetric Information Systems database, Hospital Activity Analysis databases, National Cancer Register and the Hypothyroid Screening Programme.

**Exposure information:**
Information regarding maternal exposure to medicinal drugs, smoking, alcohol and drug abuse as well as parental occupation are collected for
all malformed infants and fetuses.

**Background information:**
Epidemiological background data on all births are available from the National Obstetric Information Systems database and vital statistics**.**

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# Norway: MBRN

## Medical Birth Registry of Norway

**History:**
The Programme was started in 1967. The Programme was a founding member of the ICBDSR and is a full member.

**Size and coverage:**
The programme covers all births in Norway, approximately 60,000 annual births. Notification to MBRN is compulsory for births and pregnancy terminations after 12 weeks of gestation. Reporting to Clearinghouse includes:

- All live births
- Stillbirths from 20 weeks of gestation or birthweight 300 grams
- Pregnancy terminations from 12 weeks of gestation.

**Legislation and funding:**
The Programme is run and funded by the governmental Norwegian Institute of Public Health. Reporting is compulsory.

**Sources of ascertainment:**
The registry is based on the notification of births from the delivery units and since 1999 also from the neonatal units.

**Exposure information:**
Some basic information, such as maternal disease and since 1999, smoking and occupation, is collected on all infants, malformed or not.

**Background information:**
All information available for the reported malformed infants is also available for the total population of births.

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# Sweden

## The Swedish Birth Defects Register and the Medical Birth Registry

**History:**
The Swedish Registry of Congenital Malformations started in 1964 and changed name to The Swedish Birth Defects Register in 2007. The Swedish Medical Registry started in 1973. The programme was a founding member of the ICBDSR and contributed with data until 1994. The register has a new regime from 1999 and is since then again a full member of the ICBDSR.

**Size and coverage:**
All births in Sweden are included, approximately 100,000 – 120,000 annual births. The definition of a child is all children born alive and foetal deaths after 22 weeks gestation. In 1999 a special fetal surveillance system was started to include those fetuses with congenital anomalies who were terminated as a result of prenatal diagnosis.

**Legislation and funding:**
Reporting of birth defects in live- and stillborn infants is compulsory. Reporting of terminated pregnancies because of birth defects of the fetuses is, however, not compulsory. The registers are run by and funded by the National Board of Health and Social Welfare (Govermental).

**Sources of ascertainment:**
Reports are received from delivery units, paediatric clinics, pathology departments, child cardiology clinics, and cytogenetic laboratories.

**Exposure information:**
Some exposure information for all births is available in the Medical Birth Registry: maternal occupation, civic status, maternal smoking, drug use during pregnancy, contraceptive usage, and maternal diseases.

**Background information:**
Epidemiological background data are available on all birth in the Medical Birth Registry.

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Iowa Registry for Congenital and Inherited

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