# ANNUAL REPORT 2007 with data for 2005

INTERNATIONAL CLEARINGHOUSE FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH

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THE CENTRE OF THE INTERNATIONAL CLEARINGHOUSE FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH
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# INTERNATIONAL CLEARINGHOUSE FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH (ICBDSR)

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

# Annual Report

# 2007

with data for 2005

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# INTERNATIONAL CLEARINGHOUSE FOR BIRTH DEFECTS SURVEILLANCE AND RESEARCH
# ANNUAL REPORT 2007 (WITH DATA FOR 2005)

## Contents

# Collaborative Research Projects

## Multiple Congenital Anomalies 2005 Annual Report

*by Monica Rittler and Jorge Lopez-Camelo*

**Introduction**

For the year 2005, we received data from 8 programmes, for a total of 2302 reported cases, among 589,452 births (Table 1). Of these, 506 were reported as syndromes and 1061 had at least two major, unrelated congenital anomalies, which is our current case definition of multiple congenital anomaly (MCA). Coding was done by Monica Rittler, statistical analyses and report writing by Jorge Lopez-Camelo

**Main findings:**

This year, among the 47 defect groups, all except 3 (exstrophy of cloaca, sacrum anomalies, and gastroschisis) were associated with an O/E ratio greater than 1. Thirty four of them (72.3%) reached statistical significance at a $p<0.001$ level, and are shown in Table 2.

A significant excess, at a $p<0.001$ level, was found for 20 two-defects (Table 3) and for 7 three-defects combinations (Table 4).

For all comparisons, the data reported from 1992 through 2000, over 2,571,187 births were used as baseline.

*Table 1: Cases of reported congenital anomalies, by programme and number of defects (2005).*

| PROGRAMME | Births | Total cases Reported | Known etiology (syndromes) | < 2 major unrelated defects | 2 + defects | Rate |
|---|---|---|---|---|---|---|
| Canada: British Columbia | 40,974 | 581 | 76 | 361 | 144 | 35.1 |
| Finland | 57,927 | 336 | 155 | 35 | 146 | 25.2 |
| France: Central East/REMERA | 106,988 | 95 | 11 | 12 | 72 | 6.7 |
| Israel: IBDSP | 36,929 | 78 | 0 | 16 | 62 | 16.8 |
| Japan: JAOG | 72,229 | 517 | 175 | 187 | 155 | 21.5 |
| Mexico | 29,463 | 50 | 10 | 10 | 30 | 10.2 |
| South America | 192,882 | 609 | 62 | 113 | 434 | 22.5 |
| USA: Atlanta | 52,060 | 36 | 17 | 1 | 18 | 3.5 |
| **TOTAL** | **589,452** | **2,302** | **506** | **735** | **1,061** | **18.0** |

*Table 2: Association rates of defects, among cases with multiple congenital anomalies.*

| Defect group | Observed | Expected | Rate ratio | Excess | Poisson |
|---|---|---|---|---|---|
| Congenital heart defects | 441 | 174.57 | 2.53 | 266.43 | *** |
| Club foot/arthrogryposis | 159 | 75.74 | 2.10 | 83.26 | *** |
| Other CNS defects | 115 | 37.15 | 3.10 | 77.85 | *** |
| Anorectal atresia/stenosis | 134 | 56.37 | 2.38 | 77.63 | *** |
| Other urinary tract anomalies | 138 | 67.13 | 2.06 | 70.87 | *** |
| Cleft Palate | 105 | 38.59 | 2.72 | 66.41 | *** |
| Cleft lip with or without cleft palate | 111 | 54.08 | 2.05 | 56.92 | *** |
| Esophageal atresia/stenosis | 86 | 44.47 | 1.93 | 41.53 | *** |
| Hydrocephaly | 88 | 47.05 | 1.87 | 40.95 | *** |
| Axial skeleton anomalies | 85 | 47.05 | 1.81 | 37.95 | *** |
| Syndactyly | 80 | 43.61 | 1.83 | 36.39 | *** |
| Omphalocele | 60 | 25.96 | 2.31 | 34.04 | *** |
| Severe ear defects | 58 | 27.4 | 2.12 | 30.60 | *** |
| Genitalia defects (ambiguous and other) | 64 | 35.29 | 1.81 | 28.71 | *** |
| Kidney agenesis | 56 | 28.12 | 1.99 | 27.88 | *** |
| Other limb reduction defects | 44 | 17.07 | 2.58 | 26.93 | *** |
| Microcephaly | 49 | 22.81 | 2.15 | 26.19 | *** |
| Spleen anomalies | 32 | 6.31 | 5.07 | 25.69 | *** |
| Hypospadias | 52 | 26.97 | 1.93 | 25.03 | *** |
| Transverse limb reduction defects | 33 | 10.9 | 3.03 | 22.10 | *** |
| An/microphthalmia | 39 | 17.93 | 2.18 | 21.07 | *** |
| Situs inversus | 25 | 5.45 | 4.59 | 19.55 | *** |
| Other gut defects | 40 | 20.66 | 1.94 | 19.34 | *** |
| Dysplastic ears | 21 | 3.59 | 5.85 | 17.41 | *** |
| Lung defects | 37 | 21.66 | 1.71 | 15.34 | *** |
| Anencephaly | 23 | 8.61 | 2.67 | 14.39 | *** |
| Diaphragmatic hernia | 35 | 20.66 | 1.69 | 14.34 | *** |
| Polydactyly | 30 | 16.21 | 1.85 | 13.79 | *** |
| Other eye malformations | 30 | 16.35 | 1.83 | 13.65 | *** |
| Cystic kidneys | 32 | 19.22 | 1.66 | 12.78 | |
| Spina bifida aperta | 31 | 18.36 | 1.69 | 12.64 | |
| Preaxial limb reduction defects | 28 | 15.49 | 1.81 | 12.51 | *** |
| Duodenal atresia | 17 | 6.74 | 2.52 | 10.26 | *** |
| Holoprosencephaly | 15 | 6.31 | 2.38 | 8.69 | *** |
| Pterygium colli, cystic hygroma | 15 | 6.46 | 2.32 | 8.54 | *** |
| Vessel anomalies | 9 | 0.72 | 12.50 | 8.28 | *** |
| Encephalocele | 18 | 10.76 | 1.67 | 7.24 | |
| Other gut atresia/stenosis | 13 | 6.74 | 1.93 | 6.26 | |
| Malrotation of gut | 12 | 6.89 | 1.74 | 5.11 | |
| Choanal atresia/stenosis | 10 | 5.45 | 1.83 | 4.55 | |
| Craniostenosis | 8 | 5.59 | 1.43 | 2.41 | |
| Sirenomelia | 5 | 3.3 | 1.52 | 1.70 | |
| Tracheo-bronchial atresia/stenosis | 5 | 3.44 | 1.45 | 1.56 | |
| Other clefts and facial anomalies | 11 | 10.76 | 1.02 | 0.24 | |
| Exstrophy of cloaca | 6 | 6.02 | 1.00 | -0.02 | |
| Sacrum anomalies | 2 | 3.16 | 0.63 | -1.16 | |
| Gastroschisis | 9 | 10.61 | 0.85 | -1.61 | |

***= $p<0.001$

*Table 3: Two-defect combinations.*

| Defect combination | Observed | Expected | O/E | Poisson |
|---|---|---|---|---|
| Other CNS anomalies + Other limb reduction defects | 5 | 0.14 | 35.7 | ** |
| Anencephaly + Transverse limb reduction defects | 3 | 0.14 | 21.4 | *** |
| Other CNS anomalies + Transverse limb reduction defects | 4 | 0.29 | 13.8 | ** |
| Omphalocele + Transverse limb reduction defects | 4 | 0.29 | 13.8 | ** |
| Congenital heart defects + Situs inversus | 15 | 1.29 | 11.6 | ** |
| Choanal atresia/stenosis + Other gut defects | 4 | 0.43 | 9.3 | ** |
| Dysplastic ears + Congenital heart defects | 9 | 1.29 | 7.0 | *** |
| Cleft palate + Kidney agenesis | 6 | 0.86 | 7.0 | *** |
| Axial skeleton anomalies + situs inversus | 6 | 0.86 | 7.0 | *** |
| Congenital heart defects + Spleen anomalies | 27 | 4.16 | 6.5 | *** |
| Cleft lip with or without cleft palate + Other limb reduction defects | 11 | 1.86 | 5.9 | *** |
| Other CNS anomalies + Lung defects | 7 | 1.29 | 5.4 | *** |
| Choanal atresia/stenosis + Club foot/arthrogryposis | 3 | 0.57 | 5.3 | * |
| Other CNS + Club foot/arthrogryposis | 19 | 5.31 | 3.6 | *** |
| Severe ear defects + Club foot/arthrogryposis | 15 | 4.45 | 3.4 | *** |
| Microcephaly + Congenital heart defects | 18 | 5.88 | 3.1 | *** |
| Other CNS anomalies + Congenital heart defects | 42 | 13.63 | 3.1 | *** |
| Cleft palate + Congenital heart defects | 31 | 10.47 | 3.0 | *** |
| Anorectal atresia/stenosis + Congenital heart defects | 37 | 12.62 | 2.9 | *** |
| Congenital heart defects + Polydactyly | 23 | 8.32 | 2.8 | *** |
| Anorectal atresia/stenosis + Axial skeleton anomalies | 23 | 9.18 | 2.5 | *** |
| Cleft palate + Club foot/arthrogryposis | 21 | 8.89 | 2.4 | *** |
| Esophageal atresia/stenosis + anorectal atresia/stenosis | 28 | 11.48 | 2.4 | *** |
| Other gut defects + Congenital heart defects | 23 | 9.75 | 2.4 | *** |
| Cleft lip with or without cleft palate + Congenital heart defects | 33 | 14.06 | 2.3 | *** |
| Congenital heart defects + Club foot/arthrogryposis | 33 | 15.78 | 2.1 | *** |

***=p<0.001; **=p<0.001; *=p<0.005

*Table 4: Three-defects combinations.*

| Defect combination | Observed | Expected | O/E | Poisson |
|---|---|---|---|---|
| | | | | |
| Anorectal atresia/stenosis + Other gut defects + Axial skeleton anomalies | 4 | 0.14 | 28.6 | *** |
| Encephalocele + Cleft lip with or without cleft palate + Club foot/arthrogryposis | 3 | 0.14 | 21.4 | *** |
| Dysplastic ears + Anorectal atresia/stenosis + Congenital heart defects | 3 | 0.14 | 21.4 | *** |
| Congenital heart defects + Axial skeleton anomalies + Other limb reduction defects | 3 | 0.14 | 21.4 | *** |
| Anorectal atresia/stenosis + Congenital heart defects + Axial skeleton anomalies | 14 | 2.01 | 7.0 | *** |
| Other CNS defects + Lung defects + Club foot/arthrogryposis | 3 | 0.00 | .*** | *** |
| Anorectal atresia/stenosis + Other gut defects + Cystic kidney | 3 | 0.00 | .*** | *** |

The high number of defects and combinations with a significant excess might be due to an inadequate baseline which should be adjusted for future monitoring.

## Prenatal Diagnosis and Down Syndrome, 2005

*Guido Cocchi (Italy: IMER)*
*Silvia Gualdi (Italy: IMER)*

**Introduction**

Aim of the survey was to assess in time and in the Programme the variability in the use and the spread of prenatal diagnostic techniques and to analyse the impact of elective termination on prevalence rates at birth of Down Syndrome (DS), in Countries where elective abortions are legally performed.

Participation in the Clearinghouse programmes worldwide provides a unique opportunity to analyse international variations in the use of prenatal diagnosis (Chorion Villus Sampling = CVS, Amniocentesis = AC, Cordocentesis= CC), and access to screening, as well as differences in advice and abortion legislation. In addition, repeating this study over time has made it possible to follow the evolution of these techniques and to evaluate the impact of each practice on the prevalence of DS.

**2005 Data**

During 2005, 21 programmes (Cuba joined the survey for the first time) provided data on 3055 DS cases, 1601 of them (52.4 %) were prenatally diagnosed and terminated (Table 1).

The total number of births under surveillance in the 21 programmes was 1,745,248.

The percentage of terminations of pregnancy (ToP) ranged from the lowest values in USA: Utah (4.2%), Russia: Moscow Region (7.1%), and USA: Atlanta (16.3%), to the highest in the registries of French and Italian regions: France: Paris(80.1%), France: REMERA(79.9%) and France: Strasbourg(74.2%). For the Italian registries the percentages of terminations range from the highest of Italy: BDRCam(75.7%) to the lowest of Italy:North East (47.8%) with a medium value of 61.8%. Other Registries show percentages of terminations over 60%: Czech Republic (73.8%), Germany: Saxony-Anhalt (73.7%) and Australia: VBDR (61.2%).

In the European registries that provided a data set of 13 years (1993-2005), a regular increase in the percentage of ToP has been observed, passing from the lowest values of the first three years 41.5% in 1993, 45.9% in 1994, 48.5% in 1995, to the values of the last three years : 68.9% in 2003, 69.7% in 2004 and 65.4% in 2005 .

The comparison of the rate of ToPs in 2005, between European Countries and extra-European Countries (i.e. Australia:VBDR, Canada: Alberta, Cuba, USA: Atlanta and USA: Utah ) is significantly different (59.2% vs 32.3% , $^{-}2 = 133.4$ $p< 0.0001$).

Terminations are related to the maternal age as shown in Table 2: the percentage of ToPs is lower in the lowest maternal age class (≤29 years). On the contrary in the higher maternal age classes: i.e. over 35 years (38-39 and ? 40) the percentage of terminations is higher.

The percentage of mothers aged over 35 years (Table3) , has increased year by year and in many registries is over 20%: Sweden: 20.6%; Australia:Victoria: 23.5%; all the three Italian Registries: BDRCam: 22.2%, IMER: 26.4% and Tuscany: 27.4%; while the Register of Paris shows the highest values: 29.67%.

The greater percentages of termination are typically detected in the registries that show the higher values of higher aged mothers. In fact overall, the proportion of DS pregnancies which were terminated among women at higher risk (?35 years old), was over 80% in three Registries: France: REMERA and France: Paris (96% and 85.1% respectively); and in Czech Republic the percentage of terminations is 87.5% . Percentages of ToPs less than 40% were observed in many registries: Russia: Moscow region (7.6%); the two registries of United States of America: Utah and Atlanta (10.78% and 22.2 respectively), Italy: BDRCam (26.7% ), Canada: Alberta(28.1%), and Northern Netherlands(33.3%).

The most common technique of prenatal diagnosis remained AC in 2005 (Table 4), with a mean value of 64.7%. CVS, with a mean value of 35%, shows a progressive increase year by year: 18.3% in 1995, 19.0% in 1996, 19.3% in 1997, 18.2% in 1998, 20.2% in 1999, 21.8% in 2000, 22.9% in 2001, 28.6 in 2002 and in 2003, 30% in 2004.

In Australia: VBDR CVS is the first technique of prenatal detection used with a rate of 63.5%. CVS has been used mainly in Finland (55%) and in Northern Netherlands (83.3%) and in Italy: IMER (51.2%). England and Wales and France: Strasbourg show a percentage slightly under 50%: 48.9% and 47.8% respectively. In the Registries of France the mean percentage is 41.1% vs 23.6% of 2004 and 16% of 2003, while the mean value in Italy has increased in the last few years passing from 9.4% in 2003, to 16% in 2004 till 38.1% this year (Table 4).

The programmes, where CVS is most frequently used, show –as expected- the lowest mean gestational ages at pregnancy termination in the older maternal age group (>35) as in Australia: VBDR (14.8±2.3 wks) in Finland (15.7±2.2 wks) and in Northern Netherlands and in Italy: IMER (16±3.2

and 14.2±2.7 wks respectively) (Table 5). The mean age (wks) of terminations after CVS diagnosis is heterogeneous and significantly different in the programs in both maternal age groups. In the younger group (<=34 years) there is a lower limit of around 12 and 13 wks in many Registries such as in Northern Netherlands and in Wales (12wks), in Italy: Tuscany (12.5wks) and in Russia: Moscow Region, in Australia: VBDR and in Italy: IMER (13, 13.3 and 13.4 wks respectively) to an upper limit of 22.3 wks in USA: Atlanta. (Table 5).

The prevalence at birth of DS has decreased in the majority of the 14 programmes that can provide the rates for all the 13 year period.
A significant negative temporal trend was observed above all in the registries that showed, as expected, an increase in the termination of pregnancies: the Czech Republic, all the three registries of France (REMERA, Paris and Strasbourg), all the four Italian registries (BDRCam, IMER, North East and Tuscany) and Germany:Saxony-Anhalt (Table 6). These programmes are the same ones that showed the highest rates of ToPs and an increase in the terminations year by year.
Likewise, the highest rates of prevalence at birth were observed in the Programmes where ToPs were lowest i.e Canada: Alberta (20.5 x 10,000) and the two registries of USA: Atlanta(12.86) and USA: Utah(13.19). The Canada: Alberta register has shown a significant increase over the years (regression analysis $r = +0.799$ $p<0.001$) till the rate of 20.5 this year. Controversial data are shown by the Finnish register where despite quite high rates of ToP (54.7% see Table 2) it has shown a high rate of prevalence at birth i.e 11.73 per 10,000.

*Table 1. List of the programs participating in the Prenatal Diagnosis Study in the years.*

| | 1993 | 1994 | 1995 | 1996 | 1997 | 1998 | 1999 | 2000 | 2001 | 2002 | 2003 | 2004 | 2005 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| AUSTRALIA | X | X | X | X | X | X | | | | X | X | X | X |
| CANADA: Alberta | | | | | X | X | X | X | X | X | X | X | X |
| CZECH REPUBLIC | X | X | X | X | X | X | X | X | X | X | X | X | X |
| CUBA | | | | | | | | | | | | | X |
| ENGLAND & WALES | X | X | X | X | X | X | X | X | X | X | | X | X |
| FINLAND | X | X | X | X | X | X | X | X | | X | X | X | X |
| FRANCE: CE/REMERA | X | X | X | X | X | X | X | X | X | X | X | X | X |
| FRANCE: Paris | X | X | X | X | X | X | X | X | X | X | X | X | X |
| FRANCE: Strasbourg | X | X | X | X | X | X | X | X | X | X | X | X | X |
| GERMANY: Saxony -Anhalt | | | | | | | | X | X | X | X | X | X |
| ISRAEL: IBDSP | X | X | X | X | X | X | X | X | X | X | X | X | X |
| ITALY: BDRCam | X | X | X | X | X | X | X | X | X | X | X | X | X |
| ITALY: IMER | X | X | X | X | X | X | X | X | X | X | X | X | X |
| ITALY: North-East | X | X | X | X | X | X | X | X | X | X | | X | X |
| ITALY: Tuscany | X | X | X | X | X | X | X | X | X | X | X | X | X |
| RUSSIA: Moscow Region | | | | | | | | | | | X | X | X |
| NORTHERN NETHERLAND | X | X | X | X | X | X | X | X | X | X | X | X | X |
| SLOVAK | | | | | | | | | | | | X | |
| SWEDEN | | | | | | | | X | X | X | X | X | X |
| USA: Atlanta | X | X | X | X | X | X | X | X | X | X | X | X | X |
| USA: UTAH | | | | | | | | | | | | X | X |
| WALES | | | | | | | | | | | | X | X |

*Table 2. Percentage (%) of terminations (TOP) among the total number of cases recorded in 2005.*

| Monitoring Program | Maternal Age (years) | | | | | |
|---|---|---|---|---|---|---|
| | <= 29 | 30 – 34 | 35 – 37 | 38 – 39 | >= 40 | Total |
| Australia: VDBR | 29.4 | 54.8 | 58.5 | 80.8 | 56.7 | 61.6 |
| Canada: Alberta | 13.8 | 20.0 | 26.3 | 25.0 | 31.8 | 22.5 |
| Czech Republic | 57.8 | 76.0 | 81.6 | 94.7 | 80.0 | 73.8 |
| Cuba | 7.3 | 3.4 | 16.7 | 76.9 | 47.8 | 24.0 |
| England and Wales | 28.9 | 49.7 | 53.3 | 53.3 | 60.8 | 48.8 |
| Finland | 40.7 | 50.0 | 55.2 | 52.9 | 68.3 | 54.7 |
| France: REMERA | 90.0 | 92.5 | 83.8 | 95.2 | 96.3 | 93.5 |
| France: Paris | 63.2 | 73.2 | 90.0 | 91.3 | 79.3 | 80.1 |
| France: Strasbourg | 60.0 | 77.8 | 75.0 | 100.0 | 33.3 | 74.2 |
| Germany: Saxony-Anhalt | 50.0 | 100.0 | 50.0 | 100.0 | 100.0 | 73.7 |
| Israel: IBDSP | 11.1 | 55.6 | 55.6 | 0.0 | 33.3 | 37.8 |
| Italy: BDRCam | 40.0 | 81.8 | 83.3 | 69.2 | 85.7 | 75.7 |
| Italy: IMER | 28.6 | 62.5 | 58.8 | 58.3 | 63.2 | 59.2 |
| Italy: North-East | 60.0 | 42.9 | 58.3 | 42.9 | 77.8 | 47.8 |
| Italy: Tuscany | 33.3 | 60.0 | 66.7 | 85.7 | 81.8 | 69.2 |
| Russia: Moscow Region | 6.3 | 7.7 | 7.7 | 0.0 | 13.3 | 7.2 |
| Northern Netherlands | 14.3 | 11.1 | 25.0 | 100.0 | 33.3 | 21.4 |
| Sweden | 19.5 | 37.5 | 63.6 | 63.6 | 77.9 | 54.3 |
| USA: Atlanta | 6.7 | 10.0 | 11.8 | 33.3 | 25.0 | 16.3 |
| USA: Utah | 0.0 | 0.0 | 12.5 | 20.0 | 0.0 | 4.2 |
| Wales | 23.5 | 50.0 | 35.3 | 66.7 | 57.1 | 44.0 |

*Table 3. Percentage of mothers aged 35 and over in the monitoring programs participating in the study and percentage of terminations (ToP) in the same group of mothers.Prevalence rate in live and stillbirths (per 10,000) and comparison with the rate after inclusion of ToP.*

| Monitoring Program | % of mothers | % of ToP in mothers | Prevalence rate (* 10,000) | |
|---|---|---|---|---|
| | aged >=35 | aged >=35 | L+S | L+S+ToP |
| Australia:VDBR | 23.5 | 63.3 | 26.0 | 70.8 |
| Canada Alberta | 15.2 | 28.1 | 64.6 | 89.8 |
| Czech Republic | 8.7 | 87.5 | 12.3 | 98.6 |
| Cuba | - | 44.4 | - | - |
| England and Wales | 19.6 | 56.2 | 17.7 | 40.4 |
| Finland | 19.1 | 58.6 | 30.7 | 78.6 |
| France: REMERA | 19.2 | 96.0 | 2.4 | 62.2 |
| France: Paris | 29.3 | 85.1 | 4.6 | 30.8 |
| France: Strasbourg | 16.3 | 76.5 | 17.9 | 75.9 |
| Germany: Saxony-Anhalt | 12.3 | 76.9 | 14.2 | 61.6 |
| Israel IBDMS | 16.4 | 42.1 | 18.2 | 31.4 |
| Italy: BDRCam | 22.2 | 26.7 | 6.8 | 34.0 |
| Italy: IMER | 27.4 | 60.4 | 18.4 | 46.5 |
| Italy: North-East | - | 61.4 | - | - |
| Italy: Tuscany | 29.6 | 76.7 | 8.0 | 34.4 |
| Russia: Moscow Region | 7.6 | 7.6 | 83.0 | 89.9 |
| Northern Netherlands | 19.4 | 33.3 | 22.2 | 33.3 |
| Sweden | 20.6 | 70.3 | 23.5 | 79.0 |
| USA Atlanta | 16.7 | 22.2 | 40.4 | 51.9 |
| USA:Utah | 9.1 | 10.7 | 53.4 | 59.8 |
| Wales | - | 51.8 | - | - |

** estimated*

*Table 4 . Down Syndrome techniques of prenatal diagnosis (number of cases) registered in 2005 grouped in maternal age classes.*

| Monitoring Program | <35 | | | | 35-39 | | | | >39 | | | | Tot* | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | CVS | AC | CC | UK | CVS | AC | CC | UK | CVS | AC | CC | UK | CVS | AC | CC | UK |
| Australia: VBDR | 14 | 8 | 0 | 5 | 25 | 20 | 0 | 6 | 13 | 3 | 0 | 1 | 54 | 31 | 0 | 27 |
| Canada: Alberta | 0 | 8 | 0 | 1 | 0 | 9 | 0 | 0 | 3 | 4 | 0 | 0 | 3 | 21 | 0 | 1 |
| Czech Republic | 9 | 66 | 0 | 0 | 9 | 40 | 0 | 0 | 5 | 23 | 0 | 0 | 23 | 129 | 0 | 0 |
| Cuba | 0 | 4 | 0 | 0 | 0 | 13 | 0 | 0 | 0 | 11 | 0 | 0 | 0 | 28 | 0 | 3 |
| England and Wales | 49 | 67 | 0 | 0 | 76 | 72 | 0 | 0 | 56 | 50 | 0 | 0 | 181 | 189 | 0 | 0 |
| Finland | 19 | 8 | 0 | 0 | 12 | 13 | 0 | 0 | 13 | 15 | 0 | 0 | 44 | 36 | 0 | 0 |
| France: REMERA | 16 | 52 | 0 | 8 | 12 | 49 | 0 | 10 | 11 | 36 | 0 | 5 | 39 | 137 | 0 | 23 |
| France : Paris | 12 | 30 | 0 | 0 | 20 | 37 | 0 | 0 | 18 | 27 | 1 | 1 | 50 | 94 | 1 | 1 |
| France: Strasbourg | 3 | 7 | 0 | 0 | 7 | 5 | 0 | 0 | 1 | 0 | 0 | 0 | 11 | 12 | 0 | 0 |
| Germany: Saxony-Anhalt | 0 | 3 | 0 | 1 | 0 | 5 | 0 | 1 | 0 | 4 | 0 | 0 | 0 | 12 | 0 | 2 |
| Israel: IBDSP | 0 | 6 | 0 | 0 | 0 | 5 | 0 | 0 | 0 | 3 | 0 | 0 | 0 | 14 | 0 | 0 |
| Italy: BDRCam | 1 | 10 | 0 | 0 | 5 | 19 | 0 | 0 | 0 | 12 | 0 | 0 | 6 | 47 | 0 | 0 |
| Italy: IMER | 5 | 6 | 0 | 0 | 8 | 9 | 0 | 0 | 8 | 4 | 0 | 0 | 21 | 20 | 0 | 0 |
| Italy: North-East | 2 | 2 | 0 | 2 | 4 | 5 | 0 | 4 | 4 | 8 | 0 | 2 | 10 | 15 | 0 | 8 |
| Italy: Tuscany | 2 | 2 | 0 | 0 | 1 | 13 | 0 | 0 | 0 | 8 | 0 | 0 | 3 | 23 | 0 | 0 |
| Russia: Moscow region | 1 | 0 | 1 | 0 | 0 | 0 | 1 | 0 | 2 | 0 | 1 | 0 | 3 | 0 | 3 | 0 |
| Northern Netherlands | 2 | 0 | 0 | 0 | 2 | 1 | 0 | 0 | 1 | 0 | 0 | 0 | 5 | 1 | 0 | 0 |
| USA : Atlanta | 0 | 3 | 0 | 0 | 1 | 4 | 0 | 0 | 0 | 5 | 0 | 0 | 1 | 12 | 0 | 0 |
| USA: Utah | 0 | 0 | 0 | 0 | 0 | 3 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 0 | 0 |
| Wales | 2 | 8 | 0 | 1 | 1 | 12 | 0 | 1 | 3 | 5 | 0 | 0 | 6 | 25 | 0 | 2 |
| **Total** | **137** | **290** | **1** | **10** | **183** | **334** | **1** | **22** | **138** | **218** | **2** | **9** | **460** | **849** | **4** | **67** |

CVS = Chorion Villus sampling
AC = Amniocentesis
CC = Chordocentesis
UK = Unknown
* maternal age unknown

*Table 5. Mean gestational age (weeks) and Standard Deviation of induced abortions by maternal age group and by type of prenatal diagnosis.*

| Monitoring Program | <=34 | | | >=35 | | |
|---|---|---|---|---|---|---|
| | CVS | AC | Total | CVS | AC | Total |
| Australia: VBDR | 13.29±0.99 | 17.13±1.73 | 14.68±2.28 | 13.37±1.42 | 17.09±1.20 | 14.84±2.26 |
| Canada: Alberta | - | 17.38±1.30 | 17.38±1.30 | 14.00±1.00 | 17.85±0.99 | 17.13±1.82 |
| Czech Republic | 14.22±2.22 | 19.67±2.11 | 19.01±2.76 | 14.50±2.03 | 19.29±2.01 | 18.42±2.73 |
| Finland | 14.79±1.08 | 18.13±0.99 | 15.78±1.87 | 14.00±1.61 | 17.29±1.21 | 15.74±2.17 |
| France: REMERA | 14.19±1.68 | 19.35±2.86 | 18.13±3.42 | 14.74±1.29 | 20.08±4.38 | 18.94±4.49 |
| France: Paris | 14.08±1.68 | 21.33±4.94 | 19.26±5.38 | 14.08±1.08 | 19.47±3.14 | 17.45±3.63 |
| France: Strasbourg | 17.33±2.31 | 20.86±1.86 | 19.80±2.53 | 16.00±2.14 | 21.80±3.27 | 18.23±3.85 |
| Germany: Saxony-Anhalt | - | 19.25±3.20 | 19.25±3.20 | - | 19.50±2.01 | 19.50±2.01 |
| Israel IBDSP | - | 22.00±1.26 | 22.00±1.26 | - | 24.63±3.78 | 24.63±3.78 |
| Italy: BDRCam | 16.00±0.00 | 21.50±1.41 | 20.89±2.26 | 16.20±2.17 | 20.77±2.14 | 20.04±2.62 |
| Italy: IMER | 13.40±0.89 | 20.20±1.30 | 16.80±3.74 | 14.06±1.18 | 19.00±0.74 | 16.18±2.68 |
| Italy: North-East | 15.00±0.00 | 18.50±2.12 | 16.75±2.36 | 14.88±1.46 | 19.54±1.27 | 17.76±2.66 |
| Italy: Tuscany | 12.50±0.71 | 18.50±0.71 | 15.50±3.51 | 12.00±0.00 | 17.53±1.39 | 17.25±1.83 |
| Russia: Moscow Region | 13.00±0.00 | 24.00±0.00 | 18.50±7.78 | 12.00±0.00 | 24.00±0.00 | 18.00±6.93 |
| Northern Netherlands | 12.00±0.00 | - | 12.00±0.00 | 14.67±2.08 | 20.00±0.00 | 16.00±3.16 |
| USA: Atlanta | 22.33±1.15 | - | 22.33±1.15 | - | 19.50±2.74 | 19.50±2.74 |
| USA: Utah | - | - | - | - | 18.33±2.08 | 18.33±2.08 |
| Wales | 12.00±0.00 | 21.38±6.21 | 19.50±6.75 | 13.00±0.00 | 18.18±1.88 | 17.19±2.68 |

*Table 6. Prevalence at birth (x 10,000) in the years of DS in the programs participating in the survey.*

| Programme | 1993 | 1994 | 1995 | 1996 | 1997 | 1998 | 1999 | 2000 | 2001 | 2002 | 2003 | 2004 | 2005 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Australia: VBDR | | | | | | | | | | 9.98 | 8.49 | 11.03 | 10.85 |
| Canada: Alberta | 11.45 | 11.07 | 13.15 | 8.49 | 11.14 | 14.02 | 11.56 | 14.65 | 15.2 | 12.71 | 19.2 | 16.52 | 20.54 |
| Czech Republic | 7.52 | 7.67 | 7.26 | 5.51 | 5.06 | 6.72 | 6.57 | 5.37 | 5.51 | 5.37 | 6.38 | 5.51 | 5.26 |
| Cuba | | | | | | | | | | | | | 8.31 |
| England and Wales | 4.59 | 4.73 | 4.91 | 5.50 | 6.39 | 7.18 | 6.71 | 6.60 | 6.27 | 5.9 | | 7.06 | 6.94 |
| Finland | 13.21 | 12.83 | 12.94 | 10.33 | 10.07 | 11.33 | 10.04 | 11.76 | 14.18 | 14.16 | 12.32 | 12.25 | 11.73 |
| France: CE/REMERA | 10.98 | 10.43 | 8.91 | 9.47 | 9.01 | 6.83 | 4.86 | 5.83 | 5.85 | 5.51 | 4.86 | 5.86 | 4.77 |
| France: Paris | 10.61 | 9.19 | 7.05 | 9.67 | 7.78 | 10.48 | 5.24 | 7.87 | 7.79 | 6.20 | 4.69 | 5.31 | 9.15 |
| France: Strasbourg | 16.75 | 17.87 | 24.04 | 17.44 | 27.95 | 2.20 | 4.34 | 5.62 | 2.23 | 2.96 | 5.18 | 8.2 | 5.81 |
| Germany: Saxony-Anhalt | 5.79 | 6.33 | 7.43 | 7.86 | 8.33 | 13.65 | 6.09 | 6.38 | 8.26 | 9.08 | 5.30 | 9.18 | 2.90 |
| Israel IBDSP | 5.06 | 5.03 | 6.32 | 4.87 | 9.13 | 3.28 | 6.01 | 4.74 | 6.15 | 4.75 | 6.45 | 6.66 | 6.22 |
| Italy: BDRCam | 10.94 | 7.63 | 10.01 | 9.22 | 6.74 | 8.73 | 6.33 | 2.99 | 6.83 | 5.42 | 5.17 | 4.76 | 2.85 |
| Italy: IMER | 8.97 | 9.27 | 10.24 | 7.97 | 7.27 | 9.36 | 9.58 | 6.47 | 6.33 | 6.15 | 8.11 | 5.72 | 8.24 |
| Italy: North East | 12.87 | 10.31 | 11.46 | 9.14 | 7.15 | 7.23 | 7.17 | 6.90 | 7.83 | 9.04 | | 6.93 | 6.41 |
| Italy: Tuscany | 11.83 | 9.80 | 11.42 | 6.91 | 7.34 | 6.28 | 6.14 | 4.90 | 5.70 | 3.76 | 4.00 | 4.14 | 4.08 |
| Russia: Moscow Region | | | | | | | | | | | 11.27 | 10.66 | 13.64 |
| Northern Netherlands | 9.86 | 5.74 | 9.38 | 13.74 | 11.91 | 10.03 | 8.43 | 6.35 | 9.32 | 13.31 | 5.99 | 9.4 | 11.87 |
| Sweden | | | | | | | 14.01 | 11.01 | 14.59 | 13.31 | 15.,47 | 10.56 | 12.69 |
| USA: Atlanta | 12.02 | 13.81 | 10.93 | 11.98 | 10.49 | 11.46 | 12.00 | 11.08 | 13.25 | 5.66 | 13.01 | 12.98 | 12.86 |
| USA: Utah | | | | | | | | | | | | 14.01 | 13.19 |
| Wales | | | | | | | | | | | | 11.69 | 13.12 |

***Alessandra Lisi*** was a researcher statistician at the ICBDSR Centre in the years 2002 – 2006. Over the years Alessandra's working skill, ethic, grace and kindness made her an increasingly central part of the ICBDSR Centre. Nothing was done at the Centre without her valid help.
She was the only victim of an Underground accident that occurred in Rome on October 17, 2006.
We mourn her loss, miss her beyond words and we want remember her with a Prize for a young researcher involved in the field of birth defects and working in one of the ICBDSR Member Registries.

**Aim**
To recognise a high quality recently published, original peer-reviewed article written in English by a junior researcher and based on research conducted using data from a Clearinghouse Program.

**Eligibility**
Prize is be open to all junior researchers who are no more than two-years post-doctoral level, including those without post-graduate qualifications and will be based on research using data from a Clearinghouse Program.

**The prize**
One prize of $500, a plaque/ certificate and the summary published in the Annual Report. The winner will be invited to give a presentation at the Annual and Scientific Meeting of the Clearinghouse, in order to present the work on which the Award was based and also to present the work they are doing at present.

In 2007 there were two winners because of an exceptional situation with two very good papers.

**Further information about the Prize application (award criteria, application process, deadline) can be requested to centre@icbdsr.org**

## Winners of 2007 Award

The Award of the 2007 "Alessandra Lisi Memorial Prize", given by the International Clearinghouse for Birth Defects Monitoring Systems (ICBDSR) to a junior researcher from a Monitoring Program of the ICBDSR, goes to **Natasha Nassar**, as Author of the high quality, original peer-reviewed article: "*Increasing Prevalence of Hypospadias in Western Australia, 1980-2000*", published in the *Archives of Disease in Childhood, Vol 92, Issue 7, pages 580-584, 2007*.

Increasing prevalence of hypospadias in Western Australia, 1980-2000.
Arch Dis Child. 2007 Jul;92(7):580-4
Nassar N, Bower C, Barker A.
Telethon Institute for Child Health Research, Centre for Child Health Research, The University of Western Australia, Perth, Australia. natashan@ichr.uwa.edu.au

OBJECTIVES: Hypospadias, a common birth defect, has shown widespread variation in reported rates and temporal trends across countries over the last 30 years. The aim of this study was to determine the prevalence and trends of hypospadias in an Australian population.
DESIGN: Population-based study of all male infants born in Western Australia (WA) between 1980 and 2000 diagnosed with hypospadias and notified to the WA Birth Defects Registry.
MAIN OUTCOME MEASURES: Prevalence of hypospadias, birth outcome and association with other congenital anomalies, stratified by degree-of-severity.
RESULTS: 1788 cases of hypospadias were registered in WA in 1980-2000 with an overall prevalence of 34.8 (95% confidence interval (CI): 33.2 to 36.4) cases per 10 000 births. The prevalence increased by 2.0% per annum (95% CI: 1.2% to 2.8%) from 27.9 in 1980 to 43.2 per 10 000 births in 2000 ($p<0.001$). Hypospadias was mild in 84% of cases, moderate-severe in 11% and unspecified in 5%, with the number of moderate-severe hypospadias almost doubling over time ($p<0.01$). There were 1465 (82%) cases of isolated hypospadias and 323 (18%) had co-existing anomalies. Infants with co-existing genital (relative risk (RR) 4.5; 95% CI: 3.3 to 6.1) or non-genital (RR 1.5; 95% CI: 1.0 to 2.2) anomalies were more likely to have moderate-severe hypospadias compared with isolated cases.
CONCLUSION: Hypospadias affects one in 231 births and has been reported to have increased significantly over the last 20 years. Future investigation of the aetiology of hypospadias is important to identify potentially modifiable risk factors and ensure optimal male reproductive health in the future.

The Award of the 2007 "Alessandra Lisi Memorial Prize", given by the International Clearinghouse for Birth Defects Monitoring Systems (ICBDSR) to a junior researcher from a Monitoring Program of the ICBDSR, goes to **Fernando A. Poletta**, as Author of the high quality, original peer-reviewed artiche: "*Regional Analysis on the Occurrence of Oral Clefts in South America*",published in the *American Journal of Medical Genetics, Vol 143A, Issue 24, pages 3216-3217, 2007*.

Regional analysis on the occurrence of oral clefts in South America.
Am J Med Genet A. 2007 Dec 15;143A(24):3216-27
Poletta FA, Castilla EE, Orioli IM, Lopez-Camelo JS.
ECLAMC (Latin American Collaborative Study of Congenital Malformations) at CEMIC (Center for Medical Education and Clinical Research), Buenos Aires, Argentina.

The aim of this work was to search for unequal birth prevalence rates (BPRs) of cleft lip +/- cleft palate (CL/P), and cleft palate only (CPO), among different geographic areas in South America, and to analyze phenotypic characteristics and associated risk factors in each identified cluster. Included were 5,128 CL/P cases, 1,745 CPO cases, and 3,712 controls (like-sexed, non-malformed liveborn infant, born immediately after a malformed one, in the same hospital), over 4,199,630 consecutive births.
They were ascertained between 1967 and 2004, in 190 maternity hospitals of the ECLAMC (Estudio Colaborativo Latinoamericano de Malformaciones Congénitas) network, in 102 cities of all 10 South American countries.

Non-predefined geographical areas with significantly unusual cleft BPRs were identified with Kulldorf and Nagarwalla's spatial scan statistic, employing number of cases and births, and exact location of each hospital. Expected values were cleft BPRs registered for the entire ECLAMC hospital network. Syndromic and non-syndromic clefts were considered for cluster analysis, and phenotypic characterization, while only non-syndromic for risk factor analysis. Seven clusters for CL/P, and four for CPO, with unusual BPRs were identified. CL/P cases in high BPR areas were more severe than elsewhere in the sample, similar to a previous ECLAMC report on microtia. For CL/P, high BPR clusters were associated with high altitude above sea level, Amerindian ancestry, and low socioeconomic strata; low BPR clusters showed association with African Black ancestry. Advanced maternal age, a recognized risk factor for CPO, was also associated with the only identified geographic cluster for CPO.

| Monitoring Program | Coverage | Year Joined ICBDMS | Maximum age at diagnosis | Criteria defining stillbirths | Termination of Pregnancy (ToP) |
|---|---|---|---|---|---|
| Australia: National | Population-based National | 1981 | 1 year | 20 weeks or 400 grams | Permitted<br>Reported |
| Australia: VBDR | Population-based Statewide | 2002 | Up to 15 years | 20 weeks or 400 grams | Permitted<br>Reported |
| Australia: WABDR | Population-based, Statewide | 2002 | Up to 6 years | 20 weeks or 400 grams | Permitted<br>Reported |
| Canada: Alberta-ACASS | Population-based Provincial | 1996 | 1 year | 20 weeks or 500 grams | Permitted<br>Reported |
| Canada British Columbia | Population-based Provincial | 2001 | No limit | At least 20 weeks or 500 grams | Permitted,<br>Not reported |
| Canada: CCASNI | Population-based National | 1996 | 1 year | 20 weeks or 500 grams | Permitted<br>Not reported |
| Chile-Maule: RRMC-SSM | Hospital-based Regional | 2003 | Hospital discharge | 500 grams | Not permitted<br>Not reported |
| China: BDSS.Beijing | Population-based Four Provinces | 1997 | 6 weeks | 20 weeks | Permitted<br>Not reported |
| China: CBDMN | Hospital-based | 1985 | 7 days | 28 weeks | Permitted<br>Not reported |
| Costa Rica: CREC | Population-based National | 2003 | 3 days | 22 weeks or 500 grams | Not permitted |
| Cuba: RECUMAC | Hospital based National | 2003 | Hospital discharge | 500 grams | Permitted<br>Reported |
| Czech Republic | Population-based National | 1974 | Up to 15 years | non-viable fetuses, 28 weeks or >1000 grams | Permitted<br>Reported |
| England and Wales | Population-based National | 1974 | 1995 onwards no limit | 24 weeks | Permitted<br>Reported only for a few selected malformations |
| Finland | Population-based National | 1974 | 1 year | 22 weeks or 500 grams | Permitted<br>Reported |
| France-Rhône Alpes: REMERA | Population-based Regional | 1974 | 1 year | 22 weeks | Permitted<br>Reported |
| France: Paris | Population-based Regional | 1982 | Hospital discharge | 22 weeks | Permitted<br>Reported |
| France: Strasbourg | Population-based Regional | 1982 | 2 years | 22 weeks or 500 grams | Permitted<br>Reported |
| Germany: Saxony-Anhalt | Population-based (Federal State) | 2001 | Hospital discharge (almost first week of life) – up to 1 year | >/= 500 grams | Permitted<br>Reported |
| Hungary | Population-based National | 1974 | 1 year | 24 weeks or 500 grams | Permittted<br>Reported |
| Iran: TROCA | Hospital-based Regional | 2006 | 1 year | 20 weeks or 400 grams | Permitted<br>Reported only for a few selected malformations |
| Ireland: Dublin | Population-based Regional | 1997 | 5 years | 24 weeks or 500 grams | Not permitted |
| Israel: IBDSP | Hospital-based Regional | 1974 | Hospital discharge 2-5 days | 20weeks or 500 grams | Permitted<br>Reported |
| Italy: BDRCam | Population-based Regional | 1996 | 7 days | 180 days (25 weeks + 5 days) | Permitted<br>Reported |
| Italy: IMER | Population-based Regional | 1985 | 7 days | 180 days (25 weeks + 5 days) | Permitted<br>Reported |
| Italy: ISMAC | Hospital-based Regional | 1991 | 1 year | 180 days (25 weeks + 5 days) | Permitted<br>Reported |
| Italy: North East | Population-based Regional | 1997 | 7 days | 180 days (25 weeks + 5 days) | Permitted<br>Reported |

# Synopsis of Contributing Monitoring Systems

| Monitoring Program | Coverage | Year Joined ICBDMS | Maximum age at diagnosis | Criteria defining stillbirths | Termination of Pregnancy (ToP) |
|---|---|---|---|---|---|
| Italy-Tuscany:RTDC | Population-based Regional | 1998 | 1 year | 180 days (25 weeks + 5 days) | Permitted<br>Reported |
| Japan: JAOG | Hospital-based National | 1988 | 7 days | 22 weeks | Permitted<br>Not reported |
| Malta: MCAR | Population-based National | 2000 | 1 year | 20 weeks | Not permitted |
| Mexico: RYVEMCE | Hospital based National | 1980 | 72 hours | 20 weeks or 500 grams | Not permitted |
| New Zealand | Population-based National | 1979 | No limit | 20 weeks or 400 grams | Permitted<br>Not reported |
| Northern Netherlands | Population-based Regional | 1993 | Up to 15 years | 24 weeks | Permitted<br>Reported |
| Norway: MBRN | Population-based National | 1974 | Hospital discharge Lifelong for mortality (from 2002 1 year) | 16 weeks (12 weeks from 1999) | Permitted<br>Reported |
| Russia-Moscow Region: MRRCM | Population-based Regional | 2001 | 1 year | 28 weeks | Permitted<br>Reported |
| Slovak Republic | Population-based Regional | 2003 | 1 year | Non-viable fetuses, 28 weeks or >1000 grams | Permitted<br>Reported |
| South America: ECLAMC | Hospital-based Multinational | 1977 | 3 days | 500 grams | Not permitted |
| Spain: ECEMC | Hospital-based National | 1979 | 3 days | 24 weeks or 500 grams | Permitted<br>Not reported |
| Sweden | Population-based National | 1974 | 28 days | 22 weeks | Permitted<br>Reported |
| Ukraine: UABDP | Population-based Regional | 2001 | 28 days | 500 grams | Permitted<br>Reported only for selected malformations |
| United Arab Emirates | Hospital-based Regional | 1995 | 7 days | 23 weeks | Not permitted |
| USA-Atlanta: MACDP | Population-based Regional | 1974 | 6 years | 20 weeks | Permitted<br>Reported |
| USA-Texas: BDES | Population-based Regional | 2004 | 1 year | Before 2001: 20 weeks. Since 2001: All stillbirths with documented birth defects included | Permitted<br>Reported |
| USA-Utah UBDN | Population-based Regional | 2005 | 2 years | 20 weeks | Permitted<br>Reported |
| Wales: CARIS | Population-based Regional | 2005 | 1 year | 24 weeks | Permitted<br>Reported |

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. Incl. craniorachischisis. Incl. infants with iniencephaly and other neural tube defects as encephalocele or open spina bifida, when associated with anencephaly.Excl. 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. Incl. meningocele, meningomyelocele, myelocele, myelomeningocele, rachischisis. Spina bifida is not counted when present with anencephaly. Excl. 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 occipitofrontal circumference (OFC) 3 standard deviation below the age and sexappropriate distribution curves. [If using a different definition or cutoff point (e.g., 2 standard deviations), report but specify criteria]. Excl. 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. Excl. 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 anteroposterior 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 (IIV) of which the extreme form (grade IV) is anotia, absence of pinna.
Excl. 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. Incl. double outlet ventricle socalled 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. Excl. 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. Incl. submucous cleft palate. Excl. 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. Excl. 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. Incl. 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. Excl. 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. Excl. 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. Excl. 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. Incl. penile, scrotal, and perineal hypospadias. Excl. glandular or firstdegree 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. Incl. 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. Incl. infantile polycystic kidney, multicystic kidney, other forms of cystic kidney and unspecified cystic kidney. Excl. 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. Incl. femoral hypoplasia. Excl. 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. Incl. total absence of the diaphragm. Excl. 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. Excl. gastroschisis (paraumbilical hernia), a or hypoplasia of abdominal muscles, skincovered 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. Excl. aor hypoplasia of abdominal muscles, skincovered 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.

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

**34. Trisomy 18:** a congenital chromosomal malformation syndrome associated with extra chromosome 18 material.
Incl. 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. Incl. trisomy mosaicism and translocations of chromosome 21.

## Deviations from the ICBDSR Definitions by Registry

| | Encephalocele | Microcephaly | 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 sequenze | Trisomy 13 | Trisomy 18 | Down syndrome |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Australia: National | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Australia: VBDR | | | | | | | 8 | | 11 | 14 | | | 18 | | | 25 | | | 28 | 30 | 35 | | | | | |
| Australia: WABDR | | | | | | | | | 11 | | | | | | | 25 | | | 28 | | 35 | | | | | |
| Canada: Alberta | | 2 | | | 2 | 7 | 8 | | 11\12 | | | | | | | 25 | | | | | 35 | | | | | 2 |
| Canada: British Columbia | 1 | 2 | 4 | 6 | 2 | 7 | 8 | 10 | 11\12 | 13 | 15 | | 18\19 | | | 25 | 25\26 | 27 | 28 | 30 | 35 | 37 | | 2 | 2 | 2 |
| Canada: National | 1 | 2 | | 6 | 2 | | | | 11\12 | 14 | | | 18 | 21 | 23 | 25 | 26 | | 28 | 31 | 35 | | 40 | 2 | 2 | 2 |
| China: Beijing | | | | | | | | | | | | | | | | | | | | | 35 | | | | | |
| China: CBDMN | 1 | 2 | | 6 | 2 | 7 | 9 | | 12 | | | | 18 | | | 25 | | 27 | 28 | 31 | 35 | 37 | | 2 | 2 | 2 |
| 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 | | | | | |
| England and Wales | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Finland | | 2 | | | 2 | | 8 | | 11\12 | | | | | | | | | 27 | | 32 | | | | 2 | 2 | 2 |
| France: Central East | | | | | | | | | | | | | | | | 25 | | | | | | | | | | 2 |
| France: Paris | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| France: Strasbourg | | 2 | | | 2 | | 9 | | | | | | 18 | | | | | | 28\29 | 30 | | | | | | |
| Germany: Saxony-Anhalt | | 2\3 | | | | | 9 | | 11 | | | | 19 | | | 25 | | | 28 | 32 | 36 | 38 | | 2 | 2 | 2 |
| Hungary | 1 | 2 | | | 2 | | 9 | | | | | | | | | 25 | 26 | | | | 35 | 38\39 | | 2 | 2 | 2 |
| Ireland: Dublin | | 2 | | | 2 | | | | 11 | | | | 18\19 | | 24 | 25 | 26 | | | | 35 | | | 2 | 2 | 2 |
| Israel: IBDMS | | | | | | | 8 | | | | | | | | | 25 | | | | 33 | | | | | | |
| Italy: BDRCAM | | | | | | | | | | | | | | | | | | | | | | | | 2 | 2 | 2 |
| Italy: IMER | | | | | | | | | | | | | | | | 25 | | | | | 35 | | | | | |
| Italy: ISMAC | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| Italy: North East | | | 5 | | 2 | | | | | 13 | 15 | 17 | 18\20 | 22 | | | | | 29 | | 35 | | | | | 2 |
| Italy: Tuscany | | | | | | | 8 | | | | | | | | | | | | | | | | | | | |
| 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 |
| New Zealand | | | | | 2 | | | | | | | | | | | 25 | 26 | | | | | | | 2 | 2 | 2 |
| Northern Netherlands | | | | | | | | | | | | | | | 24 | 25 | | | | | 35 | | | | | |
| Norway | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Russia: Moscow region | | 2 | | | 2 | | 9 | | | | | | 18 | | | 25 | | 27 | 28 | 31 | 35 | | | 2 | 2 | 2 |
| Slovak Republic | | | | | | | | | | | 15 | | | | | 25 | | | | | 35 | | | | | 2 |
| South Africa: SABDSS | 1 | 2 | | | 2 | | | | 11\12 | | | | | | | 25 | | 27 | | 31 | 35 | 37 | | 2 | 2 | 2 |
| South America: ECLAMC | | | | | | | | | | | | | | | | 25 | | | | | | | | | | |
| Spain: ECEMC | | 3 | | | 2 | | | | | | | | | | | | | 27 | | | | 37 | | | | 2 |
| Sweden | | 2 | | | 2 | | | | 11 | | | | | | | 25 | | | 28 | 32 | | | | | | 2 |
| Ukraine | | 2\3 | | 6 | 2 | 7 | 9 | | | | 16 | | | | | | | 27 | | | | | | 2 | 2 | 2 |
| United Arab Emirates | | 2 | | | 2 | 7 | 8 | 10 | 11 | | | | 18 | | | | | | 28/29 | 31 | | | | | | |
| USA: Atlanta | | | | | | | | | 12 | | 16 | | | | | | | | | | | | | | | |
| Wales | 1 | 2 | | | 2 | 7 | | | | | | | | | 24 | 25 | | | | | | | | 2 | 2 | 2 |

## ICBDSR Definitions of the Reported Malformations

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
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

## Australia: VBDR
Victoria Birth Defects Registry

**History:**
In 1979 the Commonwealth Government agreed in principle to collect more information about births and birth defects. It was decided that the States would be responsible for setting up their own systems and the Commonwealth would establish a National Perinatal Statistics Unit, to collate information from all the states and provide an overall picture. The Victorian Perinatal Data Collection Unit (VPDCU), established under the Health Act of 1958, operates under the aegis of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (the Council). One of the fundamental purposes of the VPDCU was the establishment and maintenance of the Victorian Birth Defects Register (VBDR). The VPDCU and VBDR were established in 1982.

**Size and coverage:**
The VBDR collects information on all birth defects for livebirths, stillbirths and terminations of pregnancy pre 20 wks gestation and children up to 15 yrs of age (irrespective of the age at diagnosis). Approximately 3.8% of babies are born with a birth defect at or after 20 weeks gestation. We also follow up terminations for birth defects before 20 weeks, once these are included the overall prevalence is approximately 4%. Birth defects are notified to the register for those babies/fetus' who were born in Victoria

**Legislation and funding:**
The ongoing maintenance of the VBDR is enshrined in the legislation pertaining to the VPDCU (Health Act 1958) and is an ongoing function of the VPDCU, however notification of birth defects outside the reporting period on the Perinatal Morbidity Statistics form (28 days) is a voluntary process. There is a section for reporting of birth defects on the Perinatal form which is completed at the time of birth. Several measures are taken to ensure the ascertainment of birth defects outside this reporting period which will be specified in 'sources of ascertainment'. The VPDCU & VBDR are funded by the Department of Human Services (State Government)

**Sources of ascertainment:**

Perinatal forms (approx 48.8%)
Hospital listings* (aPprox 28.8%)
Perinatal death certificates/autopsy reports (approx 7.8%)
Cytogenetic reports (approx 9.3%)
Maternal & Child Health Nurse (approx 4.2%)
Other professionals/parents (approx 1.1%)

* These include obtaining annual inpatient listings from the two major paediatric teaching hospitals detailing all children up to the age of five years who have been subsequently admitted to these hospitals each year with a birth defect. We also obtain annual listings from specialist clinics at these hospital for all children up to the age of five years who have visited either as an inpatient or an outpatient. This procedure has also been adopted for Monash Medical Centre. Other listings are also received from Newborn Screening Services and Genetic Health Services Victoria.

**Exposure information:**
No exposure information is available

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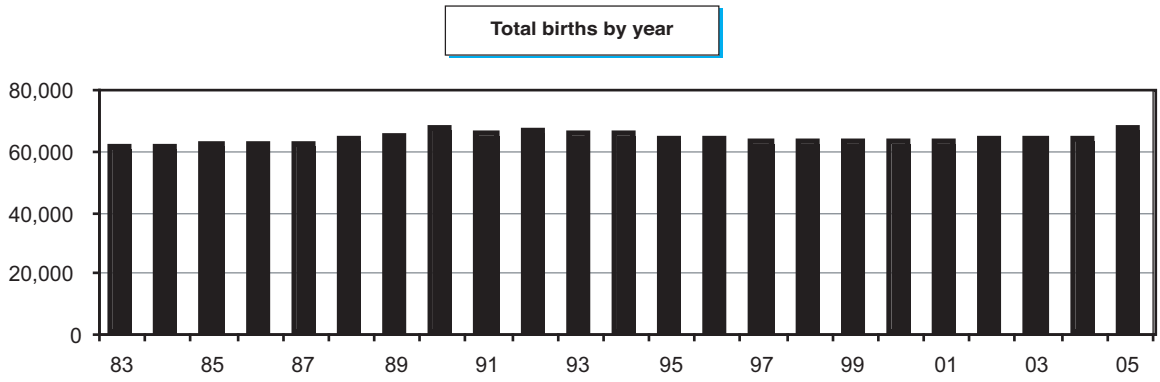
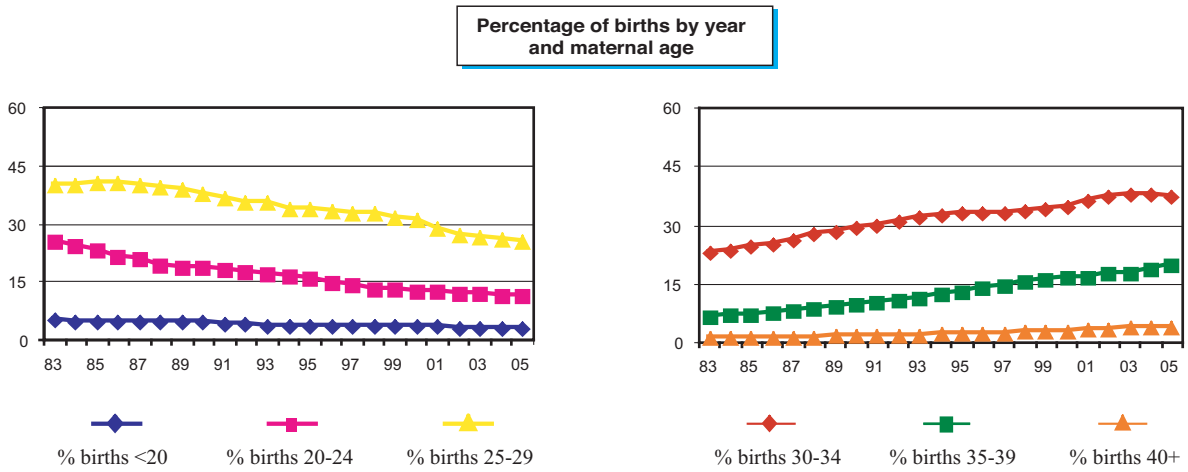
## Australia: VBDR




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 93 | 80.2 | Cystic kidney | 5 | 4.0 |
| Spina bifida | 40 | 35.1 | Limb reduction defects | 6 | 5.4 |
| Encephalocele | 10 | 40.0 | Diaphragmatic hernia | 6 | 9.8 |
| Holoprosencephaly | 13 | 36.1 | Omphalocele | 33 | 50.0 |
| Hydrocephaly | 37 | 19.3 | Gastroschisis | 6 | 15.0 |
| Hypoplastic left heart syndrome | 13 | 15.9 | Trisomy 13 | 44 | 71.0 |
| Cleft palate without cleft lip | 6 | 3.2 | Trisomy 18 | 138 | 74.2 |
| Cleft lip with or without cleft palate | 12 | 6.1 | Down syndrome | 351 | 63.8 |
| Renal agenesis | 16 | 11.7 | | | |

Total ToPs with births defects = 1,003 (Ratio ToPs/Births: 5.19 per 1,000)
*ToPs/ToPs+Births

# Australia: VBDR, 2005

| | |
|---|---|
| Live births (LB) | 66,041 |
| Stillbirths (SB) | 613 |
| Total births | 66,654 |
| Number of terminations of pregnancy (ToP) for birth defects | 333 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 6 | 28 | 5.25 |
| Spina bifida | 16 | 10 | 13 | 5.85 |
| Encephalocele | 3 | 2 | 3 | 1.20 |
| Microcephaly | 11 | 0 | 0 | 1.65 |
| Holoprosencephaly | 8 | 2 | 7 | 2.55 |
| Hydrocephaly | 33 | 15 | 11 | 8.85 |
| Anophthalmos | 0 | 0 | 3 | 0.45 |
| Microphthalmos | 1 | 0 | 0 | 0.15 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 6 | 0 | 0 | 0.90 |
| Microtia | 5 | 0 | 0 | 0.75 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 39 | 3 | 2 | 6.60 |
| Tetralogy of Fallot | 19 | 1 | 0 | 3.00 |
| Hypoplastic left heart syndrome | 24 | 5 | 3 | 4.80 |
| Coarctation of aorta | 29 | 0 | 1 | 4.50 |
| Choanal atresia, bilateral | 14 | 0 | 0 | 2.10 |
| Cleft palate without cleft lip | 54 | 0 | 1 | 8.25 |
| Cleft lip with or without cleft palate | 49 | 7 | 5 | 9.15 |
| Oesophageal atresia / stenosis with or without fistula | 22 | 1 | 0 | 3.45 |
| Small intestine atresia / stenosis | 20 | 1 | 0 | 3.15 |
| Anorectal atresia / stenosis | 22 | 3 | 4 | 4.35 |
| Undescended testis (36 weeks of gestation or later) | 364 | 1 | 0 | 54.76 |
| Hypospadias | 223 | 0 | 0 | 33.46 |
| Epispadias | 4 | 0 | 0 | 0.60 |
| Indeterminate sex | 10 | 0 | 1 | 1.65 |
| Renal agenesis | 35 | 3 | 6 | 6.60 |
| Cystic kidney | 45 | 2 | 2 | 7.35 |
| Bladder exstrophy | 2 | 1 | 0 | 0.45 |
| Polydactyly, preaxial | 60 | 4 | 6 | 10.50 |
| Total Limb reduction defects (include unspecified) | 26 | 8 | 3 | 5.55 |
| 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 | 16 | 1 | 0 | 2.55 |
| Omphalocele | 8 | 6 | 15 | 4.35 |
| Gastroschisis | 13 | 0 | 4 | 2.55 |
| Unspecified Omphalocele/Gastroschisis | 1 | 0 | 1 | 0.30 |
| Prune belly sequence | 1 | 0 | 1 | 0.30 |
| Trisomy 13 | 6 | 2 | 16 | 3.60 |
| Trisomy 18 | 9 | 6 | 46 | 9.15 |
| Down syndrome, all ages (include age unknown) | 62 | 9 | 114 | 27.76 |
| <20 | 1 | 0 | 0 | 5.45 |
| 20-24 | 2 | 0 | 3 | 6.74 |
| 25-29 | 7 | 2 | 2 | 6.62 |
| 30-34 | 16 | 3 | 23 | 17.38 |
| 35-39 | 24 | 3 | 52 | 60.91 |
| 40-44 | 12 | 1 | 15 | 119.91 |
| 45+ | 0 | 0 | 2 | 215.05 |
| unknown | 0 | 0 | 17 | --- |

nr = not reported

# Australia: VBDR, Previous years rates 1983 - 2005

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

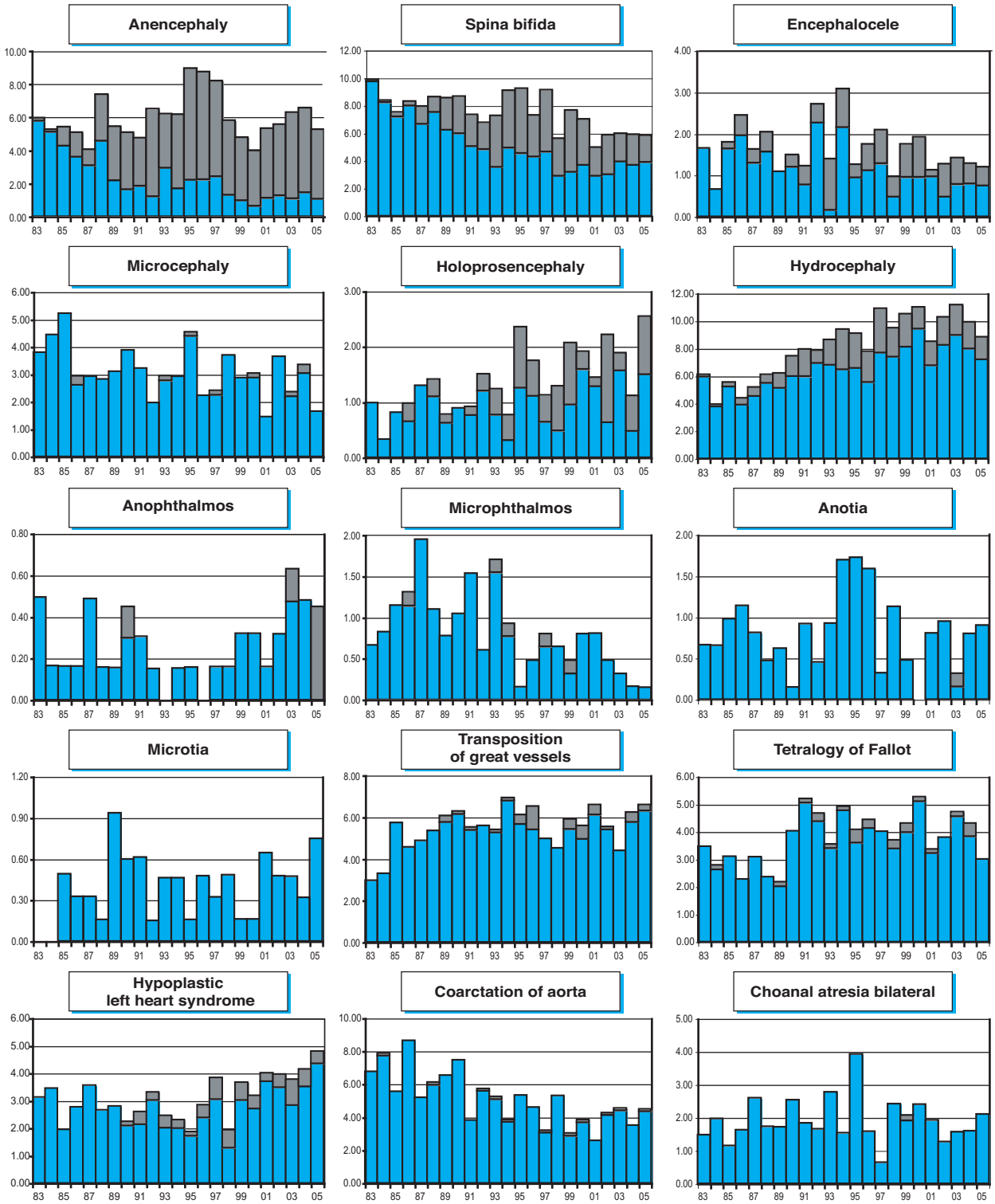
| | 1974-1980 | 1981-1985* | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | **182,554** | **317,619** | **324,939** | **312,603** | **317,966** |
| Anencephaly | | 5.53 | 5.42 | 6.49 | 6.30 | 5.79 |
| Spina bifida | | 8.60 | 8.44 | 7.94 | 7.36 | 5.72 |
| Encephalocele | | 1.37 | 1.73 | 1.94 | 1.70 | 1.26 |
| Microcephaly | | 4.49 | 3.15 | 3.11 | 2.85 | 2.48 |
| Holoprosencephaly | | 0.71 | 1.07 | 1.35 | 1.63 | 1.86 |
| Hydrocephaly | | 5.20 | 5.92 | 8.59 | 9.95 | 9.75 |
| Anophthalmos | | 0.27 | 0.28 | 0.15 | 0.19 | 0.41 |
| Microphthalmos | | 0.88 | 1.23 | 0.98 | 0.64 | 0.38 |
| Unspecified Anophthalmos / Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 0.77 | 0.63 | 1.14 | 0.70 | 0.75 |
| Microtia | | 0.16 | 0.47 | 0.37 | 0.32 | 0.53 |
| Unspecified Anotia / Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 4.00 | 5.45 | 5.91 | 5.50 | 5.88 |
| Tetralogy of Fallot | | 3.12 | 2.80 | 4.49 | 4.35 | 3.84 |
| Hypoplastic left heart syndrome | | 2.85 | 2.80 | 2.52 | 3.10 | 4.15 |
| Coarctation of aorta | | 6.74 | 6.80 | 4.80 | 4.00 | 3.90 |
| Choanal atresia, bilateral | | 1.53 | 2.05 | 2.34 | 1.82 | 1.70 |
| Cleft palate without cleft lip | | 8.16 | 7.43 | 7.72 | 8.22 | 8.84 |
| Cleft lip with or without cleft palate | | 11.07 | 10.01 | 10.56 | 10.94 | 10.00 |
| Oesophageal atresia / stenosis with or without fistula | | 4.38 | 2.99 | 3.97 | 3.90 | 2.67 |
| Small intestine atresia / stenosis | | 2.03 | 2.71 | 2.22 | 3.39 | 3.14 |
| Anorectal atresia / stenosis | | 3.94 | 3.27 | 4.89 | 4.73 | 3.81 |
| Undescended testis (36 weeks of gestation or later) | | 5.92 | 27.42 | 44.75 | 48.40 | 49.82 |
| Hypospadias | | 17.15 | 23.30 | 32.68 | 34.61 | 34.44 |
| Epispadias | | 0.16 | 0.41 | 0.34 | 0.67 | 0.44 |
| Indeterminate sex | | 1.26 | 2.74 | 2.80 | 1.57 | 1.64 |
| Renal agenesis | | 4.55 | 4.97 | 5.54 | 6.94 | 6.51 |
| Cystic kidney | | 2.90 | 4.03 | 5.48 | 7.26 | 6.48 |
| Bladder exstrophy | | 0.49 | 0.38 | 0.40 | 0.35 | 0.53 |
| Polydactyly, preaxial | | 7.61 | 8.12 | 10.22 | 10.27 | 11.04 |
| Total Limb reduction defects (include unspecified) | | 5.70 | 5.95 | 6.65 | 7.13 | 5.44 |
| 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 | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 3.12 | 3.05 | 4.34 | 3.58 | 3.14 |
| Omphalocele | | 2.25 | 3.27 | 3.02 | 3.55 | 3.46 |
| Gastroschisis | | 0.71 | 1.35 | 1.82 | 2.91 | 2.11 |
| Unspecified Omphalocele / Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | 0.27 | 0.31 | 0.31 | 0.35 | 0.13 |
| Trisomy 13 | | 0.99 | 1.29 | 2.12 | 2.72 | 3.18 |
| Trisomy 18 | | 2.08 | 3.31 | 4.99 | 6.62 | 8.49 |
| Down syndrome, all ages (include age unknown) | | 11.89 | 16.78 | 18.93 | 23.10 | 28.87 |
| <20 | | 12.67 | 5.81 | 5.90 | 4.83 | 4.25 |
| 20-24 | | 6.55 | 8.99 | 7.31 | 6.91 | 9.39 |
| 25-29 | | 8.38 | 8.53 | 8.82 | 8.68 | 9.34 |
| 30-34 | | 13.73 | 16.07 | 16.52 | 16.91 | 17.80 |
| 35-39 | | 30.47 | 50.72 | 50.54 | 48.34 | 58.43 |
| 40-44 | | 81.66 | 127.34 | 143.34 | 203.89 | 170.72 |
| 45+ | | 107.53 | 74.63 | 485.44 | 362.32 | 320.99 |
| unknown | | --- | --- | --- | --- | --- |

* data include less than 5 years

nr = not reported

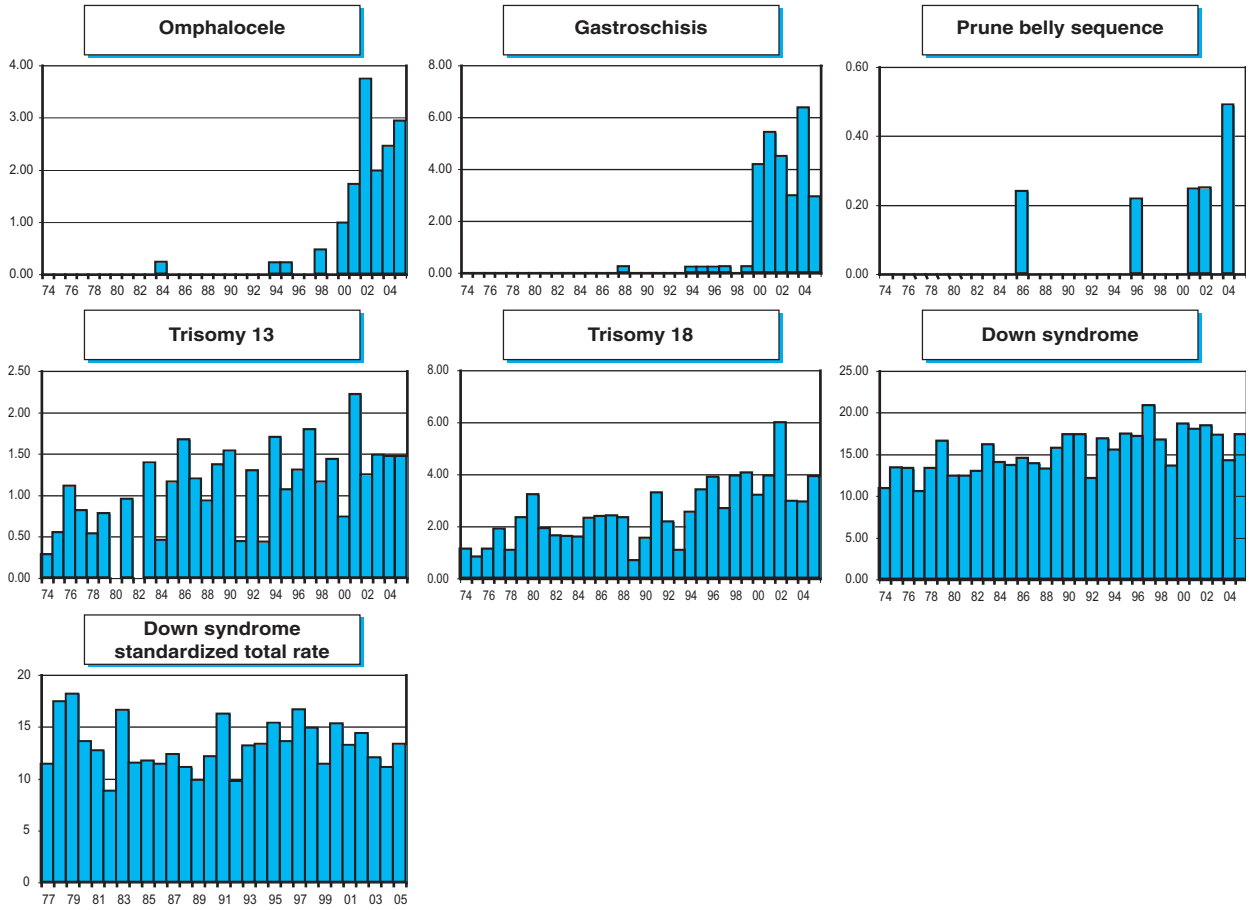
## Australia: VBDR

'Time trends 1983-2005 (Birth prevalence rates per 10,000)



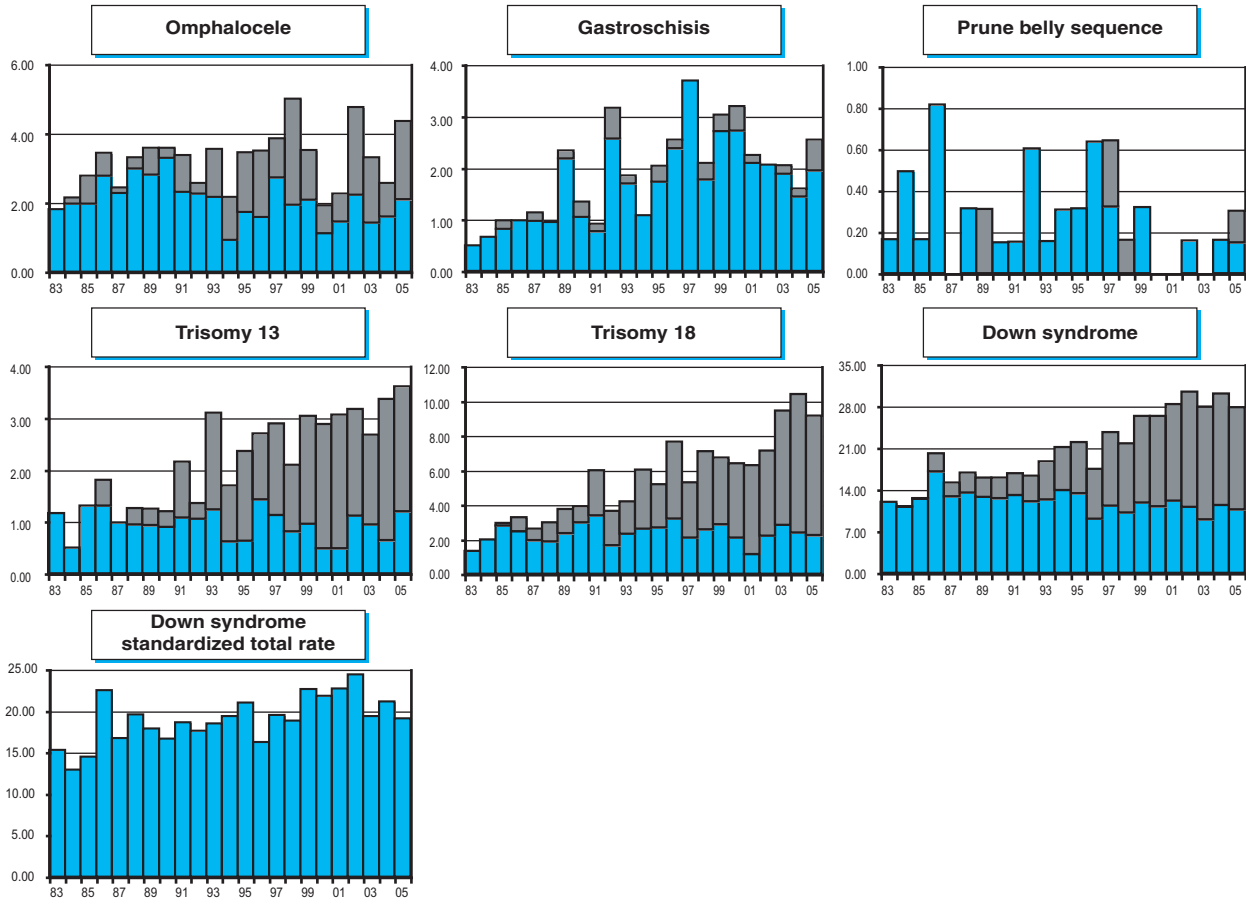

**Note:** L+S rates, ToP rates

## Canada: British Columbia




**Note:** L+S rates, ToP rates

## Australia: VBDR




**Note:** L+S rates, ToP rates

## Australia: WABDR
Western Australian Birth Defects Registry

**History:**
The Registry was established in 1980, and is currently located in a teaching obstetric hospital. The objectives of the Registry have always been to establish how often birth defects occur, to conduct research into causes and prevention of birth defects, to provide health professionals and the public with information about birth defects, and to monitor and evaluate screening, treatment and prevention programs.

**Size and coverage:**
Population-based in the state of Western Australia. 25,000 birth a year, ~6% reported with a birth defect. Birth defects diagnosed prenatally and up to the age of 6 years, in stillbirths, terminations of pregnancy and livebirths are included.

**Legislation and funding:**
Following a period of short term funding from both Federal and State sources, the Registry is now wholly funded by the Western Australian Department of Health. There are several statutory sources of information (birth, death and hospital data collections), and a large number of voluntary sources. Statutory notification is being considered by the Department of Health.

**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).
Voluntary sources: Maternity and paediatric hospitals, Obstetricians, paediatricians, orthopaedic surgeons, Community and Child Health Nurses, Cytogenetic laboratories, Pathology services (including prenatal screening services), Ultrasound practices Genetic Services, Disability services.

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

**Background information:**
The data on the Registry 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 birth defects (numerators) and all births in Western Australia (denominators).

Data from the Registry are provided to the National Perinatal Statistics Unit for monitoring birth defects in Australia as a whole.

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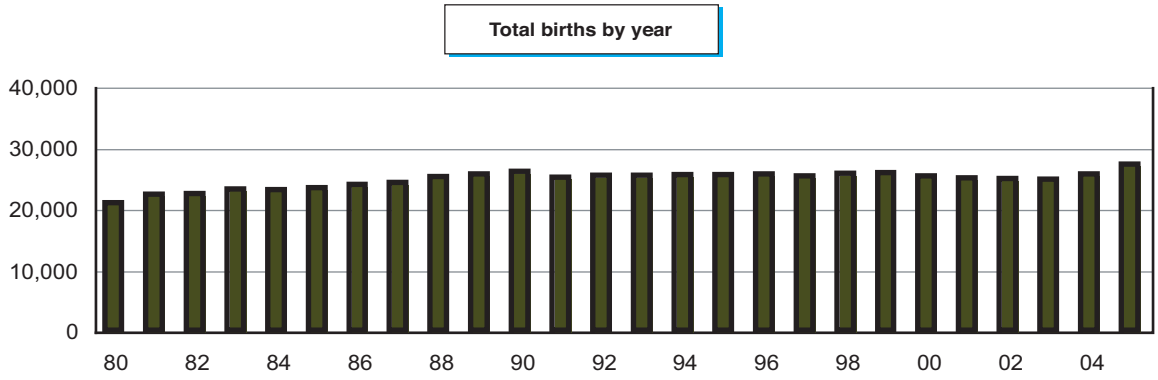
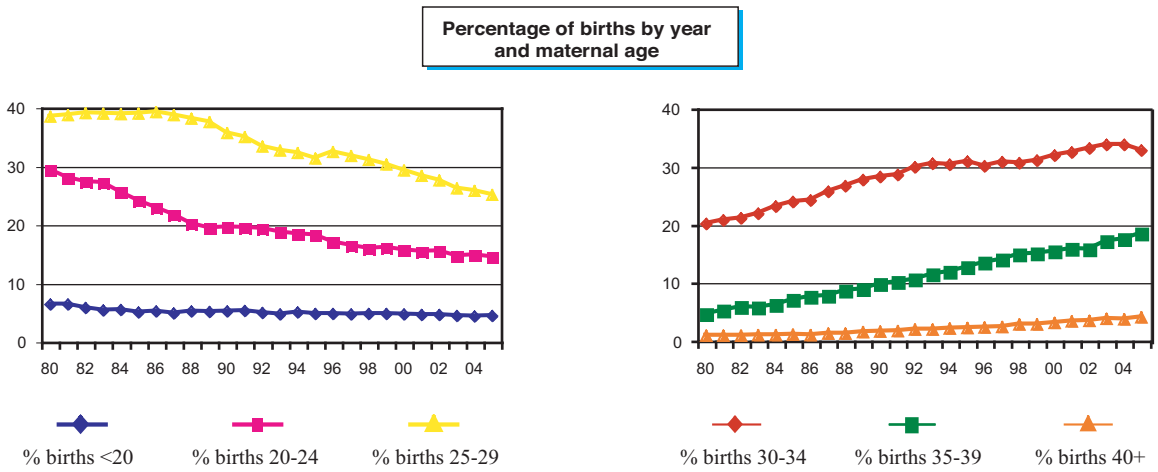
**Website:**
http://www.kemh.health.wa.gov.au/services/birth_defects/index.htm

## Australia: WABDR




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 38 | 92.7 | Cystic kidney | 19 | 31.1 |
| Spina bifida | 40 | 64.5 | Limb reduction defects | 15 | 37.5 |
| Encephalocele | 6 | 60.0 | Diaphragmatic hernia | 4 | 16.7 |
| Holoprosencephaly | 12 | 70.6 | Omphalocele | 25 | 62.5 |
| Hydrocephaly | 28 | 52.8 | Gastroschisis | 5 | 17.9 |
| Hypoplastic left heart syndrome | 9 | 75.0 | Trisomy 13 | 24 | 77.4 |
| Cleft palate without cleft lip | 5 | 6.0 | Trisomy 18 | 47 | 81.0 |
| Cleft lip with or without cleft palate | 14 | 13.3 | Down syndrome | 130 | 61.9 |
| Renal agenesis | 20 | 54.1 | | | |

Total ToPs with births defects = 503 (Ratio ToPs/Births: 6.50 per 1,000 births)
*ToPs/ToPs+Births

# Australia: WABDR, 2005

| | |
|---|---|
| Live births (LB) | 26,992 |
| Stillbirths (SB) | 200 |
| Total births | 27,192 |
| Number of terminations of pregnancy (ToP) for birth defects | 185 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 2 | 12 | 5.52 |
| Spina bifida | 8 | 0 | 15 | 8.46 |
| Encephalocele | 2 | 0 | 2 | 1.47 |
| Microcephaly | 13 | 1 | 0 | 5.15 |
| Holoprosencephaly | 1 | 2 | 4 | 2.57 |
| Hydrocephaly | 7 | 2 | 14 | 8.46 |
| Anophthalmos | 0 | 1 | 0 | 0.37 |
| Microphthalmos | 2 | 0 | 0 | 0.74 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 3 | 0 | 0 | 1.10 |
| Microtia | 3 | 0 | 0 | 1.10 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 5 | 1 | 7 | 4.78 |
| Tetralogy of Fallot | 10 | 0 | 0 | 3.68 |
| Hypoplastic left heart syndrome | 2 | 0 | 3 | 1.84 |
| Coarctation of aorta | 11 | 0 | 4 | 5.52 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 27 | 1 | 4 | 11.77 |
| Cleft lip with or without cleft palate | 22 | 0 | 6 | 10.30 |
| Oesophageal atresia / stenosis with or without fistula | 12 | 0 | 4 | 5.88 |
| Small intestine atresia / stenosis | 5 | 0 | 3 | 2.94 |
| Anorectal atresia / stenosis | 12 | 1 | 4 | 6.25 |
| Undescended testis (36 weeks of gestation or later) | 45 | 0 | 0 | 16.55 |
| Hypospadias | 67 | 0 | 0 | 24.64 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 3 | 2 | 3 | 2.94 |
| Cystic kidney | 11 | 1 | 4 | 5.88 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 21 | 1 | 4 | 9.56 |
| Total Limb reduction defects (include unspecified) | 8 | 0 | 7 | 5.52 |
| 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 | 9 | 0 | 0 | 3.31 |
| Omphalocele | 1 | 0 | 8 | 3.31 |
| Gastroschisis | 7 | 0 | 1 | 2.94 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 2 | 1 | 8 | 4.05 |
| Trisomy 18 | 4 | 1 | 20 | 9.19 |
| Down syndrome, all ages (include age unknown) | 56 | 0 | 100 | 57.37 |
| <20 | 4 | 0 | 0 | 33.06 |
| 20-24 | 4 | 0 | 4 | 20.36 |
| 25-29 | 6 | 0 | 2 | 11.71 |
| 30-34 | 18 | 0 | 25 | 48.25 |
| 35-39 | 16 | 0 | 43 | 117.79 |
| 40-44 | 8 | 0 | 26 | 321.67 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | --- |

nr = not reported

# Australia: WABDR, Previous years rates 1980 - 2005

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

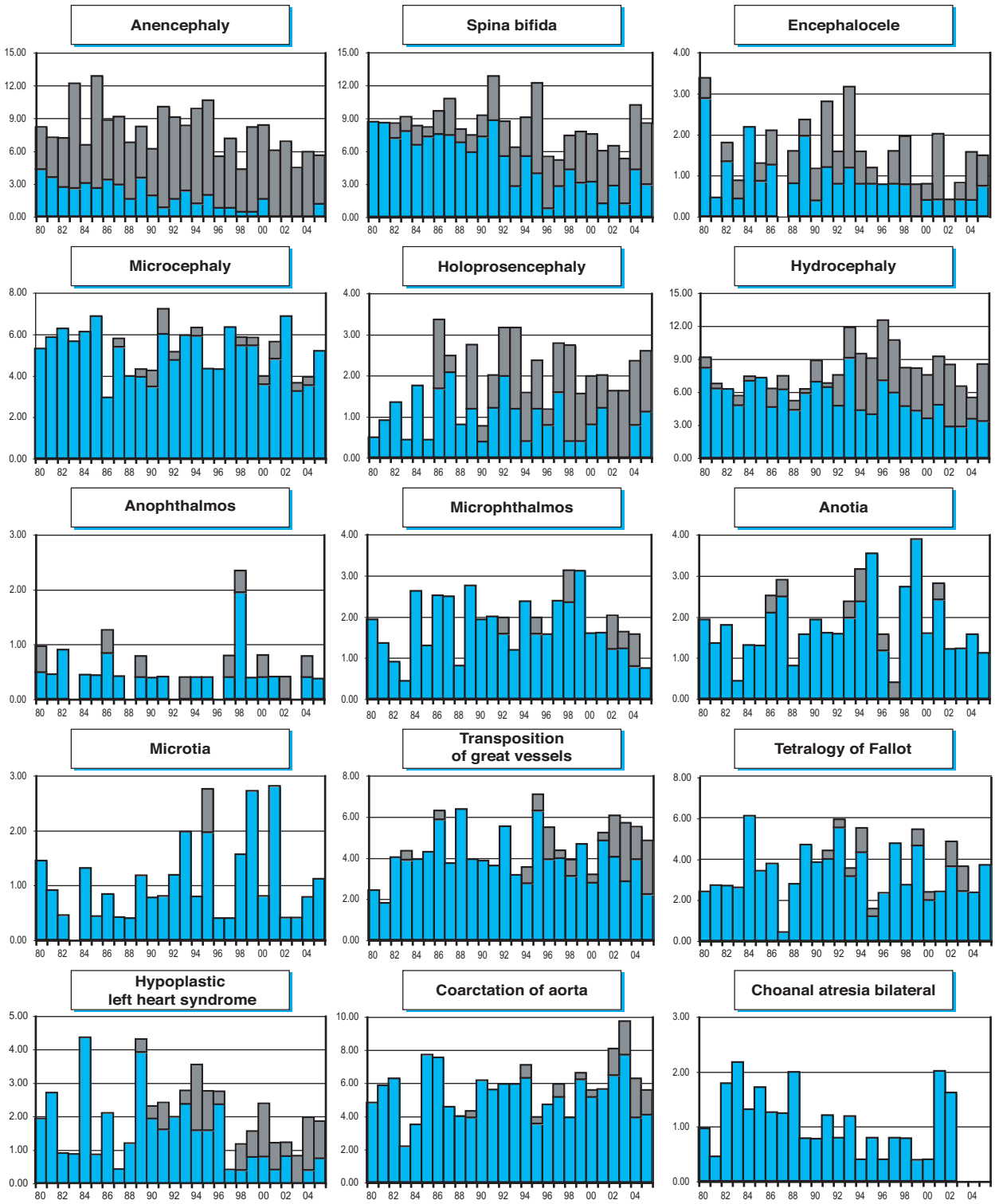
| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **20,815** | **114,004** | **124,813** | **126,534** | **127,524** | **126,924** |
| Anencephaly | 8.17 | 9.21 | 7.77 | 9.56 | 6.67 | 5.75 |
| Spina bifida | 8.65 | 8.51 | 8.97 | 9.80 | 6.67 | 7.33 |
| Encephalocele | 3.36 | 1.32 | 1.44 | 2.05 | 1.18 | 1.26 |
| Microcephaly | 5.28 | 6.14 | 4.25 | 5.77 | 5.25 | 5.04 |
| Holoprosencephaly | 0.48 | 0.96 | 2.00 | 2.45 | 2.04 | 2.05 |
| Hydrocephaly | 9.13 | 6.67 | 6.81 | 8.93 | 9.41 | 7.64 |
| Anophthalmos | 0.96 | 0.44 | 0.56 | 0.32 | 0.86 | 0.39 |
| Microphthalmos | 1.92 | 1.32 | 2.08 | 1.90 | 2.35 | 1.50 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 1.92 | 1.23 | 1.92 | 2.45 | 2.04 | 1.58 |
| Microtia | 1.44 | 0.61 | 0.72 | 1.50 | 1.18 | 1.10 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 2.40 | 3.68 | 4.81 | 4.58 | 4.31 | 5.44 |
| Tetralogy of Fallot | 2.40 | 3.51 | 3.12 | 4.19 | 3.53 | 3.39 |
| Hypoplastic left heart syndrome | 1.92 | 1.93 | 2.08 | 2.69 | 1.65 | 1.42 |
| Coarctation of aorta | 4.80 | 5.09 | 5.29 | 5.69 | 5.33 | 7.01 |
| Choanal atresia, bilateral | 0.96 | 1.49 | 1.20 | 0.87 | 0.55 | 0.71 |
| Cleft palate without cleft lip | 7.21 | 8.95 | 9.13 | 10.35 | 12.31 | 11.66 |
| Cleft lip with or without cleft palate | 14.41 | 11.75 | 13.62 | 10.35 | 12.31 | 13.00 |
| Oesophageal atresia / stenosis with or without fistula | 1.44 | 3.07 | 3.69 | 3.16 | 3.29 | 4.18 |
| Small intestine atresia / stenosis | 1.92 | 3.07 | 2.80 | 2.13 | 3.29 | 2.84 |
| Anorectal atresia / stenosis | 8.65 | 4.91 | 5.37 | 6.72 | 5.88 | 6.78 |
| Undescended testis (36 weeks of gestation or later) | 71.10 | 64.73 | 67.06 | 64.57 | 55.99 | 35.45 |
| Hypospadias | 24.98 | 28.07 | 30.13 | 36.04 | 37.01 | 31.91 |
| Epispadias | 0.00 | 0.26 | 0.40 | 0.16 | 0.16 | 0.16 |
| Indeterminate sex | 0.00 | 0.18 | 0.40 | 0.32 | 0.16 | 0.32 |
| Renal agenesis | 3.36 | 4.12 | 3.45 | 4.74 | 5.02 | 4.88 |
| Cystic kidney | 3.36 | 2.54 | 4.73 | 7.35 | 7.61 | 9.38 |
| Bladder exstrophy | 0.00 | 0.18 | 0.40 | 0.16 | 0.63 | 0.32 |
| Polydactyly, preaxial | 8.17 | 10.09 | 10.42 | 10.51 | 11.92 | 10.16 |
| Total Limb reduction defects (include unspecified) | 3.84 | 4.21 | 5.13 | 5.69 | 8.08 | 6.46 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 4.32 | 2.81 | 2.56 | 3.24 | 4.39 | 2.68 |
| Omphalocele | 2.88 | 2.02 | 2.96 | 3.40 | 3.53 | 4.81 |
| Gastroschisis | 0.48 | 1.58 | 1.68 | 2.77 | 4.16 | 3.62 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.48 | 0.70 | 0.48 | 0.63 | 0.24 | 0.00 |
| Trisomy 13 | 0.96 | 0.61 | 1.12 | 1.82 | 2.04 | 3.23 |
| Trisomy 18 | 0.96 | 1.75 | 2.16 | 3.95 | 6.19 | 7.33 |
| Down syndrome, all ages (include age unknown) | 11.05 | 12.37 | 15.38 | 18.49 | 21.33 | 26.16 |
| <20 | 0.00 | 4.71 | 7.86 | 6.41 | 11.63 | 10.53 |
| 20-24 | 4.90 | 4.65 | 8.13 | 6.30 | 7.24 | 6.84 |
| 25-29 | 8.74 | 9.02 | 8.05 | 8.16 | 11.40 | 10.35 |
| 30-34 | 23.84 | 11.87 | 14.19 | 20.25 | 16.00 | 19.44 |
| 35-39 | 20.94 | 54.17 | 39.47 | 41.87 | 44.25 | 54.24 |
| 40-44 | 0.00 | 138.20 | 230.18 | 167.08 | 172.15 | 152.36 |
| 45+ | 588.24 | 500.00 | 434.78 | 476.19 | 251.57 | 516.43 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 7 years

nr = not reported

## Australia: WABDR

'Time trends 1980-2005 (Birth prevalence rates per 10,000)

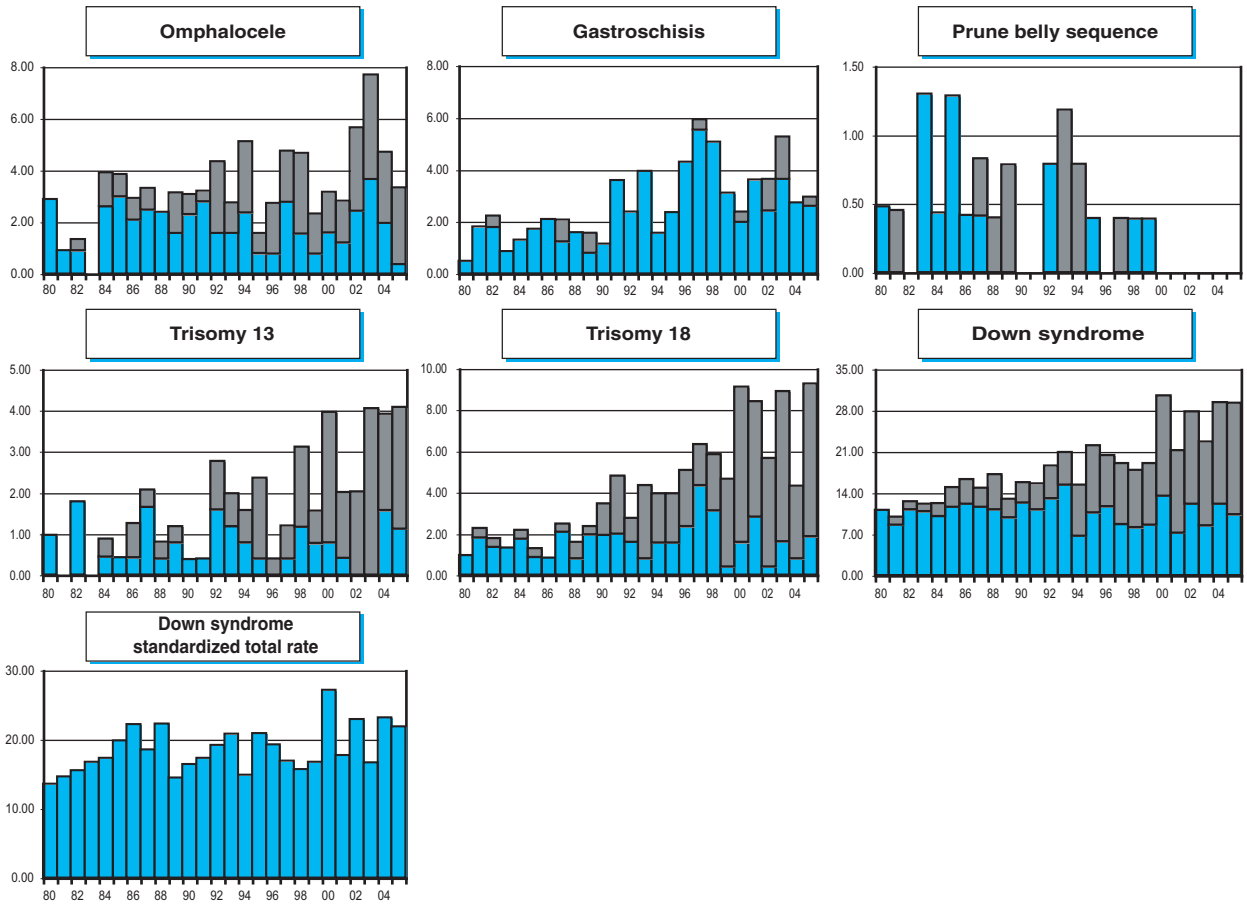



**Note:** L+S rates, ToP rates

## Australia: WABDR


**Note:** L+S rates, ToP rates

## Australia: VBDR




**Note:** L+S rates, ToP rates

## **Canada-Alberta:** ACASS
## Alberta Congenital Anomalies Surveillance System

**History:**
The Programme began in 1966 as a general Registry for Handicapped Children. This was disbanded in 1980 and continued as a surveillance Programme 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 comprises about 40,000 births per year. The definition of stillbirth is 20 weeks or more or 500 grams or more. The vast majority of births occur in hospital (approximately 97%). In 1997 a special fetal congenital anomalies surveillance system was started to include those fetuses with congenital anomalies who were either spontaneously lost prior to 20 weeks or where there was termination as a result of prenatal diagnosis.

**Legislation and funding:**
Reporting is voluntary. The system is run by members of the Department of Medical Genetics, Alberta Children's Hospital/University of Calgary reporting to Alberta Vital Statistics and Alberta Health. Funding is from Alberta Ministry of Health.

**Sources of ascertainment:**
Reports are obtained from physician's notice of birth, live birth and stillbirth registrations, death registrations and a special congenital anomalies reporting form (CARF) from hospitals. This is based on discharge diagnosis, including readmissions for any reason up to one year of age. Additional sources are speciality clinics, such as medical genetics and cytogenetics laboratories.

**Exposure information:**
None is routine.

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

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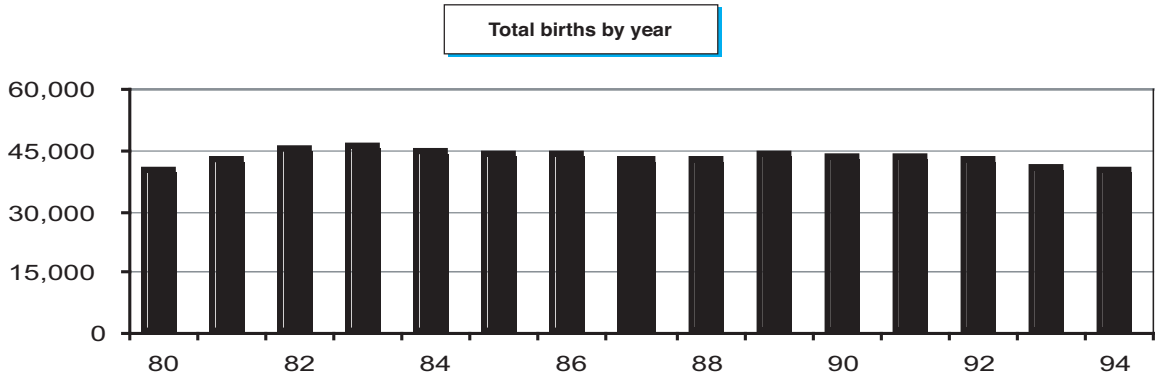
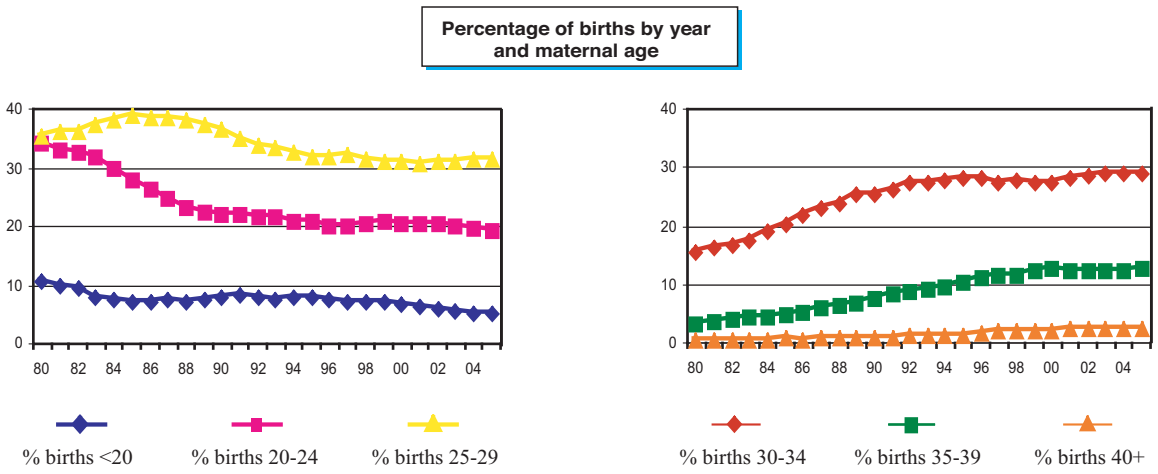
## Canada-Alberta: ACASS




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 12 | 38.7 | Cystic kidney | 9 | 8.4 |
| Spina bifida | 10 | 24.4 | Limb reduction defects | 32 | 23.2 |
| Encephalocele | 6 | 33.3 | Diaphragmatic hernia | 5 | 14.3 |
| Holoprosencephaly | 9 | 39.1 | Omphalocele | 9 | 31.0 |
| Hydrocephaly | 3 | 4.5 | Gastroschisis | 2 | 4.0 |
| Hypoplastic left heart syndrome | 1 | 2.9 | Trisomy 13 | 12 | 42.9 |
| Cleft palate without cleft lip | 2 | 2.3 | Trisomy 18 | 24 | 40.7 |
| Cleft lip with or without cleft palate | 12 | 8.1 | Down syndrome | 59 | 20.8 |
| Renal agenesis | 9 | 14.5 | | | |

Total ToPs with births defects = 252 (Ratio ToPs/Births: 2.06 per 1,000 births)
*ToPs/ToPs+Births

# Canada-Alberta: ACASS, 2005

| | |
|---|---|
| Live births (LB) | 41,551 |
| Stillbirths (SB) | 305 |
| Total births | 41,856 |
| Number of terminations of pregnancy (ToP) for birth defects | 84 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 5 | 2 | 2 | 2.15 |
| Spina bifida | 6 | 2 | 4 | 2.87 |
| Encephalocele | 5 | 1 | 3 | 2.15 |
| Microcephaly | 15 | 4 | 1 | 4.78 |
| Holoprosencephaly | 4 | 1 | 3 | 1.91 |
| Hydrocephaly | 15 | 5 | 2 | 5.26 |
| Anophthalmos | 1 | 0 | 0 | 0.24 |
| Microphthalmos | 9 | 1 | 0 | 2.39 |
| Unspecified Anophthalmos/ Microphthalmos | nr | nr | nr | nr |
| Anotia | 2 | 1 | 0 | 0.72 |
| Microtia | 9 | 0 | 0 | 2.15 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 20 | 1 | 2 | 5.50 |
| Tetralogy of Fallot | 8 | 1 | 0 | 2.15 |
| Hypoplastic left heart syndrome | 14 | 5 | 0 | 4.54 |
| Coarctation of aorta | 16 | 0 | 0 | 3.82 |
| Choanal atresia, bilateral | 5 | 0 | 0 | 1.19 |
| Cleft palate without cleft lip | 26 | 2 | 1 | 6.93 |
| Cleft lip with or without cleft palate | 41 | 4 | 3 | 11.47 |
| Oesophageal atresia / stenosis with or without fistula | 5 | 2 | 1 | 1.91 |
| Small intestine atresia / stenosis | 8 | 0 | 0 | 1.91 |
| Anorectal atresia / stenosis | 12 | 2 | 2 | 3.82 |
| Undescended testis (36 weeks of gestation or later) | 94 | 1 | 0 | 22.70 |
| Hypospadias | 92 | 2 | 0 | 22.46 |
| Epispadias | 5 | 0 | 0 | 1.19 |
| Indeterminate sex | 3 | 1 | 1 | 1.19 |
| Renal agenesis | 13 | 2 | 3 | 4.30 |
| Cystic kidney | 26 | 5 | 3 | 8.12 |
| Bladder exstrophy | 1 | 0 | 0 | 0.24 |
| Polydactyly, preaxial | 12 | 1 | 0 | 3.11 |
| Total Limb reduction defects (include unspecified) | 30 | 6 | 13 | 11.71 |
| 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 | nr | nr | nr | nr |
| Diaphragmatic hernia | 11 | 2 | 1 | 3.34 |
| Omphalocele | 3 | 1 | 6 | 2.39 |
| Gastroschisis | 20 | 2 | 2 | 5.73 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | 4 | 1 | 0 | 1.19 |
| Trisomy 13 | 0 | 4 | 5 | 2.15 |
| Trisomy 18 | 9 | 9 | 10 | 6.69 |
| Down syndrome, all ages (include age unknown) | 71 | 15 | 25 | 26.52 |
| <20 | 2 | 1 | 1 | 18.45 |
| 20-24 | 3 | 1 | 1 | 6.23 |
| 25-29 | 17 | 1 | 2 | 15.18 |
| 30-34 | 16 | 4 | 5 | 20.59 |
| 35-39 | 19 | 7 | 9 | 66.15 |
| 40-44 | 12 | 1 | 6 | 187.38 |
| 45+ | 2 | 0 | 1 | 731.71 |
| unknown | nr | nr | nr | nr |

nr = not reported

# Canada-Alberta: ACASS, Previous years rates 1980 - 2005

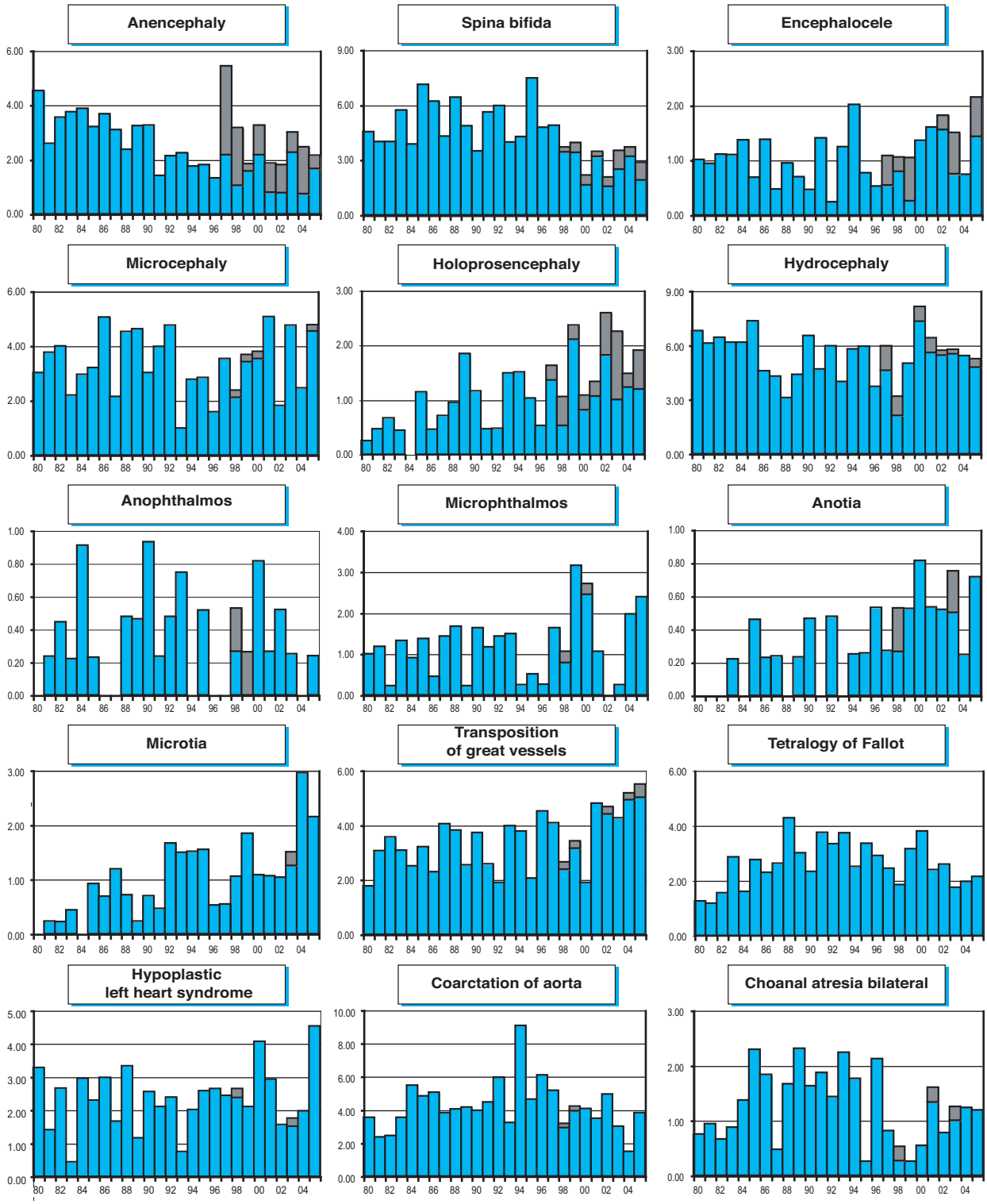
Birth prevalence rates: (LB+SB) * 10,000 until 1996
Birth prevalence rates: (LB+SB+TOP) * 10,000 since 1997

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **39,655** | **220,260** | **213,597** | **203,286** | **187,180** | **198,259** |
| Anencephaly | 4.54 | 3.41 | 3.14 | 1.87 | 2.99 | 2.27 |
| Spina bifida | 4.54 | 4.95 | 5.06 | 5.46 | 3.90 | 3.13 |
| Encephalocele | 1.01 | 1.04 | 0.80 | 1.13 | 1.02 | 1.56 |
| Microcephaly | 3.03 | 3.22 | 3.89 | 3.10 | 2.99 | 3.78 |
| Holoprosencephaly | 0.25 | 0.54 | 1.03 | 0.98 | 1.34 | 1.92 |
| Hydrocephaly | 6.81 | 6.45 | 4.59 | 5.26 | 5.18 | 5.70 |
| Anophthalmos | 0.00 | 0.41 | 0.37 | 0.39 | 0.32 | 0.25 |
| Microphthalmos | 1.01 | 1.00 | 1.08 | 0.98 | 1.76 | 1.16 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.00 | 0.14 | 0.23 | 0.20 | 0.53 | 0.55 |
| Microtia | 0.00 | 0.36 | 0.70 | 1.33 | 1.02 | 1.77 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 1.77 | 3.09 | 3.28 | 2.85 | 3.31 | 4.89 |
| Tetralogy of Fallot | 1.26 | 2.00 | 2.90 | 3.35 | 2.83 | 2.17 |
| Hypoplastic left heart syndrome | 3.28 | 1.95 | 2.34 | 1.97 | 2.78 | 2.57 |
| Coarctation of aorta | 3.53 | 3.72 | 4.21 | 5.46 | 4.54 | 3.33 |
| Choanal atresia, bilateral | 0.76 | 1.23 | 1.59 | 1.52 | 0.85 | 1.21 |
| Cleft palate without cleft lip | 6.30 | 6.22 | 8.05 | 7.72 | 8.49 | 7.82 |
| Cleft lip with or without cleft palate | 11.10 | 10.22 | 12.36 | 11.86 | 11.49 | 12.46 |
| Oesophageal atresia / stenosis with or without fistula | 0.50 | 2.95 | 3.23 | 2.31 | 2.40 | 2.12 |
| Small intestine atresia / stenosis | 0.50 | 0.77 | 1.22 | 1.48 | 1.87 | 1.92 |
| Anorectal atresia / stenosis | 3.53 | 3.50 | 5.34 | 4.92 | 5.34 | 6.41 |
| Undescended testis (36 weeks of gestation or later) | 22.70 | 27.10 | 29.45 | 25.38 | 23.45 | 25.77 |
| Hypospadias | 17.65 | 17.43 | 26.22 | 22.19 | 18.38 | 20.78 |
| Epispadias | 0.76 | 0.45 | 0.23 | 0.44 | 0.48 | 0.76 |
| Indeterminate sex | 0.25 | 0.36 | 0.70 | 1.13 | 1.12 | 1.31 |
| Renal agenesis | 2.27 | 3.41 | 5.15 | 4.97 | 4.65 | 6.00 |
| Cystic kidney | 0.25 | 2.81 | 4.35 | 4.72 | 5.61 | 8.02 |
| Bladder exstrophy | 0.00 | 0.36 | 0.23 | 0.34 | 0.21 | 0.40 |
| Polydactyly, preaxial | 13.37 | 9.40 | 15.82 | 14.81 | 12.93 | 13.47 |
| Total Limb reduction defects (include unspecified) | 6.56 | 6.36 | 9.88 | 9.84 | 10.47 | 11.45 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 2.77 | 3.81 | 2.86 | 2.41 | 3.53 | 3.58 |
| Omphalocele | 1.26 | 1.86 | 2.29 | 1.82 | 2.30 | 2.37 |
| Gastroschisis | 1.51 | 1.41 | 1.45 | 1.72 | 2.56 | 3.93 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.50 | 0.45 | 0.33 | 0.10 | 0.48 | 0.50 |
| Trisomy 13 | 0.50 | 0.82 | 0.80 | 1.38 | 1.50 | 1.92 |
| Trisomy 18 | 1.26 | 1.59 | 1.92 | 2.36 | 4.01 | 4.79 |
| Down syndrome, all ages (include age unknown) | 12.10 | 8.67 | 10.16 | 11.26 | 15.49 | 21.29 |
| <20 | nr | nr | 6.35 | 3.15 | 7.67 | 10.93 |
| 20-24 | nr | nr | 5.54 | 6.68 | 4.98 | 6.10 |
| 25-29 | nr | nr | 6.33 | 7.83 | 8.20 | 11.19 |
| 30-34 | nr | nr | 12.57 | 13.88 | 14.11 | 16.70 |
| 35-39 | nr | nr | 34.79 | 24.83 | 39.29 | 58.83 |
| 40-44 | nr | nr | 101.56 | 72.63 | 146.58 | 160.90 |
| 45+ | nr | nr | 178.57 | 379.75 | 306.12 | 211.64 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 7 years
nr = not reported

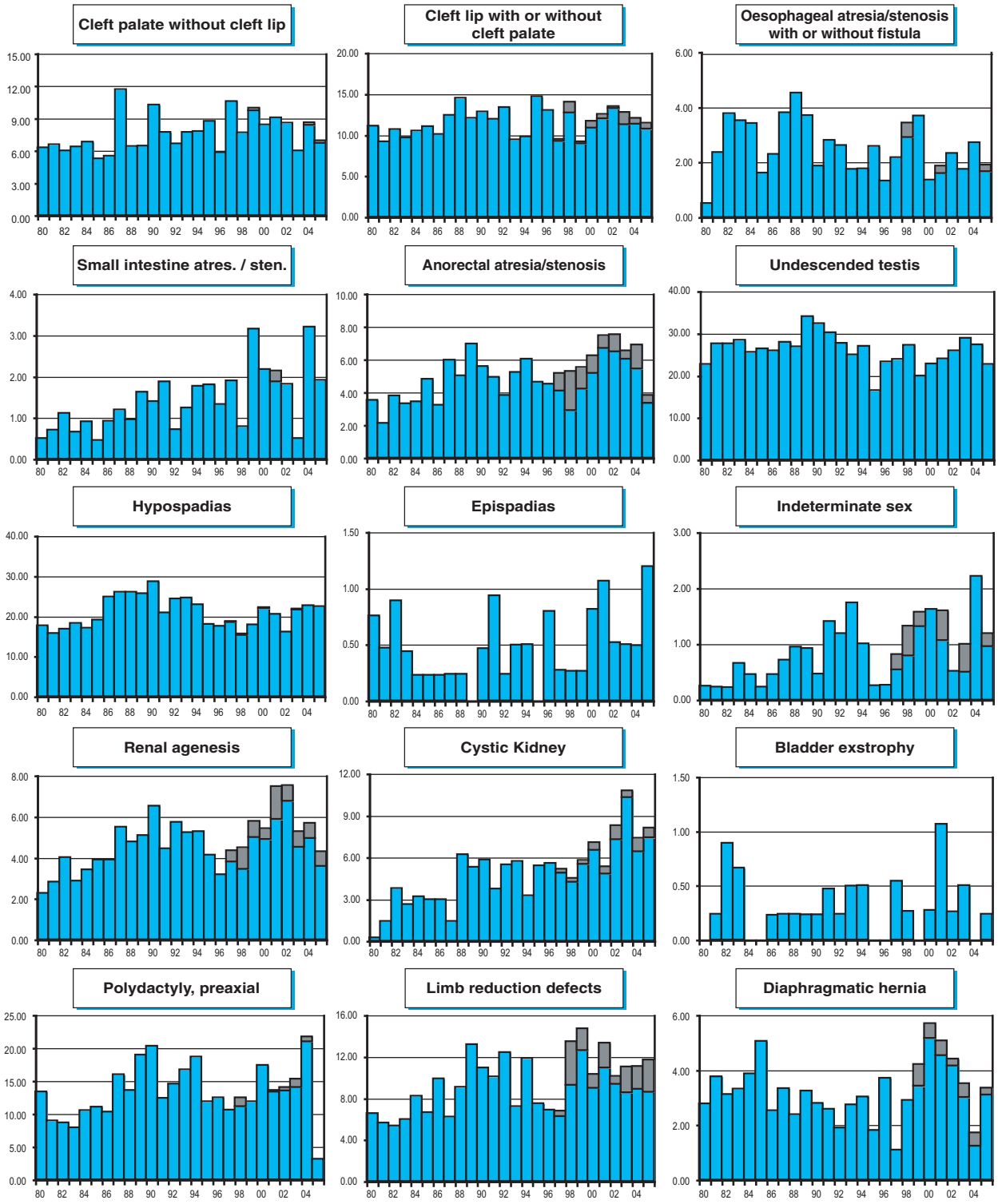
## Canada-Alberta: ACASS

'Time trends 1980-2005 (Birth prevalence rates per 10,000)



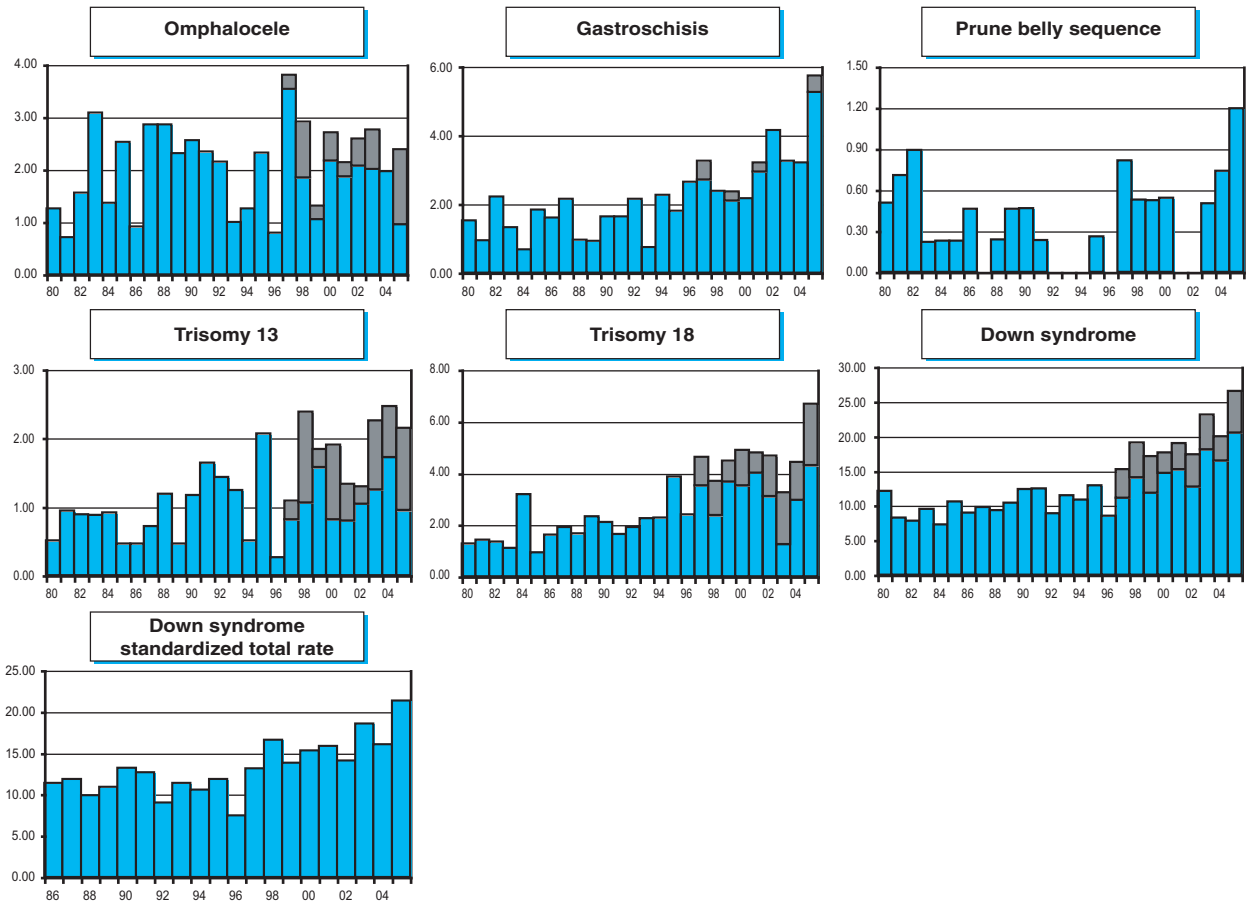

**Note:** L+S rates, ToP rates

## Canada-Alberta: ACASS




**Note:** L+S rates, ToP rates

## Canada-Alberta: ACASS




**Note:** L+S rates, ToP rates

## Canada: British Columbia

British Columbia Health Status Registry ( BCHSR )
Congenital Anomalies Surveillance Programme

**History:**
The Programme was established in 1952 as the Crippled Children's Registry. Until 1959 the Programme had an age limit of 21, but this was removed in 1960 and the name was changed to the Registry for Handicapped Children and Adults and included all familial conditions and congenital malformations. In 1975, the Registry's name was changed to the Health Surveillance Registry as risk registers for amniocentesis, rubella, hyaline membrane disease, and fetal alcohol syndrome were added. In 1991, the Royal Commission Report on Health Care and Costs contained a recommendation that Vital Statistics should develop and maintain a registry of individuals with disabilities to assist in the development of long-range plans and to monitor the changing needs of the population. Subsequently, in September 1992, amendments to the Health Act established the legislative mandate and responsibilities for the HSR. The Registry's current name, Health Status Registry, was acquired in 1992. In order to refocus the Registry's emphasis on children, the criteria for registration of individuals with long-term physical, mental and/or emotional problems was restricted to persons under the age of 20 years old, however registration of persons with genetic conditions was not age limited. By 2000 there were approximately 215,000 records in the Registry.

**Size and coverage:**
The registry covers all births in the province approximately 45,000 births annually including stillbirths with at least 20 weeks gestation or birth weight 500 grams or more.

**Legislation and funding:**
In 1992, amendments to the Health Act established the legislative mandate and responsibilities for the BC HSR. Funding comes from the British Columbia Vital Statistics Agency.

**Sources of ascertainment:**
Sources include: Notice of Live and Stillbirth, Death registrations, Hospital Admission/Discharge Abstracts, Children's Hospital, Sunnyhill Hospital, UBC and Victoria General Medical Genetics Clinics, Child Development Centres, Health Regions, the Asante Centre for Fetal Alcohol Syndrome.

**Exposure information:**
Information on complications of pregnancy, labour or delivery is available on Vital Statistics birth registrations and environmental/occupational and drug/alcohol/smoking lifestyle related information can be obtained from the death registrations for the deceased.

**Background information:**
The registry data are regularly matched to Vital Statistics birth registrations to obtain birth particulars of the registrants and maternal/paternal information, and also matched to death registrations to get the date of death and causes of death if the registered person was deceased. The registry also registers cases of medically terminated pregnancies due to congenital anomalies.

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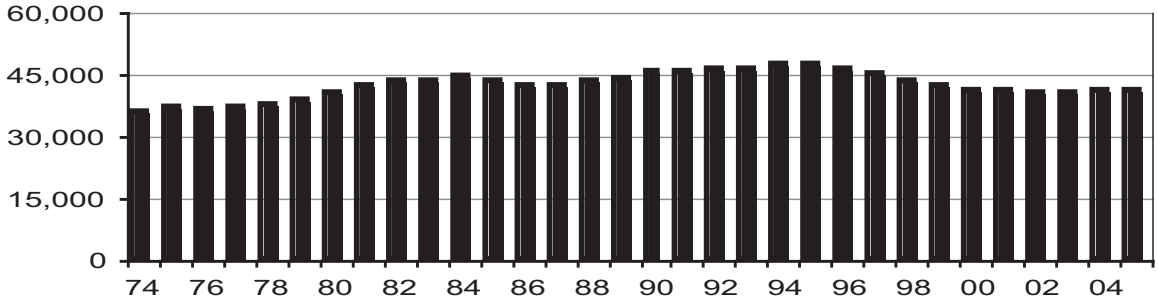
**E-mail:** Andrea.Coppard@gov.bc.ca

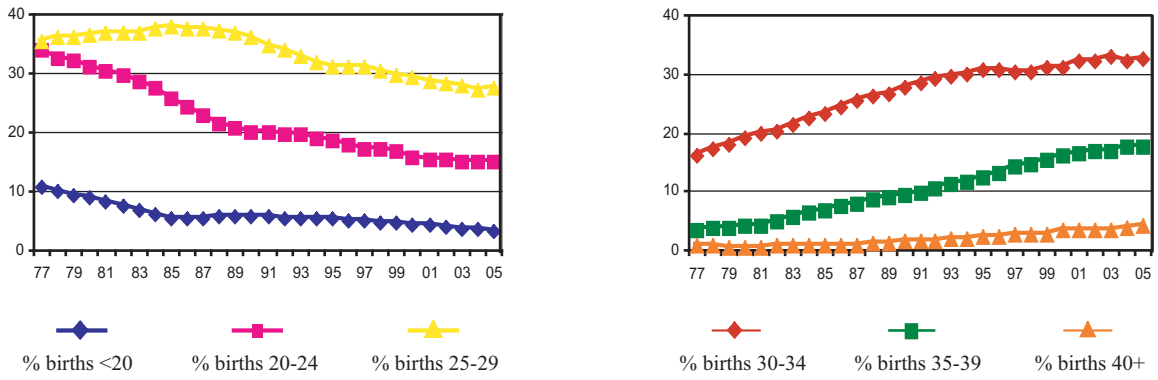
**Web:** http://www.vs.gov.bc.ca/stats/hsr/index.html

## Canada: British Columbia

**Total births by year**




**Percentage of births by year and maternal age**



# Canada: British Columbia, 2005

| | |
|---|---|
| Live births (LB) | 40,660 |
| Stillbirths (SB) | 314 |
| Total births | 40,974 |
| 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 | 5 | nr | 1.22 |
| Spina bifida | 10 | 5 | nr | 3.66 |
| Encephalocele | 2 | 1 | nr | 0.73 |
| Microcephaly | 34 | 0 | nr | 8.30 |
| Holoprosencephaly | 54 | 17 | nr | 17.33 |
| Hydrocephaly | 17 | 2 | nr | 4.64 |
| Anophthalmos | 1 | 0 | nr | 0.24 |
| Microphthalmos | 4 | 0 | nr | 0.98 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 2 | 0 | nr | 0.49 |
| Unspecified Anotia/Microtia | 4 | 0 | nr | 0.98 |
| Transposition of great vessels | 11 | 0 | nr | 2.68 |
| Tetralogy of Fallot | 18 | 0 | nr | 4.39 |
| Hypoplastic left heart syndrome | 14 | 3 | nr | 4.15 |
| Coarctation of aorta | 13 | 0 | nr | 3.17 |
| Choanal atresia, bilateral | 4 | 0 | nr | 0.98 |
| Cleft palate without cleft lip | 40 | 0 | nr | 9.76 |
| Cleft lip with or without cleft palate | 18 | 4 | nr | 5.37 |
| Oesophageal atresia / stenosis with or without fistula | 5 | 0 | nr | 1.22 |
| Small intestine atresia / stenosis | 17 | 0 | nr | 4.15 |
| Anorectal atresia / stenosis | 29 | 0 | nr | 7.08 |
| Undescended testis (36 weeks of gestation or later) | 124 | 0 | nr | 30.26 |
| Hypospadias | 75 | 1 | nr | 18.55 |
| Epispadias | 1 | 0 | nr | 0.24 |
| Indeterminate sex | 0 | 0 | nr | 0.00 |
| Renal agenesis | 0 | 1 | nr | 0.24 |
| Cystic kidney | 3 | 0 | nr | 0.73 |
| Bladder exstrophy | 0 | 0 | nr | 0.00 |
| Polydactyly, preaxial | 32 | 0 | nr | 7.81 |
| Total Limb reduction defects (include unspecified) | 14 | 3 | nr | 4.15 |
| 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 | 12 | 1 | nr | 3.17 |
| Omphalocele | 9 | 3 | nr | 2.93 |
| Gastroschisis | 10 | 2 | nr | 2.93 |
| Unspecified Omphalocele/Gastroschisis | 5 | 0 | nr | 1.22 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13 | 4 | 2 | nr | 1.46 |
| Trisomy 18 | 6 | 10 | nr | 3.90 |
| Down syndrome, all ages (include age unknown) | 54 | 17 | nr | 17.33 |
| <20 | 0 | 0 | nr | 0.00 |
| 20-24 | 4 | 1 | nr | 8.08 |
| 25-29 | 8 | 2 | nr | 8.89 |
| 30-34 | 14 | 5 | nr | 14.23 |
| 35-39 | 17 | 5 | nr | 30.62 |
| 40-44 | 8 | 4 | nr | 75.28 |
| 45+ | 3 | 0 | nr | 416.67 |
| unknown | 0 | 0 | nr | --- |

nr = not reported

# Canada: British Columbia, Previous years rates 1974 - 2005

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

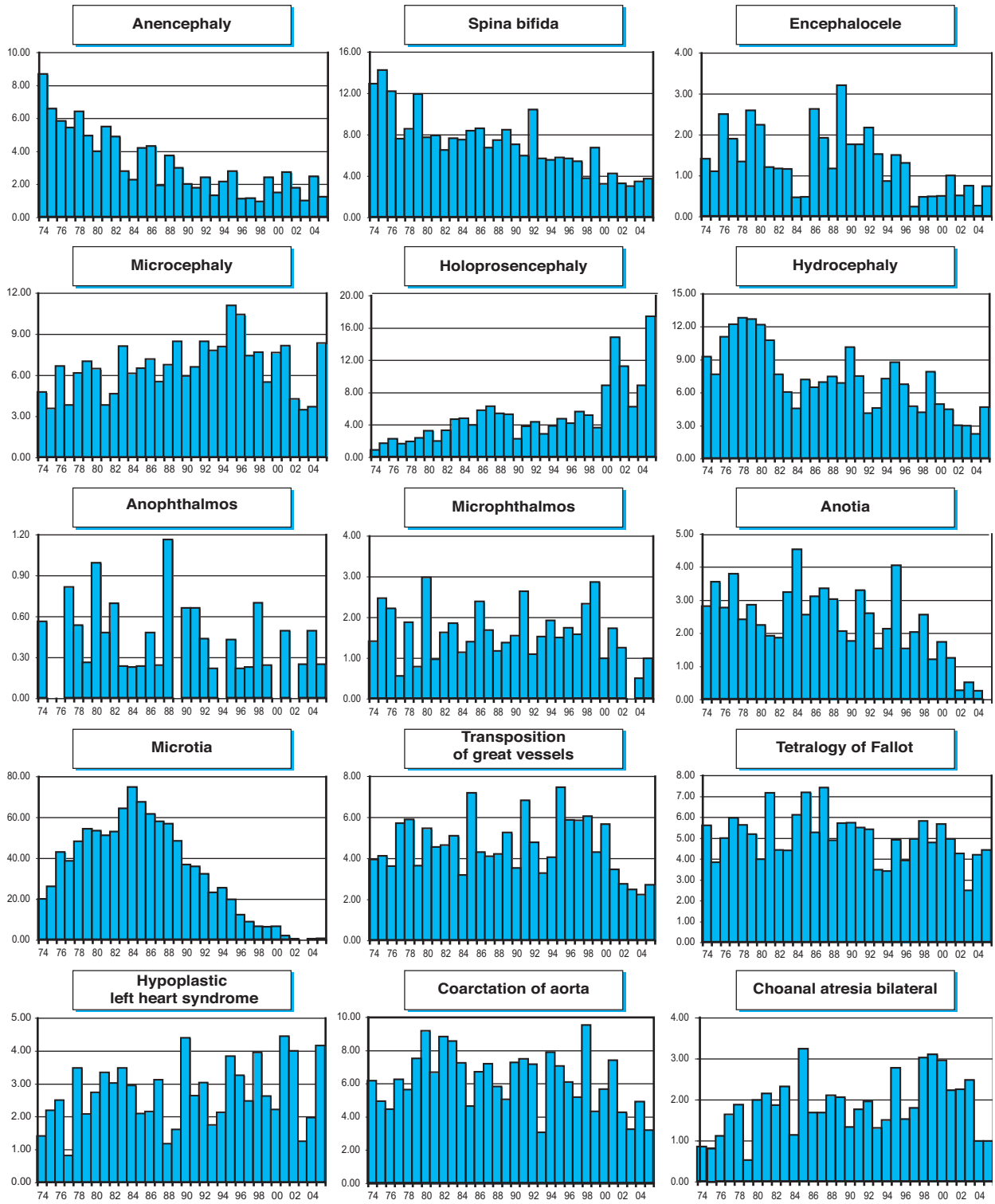
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **262,466** | **216,331** | **216,626** | **232,413** | **216,938** | **203,294** |
| Anencephaly | 5.91 | 3.88 | 2.95 | 2.07 | 1.38 | 1.82 |
| Spina bifida | 10.63 | 7.53 | 7.62 | 6.63 | 4.93 | 3.49 |
| Encephalocele | 1.87 | 0.88 | 2.12 | 1.55 | 0.60 | 0.64 |
| Microcephaly | 5.49 | 5.82 | 6.74 | 8.39 | 7.74 | 5.56 |
| Holoprosencephaly | 1.98 | 3.70 | 4.89 | 3.87 | 5.39 | 11.66 |
| Hydrocephaly | 11.13 | 7.16 | 7.57 | 6.41 | 5.67 | 3.44 |
| Anophthalmos | 0.46 | 0.37 | 0.51 | 0.34 | 0.28 | 0.30 |
| Microphthalmos | 1.75 | 1.39 | 1.62 | 1.72 | 1.89 | 0.89 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 2.90 | 2.82 | 2.63 | 2.71 | 1.80 | 0.44 |
| Microtia | 40.69 | 62.03 | 51.84 | 27.02 | 8.02 | 0.54 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 4.61 | 4.90 | 4.25 | 5.25 | 5.53 | 2.71 |
| Tetralogy of Fallot | 4.99 | 5.82 | 5.77 | 4.52 | 4.98 | 4.03 |
| Hypoplastic left heart syndrome | 2.17 | 2.96 | 2.49 | 2.67 | 2.90 | 3.15 |
| Coarctation of aorta | 6.32 | 7.16 | 6.37 | 6.50 | 6.13 | 4.57 |
| Choanal atresia, bilateral | 1.26 | 2.13 | 1.75 | 1.85 | 2.44 | 1.77 |
| Cleft palate without cleft lip | 10.48 | 11.79 | 12.97 | 13.12 | 11.06 | 7.72 |
| Cleft lip with or without cleft palate | 13.72 | 16.73 | 13.99 | 13.73 | 13.55 | 8.41 |
| Oesophageal atresia / stenosis with or without fistula | 3.31 | 4.02 | 3.37 | 3.18 | 3.32 | 2.51 |
| Small intestine atresia / stenosis | 2.44 | 3.51 | 3.19 | 3.66 | 4.52 | 3.98 |
| Anorectal atresia / stenosis | 4.95 | 4.76 | 4.15 | 5.59 | 4.98 | 5.56 |
| Undescended testis (36 weeks of gestation or later) | 73.50 | 73.78 | 73.54 | 64.71 | 54.67 | 24.84 |
| Hypospadias | 28.38 | 33.37 | 33.01 | 38.34 | 33.93 | 17.81 |
| Epispadias | 0.00 | 0.05 | 0.00 | 0.00 | 0.00 | 0.54 |
| Indeterminate sex | 1.14 | 1.39 | 0.74 | 1.25 | 1.24 | 0.05 |
| Renal agenesis | 5.03 | 6.56 | 6.51 | 6.84 | 5.99 | 0.69 |
| Cystic kidney | 3.35 | 5.18 | 5.35 | 6.32 | 7.19 | 0.59 |
| Bladder exstrophy | 0.38 | 0.51 | 0.60 | 0.43 | 0.37 | 0.39 |
| Polydactyly, preaxial | 22.94 | 21.31 | 21.51 | 22.33 | 22.13 | 8.85 |
| Total Limb reduction defects (include unspecified) | 10.17 | 8.14 | 7.48 | 6.93 | 6.18 | 2.75 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 4.42 | 4.48 | 3.46 | 3.87 | 4.19 | 3.25 |
| Omphalocele | 0.00 | 0.05 | 0.00 | 0.09 | 0.28 | 2.56 |
| Gastroschisis | 0.00 | 0.00 | 0.05 | 0.09 | 0.92 | 4.43 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.00 | 0.00 | 0.05 | 0.00 | 0.05 | 0.20 |
| Trisomy 13 | 0.57 | 0.79 | 1.34 | 0.99 | 1.29 | 1.57 |
| Trisomy 18 | 1.68 | 1.80 | 1.85 | 2.50 | 3.55 | 3.94 |
| Down syndrome, all ages (include age unknown) | 12.92 | 13.82 | 14.96 | 15.83 | 17.38 | 17.02 |
| <20 | 5.38* | 9.54 | 7.47 | 11.76 | 9.69 | 5.24 |
| 20-24 | 6.04* | 5.86 | 7.18 | 8.91 | 8.96 | 6.82 |
| 25-29 | 7.94* | 7.97 | 5.86 | 7.99 | 11.24 | 8.49 |
| 30-34 | 17.33* | 12.73 | 15.07 | 15.45 | 14.60 | 12.89 |
| 35-39 | 39.44* | 26.44 | 19.17 | 25.63 | 26.44 | 32.37 |
| 40-44 | 128.37* | 82.64 | 71.14 | 70.06 | 62.16 | 73.28 |
| 45+ | 188.68* | 185.19 | 117.65 | 372.67 | 424.71 | 173.91 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 7 years

nr = not reported

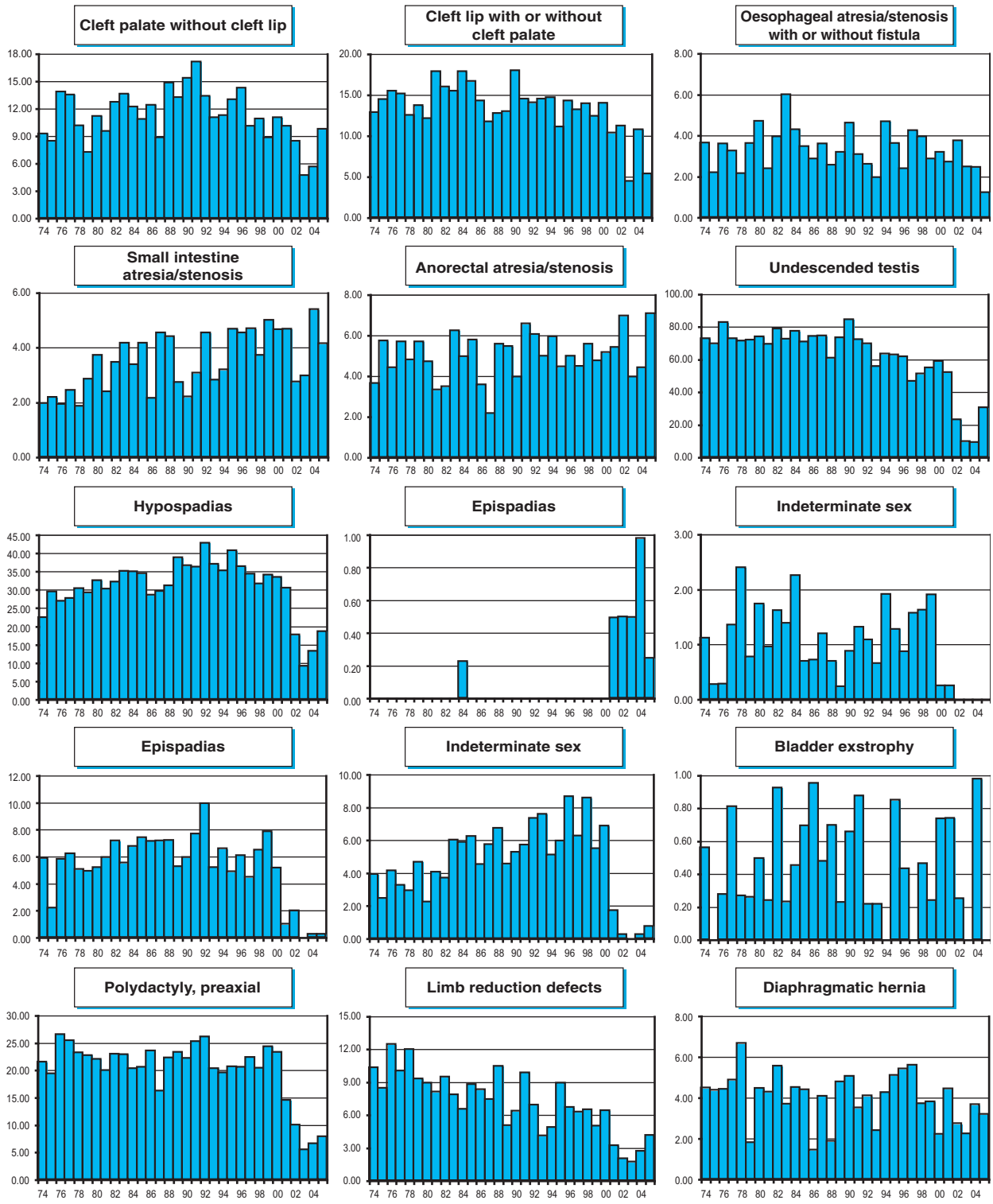
## Canada: British Columbia

Time trends 1974-2005 (Birth prevalence rates per 10,000)



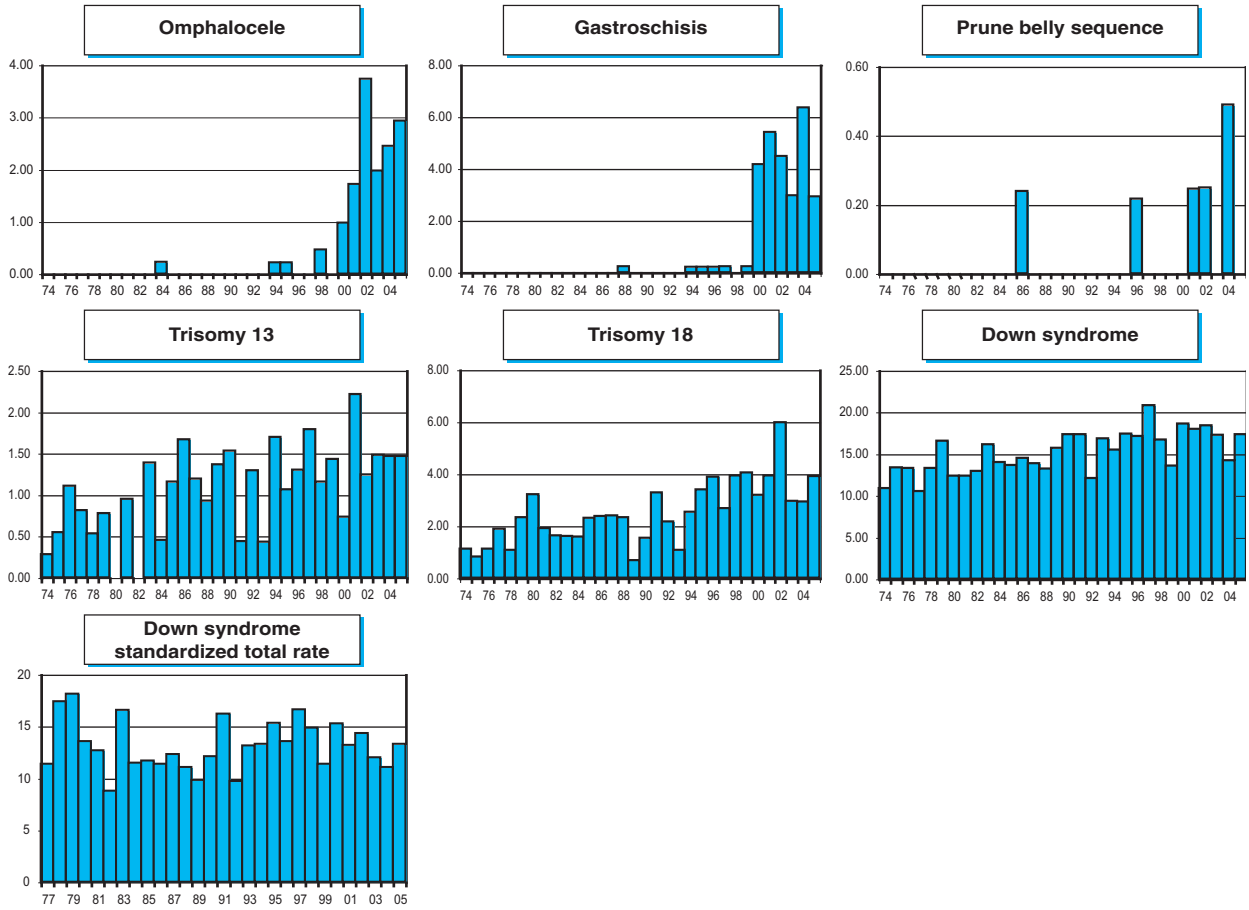

**Note:** L+S rates

## Canada: British Columbia




**Note:** L+S rates

## Canada: British Columbia




**Note:** L+S rates

## Canada: National
Canadian Congenital Anomalies Surveillance Network (CCASN)

**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 associate member status in 1996.

**Size and coverage:**
This system presently monitors about 330,000 births annually, which captures virtually all births in the 10 provinces and 3 territories of Canada. Live births to 1 year of age and registered stillbirths (a birth weight of greater or equal to 500 grams, or greater than or equal to 20 weeks in pregnancy) were captured until 2000. Since 2001, all data provided by Canadian Institute for Health Information (CIHI) only include a 30 days followup period.

**Legislation and funding:**
Reporting is based on an agreement between the Canadian Institute for Health Information (CIHI), a non-profit organization, which collects and disseminates data on hospital admission /separation in Canada, and the central registry, which is run and funded by the Public Health Agency of Canada. The Alberta Congenital Anomalies Surveillance System and Med-Echo (Système de maintenance et d'exploitation des données pour l'étude de la clientèle hospitalière) for the province of Québec provide their data separately.

**Sources of ascertainment:**
Cases from most provinces and territories are ascertained from hospital admission/separation summary records collected by the Canadian Institute for Health Information (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 Canadian Institute for Health Information (CIHI) only include a 30 days followup period.

**Exposure information:**
No exposure information is routinely collected in the central registry.

**Background information:**
Background information is based on hospital admission/separation summary records from the Canadian Institute for Health Information (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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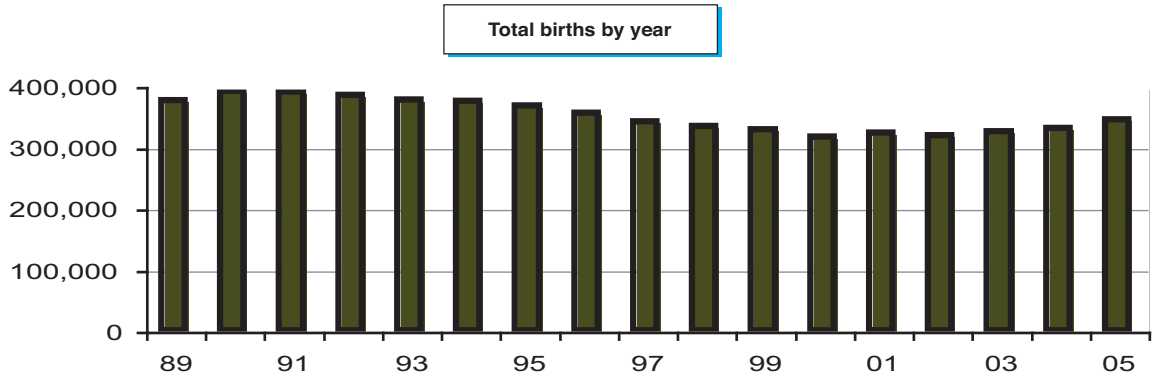
## Canada: National



# Canada: National, 2005

| | |
|---|---|
| Live births (LB) | 342,805 |
| Stillbirths (SB) | 2,094 |
| Total births | 344,899 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 17 | 16 | nr | 0.96 |
| Spina bifida | 86 | 20 | nr | 3.07 |
| Encephalocele | 21 | 3 | nr | 0.70 |
| Microcephaly | 169 | 1 | nr | 4.93 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | 144 | 15 | nr | 4.61 |
| Anophthalmos | 20 | 1 | nr | 0.61 |
| Microphthalmos | 10 | 0 | nr | 0.29 |
| 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 | 181 | 3 | nr | 5.33 |
| Tetralogy of Fallot | 148 | 3 | nr | 4.38 |
| Hypoplastic left heart syndrome | 88 | 7 | nr | 2.75 |
| Coarctation of aorta | 167 | 0 | nr | 4.84 |
| Choanal atresia, bilateral | 72 | 0 | nr | 2.09 |
| Cleft palate without cleft lip | 248 | 1 | nr | 7.22 |
| Cleft lip with or without cleft palate | 300 | 8 | nr | 8.93 |
| Oesophageal atresia / stenosis with or without fistula | 97 | 0 | nr | 2.81 |
| Small intestine atresia / stenosis | 120 | 3 | nr | 3.57 |
| Anorectal atresia / stenosis | 139 | 1 | nr | 4.06 |
| Undescended testis (36 weeks of gestation or later) | 1257 | 0 | nr | 36.45 |
| Hypospadias** | 963 | 1 | nr | 27.95 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | 39 | 0 | nr | 1.13 |
| Renal agenesis | 163 | 22 | nr | 5.36 |
| Cystic kidney | 218 | 16 | nr | 6.78 |
| Bladder exstrophy | 13 | 0 | nr | 0.38 |
| Polydactyly, preaxial | 488 | 4 | nr | 14.27 |
| Total Limb reduction defects (include unspecified) | 123 | 6 | nr | 3.74 |
| 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 | 123 | 5 | nr | 3.71 |
| Omphalocele*** | 201 | 11 | nr | 6.15 |
| Gastroschisis | nr | nr | nr | nr |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 29 | 12 | nr | 1.19 |
| Trisomy 18 | 54 | 44 | nr | 2.84 |
| Down syndrome, all ages (include age unknown) | 462 | 56 | nr | 15.02 |
| <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 |

** = include Epispadias
*** = include Gastroschisis and Unspecified Omphalocele/Gastroschisis
nr = not reported

# Canada: National, Previous years rates 1989 - 2005

Prevalence rates: (LB+SB) * 10,000

| | 1974-1980 | 1981-19851986-1990* | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|
| **Births** | | **766,679** | **1,895,384** | **1,730,578** | **1,691,365** |
| Anencephaly | | 2.19 | 1.81 | 1.13 | 1.01 |
| Spina bifida | | 7.94 | 6.58 | 4.56 | 3.06 |
| Encephalocele | | 1.32 | 1.37 | 0.88 | 0.64 |
| Microcephaly | | 6.07 | 5.08 | 5.70 | 5.04 |
| Holoprosencephaly | | nr | nr | nr | nr |
| Hydrocephaly | | 7.40 | 6.86 | 6.85 | 5.04 |
| Anophthalmos | | 0.30 | 0.32 | 0.26 | 0.59 |
| Microphthalmos | | 1.04 | 0.98 | 1.05 | 0.40 |
| Unspecified Anophthalmos / Microphthalmos | | --- | --- | --- | --- |
| Anotia | | nr | nr | nr | nr |
| Microtia | | nr | nr | nr | nr |
| Unspecified Anotia / Microtia | | nr | nr | nr | nr |
| Transposition of great vessels | | 4.38 | 4.76 | 5.63 | 4.93 |
| Tetralogy of Fallot | | 4.73 | 4.67 | 5.32 | 3.98 |
| Hypoplastic left heart syndrome | | 3.14 | 2.84 | 2.81 | 2.74 |
| Coarctation of aorta | | 5.40 | 5.61 | 5.93 | 5.13 |
| Choanal atresia, bilateral | | 2.06 | 2.06 | 2.64 | 2.65 |
| Cleft palate without cleft lip | | 7.07 | 6.94 | 7.62 | 7.11 |
| Cleft lip with or without cleft palate | | 11.50 | 11.14 | 10.92 | 9.39 |
| Oesophageal atresia / stenosis with or without fistula | | 3.61 | 3.26 | 3.50 | 2.98 |
| Small intestine atresia / stenosis | | 3.56 | 3.45 | 3.64 | 3.84 |
| Anorectal atresia / stenosis | | 5.73 | 4.94 | 4.98 | 4.36 |
| Undescended testis (36 weeks of gestation or later) | | 36.26 | 33.10 | 32.50 | 36.78 |
| Hypospadias** | | 27.17 | 26.06 | 27.45 | 29.57 |
| Epispadias | | nr | nr | nr | nr |
| Indeterminate sex | | 0.81 | 0.65 | 0.69 | 1.08 |
| Renal agenesis | | 5.18 | 4.83 | 5.11 | 5.06 |
| Cystic kidney | | 4.38 | 5.18 | 6.29 | 6.92 |
| Bladder exstrophy | | 0.44 | 0.41 | 0.36 | 0.41 |
| Polydactyly, preaxial | | 12.37 | 11.57 | 12.32 | 14.13 |
| Total Limb reduction defects (include unspecified) | | 4.59 | 4.64 | 4.07 | 3.83 |
| 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 | | --- | --- | --- | --- |
| Diaphragmatic hernia | | 3.77 | 3.63 | 3.75 | 3.32 |
| Omphalocele*** | | 3.91 | 6.09 | 6.19 | 6.82 |
| Gastroschisis | | --- | --- | --- | --- |
| Unspecified Omphalocele/Gastroschisis | | --- | --- | --- | --- |
| Prune belly sequence | | nr | nr | nr | nr |
| Trisomy 13 | | 1.21 | 1.16 | 1.11 | 1.18 |
| Trisomy 18 | | 2.15 | 2.20 | 2.39 | 2.28 |
| Down syndrome, all ages (include age unknown) | | 13.34 | 12.98 | 14.04 | 14.53 |
| <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 |

* data include less than 5 years

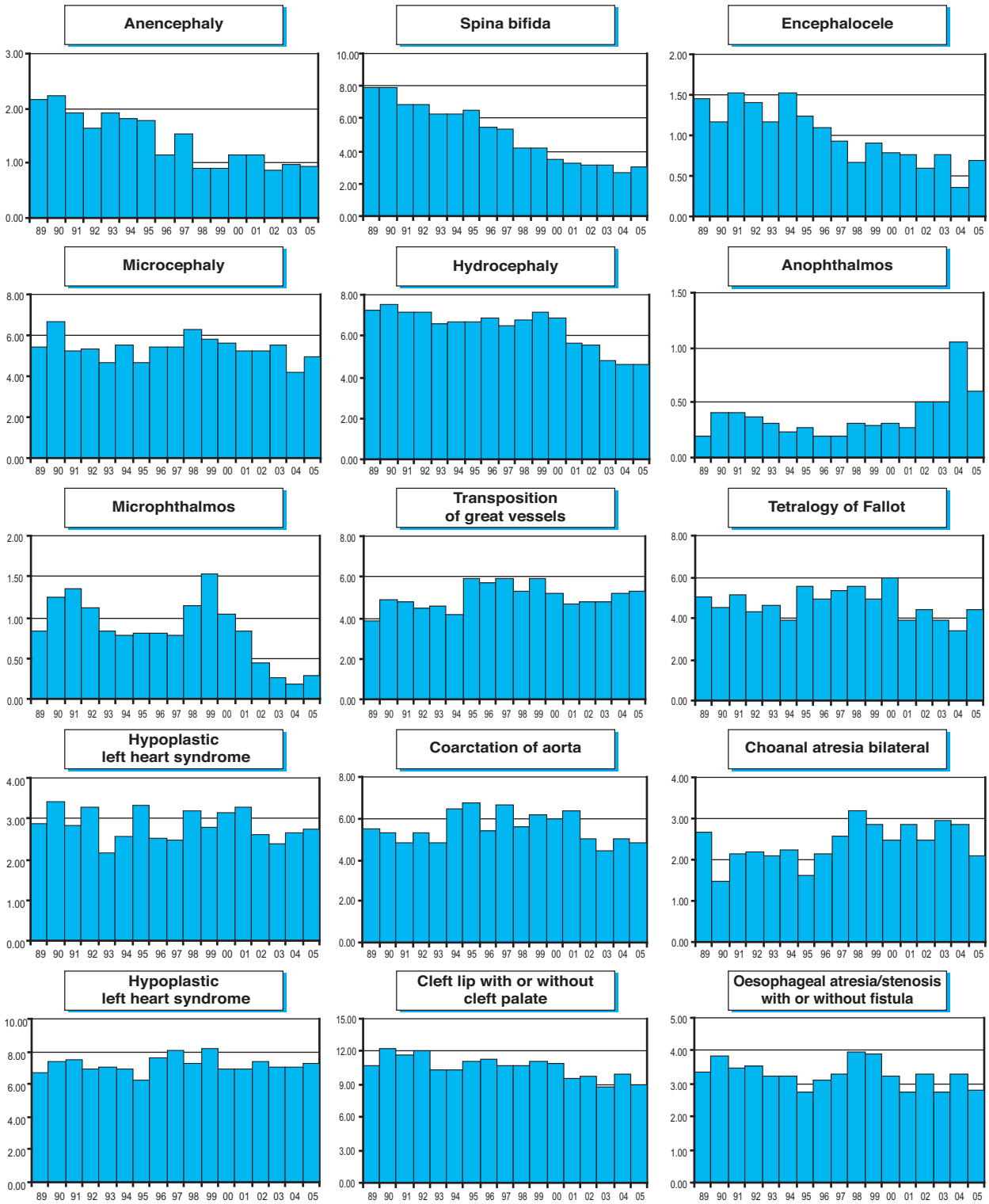
** = include Epispadias

*** = include Gastroschisis and Unspecified Omphalocele/Gastroschisis

nr = not reported

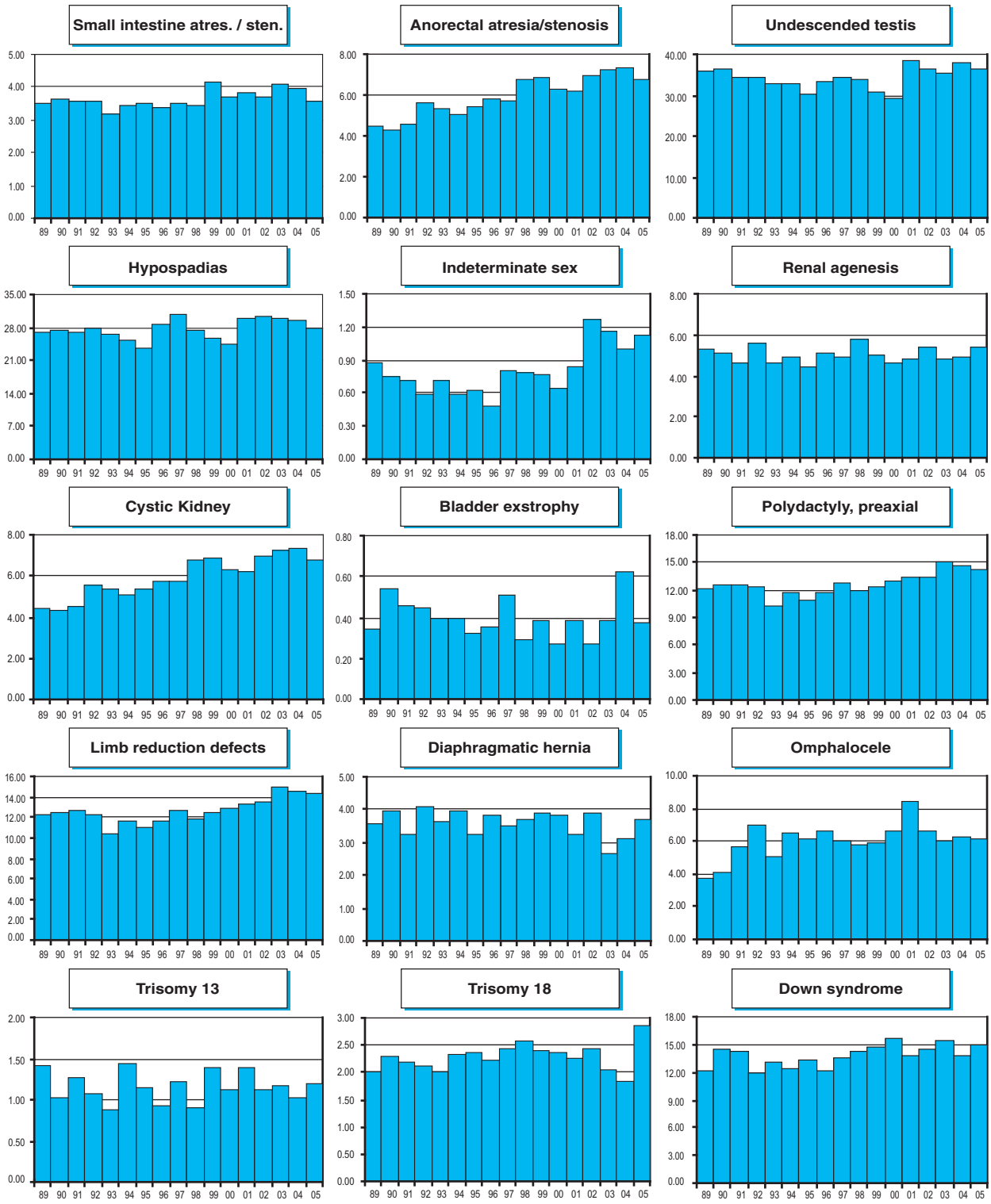
## Canada: National

Time trends 1989-2005 (Birth prevalence rates per 10,000)




**Note:** L+S rates

## Canada: National




**Note:** L+S rates

## 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 assesored for South America.ECLAMC, Eduardo Castilla. RRMC-SSM became an associated 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 34° 41' & 36° 33' S and 70° 20' & 72° 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 2005 will included the unique private maternity of the region. There are around 13.130 births annually (2005).
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 partcipating 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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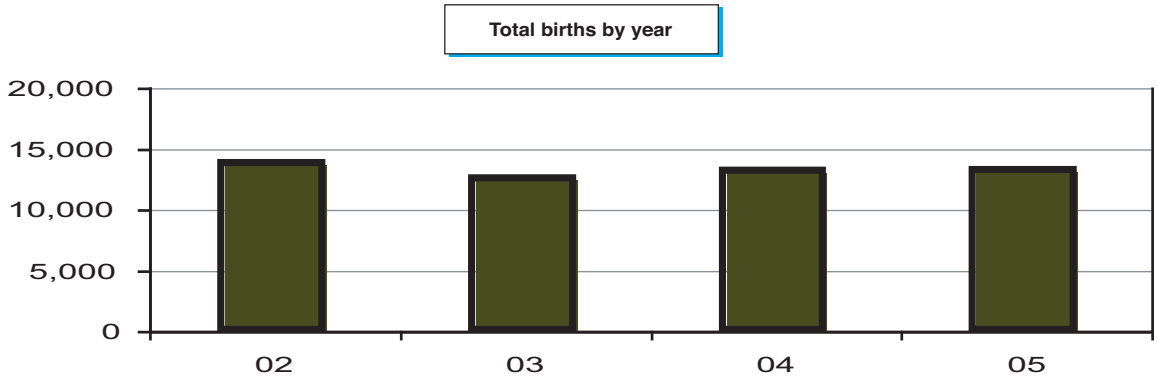
## Chile: Maule



# Chile-Maule: RRMC-SSM, 2005

| | |
|---|---|
| Live births (LB) | 13,049 |
| Stillbirths (SB) | 80 |
| Total births | 13,129 |
| 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 | 1 | 2 | | 2.29 |
| Spina bifida | 4 | 0 | | 3.05 |
| Encephalocele | 1 | 0 | | 0.76 |
| Microcephaly | 1 | 1 | | 1.52 |
| Holoprosencephaly | 0 | 0 | | 0.00 |
| Hydrocephaly | 2 | 0 | | 1.52 |
| Anophthalmos | 0 | 0 | | 0.00 |
| Microphthalmos | 1 | 0 | | 0.76 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 0 | 0 | | 0.00 |
| Microtia | 4 | 0 | | 3.05 |
| Unspecified Anotia/Microtia | 0 | 0 | | 0.00 |
| Transposition of great vessels | 1 | 0 | | 0.76 |
| Tetralogy of Fallot | 2 | 0 | | 1.52 |
| Hypoplastic left heart syndrome | 0 | 0 | | 0.00 |
| Coarctation of aorta | 0 | 0 | | 0.00 |
| Choanal atresia, bilateral | 0 | 0 | | 0.00 |
| Cleft palate without cleft lip | 4 | 0 | | 3.05 |
| Cleft lip with or without cleft palate | 12 | 0 | | 9.14 |
| Oesophageal atresia / stenosis with or without fistula | 4 | 0 | | 3.05 |
| Small intestine atresia / stenosis | 2 | 0 | | 1.52 |
| Anorectal atresia / stenosis | 2 | 0 | | 1.52 |
| Undescended testis (36 weeks of gestation or later) | 12 | 0 | | 9.14 |
| Hypospadias | 9 | 0 | | 6.86 |
| Epispadias | 1 | 0 | | 0.76 |
| Indeterminate sex | 0 | 0 | | 0.00 |
| Renal agenesis | 1 | 0 | | 0.76 |
| Cystic kidney | 0 | 1 | | 0.76 |
| Bladder exstrophy | 0 | 0 | | 0.00 |
| Polydactyly, preaxial | 8 | 0 | | 6.09 |
| Total Limb reduction defects (include unspecified) | 2 | 0 | | 1.52 |
| Transverse | 0 | 0 | | 0.00 |
| Preaxial | 2 | 0 | | 1.52 |
| Postaxial | 0 | 0 | | 0.00 |
| Intercalary | 0 | 0 | | 0.00 |
| Mixed | 0 | 0 | | 0.00 |
| Unspecified | 0 | 0 | | 0.00 |
| Diaphragmatic hernia | 0 | 1 | | 0.76 |
| Omphalocele | 1 | 1 | | 1.52 |
| Gastroschisis | 2 | 0 | | 1.52 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | | 0.00 |
| Prune belly sequence | 0 | 0 | | 0.00 |
| Trisomy 13 | 0 | 1 | | 0.76 |
| Trisomy 18 | 1 | 1 | | 1.52 |
| Down syndrome, all ages (include age unknown) | 35 | 0 | | 26.66 |
| <20 | 5 | 0 | | 21.21 |
| 20-24 | 3 | 0 | | 9.62 |
| 25-29 | 0 | 0 | | 0.00 |
| 30-34 | 4 | 0 | | 15.66 |
| 35-39 | 14 | 0 | | 92.65 |
| 40-44 | 7 | 0 | | 155.21 |
| 45+ | 2 | 0 | | 1000.00 |
| unknown | 0 | 0 | | 0.00 |

# Chile-Maule: RRMC-SSM, Previous years rates 2002 - 2005

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

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005* |
|---|---|---|---|---|---|---|
| **Total births** | | | | | | **52,358** |
| Anencephaly | | | | | | 3.06 |
| Spina bifida | | | | | | 3.06 |
| Encephalocele | | | | | | 1.15 |
| Microcephaly | | | | | | 1.72 |
| Holoprosencephaly | | | | | | 0.38 |
| Hydrocephaly | | | | | | 2.10 |
| Anophthalmos | | | | | | 0.19 |
| Microphthalmos | | | | | | 0.95 |
| Unspecified Anophthalmos / Microphthalmos | | | | | | --- |
| Anotia | | | | | | 0.38 |
| Microtia | | | | | | 2.10 |
| Unspecified Anotia / Microtia | | | | | | --- |
| Transposition of great vessels | | | | | | 1.53 |
| Tetralogy of Fallot | | | | | | 1.34 |
| Hypoplastic left heart syndrome | | | | | | 0.19 |
| Coarctation of aorta | | | | | | 0.00 |
| Choanal atresia, bilateral | | | | | | 0.76 |
| Cleft palate without cleft lip | | | | | | 4.77 |
| Cleft lip with or without cleft palate | | | | | | 10.70 |
| Oesophageal atresia / stenosis with or without fistula | | | | | | 1.15 |
| Small intestine atresia / stenosis | | | | | | 0.95 |
| Anorectal atresia / stenosis | | | | | | 2.10 |
| Undescended testis (36 weeks of gestation or later) | | | | | | 6.49 |
| Hypospadias | | | | | | 8.59 |
| Epispadias | | | | | | 0.19 |
| Indeterminate sex | | | | | | 0.57 |
| Renal agenesis | | | | | | 0.76 |
| Cystic kidney | | | | | | 0.38 |
| Bladder exstrophy | | | | | | 0.19 |
| Polydactyly, preaxial | | | | | | 5.35 |
| Total Limb reduction defects (include unspecified) | | | | | | 3.25 |
| Transverse | | | | | | 1.91 |
| Preaxial | | | | | | 0.38 |
| Postaxial | | | | | | 0.00 |
| Intercalary | | | | | | 0.00 |
| Mixed | | | | | | 0.00 |
| Unspecified | | | | | | --- |
| Diaphragmatic hernia | | | | | | 0.76 |
| Omphalocele | | | | | | 1.34 |
| Gastroschisis | | | | | | 1.53 |
| Unspecified Omphalocele / Gastroschisis | | | | | | --- |
| Prune belly sequence | | | | | | 0.19 |
| Trisomy 13 | | | | | | 1.53 |
| Trisomy 18 | | | | | | 0.76 |
| Down syndrome, all ages (include age unknown) | | | | | | 23.30 |
| <20 | | | | | | 6.45 |
| 20-24 | | | | | | 5.51 |
| 25-29 | | | | | | 7.90 |
| 30-34 | | | | | | 17.30 |
| 35-39 | | | | | | 70.71 |
| 40-44 | | | | | | 207.96 |
| 45+ | | | | | | 615.38 |
| unknown | | | | | | --- |

* data include less than 5 years

## China: BDSS-Beijing

Birth Defect Surveillance System in Thirty Counties of Four Provinces, People's Republic of China

**History:**
The Programme began in 1992. It became a full member of the ICBDMS in 1997.

**Size and coverage:**
This is a population based monitoring system. Reports were obtained from all hospitals and village health stations, which together cover all geographically defined population. Total number of population in these areas is around 17 millions and total number of births per year is around 150,000.

**Legislation and funding:**
Funding is from China Ministry of Health and local health authorities.

**Sources of ascertainment:**
Reports are obtained from delivery units, paediatric clinics, ultrasound departments, pathology departments and perinatal health care departments of different level hospitals, MCH institutes and village health stations in the participating counties and cities.

**Exposure information:**
Exposure information is obtained from the perinatal health care surveillance system (PHCSS) in the same areas for all women and their babies from pre-marital examination till six weeks after birth. BDSS data is linked with PHCSS data by using an ID number assigned to each woman.

**Background information:**
Background information is also obtained from PHCSS data.

**Addresses and Staff:**
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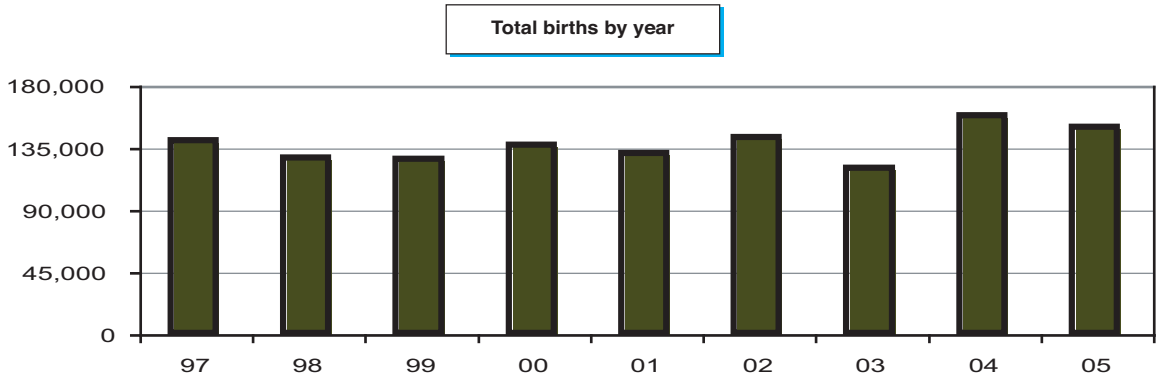
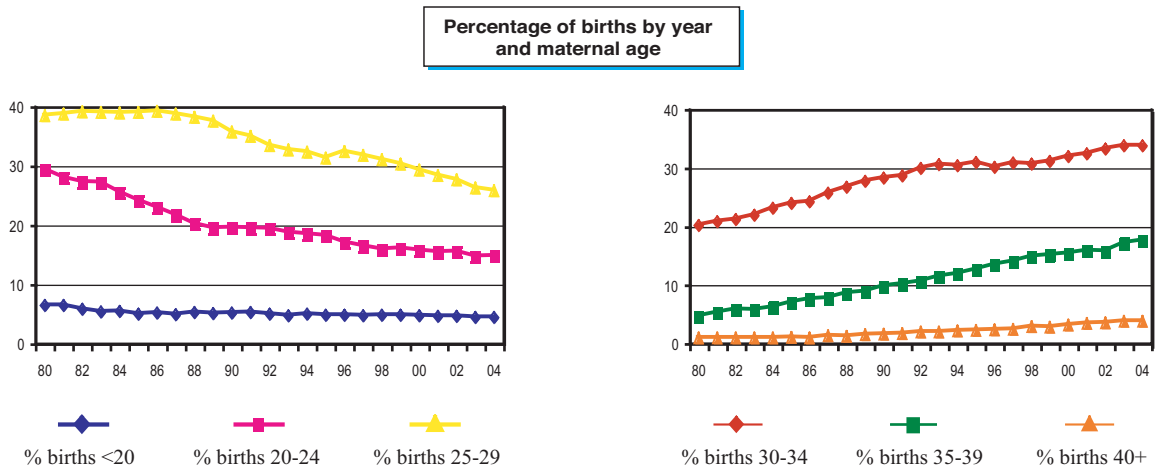
## China: BDSS-Beijing



# China: BDSS-Beijing , 2005

| | |
|---|---|
| Live births (LB) | 149,080 |
| Stillbirths (SB) | 446 |
| Total births | 149,526 |
| 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 | 35 | nr | 2.34 |
| Spina bifida | 9 | 19 | nr | 1.87 |
| Encephalocele | 3 | 10 | nr | 0.87 |
| Microcephaly | 2 | 2 | nr | 0.27 |
| Holoprosencephaly | 0 | 3 | nr | 0.20 |
| Hydrocephaly | 4 | 65 | nr | 4.61 |
| Anophthalmos | 4 | 0 | nr | 0.27 |
| Microphthalmos | 2 | 0 | nr | 0.13 |
| Unspecified Anophthalmos/ Microphthalmos | nr | nr | nr | nr |
| Anotia | 6 | 1 | nr | 0.47 |
| Microtia | 38 | 2 | nr | 2.68 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | nr | nr | nr | nr |
| Tetralogy of Fallot | nr | nr | nr | nr |
| Hypoplastic left heart syndrome | nr | nr | nr | nr |
| Coarctation of aorta | nr | nr | nr | nr |
| Choanal atresia, bilateral | nr | nr | nr | nr |
| Cleft palate without cleft lip | 28 | 1 | nr | 1.94 |
| Cleft lip with or without cleft palate | 92 | 43 | nr | 9.03 |
| Oesophageal atresia / stenosis with or without fistula | nr | nr | nr | nr |
| Small intestine atresia / stenosis | nr | nr | nr | nr |
| Anorectal atresia / stenosis | 20 | 0 | nr | 1.34 |
| Undescended testis (36 weeks of gestation or later) | 2 | 0 | nr | 0.13 |
| Hypospadias | 8 | 0 | nr | 0.54 |
| Epispadias | 0 | 0 | nr | 0.00 |
| Indeterminate sex | 4 | 1 | nr | 0.33 |
| Renal agenesis | nr | nr | nr | nr |
| Cystic kidney | nr | nr | nr | nr |
| Bladder exstrophy | 0 | 0 | nr | 0.00 |
| Polydactyly, preaxial | 77 | 1 | nr | 5.22 |
| Total Limb reduction defects (include unspecified) | 25 | 8 | nr | 2.21 |
| Transverse | 21 | 5 | nr | 1.74 |
| Preaxial | 1 | 2 | nr | 0.20 |
| Postaxial | 0 | 1 | nr | 0.07 |
| Intercalary | 3 | 0 | nr | 0.20 |
| Mixed | 0 | 0 | nr | 0.00 |
| Unspecified | nr | nr | nr | nr |
| Diaphragmatic hernia | nr | nr | nr | nr |
| Omphalocele | 3 | 10 | nr | 0.87 |
| Gastroschisis | 7 | 7 | nr | 0.94 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | 0 | 7 | nr | 0.47 |
| Trisomy 13 | nr | nr | nr | nr |
| Trisomy 18 | nr | nr | nr | nr |
| Down syndrome, all ages (include age unknown) | nr | nr | nr | nr |
| <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 = not reported

# China: BDSS-Beijing, Previous years rates 1997 - 2005

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

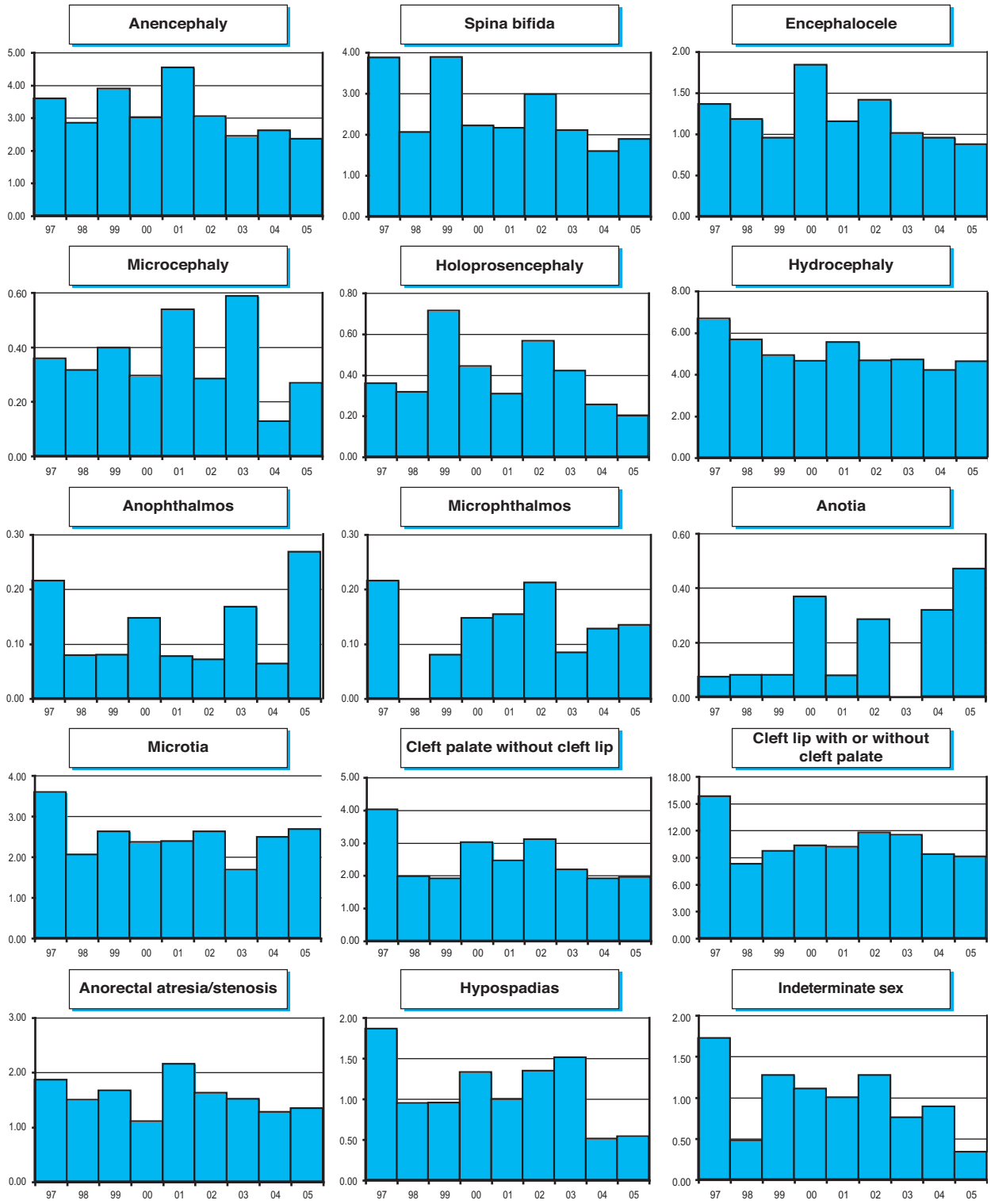
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-19951996-2000* | 2001-2005 |
|---|---|---|---|---|---|
| **Total births** | | | | **529,573** | **698,969** |
| Anencephaly | | | | 3.32 | 2.96 |
| Spina bifida | | | | 3.00 | 2.12 |
| Encephalocele | | | | 1.34 | 1.07 |
| Microcephaly | | | | 0.34 | 0.34 |
| Holoprosencephaly | | | | 0.45 | 0.34 |
| Hydrocephaly | | | | 5.48 | 4.71 |
| Anophthalmos | | | | 0.13 | 0.13 |
| Microphthalmos | | | | 0.11 | 0.14 |
| Unspecified Anophthalmos / Microphthalmos | | | | --- | --- |
| Anotia | | | | 0.15 | 0.24 |
| Microtia | | | | 2.66 | 2.39 |
| Unspecified Anotia / Microtia | | | | --- | --- |
| Transposition of great vessels | | | | nr | nr |
| Tetralogy of Fallot | | | | nr | nr |
| Hypoplastic left heart syndrome | | | | nr | nr |
| Coarctation of aorta | | | | nr | nr |
| Choanal atresia, bilateral | | | | nr | nr |
| Cleft palate without cleft lip | | | | 2.76 | 2.30 |
| Cleft lip with or without cleft palate | | | | 11.08 | 10.26 |
| Oesophageal atresia / stenosis with or without fistula | | | | nr | nr |
| Small intestine atresia / stenosis | | | | nr | nr |
| Anorectal atresia / stenosis | | | | 1.53 | 1.56 |
| Undescended testis (36 weeks of gestation or later) | | | | 0.21 | 0.24 |
| Hypospadias | | | | 1.28 | 0.94 |
| Epispadias | | | | 0.00 | 0.03 |
| Indeterminate sex | | | | 1.15 | 0.84 |
| Renal agenesis | | | | nr | nr |
| Cystic kidney | | | | nr | nr |
| Bladder exstrophy | | | | 0.04 | 0.03 |
| Polydactyly, preaxial | | | | 6.82 | 5.71 |
| Total Limb reduction defects (include unspecified) | | | | 2.61 | 2.36 |
| Transverse | | | | 1.72 | 1.85 |
| Preaxial | | | | 0.41 | 0.34 |
| Postaxial | | | | 0.00 | 0.01 |
| Intercalary | | | | 0.03 | 0.09 |
| Mixed | | | | 0.05 | 0.04 |
| Unspecified | | | | --- | --- |
| Diaphragmatic hernia | | | | nr | nr |
| Omphalocele | | | | 1.02 | 0.90 |
| Gastroschisis | | | | 1.64 | 1.77 |
| Unspecified Omphalocele / Gastroschisis | | | | --- | --- |
| Prune belly sequence | | | | 1.53 | 0.64 |
| Trisomy 13 | | | | nr | nr |
| Trisomy 18 | | | | nr | nr |
| Down syndrome, all ages (include age unknown) | | | | nr | nr |
| <20 | | | | nr | nr |
| 20-24 | | | | nr | nr |
| 25-29 | | | | nr | nr |
| 30-34 | | | | nr | nr |
| 35-39 | | | | nr | nr |
| 40-44 | | | | nr | nr |
| 45+ | | | | nr | nr |
| unknown | | | | --- | --- |

* data include less than 5 years

nr = not reported

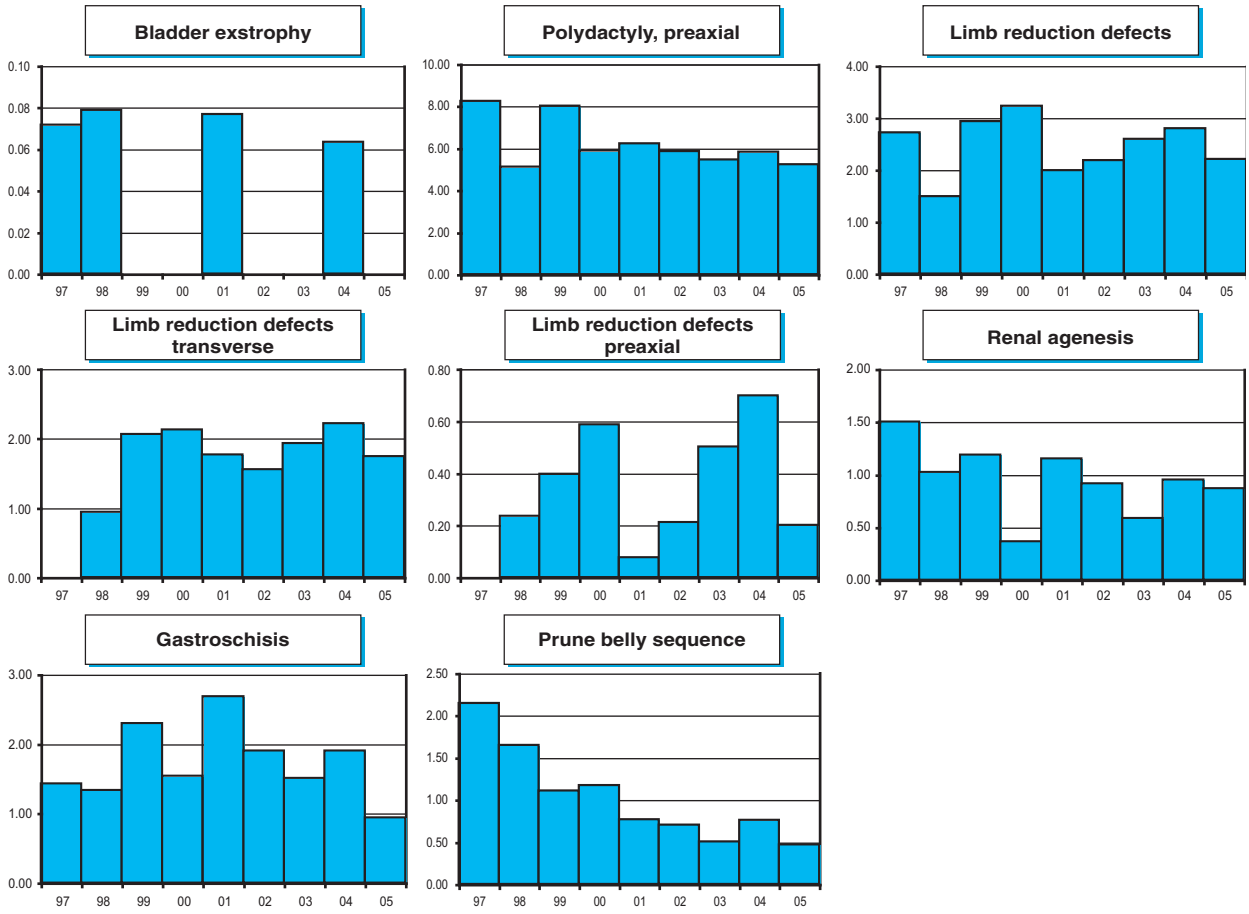
## China: BDSS-Beijing

Time trends 1997-2005 (Birth prevalence rates per 10,000)




**Note:** L+S rates

## China: BDSS-Beijing




**Note:** L+S rates

## China: CBDMN

Chinese Birth Defects Program of Sichuan Province, China (until 1994)
Chinese Birth Defects Monitoring Network

**History:**
The Programme began in 1984. It became an associate member of the ICBDSR in 1985 and a full member in 1987.

**Size and coverage:**
In 1984, reports were obtained from 100 hospitals but participation has increased. In 1985,205 hospitals participated. At present, the Programme covers approximately 260,000 births annually in 31 provinces.

Since we resumed reporting data, only one part of data ( 20 provinces, I remember apprising you by email several years ago) is sent to ICBD. The nationwide programme covers approximately 450,000~500,000 births annually in provinces.

**Legislation and funding:**
Participation is voluntary. Funding is mainly from local health authorities, also supported by Ministry of health.

**Sources of ascertainment:**
Reports are obtained from delivery units, paediatric clinics, and pathology departments of the participating hospitals.

**Exposure information:**
Exposure information is obtained by interviews of mothers of the reported malformed infants. No information is available on exposures in controls.

**Background information:**
Total number of births from each participating hospital is known.

**Addresses and Staff:**
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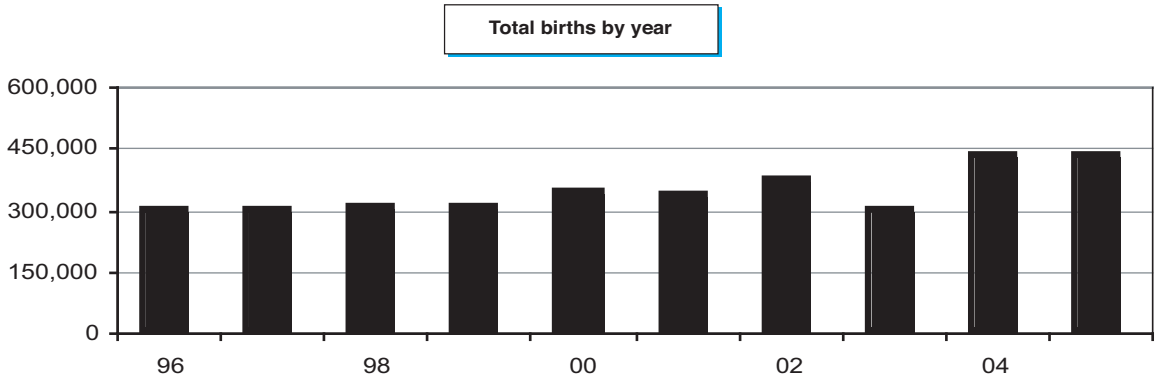
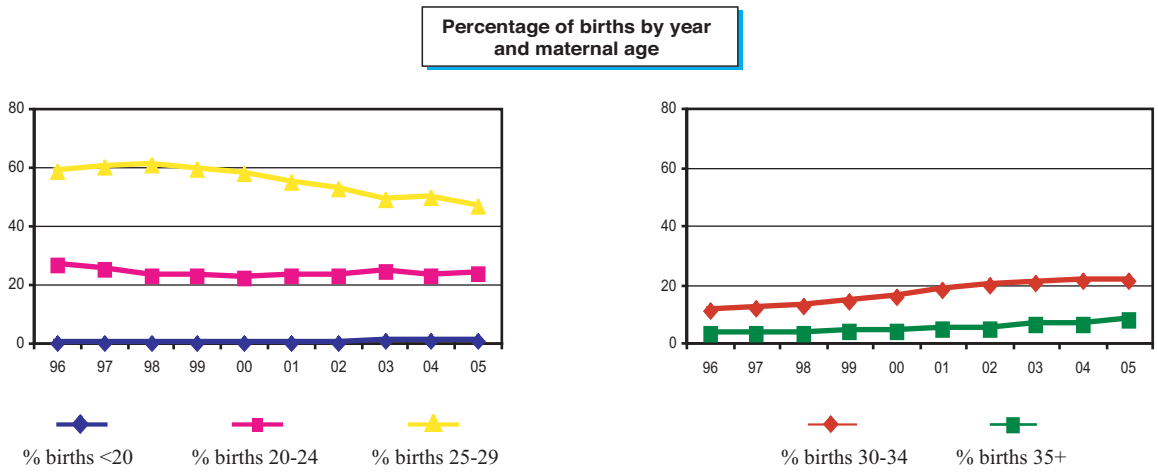
**E-mail:** cnbdms@mail.sc.cninfo.net

## China: CBDMN


Total births by year
600,000
450,000
300,000
150,000
0
96
98
00
02
04


Percentage of births by year and maternal age
80
60
40
20
0
96 97 98 99 00 01 02 03 04 05
% births <20
% births 20-24
% births 25-29
% births 30-34
% births 35+

# China: CBDMN, 2005

| | |
|---|---|
| Live births (LB) | 420,773 |
| Stillbirths (SB) | 5,733 |
| Total births | 426,506 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 11 | 108 | nr | 2.79 |
| Spina bifida | 70 | 134 | nr | 4.78 |
| Encephalocele | 23 | 40 | nr | 1.48 |
| Microcephaly | 5 | 5 | nr | 0.23 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | nr | nr | nr | nr |
| Anophthalmos* | 6 | 7 | nr | 0.30 |
| Microphthalmos | nr | nr | nr | nr |
| Unspecified Anophthalmos/ Microphthalmos | nr | nr | nr | nr |
| Anotia** | 149 | 13 | nr | 3.80 |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | nr | nr | nr | nr |
| Tetralogy of Fallot | nr | nr | nr | nr |
| Hypoplastic left heart syndrome | nr | nr | nr | nr |
| Coarctation of aorta | nr | nr | nr | nr |
| Choanal atresia, bilateral | nr | nr | nr | nr |
| Cleft palate without cleft lip | 116 | 4 | nr | 2.81 |
| Cleft lip with or without cleft palate | 405 | 158 | nr | 13.20 |
| Oesophageal atresia / stenosis with or without fistula | 32 | 15 | nr | 1.10 |
| Small intestine atresia / stenosis | nr | nr | nr | nr |
| Anorectal atresia / stenosis | 115 | 31 | nr | 3.42 |
| Undescended testis (36 weeks of gestation or later) | 55 | 1 | nr | 1.31 |
| Hypospadias | 193 | 2 | nr | 4.57 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | 26 | 18 | nr | 1.03 |
| Renal agenesis | 15 | 31 | nr | 1.08 |
| Cystic kidney | 17 | 61 | nr | 1.83 |
| Bladder exstrophy | 1 | 0 | nr | 0.02 |
| Polydactyly, unspecified | 558 | 18 | nr | 13.51 |
| Total Limb reduction defects (include unspecified) | 138 | 83 | nr | 5.18 |
| 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 | 12 | nr | 0.91 |
| Omphalocele | 33 | 25 | nr | 1.36 |
| Gastroschisis | 53 | 53 | nr | 2.49 |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | nr | nr | nr | nr |
| Trisomy 18 | nr | nr | nr | nr |
| Down syndrome, all ages (include age unknown) | 76 | 10 | nr | 2.02 |
| <20 | 0 | 0 | nr | 0.00 |
| 20-24 | 6 | 1 | nr | 0.70 |
| 25-29 | 35 | 4 | nr | 1.96 |
| 30-34 | 14 | 3 | nr | 1.86 |
| 35+ | 21 | 2 | nr | 7.05 |
| unknown | nr | nr | nr | nr |

* = include Microphthalmos and Unspecified Anophthalmos/Microphthalmos
** = include Microtia and Unspecified Anotia/Microtia
nr = not reported

# China: CBDMN, Previous years rates 1996 - 2005

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

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | | | | **1,537,995** | **1,851,341** |
| Anencephaly | | | | | 5.31 | 3.26 |
| Spina bifida | | | | | 7.50 | 6.23 |
| Encephalocele | | | | | 1.85 | 1.53 |
| Microcephaly | | | | | 0.21 | 0.23 |
| Holoprosencephaly | | | | | nr | nr |
| Hydrocephaly | | | | | 6.69 | 7.10* |
| Anophthalmos** | | | | | 0.40 | 0.30 |
| Microphthalmos | | | | | nr | nr |
| Unspecified Anophthalmos / Microphthalmos | | | | | --- | --- |
| Anotia*** | | | | | 2.97 | 3.21 |
| Microtia | | | | | nr | nr |
| Unspecified Anotia / Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | nr | nr |
| Tetralogy of Fallot | | | | | nr | nr |
| Hypoplastic left heart syndrome | | | | | nr | nr |
| Coarctation of aorta | | | | | nr | nr |
| Choanal atresia, bilateral | | | | | nr | nr |
| Cleft palate without cleft lip | | | | | 2.34 | 2.78 |
| Cleft lip with or without cleft palate | | | | | 13.99 | 14.00 |
| Oesophageal atresia / stenosis with or without fistula | | | | | 0.71 | 0.99 |
| Small intestine atresia / stenosis | | | | | nr | nr |
| Anorectal atresia / stenosis | | | | | 2.86 | 3.15 |
| Undescended testis (36 weeks of gestation or later) | | | | | 0.62 | 1.08 |
| Hypospadias | | | | | 3.45 | 4.85 |
| Epispadias | | | | | nr | nr |
| Indeterminate sex | | | | | 1.08 | 1.25* |
| Renal agenesis | | | | | 0.24* | 0.55 |
| Cystic kidney | | | | | 0.82 | 1.40 |
| Bladder exstrophy | | | | | 0.09 | 0.07 |
| Polydactyly, preaxial | | | | | nr | 13.45* |
| Total Limb reduction defects (include unspecified) | | | | | 5.29 | 5.34 |
| Transverse | | | | | nr | nr |
| Preaxial | | | | | nr | nr |
| Postaxial | | | | | nr | nr |
| Intercalary | | | | | nr | nr |
| Mixed | | | | | nr | nr |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 0.55 | 0.65 |
| Omphalocele | | | | | 1.45 | 1.46 |
| Gastroschisis | | | | | 2.73 | 2.65 |
| Unspecified Omphalocele / Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | nr | nr |
| Trisomy 13 | | | | | nr | 0.05* |
| Trisomy 18 | | | | | nr | 0.08* |
| Down syndrome, all ages (include age unknown) | | | | | 1.85 | 2.38 |
| <20 | | | | | 0.00 | 5.19 |
| 20-24 | | | | | 1.08 | 1.16 |
| 25-29 | | | | | 1.46 | 2.10 |
| 30-34 | | | | | 2.47 | 2.63 |
| 35+ | | | | | 10.67 | 8.54 |
| unknown | | | | | --- | --- |

* data include less than 5 years

** = include Microphthalmos and Unspecified Anophthalmos/Microphthalmos

*** = include Microtia and Unspecified Anotia/Microtia

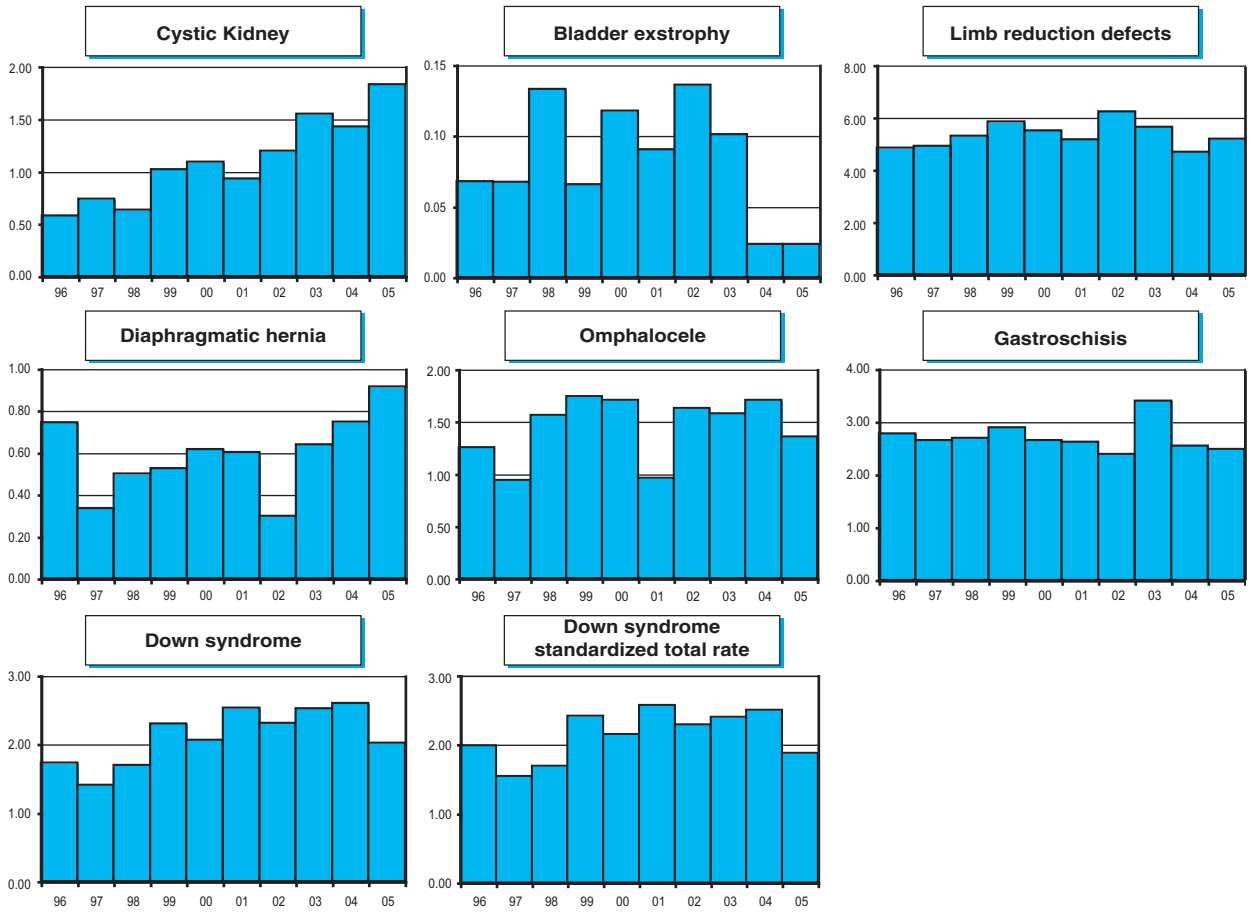
nr = not reported

## China: CBDMN

Time trends 1996-2005 (Birth prevalence rates per 10,000)


**Note:** L+S rates

## China: CBDMN




**Note:** L+S rates

## Costa Rica: CREC
Costa Rican Birth Defects Register Center

**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:**
Reporting is made by neonatologists, pediatricians and physicians before newborns discharge from maternity services, with biostatistics personal collaboration.

**Exposure information:**
None is routinely collected at present.

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

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# Costa Rica: 2005

| | |
|---|---|
| Live births (LB) | 71,020 |
| Stillbirths (SB) | 528 |
| Total births | 71,548 |
| 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 | 7 | 2 | | 1.26 |
| Spina bifida | 19 | 1 | | 2.80 |
| Encephalocele | 4 | 0 | | 0.56 |
| Microcephaly | 12 | 0 | | 1.68 |
| Holoprosencephaly | 0 | 0 | | 0.00 |
| Hydrocephaly | 7 | 2 | | 1.26 |
| Anophthalmos | 0 | 0 | | 0.00 |
| Microphthalmos | 5 | 0 | | 0.70 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 0 | 0 | | 0.00 |
| Microtia | 14 | 0 | | 1.96 |
| Unspecified Anotia/Microtia | 0 | 0 | | 0.00 |
| Transposition of great vessels | 1 | 0 | | 0.14 |
| Tetralogy of Fallot | 8 | 0 | | 1.12 |
| Hypoplastic left heart syndrome | 1 | 0 | | 0.14 |
| Coarctation of aorta | 2 | 0 | | 0.28 |
| Choanal atresia, bilateral | 1 | 1 | | 0.28 |
| Cleft palate without cleft lip | 17 | 0 | | 2.38 |
| Cleft lip with or without cleft palate | 40 | 3 | | 6.01 |
| Oesophageal atresia / stenosis with or without fistula | 16 | 1 | | 2.38 |
| Small intestine atresia / stenosis | 7 | 0 | | 0.98 |
| Anorectal atresia / stenosis | 26 | 1 | | 3.77 |
| Undescended testis (36 weeks of gestation or later) | 58 | 2 | | 8.39 |
| Hypospadias | 33 | 2 | | 4.89 |
| Epispadias | 1 | 0 | | 0.14 |
| Indeterminate sex | 11 | 2 | | 1.82 |
| Renal agenesis | 8 | 0 | | 1.12 |
| Cystic kidney | 2 | 0 | | 0.28 |
| Bladder exstrophy | 0 | 0 | | 0.00 |
| Polydactyly, preaxial | 64 | 4 | | 9.50 |
| Total Limb reduction defects (include unspecified) | 33 | 1 | | 4.75 |
| 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 | 20 | 0 | | 2.80 |
| Omphalocele | 11 | 0 | | 1.54 |
| Gastroschisis | 7 | 1 | | 1.12 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | | 0.00 |
| Prune belly sequence | 4 | 0 | | 0.56 |
| Trisomy 13 | 8 | 0 | | 1.12 |
| Trisomy 18 | 13 | 0 | | 1.82 |
| Down syndrome, all ages (include age unknown) | 56 | 2 | | 8.11 |
| <20 | 5 | 0 | | 3.51 |
| 20-24 | 6 | 0 | | 2.78 |
| 25-29 | 11 | 0 | | 63.95 |
| 30-34 | 4 | 1 | | 4.55 |
| 35-39 | 20 | 0 | | 36.54 |
| 40-44 | 10 | 1 | | 77.25 |
| 45+ | 0 | 0 | | 0.00 |
| unknown | 0 | 0 | | 0.00 |

nr = not reported

## Cuba: RECUMAC
Cuban Register of Congenital Malformation

**istory:**
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 Ministery 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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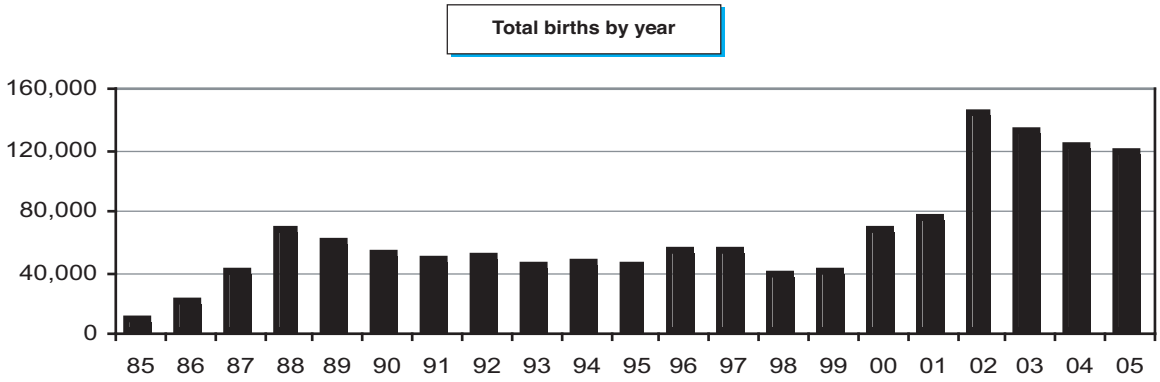
## Cuba: RECUMAC




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 160 | 98.8 | Cystic kidney | 62 | 72.1 |
| Spina bifida | 144 | 73.8 | Limb reduction defects | 25 | 33.8 |
| Encephalocele | 32 | 84.2 | Diaphragmatic hernia | 42 | 70.0 |
| Holoprosencephaly | 12 | 85.7 | Omphalocele | 62 | 84.9 |
| Hydrocephaly | 206 | 79.8 | Gastroschisis | 83 | 85.6 |
| Hypoplastic left heart syndrome | 26 | 65.0 | Trisomy 13 | 24 | 68.6 |
| Cleft palate without cleft lip | 7 | 11.3 | Trisomy 18 | 21 | 60.0 |
| Cleft lip with or without cleft palate | 26 | 16.7 | Down syndrome | 66 | 17.5 |
| Renal agenesis | 17 | 58.6 | | | |

Total ToPs with births defects = 2,078  Ratio ToPs/Births: 5.60 per 1,000 births)
*ToPs/ToPs+Births

# Cuba: 2005

| | |
|---|---|
| Live births (LB) | 116,297 |
| Stillbirths (SB) | 1,626 |
| Total births | 117,923 |
| Number of terminations of pregnancy (ToP) for birth defects | 784 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | 28 | 2.46 |
| Spina bifida | 20 | 0 | 44 | 5.43 |
| Encephalocele | 1 | 0 | 15 | 1.36 |
| Microcephaly | 4 | 0 | 7 | 0.93 |
| Holoprosencephaly | 1 | 0 | 8 | 0.76 |
| Hydrocephaly | 17 | 2 | 86 | 8.90 |
| Anophthalmos | 2 | 0 | 0 | 0.17 |
| Microphthalmos | 2 | 0 | 0 | 0.17 |
| Unspecified Anophthalmos/ Microphthalmos | 1 | 0 | 0 | 0.08 |
| Anotia | 1 | 0 | 0 | 0.08 |
| Microtia | 3 | 0 | 0 | 0.25 |
| Unspecified Anotia/Microtia | 3 | 0 | 0 | 0.25 |
| Transposition of great vessels | 15 | 2 | 11 | 2.37 |
| Tetralogy of Fallot | 12 | 1 | 5 | 1.53 |
| Hypoplastic left heart syndrome | 4 | 1 | 13 | 1.53 |
| Coarctation of aorta | 4 | 3 | 6 | 1.10 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 24 | 0 | 4 | 2.37 |
| Cleft lip with or without cleft palate | 46 | 0 | 9 | 4.66 |
| Oesophageal atresia / stenosis with or without fistula | 22 | 0 | 6 | 2.37 |
| Small intestine atresia / stenosis | 14 | 2 | 6 | 1.87 |
| Anorectal atresia / stenosis | 9 | 1 | 1 | 0.93 |
| Undescended testis (36 weeks of gestation or later) | 41 | 0 | 0 | 3.48 |
| Hypospadias | 120 | 0 | 0 | 10.18 |
| Epispadias | 4 | 0 | 0 | 0.34 |
| Indeterminate sex | 5 | 0 | 1 | 0.51 |
| Renal agenesis | 3 | 1 | 6 | 0.85 |
| Cystic kidney | 7 | 0 | 26 | 2.80 |
| Bladder exstrophy | 1 | 0 | 1 | 0.17 |
| Polydactyly, preaxial | 10 | 0 | 0 | 0.85 |
| Total Limb reduction defects (include unspecified) | 15 | 2 | 6 | 1.95 |
| Transverse | 0 | 0 | 0 | 0.00 |
| Preaxial | 0 | 0 | 0 | 0.00 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 0 | 0 | 0 | 0.00 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 15 | 2 | 6 | 1.95 |
| Diaphragmatic hernia | 11 | 2 | 16 | 2.46 |
| Omphalocele | 4 | 0 | 24 | 2.37 |
| Gastroschisis | 3 | 0 | 36 | 3.31 |
| Unspecified Omphalocele/Gastroschisis | 2 | 1 | 0 | 0.25 |
| Prune belly sequence | 0 | 0 | 1 | 0.08 |
| Trisomy 13 | 3 | 1 | 10 | 1.19 |
| Trisomy 18 | 7 | 1 | 8 | 1.36 |
| Down syndrome, all ages (include age unknown) | 98 | 0 | 31 | 10.94 |
| <20 | 14 | 0 | 2 | nr |
| 20-24 | 9 | 0 | 1 | nr |
| 25-29 | 15 | 0 | 0 | nr |
| 30-34 | 28 | 0 | 1 | nr |
| 35-39 | 18 | 0 | 13 | nr |
| 40-44 | 12 | 0 | 8 | nr |
| 45+ | 0 | 0 | 3 | nr |
| unknown | 2 | 0 | 3 | nr |

nr = not reported

# Cuba: Previous years rates 1985 - 2005

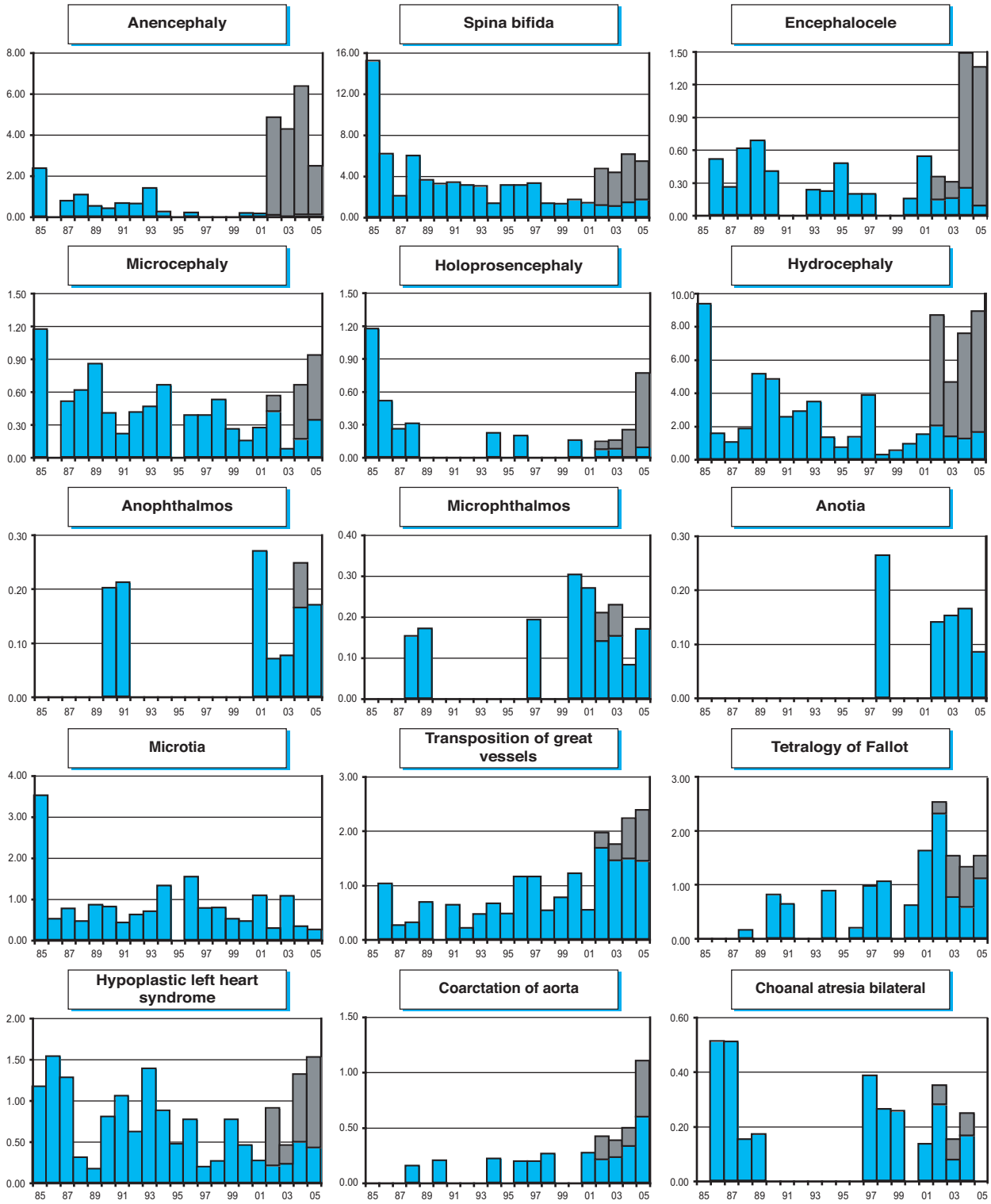
Birth prevalence rates: (LB+SB) * 10,000 until 2001
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 2002

| | 1974-1980 | 1981-1985* | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | **8,552** | **232,717** | **227,273** | **247,335** | **588,391** |
| Anencephaly | | 2.34 | 0.64 | 0.57 | 0.08 | 3.94 |
| Spina bifida | | 15.20 | 4.13 | 2.77 | 2.18 | 4.62 |
| Encephalocele | | 0.00 | 0.52 | 0.18 | 0.12 | 0.80 |
| Microcephaly | | 1.17 | 0.56 | 0.35 | 0.32 | 0.51 |
| Holoprosencephaly | | 1.17 | 0.17 | 0.04 | 0.08 | 0.27 |
| Hydrocephaly | | 9.35 | 3.14 | 2.20 | 1.46 | 6.68 |
| Anophthalmos | | 0.00 | 0.04 | 0.04 | 0.00 | 0.15 |
| Microphthalmos | | 0.00 | 0.09 | 0.00 | 0.12 | 0.19 |
| Unspecified Anophthalmos / Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 0.00 | 0.00 | 0.00 | 0.04 | 0.12 |
| Microtia | | 3.51 | 0.69 | 0.62 | 0.81 | 0.56 |
| Unspecified Anotia / Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 0.00 | 0.39 | 0.48 | 1.01 | 1.87 |
| Tetralogy of Fallot | | 0.00 | 0.21 | 0.31 | 0.57 | 1.73 |
| Hypoplastic left heart syndrome | | 1.17 | 0.64 | 0.88 | 0.49 | 0.93 |
| Coarctation of aorta | | 0.00 | 0.09 | 0.04 | 0.12 | 0.54 |
| Choanal atresia, bilateral | | 0.00 | 0.21 | 0.00 | 0.16 | 0.19 |
| Cleft palate without cleft lip | | 1.17 | 1.33 | 1.41 | 1.58 | 1.72 |
| Cleft lip with or without cleft palate | | 4.68 | 4.55 | 5.94 | 5.98 | 4.83 |
| Oesophageal atresia / stenosis with or without fistula | | 7.02 | 1.16 | 1.41 | 2.39 | 2.35 |
| Small intestine atresia / stenosis | | 0.00 | 0.73 | 0.66 | 0.81 | 1.44 |
| Anorectal atresia / stenosis | | 17.54 | 1.03 | 1.67 | 0.85 | 1.29 |
| Undescended testis (36 weeks of gestation or later) | | 0.00 | 4.51 | 4.18 | 2.43 | 2.91 |
| Hypospadias | | 0.00 | 14.87 | 12.45 | 10.67 | 8.00 |
| Epispadias | | 1.17 | 0.26 | 0.22 | 0.12 | 0.15 |
| Indeterminate sex | | 3.51 | 0.21 | 0.18 | 0.28 | 0.39 |
| Renal agenesis | | 2.34 | 0.56 | 0.22 | 0.28 | 0.78 |
| Cystic kidney | | 2.34 | 1.12 | 0.92 | 0.61 | 2.18 |
| Bladder exstrophy | | 7.02 | 0.13 | 0.13 | 0.24 | 0.10 |
| Polydactyly, preaxial | | 2.34 | 0.04 | 0.22 | 0.49 | 0.80 |
| Total Limb reduction defects (include unspecified) | | 7.02 | 2.45 | 2.90 | 2.39 | 2.24 |
| Transverse | | 2.34 | 1.07 | 0.84 | 0.69 | 0.46 |
| Preaxial | | 0.00 | 0.00 | 0.00 | 0.00 | 0.03 |
| Postaxial | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| Intercalary | | 0.00 | 0.00 | 0.00 | 0.00 | 0.10 |
| Mixed | | 0.00 | 0.00 | 0.00 | 0.00 | 0.22 |
| Unspecified | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 1.17 | 1.42 | 1.45 | 1.62 | 1.70 |
| Omphalocele | | 4.68 | 0.64 | 0.57 | 0.36 | 1.75 |
| Gastroschisis | | 0.00 | 0.39 | 0.35 | 0.44 | 2.58 |
| Unspecified Omphalocele / Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | 0.00 | 0.09 | 0.13 | 0.00 | 0.03 |
| Trisomy 13 | | 1.17 | 0.47 | 0.44 | 0.57 | 0.83 |
| Trisomy 18 | | 0.00 | 0.13 | 0.35 | 0.36 | 0.71 |
| Down syndrome, all ages (include age unknown) | | 12.86 | 7.73 | 8.05 | 7.28 | 9.82 |
| <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 | | --- | --- | --- | --- | --- |

* data include less than 5 years
nr = not reported

## Cuba: RECUMAC

'Time trends 1985-2005 (Birth prevalence rates per 10,000)

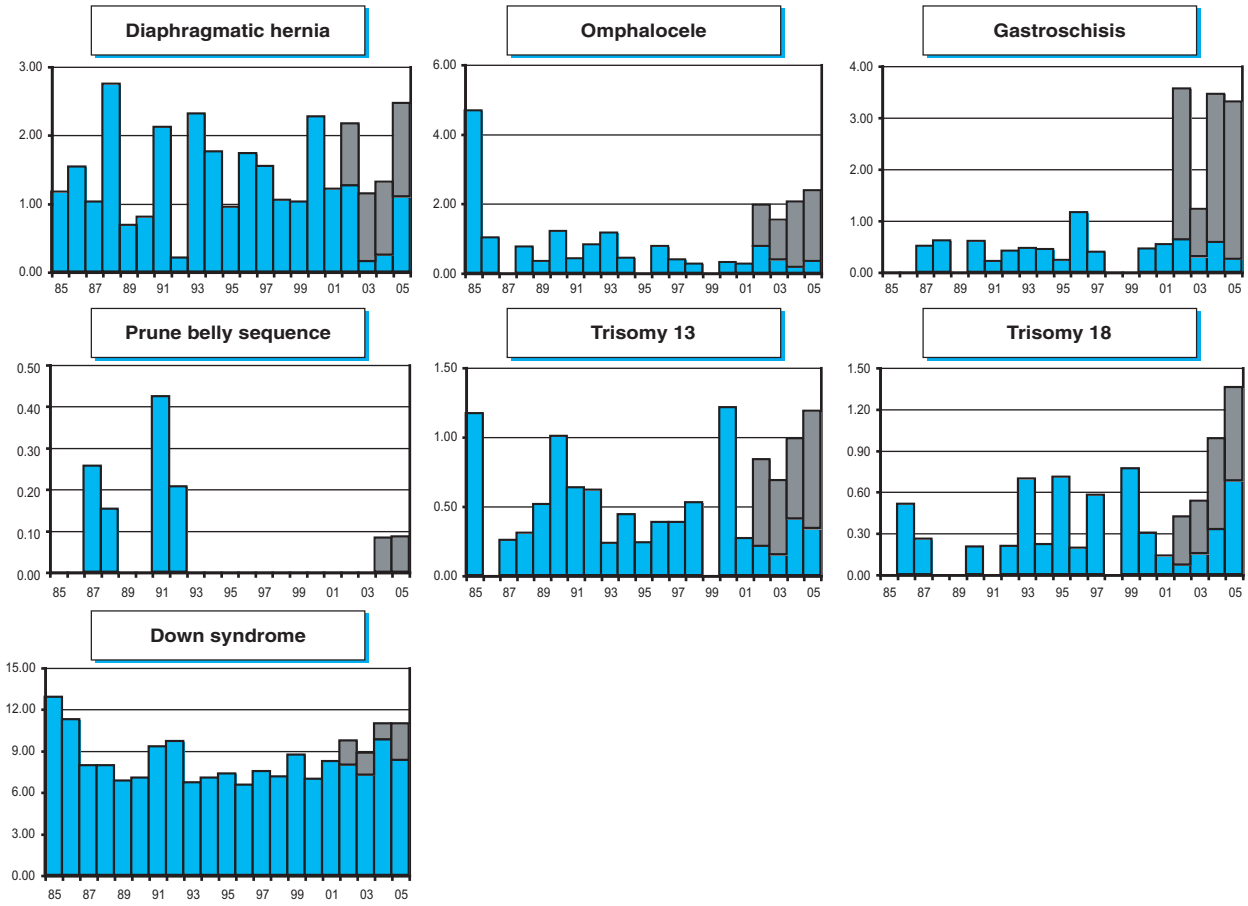



**Note:** L+S rates, ToP rates

## Cuba: RECUMAC


**Note:** L+S rates, ToP rates

## Cuba: RECUMAC




**Note:** L+S rates, ToP rates

## Czech Republic

Congenital Malformations Monitoring Program of the Czech Republic

**History:**
A registration of malformation began in 1961 and regular monitoring started in 1965. The Programme was a founding member of the Clearinghouse and is a full member.

**Size and coverage:**
All births occurring in Czech Republic (Bohemia, Moravia and Silesia regions) are covered, at present comprising about 100,000 annual births. Stillbirths weighing at least 1,000g are included.

**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.

**Sources of ascertainment:**
Reports are obtained from delivery units, neonatal, pediatric, child surgery, pathology departments, genetic departments and cytogenetic laboratories. Reporting to the central registry occurs via Regional Department of Institute of Health Information and Statistics. The bases processing of data and the analysis are performed in the Institute of Health Information and Statistics, more detailed analysis of data is performed in Department of Medical Genetics, Thomayer´s University Hospital, Prague, Czech Republic.

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

**Background information:**
Information on all births is available in the Institute of Health Information and Statistics of the Czech Republic. Selected information may be also found on the website of the programme.

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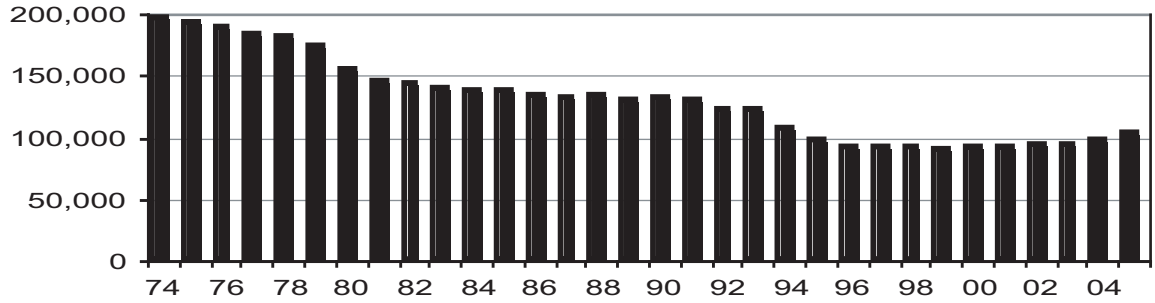
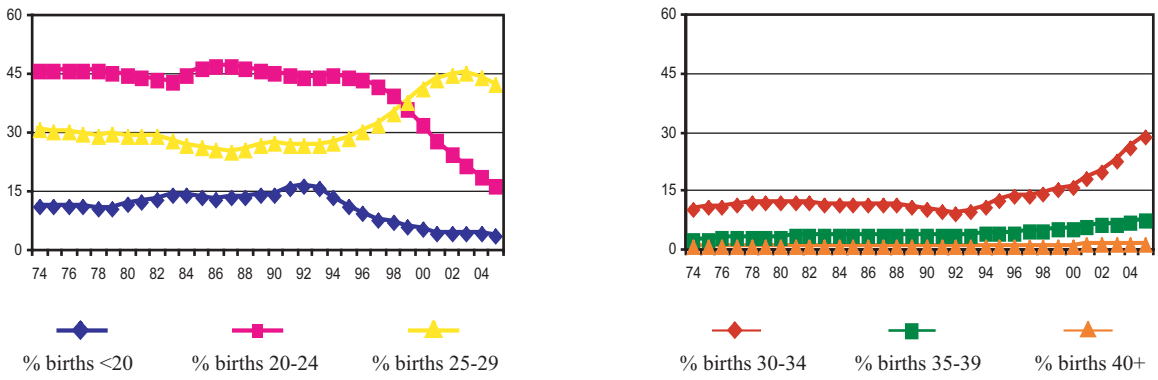
## Czech Republic

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 75 | 91.5 | Cystic kidney | 30 | 14.1 |
| Spina bifida | 77 | 70.6 | Limb reduction defects | 34 | 18.6 |
| Encephalocele | 25 | 59.5 | Diaphragmatic hernia | 18 | 23.7 |
| Holoprosencephaly | 19 | 61.3 | Omphalocele | 48 | 59.3 |
| Hydrocephaly | 71 | 48.3 | Gastroschisis | 69 | 75.0 |
| Hypoplastic left heart syndrome | 41 | 53.9 | Trisomy 13 | 43 | 67.2 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 123 | 84.2 |
| Cleft lip with or without cleft palate | 47 | 14.7 | Down syndrome | 390 | 69.9 |
| Renal agenesis | 36 | 15.7 | | | |

Total ToPs with births defects = 1,821  Ratio ToPs/Births: 6.19 per 1,000 births)
*ToPs/ToPs+Births

# Czech Republic: 2005

| | |
|---|---|
| Live births (LB) | 102,211 |
| Stillbirths (SB) | 287 |
| Total births | 102,498 |
| Number of terminations of pregnancy (ToP) for birth defects | 663 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 1 | 26 | 2.73 |
| Spina bifida | 2 | 0 | 24 | 2.54 |
| Encephalocele | 7 | 1 | 7 | 1.46 |
| Microcephaly | 22 | 3 | 1 | 2.54 |
| Holoprosencephaly | 12 | 6 | 0 | 1.76 |
| Hydrocephaly | 17 | 0 | 27 | 4.29 |
| Anophthalmos | nr | nr | nr | nr |
| Microphthalmos | nr | nr | nr | nr |
| Unspecified Anophthalmos/ Microphthalmos | 2 | 0 | 0 | 0.20 |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | 5 | 0 | 0 | 0.49 |
| Transposition of great vessels | 29 | 0 | 0 | 2.83 |
| Tetralogy of Fallot | 29 | 0 | 0 | 2.83 |
| Hypoplastic left heart syndrome | 7 | 0 | 0 | 0.68 |
| Coarctation of aorta | 42 | 0 | 0 | 4.10 |
| Choanal atresia, bilateral | nr | nr | nr | nr |
| Cleft palate without cleft lip | 70 | 0 | 0 | 6.83 |
| Cleft lip with or without cleft palate | 90 | 1 | 14 | 10.24 |
| Oesophageal atresia / stenosis with or without fistula | 30 | 1 | 0 | 3.02 |
| Small intestine atresia / stenosis | 41 | 0 | 0 | 4.00 |
| Anorectal atresia / stenosis | 35 | 0 | 0 | 3.41 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 339 | 1 | 0 | 33.17 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 69 | 0 | 6 | 7.32 |
| Cystic kidney | 77 | 0 | 12 | 8.68 |
| Bladder exstrophy | nr | nr | nr | nr |
| Polydactyly, preaxial | 135 | 0 | 0 | 13.17 |
| Total Limb reduction defects (include unspecified) | 51 | 0 | 10 | 5.95 |
| 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 | 13 | 0 | 1 | 1.37 |
| Omphalocele | 11 | 0 | 13 | 2.34 |
| Gastroschisis | 12 | 0 | 23 | 3.41 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 5 | 0 | 12 | 1.66 |
| Trisomy 18 | 7 | 0 | 52 | 5.76 |
| Down syndrome, all ages (include age unknown) | 54 | 0 | 152 | 20.10 |
| <20 | 1 | 0 | 1 | 5.34 |
| 20-24 | 8 | 0 | 8 | 9.51 |
| 25-29 | 18 | 0 | 28 | 10.61 |
| 30-34 | 12 | 0 | 38 | 16.90 |
| 35-39 | 8 | 0 | 49 | 74.47 |
| 40-44 | 3 | 0 | 26 | 236.16 |
| 45+ | 0 | 0 | 2 | 444.44 |
| unknown | 4 | 0 | 0 | 519.48 |

nr = not reported

# Czech Republic: Previous years rates 1974 - 2005

Birth prevalence rates: (LB+SB) * 10,000
Birth prevalence rates: (LB+SB+TOP) * 10,000 in different periods according to the malformation

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **1,267,450** | **699,903** | **658,624** | **575,342** | **453,465** | **478,445** |
| Anencephaly | 3.20 | 2.94 | 3.34 | 3.30 | 3.11 | 2.49 |
| Spina bifida | 3.98 | 3.87 | 4.18 | 3.30 | 4.28 | 3.64 |
| Encephalocele | 0.51 | 0.69 | 0.90 | 0.96 | 0.86 | 1.17 |
| Microcephaly | 1.02 | 1.07 | 0.97 | 0.78 | 0.77 | 1.73 |
| Holoprosencephaly | nr | nr | nr | 0.10* | 0.18 | 0.96 |
| Hydrocephaly | 2.30 | 2.99 | 3.90 | 5.14 | 3.97 | 5.25 |
| Anophthalmos | nr | nr | nr | nr | 0.04* | 0.05* |
| Microphthalmos | nr | nr | nr | nr | 0.18* | 0.32* |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | nr | 0.99* | 0.61* |
| Microtia | nr | nr | nr | nr | 0.29* | 0.53* |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 2.69 | 2.03 | 1.43 | 1.67* | 3.64 | 3.93 |
| Tetralogy of Fallot | nr | nr | nr | 2.02* | 3.18 | 3.26 |
| Hypoplastic left heart syndrome | 0.54 | 0.79 | 0.64 | 1.97* | 2.18 | 2.72 |
| Coarctation of aorta | nr | nr | nr | 3.89* | 3.62 | 4.66 |
| Choanal atresia, bilateral | nr | nr | nr | 0.25* | 0.31 | 0.27* |
| Cleft palate without cleft lip | 5.74 | 6.59 | 5.86 | 5.77 | 6.11 | 7.61 |
| Cleft lip with or without cleft palate | 9.91 | 10.37 | 10.29 | 10.59 | 9.35 | 11.68 |
| Oesophageal atresia / stenosis with or without fistula | 1.12 | 1.37 | 1.02 | 1.77 | 2.23 | 2.91 |
| Small intestine atresia / stenosis | nr | nr | nr | 1.87* | 2.12 | 3.49 |
| Anorectal atresia / stenosis | 1.36 | 1.16 | 0.62 | 2.00 | 2.87 | 3.89 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | 3.89* | 12.61 | 24.98* |
| Hypospadias | 18.60 | 20.40 | 22.36 | 24.06 | 26.40 | 32.50 |
| Epispadias | nr | nr | nr | 0.25* | 0.55 | 0.37* |
| Indeterminate sex | nr | nr | nr | 0.30* | 0.49 | 0.51* |
| Renal agenesis | 1.59 | 1.47 | 1.29 | 2.05 | 2.91 | 6.86 |
| Cystic kidney | 2.54 | 2.41 | 2.85 | 2.59 | 3.99 | 6.75 |
| Bladder exstrophy | 0.15 | 0.11 | 0.02 | 0.21* | 0.15 | 0.24* |
| Polydactyly, preaxial | nr | nr | 12.82* | 12.55 | 12.64 | 14.34 |
| Total Limb reduction defects (include unspecified) | 4.33 | 5.17 | 4.84 | 5.60 | 5.29 | 5.75 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 2.56 | 2.66 | 1.96 | 1.74 | 2.47 | 2.72 |
| Omphalocele | 2.34 | 2.14 | 2.63 | 2.10 | 2.29 | 2.74 |
| Gastroschisis | 1.06 | 1.41 | 0.99 | 1.15 | 2.87 | 3.05 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | nr | nr | nr | 0.16 |
| Trisomy 13 | nr | nr | 0.15* | 0.50 | 0.97 | 1.84 |
| Trisomy 18 | nr | nr | 0.46 | 1.51 | 3.18 | 4.62 |
| Down syndrome, all ages (include age unknown) | 8.54 | 7.66 | 8.65 | 11.19 | 14.93 | 17.85 |
| <20 | 4.65 | 5.14 | 4.08 | 5.49 | 7.41 | 5.29 |
| 20-24 | 5.61 | 4.38 | 3.37 | 5.16 | 8.79 | 7.70 |
| 25-29 | 8.61 | 6.99 | 6.00 | 8.46 | 9.69 | 11.02 |
| 30-34 | 11.73 | 8.48 | 8.59 | 12.35 | 20.99 | 18.39 |
| 35-39 | 31.77 | 29.60 | 28.69 | 45.42 | 57.02 | 67.60 |
| 40-44 | 128.08 | 74.99 | 66.29 | 226.64 | 198.62 | 200.72 |
| 45+ | 187.27 | 377.36 | 430.11 | 751.88 | 476.19 | 523.56 |
| unspecified | --- | --- | --- | --- | --- | --- |

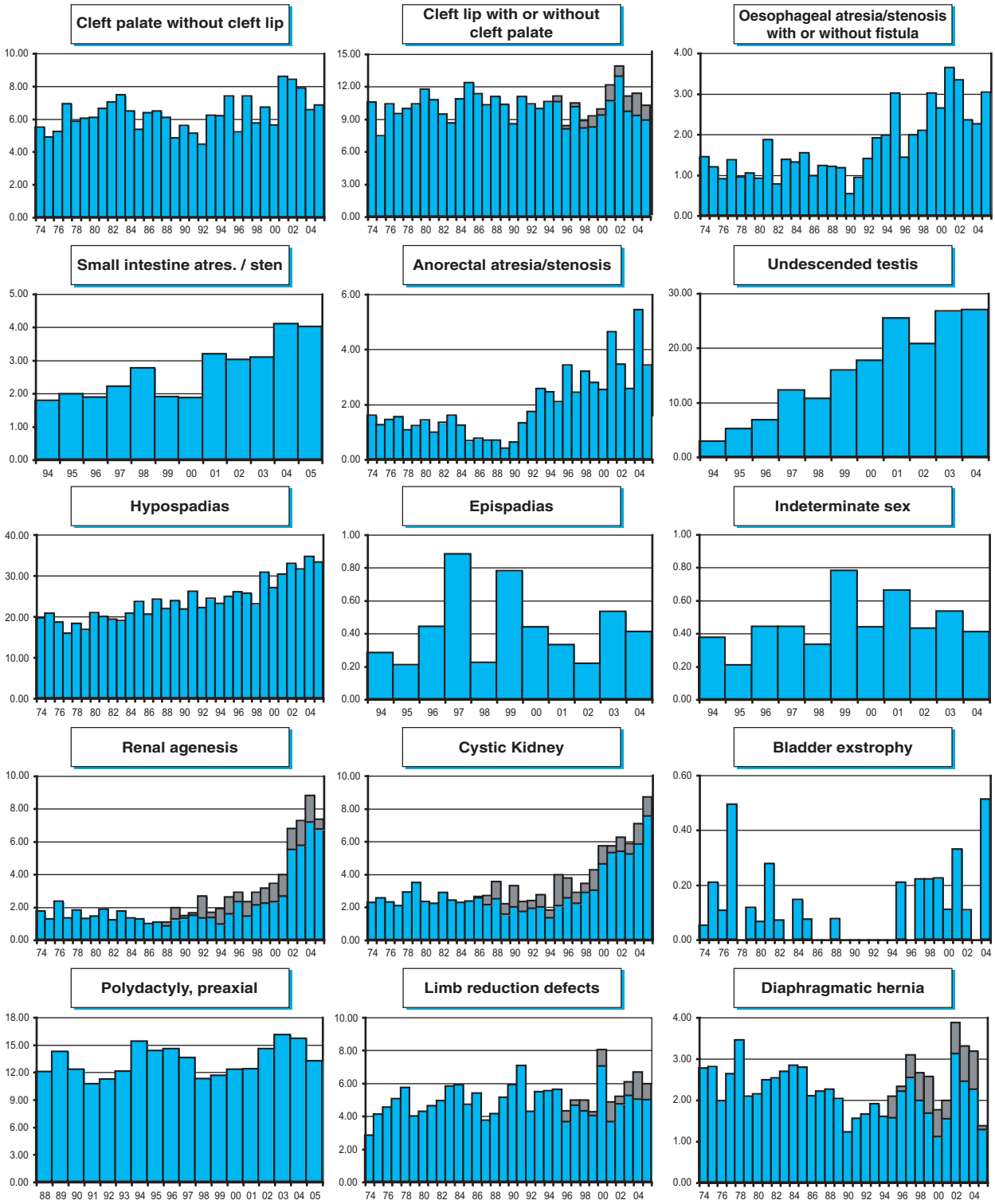
* data include less than 5 years
nr = not reported

## Czech Republic

'Time trends 1974-2005 (Birth prevalence rates per 10,000)

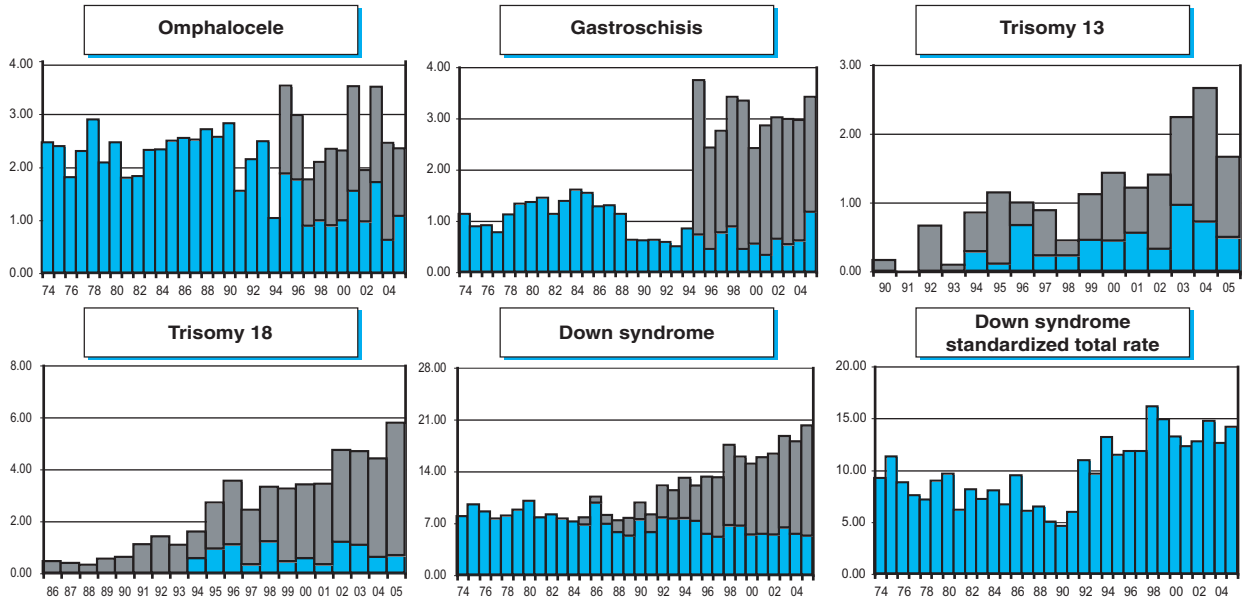

**Note:** L+S rates, ToP rates

## Czech Republic




**Note:** L+S rates, ToP rates

## Czech Republic




**Note:** L+S rates, ToP rates

## England and Wales
National Congenital Anomaly System (NCAS)

**History:**
The monitoring programme was started in 1964. It was a founding member of the Clearinghouse and is a full member.

**Size and coverage:**
All births in England and Wales are covered, at present approximately 649,000 annually. Stillbirths of 24 weeks or more gestation are registered.

**Legislation and funding:**
Reporting is voluntary. The governmental Office for National Statistics finances the National Congenital Anomaly System.

**Sources of ascertainment:**
Reports are mainly based on notifications of births prepared by attendants at birth, either physicians or midwives by means of a paper form (the SD56 form). This form contains a written description of the anomaly and details of the birth, along with some demographic information about the parents. In areas covered by local congenital anomaly registers this information is supplemented by other reports from neonatal intensive care units, special care baby units etc.

It has long been recognised, however, that there is under reporting in NCAS. Therefore NCAS has embarked on an on-going programme of improving the level of reporting to the system. Since 1998, local congenital anomaly registers have begun to provide data to NCAS in each of the years detailed below:

| | |
|---|---|
| 1998 | CARIS (Wales) |
| 1999 | East Midlands & South Yorkshire Congenital Anomaly Register |
| 2000 | North Thames (West) Congenital Malformation Register |
| 2000 | Merseyside and Cheshire Congenital Anomaly Survey |
| 2002 | Wessex Antenatally Detected Anomalies Register (WANDA) |
| 2002 | Congenital Anomaly Register for Oxfordshire, Berkshire & Buckinghamshire (Oxfordshire only prior to 2005) |
| 2003 | Northern Congenital Abnormality Survey |
| 2003 | South West Congenital Anomaly Register |

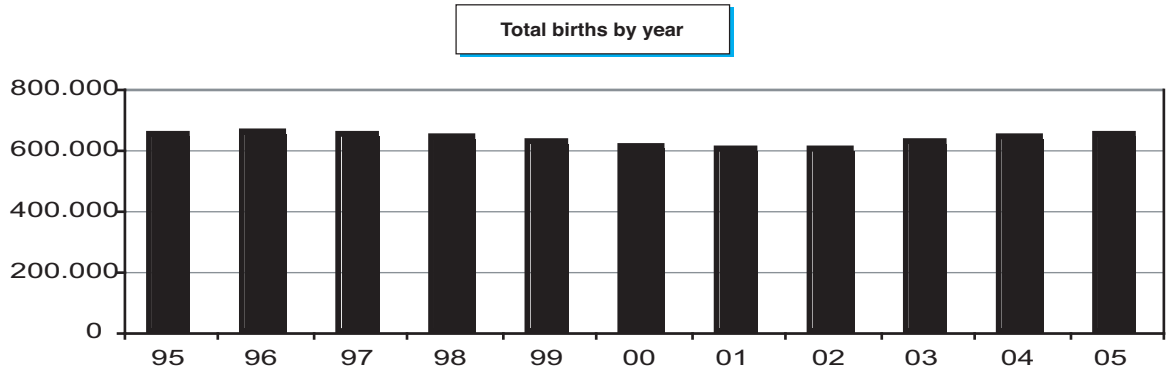
In 2005, congenital anomaly notifications are now received for all births in Wales and 45 per cent of births in England. For areas for which NCAS relies solely on SD56 notification forms recording is likely to be less complete.

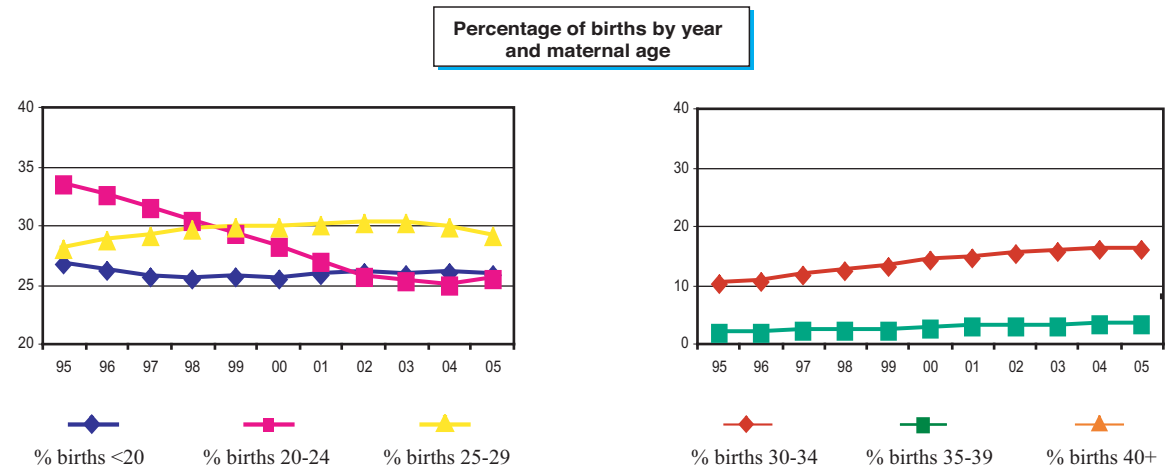
Reports of terminations of pregnancy have been compiled from notifications of abortions that are completed by the operating practitioners under the 1967 Abortion Act and are sent to the Chief Medical Officers of England and Wales. The tables sent to the International Clearinghouse only include notifications of abortions performed under Grounds E of the Act. An abortion may be performed under Grounds E if 'there is substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped'. Since April 2002, the Department of Health has been responsible for the processing of the abortions notification forms and information has been made accessible to the Office for National Statistics (ONS) for statistical purposes.

**Exposure information:**
Parents' occupation is known.

**Addresses and Staff:**
Vera Ruddock
Office for National Statistics
Room 2.101
Government Buildings
Cardiff Road
Newport, Gwent, NP10 8XG
**Phone:** 01633 455864
**E-mail:** ncas@ons.gsi.gov.uk

## England and Wales




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 443 | 84.2 | Cystic kidney | 68 | 12.0 |
| Spina bifida | 328 | 60.4 | Limb reduction defects | 22 | 3.8 |
| Encephalocele | 63 | 61.8 | Diaphragmatic hernia | 49 | 15.4 |
| Holoprosencephaly | 93 | 74.4 | Omphalocele | 24 | 12.4 |
| Hydrocephaly | 151 | 38.1 | Gastroschisis | 0 | 0.0 |
| Hypoplastic left heart syndrome | 90 | 35.3 | Trisomy 13 | 150 | 76.1 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 425 | 76.4 |
| Cleft lip with or without cleft palate | 0 | 0.0 | Down syndrome | 1249 | 48.4 |
| Renal agenesis | 75 | 21.0 | | | |

Total ToPs with births defects = 5,751 (Ratio ToPs/Births: 3.00 per 1,000 births)
*ToPs/ToPs+Births

# England and Wales: 2005

| | |
|---|---|
| Live births (LB) | 645,621 |
| Stillbirths (SB) | 3,473 |
| Total births | 649,094 |
| Number of terminations of pregnancy (ToP) for birth defects | 1,916 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 13 | 12 | 139 | 2.53 |
| Spina bifida | 47 | 9 | 120 | 2.71 |
| Encephalocele | 10 | 4 | 24 | 0.59 |
| Microcephaly | 27 | 7 | nr | 0.52 |
| Holoprosencephaly | 9 | 4 | 36 | 0.75 |
| Hydrocephaly | 53 | 27 | 44 | 1.91 |
| Anophthalmos | 7 | 1 | nr | 0.12 |
| Microphthalmos | 10 | 0 | nr | 0.15 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 13 | 0 | nr | 0.20 |
| Microtia | 7 | 0 | nr | 0.11 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 59 | 5 | nr | 0.99 |
| Tetralogy of Fallot | 67 | 4 | nr | 1.09 |
| Hypoplastic left heart syndrome | 43 | 8 | 32 | 1.28 |
| Coarctation of aorta | 73 | 5 | nr | 1.20 |
| Choanal atresia, bilateral | 15 | 0 | nr | 0.23 |
| Cleft palate without cleft lip | 202 | 4 | nr | 3.17 |
| Cleft lip with or without cleft palate | 311 | 11 | nr | 4.96 |
| Oesophageal atresia / stenosis with or without fistula | 68 | 5 | nr | 1.12 |
| Small intestine atresia / stenosis | 60 | 1 | nr | 0.94 |
| Anorectal atresia / stenosis | 62 | 3 | nr | 1.00 |
| Undescended testis (36 weeks of gestation or later) | 14 | 0 | nr | 0.22 |
| Hypospadias | 488 | 0 | nr | 7.52 |
| Epispadias | 15 | 0 | nr | 0.23 |
| Indeterminate sex | 33 | 5 | nr | 0.59 |
| Renal agenesis | 87 | 9 | 25 | 1.86 |
| Cystic kidney | 143 | 12 | 20 | 2.70 |
| Bladder exstrophy | 5 | 0 | nr | 0.08 |
| Polydactyly, preaxial | 31 | 1 | nr | 0.49 |
| Total Limb reduction defects (include unspecified) | 141 | 17 | 11 | 2.60 |
| Transverse | 48 | 4 | nr | 0.80 |
| Preaxial | 11 | 1 | nr | 0.18 |
| Postaxial | 4 | 0 | nr | 0.06 |
| Intercalary | 21 | 3 | nr | 0.37 |
| Mixed | 24 | 3 | nr | 0.42 |
| Unspecified | 33 | 6 | nr | 0.60 |
| Diaphragmatic hernia | 82 | 11 | 15 | 1.66 |
| Omphalocele | 43 | 7 | 13 | 0.97 |
| Gastroschisis | 124 | 4 | nr | 1.97 |
| Unspecified Omphalocele/Gastroschisis | 33 | 0 | nr | 0.51 |
| Prune belly sequence | 1 | 0 | nr | 0.02 |
| Trisomy 13 | 10 | 7 | 52 | 1.06 |
| Trisomy 18 | 22 | 15 | 151 | 2.90 |
| Down syndrome, all ages (include age unknown) | 419 | 32 | 429 | 13.56 |
| <25 | 55 | 1 | 15 | 4.23 |
| 25-29 | 51 | 1 | 29 | 4.90 |
| 30-34 | 92 | 5 | 96 | 10.21 |
| 35-39 | 129 | 18 | 168 | 30.09 |
| 40+ | 71 | 7 | 121 | 88.87 |
| unknown | 21 | 0 | 0 | --- |

nr = not reported
* Abortions performed under Grounds E of the 1967 Abortion Act. Data source: Department of Health. Figures less than 10 are suppressed.

# England and Wales: Previous years rates 1995 - 2005

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

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995* | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | **651,315** | **3,169,859** | **3,113,854** |
| Anencephaly | | | | 3.09 | 2.84 | 2.77 |
| Spina bifida | | | | 3.38 | 2.90 | 2.89 |
| Encephalocele | | | | 0.55 | 0.52 | 0.53 |
| Microcephaly | | | | 0.57 | 0.66 | 0.75 |
| Holoprosencephaly | | | | 0.48 | 0.50 | 0.64 |
| Hydrocephaly | | | | 2.10 | 2.15 | 2.06 |
| Anophthalmos | | | | 0.18 | 0.07 | 0.08 |
| Microphthalmos | | | | 0.21 | 0.23 | 0.21 |
| Unspecified Anophthalmos / Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | 0.20 | 0.21 | 0.19 |
| Microtia | | | | 0.09 | 0.03 | 0.09 |
| Unspecified Anotia / Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 0.51 | 0.76 | 1.14 |
| Tetralogy of Fallot | | | | 0.46 | 0.75 | 1.09 |
| Hypoplastic left heart syndrome | | | | 0.63 | 0.86 | 1.24 |
| Coarctation of aorta | | | | 0.37 | 0.72 | 1.37 |
| Choanal atresia, bilateral | | | | 0.14 | 0.13 | 0.22 |
| Cleft palate without cleft lip | | | | 3.09 | 3.13 | 3.18 |
| Cleft lip with or without cleft palate | | | | 6.10 | 6.32 | 5.70 |
| Oesophageal atresia / stenosis with or without fistula | | | | 1.09 | 0.91 | 1.12 |
| Small intestine atresia / stenosis | | | | 0.68 | 0.81 | 1.04 |
| Anorectal atresia / stenosis | | | | 1.49 | 1.41 | 1.33 |
| Undescended testis (36 weeks of gestation or later) | | | | 0.21 | 0.24 | 0.27 |
| Hypospadias | | | | 7.43 | 8.78 | 8.68 |
| Epispadias | | | | 0.43 | 0.28 | 0.20 |
| Indeterminate sex | | | | 0.58 | 0.78 | 0.59 |
| Renal agenesis | | | | 1.07 | 1.38 | 1.81 |
| Cystic kidney | | | | 2.23 | 2.10 | 2.96 |
| Bladder exstrophy | | | | 0.14 | 0.18 | 0.11 |
| Polydactyly, preaxial | | | | 0.48 | 0.68 | 0.72 |
| Total Limb reduction defects (include unspecified) | | | | 3.02 | 3.21 | 3.11 |
| Transverse | | | | 1.70 | 1.72 | 1.00 |
| Preaxial | | | | 0.17 | 0.17 | 0.23 |
| Postaxial | | | | 0.21 | 0.11 | 0.06 |
| Intercalary | | | | 0.29 | 0.56 | 0.79 |
| Mixed | | | | 0.21 | 0.18 | 0.27 |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 0.80 | 1.25 | 1.65 |
| Omphalocele | | | | 1.09 | 0.97 | 1.12 |
| Gastroschisis | | | | 1.52 | 1.75 | 2.23 |
| Unspecified Omphalocele / Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.12 | 0.04 | 0.06 |
| Trisomy 13 | | | | 0.57 | 0.80 | 1.06 |
| Trisomy 18 | | | | 1.41 | 2.00 | 2.88 |
| Down syndrome, all ages (include age unknown) | | | | 9.53 | 11.83 | 12.92 |
| <20 | | | | nr | nr | nr |
| 20-24 | | | | nr | nr | nr |
| 25-29 | | | | 4.03 | 5.64 | 5.01 |
| 30-34 | | | | 8.74 | 9.76 | 10.05 |
| 35-39 | | | | 27.16 | 29.47 | 29.51 |
| 40-45 | | | | nr | nr | nr |
| 45+ | | | | nr | nr | nr |
| unspecified | | | | --- | --- | --- |

* data include less than 5 years

nr = not reported

## England and Wales

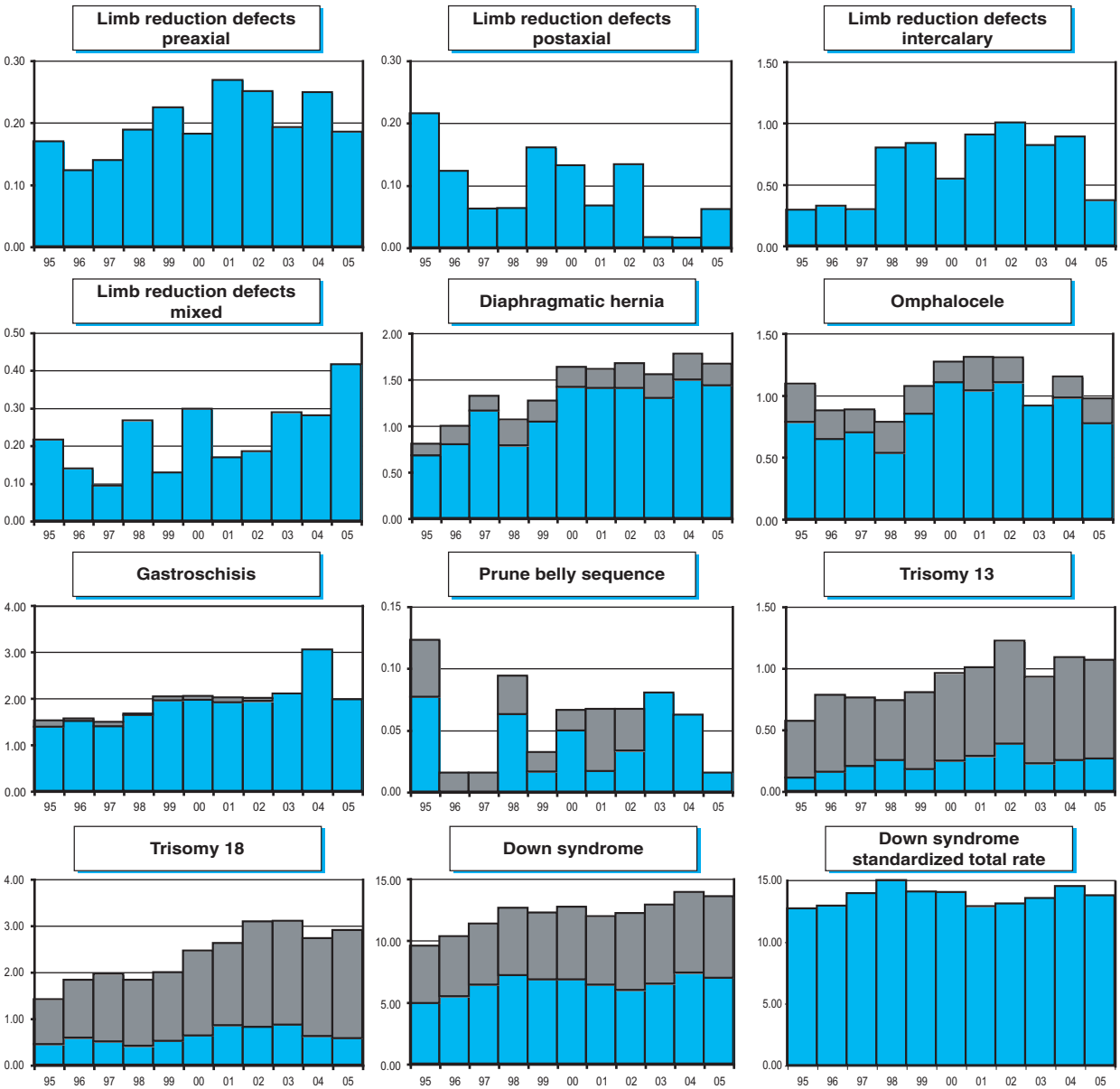
Time trends 1974-2005 (Birth prevalence rates per 10,000)


**Note:** L+S rates, ToP rates

## England and Wales


**Note:** L+S rates, ToP rates

## England and Wales




**Note:** L+S rates, ToP rates

## Finland

### The Finnish Register of Congenital Malformations

**History:**
The registry was established in 1963 and regular monitoring started in 1977. It was a founding member of the ICBDSR and is a full member. In 1998 the registry became an associate member of EUROCAT. The registry system (data collection etc.) has been changed twice, in 1985 and in 1993.

**Size and coverage:**
The registry is national and population based. All births in Finland are covered, at present approximately 58,000 annually. Stillbirths of 22 weeks / 500 g or more are registered. Information on malformations is principally collected up to 1 year of age, but later information is also included. Selective terminations of pregnancy and spontaneous abortions with malformations have been included since 1993.

**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 STAKES, the governmental National Research and Development Centre for Welfare and Health (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, Abortion Register and Hospital Discharge Register. The diagnoses of the malformation cases received from 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 malformations 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.

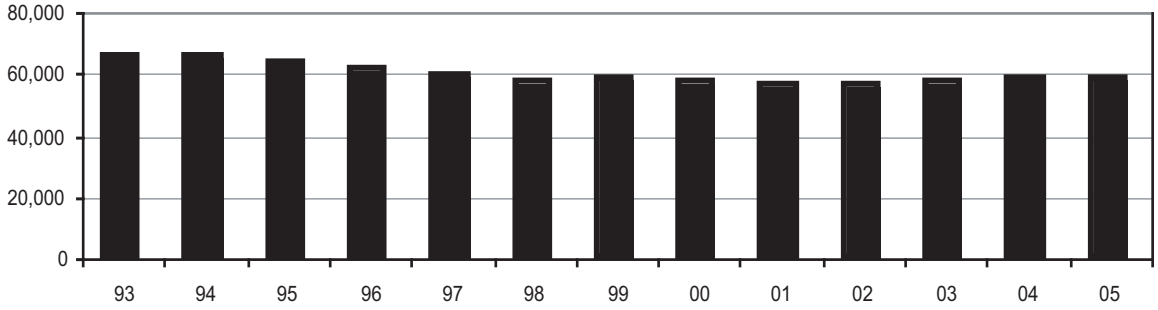
**Background information:**
Epidemiological background data are available on all births in the Medical Birth Register and in the Statistics Finland.

**Address and Staff:**
Annukka Ritvanen
The Finnish Register of Congenital Malformations
The National Research and Development Centre for Welfare and Health, STAKES
Lintulahdenkuja 4, P.O. Box 220
SF 00531-Helsinki - Finland
**Phone:** 358-9-39672376
**Fax:** 358-9-39672459
**E-mail:** annukka.ritvanen@stakes.fi

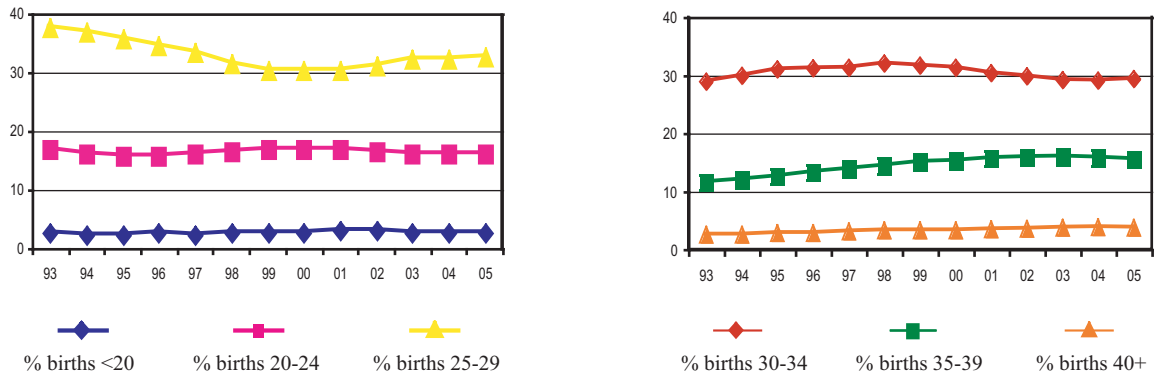
**Web:** http://www.stakes.fi/

## Finland

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 54 | 96.4 | Cystic kidney | 37 | 28.2 |
| Spina bifida | 31 | 38.3 | Limb reduction defects | 26 | 20.3 |
| Encephalocele | 22 | 71.0 | Diaphragmatic hernia | 17 | 33.3 |
| Holoprosencephaly | 14 | 48.3 | Omphalocele | 55 | 55.6 |
| Hydrocephaly | 35 | 43.8 | Gastroschisis | 25 | 43.1 |
| Hypoplastic left heart syndrome | 15 | 20.8 | Trisomy 13 | 24 | 70.6 |
| Cleft palate without cleft lip | 14 | 5.7 | Trisomy 18 | 73 | 61.3 |
| Cleft lip with or without cleft palate | 24 | 13.1 | Down syndrome | 242 | 53.7 |
| Renal agenesis | 19 | 65.5 | | | |

Total ToPs with births defects = 880 (Ratio ToPs/Births: 5.10 per 1,000)
*ToPs/ToPs+Births

# Finland: 2005

| | |
|---|---|
| Live births (LB) | 57,745 |
| Stillbirths (SB) | 182 |
| Total births | 57,927 |
| Number of terminations of pregnancy (ToP) for birth defects | 304 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 18 | 3.11 |
| Spina bifida | 13 | 1 | 15 | 5.01 |
| Encephalocele | 3 | 0 | 9 | 2.07 |
| Microcephaly | 10 | 0 | 0 | 1.73 |
| Holoprosencephaly | 7 | 0 | 3 | 1.73 |
| Hydrocephaly | 13 | 3 | 14 | 5.18 |
| Anophthalmos | 1 | 0 | 1 | 0.35 |
| Microphthalmos | 5 | 1 | 1 | 1.21 |
| Unspecified Anophthalmos / Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia / Microtia | 27 | 0 | 0 | 4.66 |
| Transposition of great vessels | 19 | 0 | 2 | 3.63 |
| Tetralogy of Fallot | 22 | 0 | 5 | 4.66 |
| Hypoplastic left heart syndrome | 16 | 1 | 9 | 4.49 |
| Coarctation of aorta | 47 | 0 | 0 | 8.11 |
| Choanal atresia, bilateral | 6 | 0 | 0 | 1.04 |
| Cleft palate without cleft lip | 75 | 1 | 7 | 14.33 |
| Cleft lip with or without cleft palate | 55 | 0 | 9 | 11.05 |
| Oesophageal atresia / stenosis with or without fistula | 20 | 1 | 1 | 3.80 |
| Small intestine atresia / stenosis | 5 | 0 | 0 | 0.86 |
| Anorectal atresia / stenosis | 23 | 0 | 4 | 4.66 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 69 | 0 | 1 | 12.08 |
| Hypospadias severe | 16 | 0 | 0 | 2.76 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 9 | 0 | 0 | 1.55 |
| Renal agenesis | 4 | 0 | 7 | 1.90 |
| Cystic kidney | 32 | 1 | 11 | 7.60 |
| Bladder exstrophy | 1 | 1 | 2 | 0.69 |
| Polydactyly, preaxial | 21 | 1 | 2 | 4.14 |
| Total Limb reduction defects (include unspecified) | 23 | 0 | 11 | 5.87 |
| Transverse | 14 | 0 | 3 | 2.93 |
| Preaxial | 5 | 0 | 3 | 1.38 |
| Postaxial | 0 | 0 | 2 | 0.35 |
| Intercalary | 3 | 0 | 1 | 0.69 |
| Mixed | 1 | 0 | 2 | 0.52 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 12 | 1 | 9 | 3.80 |
| Omphalocele | 12 | 2 | 17 | 5.35 |
| Gastroschisis | 10 | 0 | 9 | 3.28 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 1 | 0.17 |
| Prune belly sequence | 0 | 0 | 2 | 0.35 |
| Trisomy 13 | 3 | 0 | 5 | 1.38 |
| Trisomy 18 | 11 | 6 | 27 | 7.60 |
| Down syndrome, all ages (include age unknown) | 65 | 3 | 82 | 25.89 |
| <20 | 1 | 0 | 0 | 6.18 |
| 20-24 | 3 | 0 | 4 | 7.45 |
| 25-29 | 12 | 0 | 7 | 10.08 |
| 30-34 | 18 | 0 | 18 | 21.18 |
| 35-39 | 19 | 2 | 25 | 51.41 |
| 40-44 | 9 | 1 | 26 | 178.84 |
| 45+ | 3 | 0 | 2 | 446.43 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# Finland: Previous years rates 1993 - 2005

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

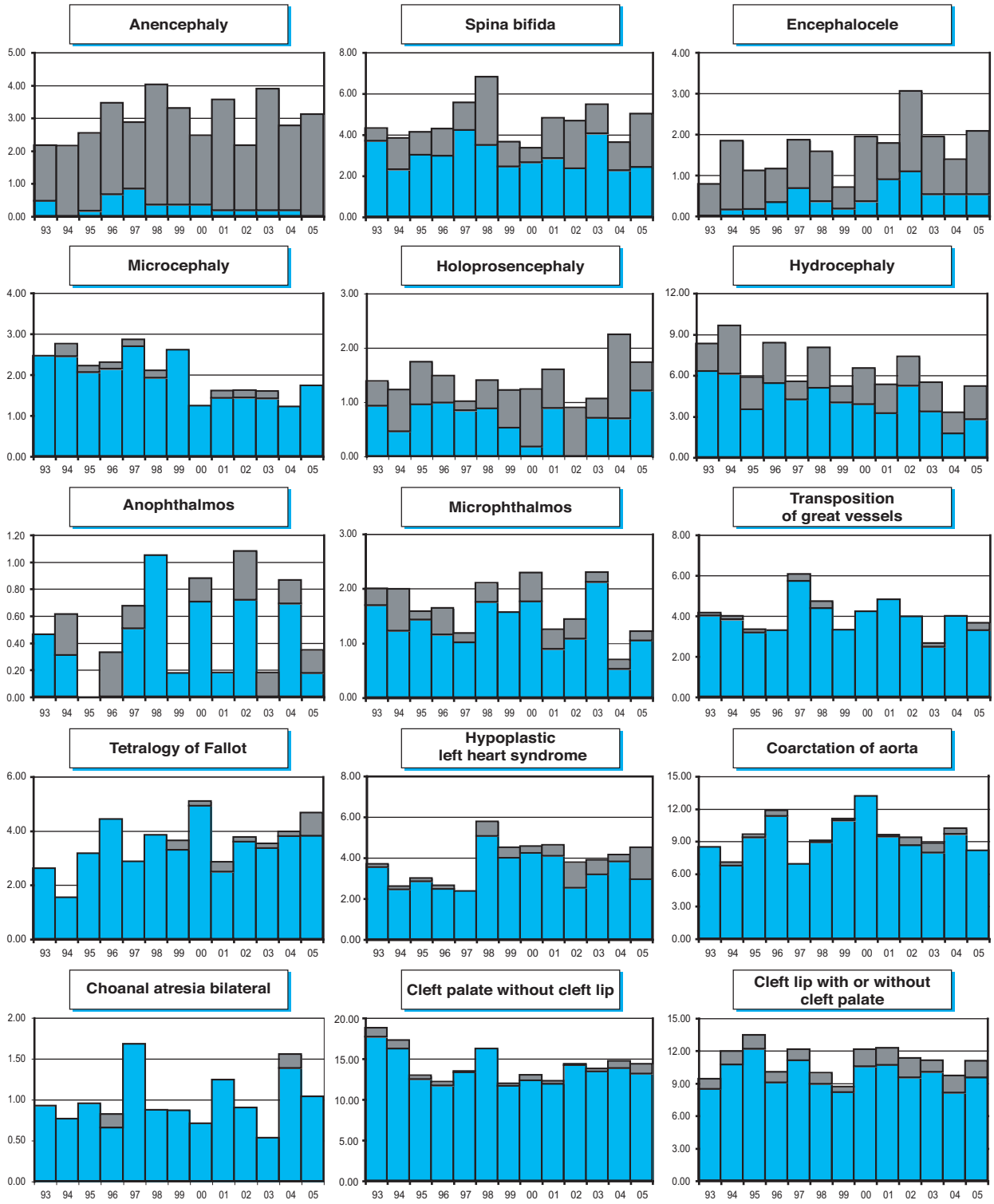
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995* | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | **193,946** | **292,630** | **284,845** |
| Anencephaly | | | | 2.27 | 3.21 | 3.09 |
| Spina bifida | | | | 4.07 | 4.72 | 4.70 |
| Encephalocele | | | | 1.24 | 1.44 | 2.04 |
| Microcephaly | | | | 2.47 | 2.22 | 1.54 |
| Holoprosencephaly | | | | 1.44 | 1.26 | 1.51 |
| Hydrocephaly | | | | 7.94 | 6.73 | 5.30 |
| Anophthalmos | | | | 0.36 | 0.62 | 0.53 |
| Microphthalmos | | | | 1.86 | 1.74 | 1.37 |
| Unspecified Anophthalmos / Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | nr | nr | nr |
| Microtia | | | | nr | nr | nr |
| Unspecified Anotia / Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 3.82 | 4.31 | 3.79 |
| Tetralogy of Fallot | | | | 2.42 | 3.96 | 3.76 |
| Hypoplastic left heart syndrome | | | | 3.09 | 3.93 | 4.18 |
| Coarctation of aorta | | | | 8.35 | 10.39 | 9.20 |
| Choanal atresia, bilateral | | | | 0.88 | 0.99 | 1.05 |
| Cleft palate without cleft lip | | | | 16.34 | 13.33 | 13.87 |
| Cleft lip with or without cleft palate | | | | 11.55 | 10.56 | 11.06 |
| Oesophageal atresia / stenosis with or without fistula | | | | 3.45 | 3.79 | 3.72 |
| Small intestine atresia / stenosis | | | | 1.29 | 1.06 | 1.12 |
| Anorectal atresia / stenosis | | | | 5.36 | 5.64 | 4.56 |
| Undescended testis (36 weeks of gestation or later) | | | | nr | nr | nr |
| Hypospadias | | | | 3.04 | 3.25 | 3.37 |
| Epispadias | | | | 0.21 | 0.31 | 0.35 |
| Indeterminate sex | | | | 0.77 | 1.44 | 1.69 |
| Renal agenesis | | | | 1.91 | 1.71 | 1.51 |
| Cystic kidney | | | | 5.88 | 6.60 | 7.76 |
| Bladder exstrophy | | | | 0.46 | 0.55 | 0.70 |
| Polydactyly, preaxial | | | | 4.43 | 4.44 | 4.00 |
| Total Limb reduction defects (include unspecified) | | | | 7.22 | 6.80 | 7.55 |
| Transverse | | | | 4.43 | 3.66 | 3.83 |
| Preaxial | | | | 1.70 | 1.74 | 2.53 |
| Postaxial | | | | 0.15 | 0.41 | 0.39 |
| Intercalary | | | | 0.41 | 0.41 | 0.35 |
| Mixed | | | | 0.41 | 0.34 | 0.28 |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 2.32 | 2.63 | 3.02 |
| Omphalocele | | | | 4.23 | 4.00 | 5.30 |
| Gastroschisis | | | | 1.70 | 2.60 | 3.41 |
| Unspecified Omphalocele / Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.21 | 0.31 | 0.28 |
| Trisomy 13 | | | | 2.42 | 1.88 | 2.04 |
| Trisomy 18 | | | | 5.36 | 5.98 | 6.71 |
| Down syndrome, all ages (include age unknown) | | | | 23.05 | 23.14 | 25.59 |
| <20 | | | | 18.54 | 6.48 | 8.30 |
| 20-24 | | | | 6.02 | 7.48 | 7.48 |
| 25-29 | | | | 12.35 | 9.39 | 10.61 |
| 30-34 | | | | 17.94 | 17.83 | 16.54 |
| 35-39 | | | | 56.41 | 53.17 | 57.15 |
| 40-44 | | | | 177.56 | 173.54 | 179.27 |
| 45+ | | | | 419.85 | 248.45 | 391.06 |
| unknown | | | | --- | --- | --- |

* data include less than 5 years

nr = not reported

## Finland

'Time trends 1974-2005 (Birth prevalence rates per 10,000)

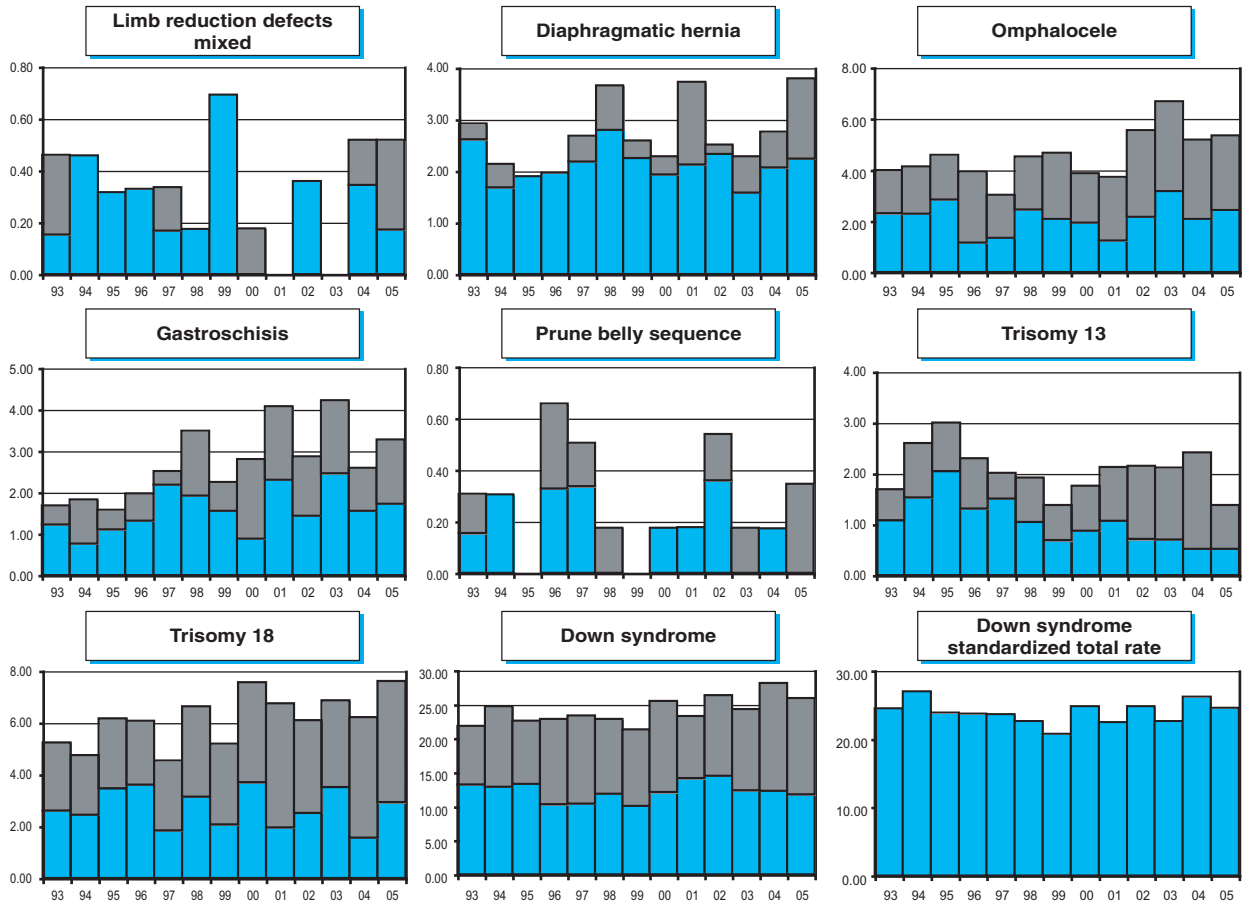



**Note:** L+S rates, ToP rates

## Finland


**Note:** L+S rates, ToP rates

## Finland




**Note:** 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:**
Reporting is volontary. The registry is part of a research unit of INSERM (National Institute of Health and Medical Research). The registry has been officially recognized by the French National Comity of Registries, and is renewed for four years (2001-2005) and 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 delivery 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).

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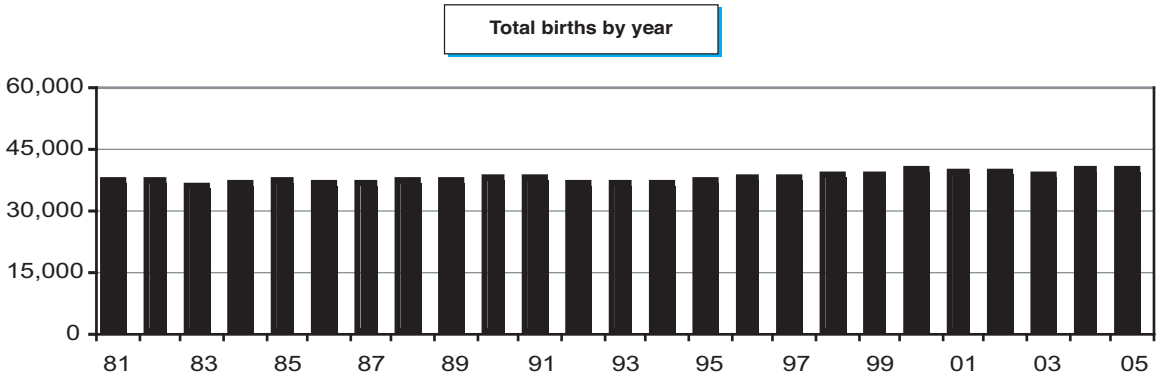
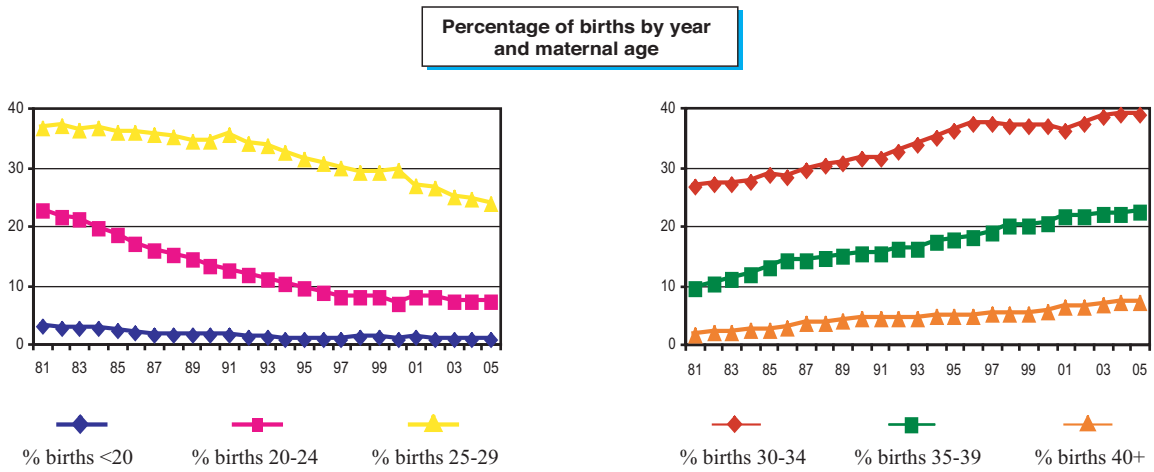
## France: Paris




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 73 | 96.1 | Cystic kidney | 34 | 29.8 |
| Spina bifida | 35 | 70.0 | Limb reduction defects | 55 | 65.5 |
| Encephalocele | 20 | 90.9 | Diaphragmatic hernia | 14 | 22.2 |
| Holoprosencephaly | 39 | 88.6 | Omphalocele | 44 | 64.7 |
| Hydrocephaly | 86 | 48.9 | Gastroschisis | 5 | 15.2 |
| Hypoplastic left heart syndrome | 34 | 66.7 | Trisomy 13 | 46 | 88.5 |
| Cleft palate without cleft lip | 14 | 21.9 | Trisomy 18 | 146 | 88.5 |
| Cleft lip with or without cleft palate | 27 | 27.6 | Down syndrome | 424 | 85.0 |
| Renal agenesis | 23 | 62.2 | | | |

Total ToPs with births defects = 1,506 (Ratio ToPs/Births: 12.86 per 1,000 births)
*ToPs/ToPs+Births

# France: Paris, 2005

| | |
|---|---|
| Live births (LB) | 38,856 |
| Stillbirths (SB) | 459 |
| Total births | 39,315 |
| Number of terminations of pregnancy (ToP) for birth defects | 487 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 2 | 0 | 25 | 6.87 |
| Spina bifida | 5 | 0 | 8 | 3.31 |
| Encephalocele | 0 | 0 | 7 | 1.78 |
| Microcephaly | 5 | 1 | 3 | 2.29 |
| Holoprosencephaly | 1 | 0 | 9 | 2.54 |
| Hydrocephaly | 31 | 2 | 28 | 15.52 |
| Anophthalmos | 1 | 0 | 0 | 0.25 |
| Microphthalmos | 1 | 0 | 2 | 0.76 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 5 | 0 | 0 | 1.27 |
| Microtia | 2 | 0 | 0 | 0.51 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 17 | 0 | 0 | 4.32 |
| Tetralogy of Fallot | 11 | 0 | 5 | 4.07 |
| Hypoplastic left heart syndrome | 6 | 0 | 8 | 3.56 |
| Coarctation of aorta | 10 | 0 | 1 | 2.80 |
| Choanal atresia, bilateral | 1 | 0 | 1 | 0.51 |
| Cleft palate without cleft lip | 21 | 1 | 5 | 6.87 |
| Cleft lip with or without cleft palate | 25 | 0 | 7 | 8.14 |
| Oesophageal atresia / stenosis with or without fistula | 16 | 1 | 1 | 4.58 |
| Small intestine atresia / stenosis | 18 | 0 | 2 | 5.09 |
| Anorectal atresia / stenosis | 8 | 0 | 6 | 3.56 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 49 | 0 | 2 | 12.97 |
| Epispadias | 3 | 0 | 0 | 0.76 |
| Indeterminate sex | 4 | 0 | 5 | 2.29 |
| Renal agenesis | 8 | 1 | 10 | 4.83 |
| Cystic kidney | 23 | 1 | 8 | 8.14 |
| Bladder exstrophy | 1 | 1 | 0 | 0.51 |
| Polydactyly, preaxial | 5 | 0 | 0 | 1.27 |
| Total Limb reduction defects (include unspecified) | 5 | 0 | 11 | 4.07 |
| Transverse | 5 | 0 | 4 | 2.29 |
| Preaxial | 0 | 0 | 5 | 1.27 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 0 | 0 | 2 | 0.51 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 16 | 2 | 4 | 5.60 |
| Omphalocele | 4 | 0 | 17 | 5.34 |
| Gastroschisis | 9 | 0 | 1 | 2.54 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 5 | 1.27 |
| Prune belly sequence | 0 | 0 | 1 | 0.25 |
| Trisomy 13 | 2 | 0 | 13 | 3.82 |
| Trisomy 18 | 4 | 2 | 47 | 13.48 |
| Down syndrome, all ages (include age unknown) | 34 | 2 | 145 | 46.04 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 2 | 10.66 |
| 25-29 | 6 | 0 | 10 | 17.04 |
| 30-34 | 11 | 0 | 30 | 26.81 |
| 35-39 | 6 | 0 | 57 | 72.23 |
| 40-44 | 9 | 2 | 42 | 203.30 |
| 45+ | 1 | 0 | 4 | 253.81 |
| uknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# France: Paris, Previous years rates 1981 - 2005

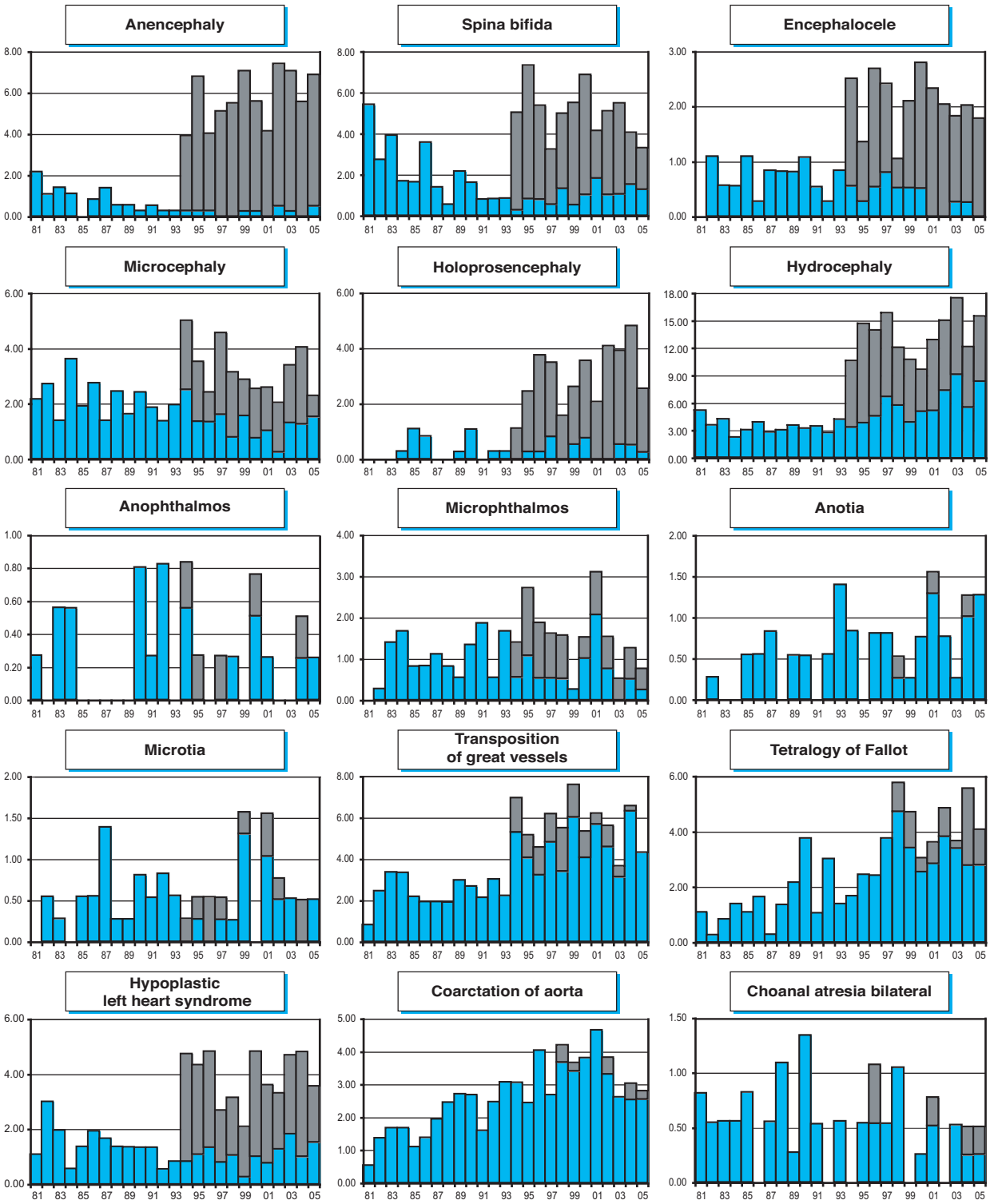
Prevalence rates: (LB+SB) * 10,000 until 1993
Prevalence rates: (LB+SB+TOP) * 10,000 since 1994

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | **181,879** | **183,304** | **182,631** | **190,269** | **195,065** |
| Anencephaly | | 1.15 | 0.71 | 2.35 | 5.47 | 6.20 |
| Spina bifida | | 3.08 | 1.85 | 2.96 | 5.20 | 4.41 |
| Encephalocele | | 0.66 | 0.76 | 1.10 | 2.21 | 2.00 |
| Microcephaly | | 2.36 | 2.13 | 2.74 | 3.10 | 2.87 |
| Holoprosencephaly | | 0.27 | 0.44 | 0.82 | 3.00 | 3.49 |
| Hydrocephaly | | 3.63 | 3.27 | 7.12 | 12.40 | 14.61 |
| Anophthalmos | | 0.27 | 0.16 | 0.44 | 0.26 | 0.21 |
| Microphthalmos | | 0.82 | 0.93 | 1.64 | 1.37 | 1.44 |
| Unspecified Anophthalmos / Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 0.16 | 0.49 | 0.55 | 0.63 | 1.03 |
| Microtia | | 0.27 | 0.65 | 0.55 | 0.58 | 0.77 |
| Unspecified Anotia / Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 2.42 | 2.29 | 3.89 | 5.83 | 5.28 |
| Tetralogy of Fallot | | 0.93 | 1.85 | 1.92 | 3.94 | 4.36 |
| Hypoplastic left heart syndrome | | 1.59 | 1.53 | 2.35 | 3.52 | 4.00 |
| Coarctation of aorta | | 1.26 | 2.24 | 2.52 | 3.68 | 3.38 |
| Choanal atresia, bilateral | | 0.66 | 0.65 | 0.33 | 0.58 | 0.46 |
| Cleft palate without cleft lip | | 3.74 | 4.42 | 6.35 | 6.99 | 6.10 |
| Cleft lip with or without cleft palate | | 6.87 | 7.69 | 9.31 | 9.67 | 7.38 |
| Oesophageal atresia / stenosis with or without fistula | | 2.47 | 2.95 | 3.39 | 4.10 | 4.25 |
| Small intestine atresia / stenosis | | 0.33 | 1.25 | 2.35 | 1.68 | 4.20 |
| Anorectal atresia / stenosis | | 3.35 | 1.91 | 3.83 | 2.73 | 4.15 |
| Undescended testis (36 weeks of gestation or later) | | 8.80 | 13.86 | 10.73 | 6.10 | 5.39 |
| Hypospadias | | 9.73 | 12.55 | 14.02 | 10.30 | 15.48 |
| Epispadias | | 0.16 | 0.71 | 0.38 | 0.37 | 0.41 |
| Indeterminate sex | | 1.43 | 1.25 | 1.42 | 1.16 | 1.49 |
| Renal agenesis | | 0.99 | 1.04 | 2.03 | 2.58 | 3.18 |
| Cystic kidney | | 1.65 | 3.27 | 6.08 | 9.83 | 10.36 |
| Bladder exstrophy | | 0.33 | 0.16 | 0.77 | 0.58 | 0.41 |
| Polydactyly, preaxial | | 0.66 | 0.98 | 1.86 | 2.31 | 1.33 |
| Total Limb reduction defects (include unspecified) | | nr | nr | 6.78* | 6.88 | 7.69 |
| Transverse | | nr | nr | 2.44* | 3.21 | 4.25 |
| Preaxial | | nr | nr | 0.54* | 1.05 | 1.38 |
| Postaxial | | nr | nr | 0.54* | 0.37 | 0.77 |
| Intercalary | | nr | nr | 0.27* | 0.58 | 0.41 |
| Mixed | | nr | nr | 0.00* | 0.47 | 0.56 |
| Unspecified | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 2.31 | 2.78 | 3.94 | 5.41 | 5.54 |
| Omphalocele | | 1.59 | 1.85 | 2.79 | 5.10 | 6.25 |
| Gastroschisis | | 0.60 | 1.04 | 2.19 | 2.89 | 3.23 |
| Unspecified Omphalocele / Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | 0.16 | 0.05 | 0.00 | 0.16 | 0.21 |
| Trisomy 13 | | 0.44 | 0.65 | 1.04 | 3.84 | 4.61 |
| Trisomy 18 | | 1.48 | 1.15 | 3.50 | 8.67 | 13.33 |
| Down syndrome, all ages (include age unknown) | | 11.66 | 12.17 | 21.25 | 35.84 | 40.91 |
| <20 | | 10.57 | 13.59 | 5.19 | 11.44 | 18.42 |
| 20-24 | | 6.92 | 6.17 | 8.48 | 16.73 | 10.99 |
| 25-29 | | 7.27 | 5.60 | 9.52 | 15.25 | 13.97 |
| 30-34 | | 11.46 | 12.50 | 14.47 | 22.59 | 22.69 |
| 35-39 | | 24.28 | 28.02 | 41.18 | 55.52 | 63.95 |
| 40-44 | | 50.30 | 24.12 | 115.23 | 201.15 | 191.37 |
| 45+ | | 183.49 | 181.16 | 206.19 | 310.42 | 381.68 |
| unspecified | | --- | --- | --- | --- | --- |

* data include less than 5 years
nr = not reported

## France: Paris

'Time trends 1981-2005 (Birth prevalence rates per 10,000)




**Note:** L+S rates, ToP rates

## France: Paris


**Note:** L+S rates, ToP rates

## France: Paris


**Note:** L+S rates, ToP rates

## France-Rhône Alpes : (REMERA)

Central-East France Register of Congenital Malformations (until 2006)

**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 and common results are published. 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 56,000 births annually, which represents about 7% of all births in France. Stillbirths of 22 weeks or more gestation are included.

**Legislation and funding:**
REMERA received agreement from the French Comité National des Registres
It has only public sources of funding: Ministry of Health, Region, Health authorities.

**Sources of ascertainment:**
The registry is population based and covers 4 French departments of Rhône-Alpes region : Rhône, Loire, Isère, Savoie.
Data collection is actively performed in private ant public maternity wards and pediatric units. Other sources of information include 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 dediacated and secured server.

The maximum age at postnatal diagnosis is 1 year. For children born in year x, notifications are taken into account until March x+2. We have no follow-up procedure.
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).
Our official stillbirth definition is 22 w (28 w before 1997), which is our lower gestational age limit to include early fetal deaths/spontaneous abortions. Terminations are registered since 1985 (TOP can be performed up to full term in case of lethal or severe foetal abnormalities).

**Exposure information:**
Our exposure data includes drug intake in 1 st trimester of pregnancy, biological, physical and chemical hazards, medically assisted procreation, occupation. Denominators information is obtained from National institute of Statistics. We collect no controls.

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

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Elisabeth Robert
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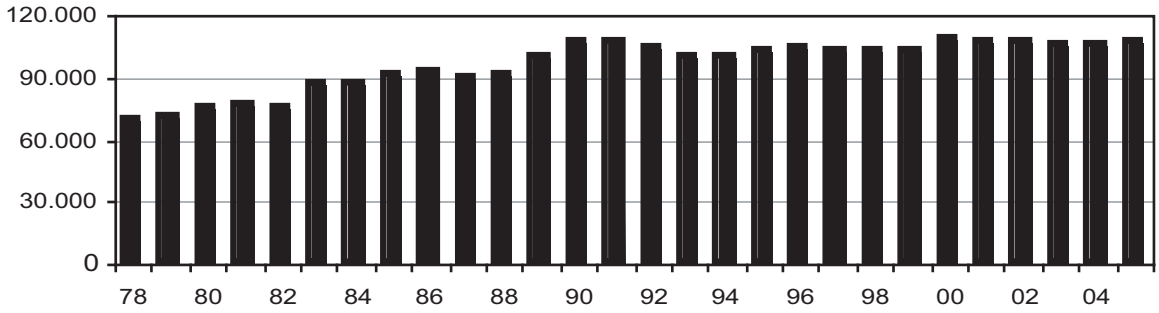
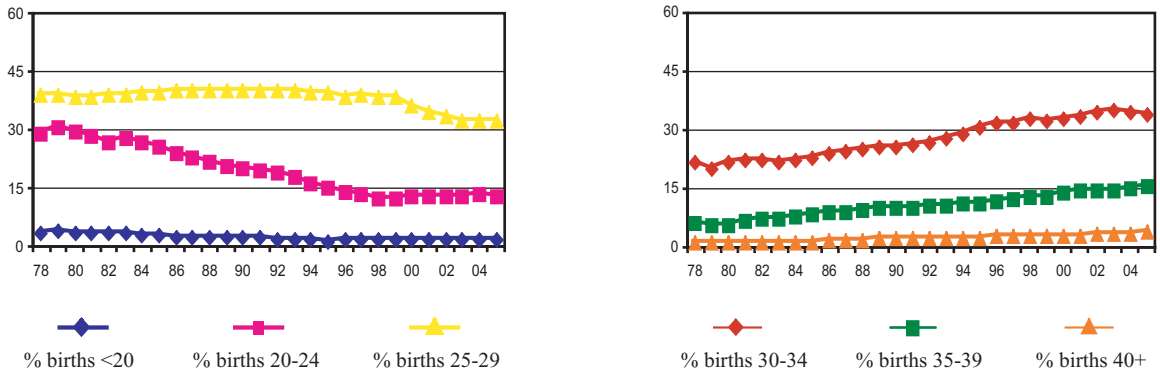
## France-Rhône Alpes : (REMERA)

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 93 | 80.17 | Cystic kidney | 5 | 4.03 |
| Spina bifida | 40 | 35.09 | Limb reduction defects | 6 | 5.36 |
| Encephalocele | 10 | 40.00 | Diaphragmatic hernia | 6 | 9.84 |
| Holoprosencephaly | 13 | 36.11 | Omphalocele | 33 | 50.00 |
| Hydrocephaly | 37 | 19.27 | Gastroschisis | 6 | 15.00 |
| Hypoplastic left heart syndrome | 13 | 15.85 | Trisomy 13 | 44 | 70.97 |
| Cleft palate without cleft lip | 6 | 3.17 | Trisomy 18 | 138 | 74.19 |
| Cleft lip with or without cleft palate | 12 | 6.06 | Down syndrome | 351 | 63.82 |
| Renal agenesis | 16 | 11.68 | | | |

Total ToPs with births defects = 1,770 (Ratio ToPs/Births: 5.57 per 1,000 births)
*ToPs/ToPs+Births

# France-Rhône Alpes: REMERA, 2005

| | |
|---|---|
| Live births (LB) | 106,111 |
| Stillbirths (SB) | 877 |
| Total births | 106,988 |
| Number of terminations of pregnancy (ToP) for birth defects | 649 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | 24 | 2.34 |
| Spina bifida | 7 | 2 | 42 | 4.77 |
| Encephalocele | 0 | 1 | 20 | 1.96 |
| Microcephaly | 9 | 0 | 14 | 2.15 |
| Holoprosencephaly | 2 | 0 | 23 | 2.34 |
| Hydrocephaly | 23 | 1 | 38 | 5.80 |
| Anophthalmos | 1 | 0 | 4 | 0.47 |
| Microphthalmos | 5 | 0 | 3 | 0.75 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 2 | 0 | 0 | 0.19 |
| Microtia | 4 | 0 | 3 | 0.65 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 46 | 0 | 8 | 5.05 |
| Tetralogy of Fallot | 36 | 0 | 5 | 3.83 |
| Hypoplastic left heart syndrome | 9 | 3 | 19 | 2.90 |
| Coarctation of aorta | 30 | 0 | 0 | 2.80 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 53 | 0 | 7 | 5.61 |
| Cleft lip with or without cleft palate | 63 | 1 | 17 | 7.57 |
| Oesophageal atresia / stenosis with or without fistula | 27 | 2 | 3 | 2.99 |
| Small intestine atresia / stenosis | 20 | 1 | 4 | 2.34 |
| Anorectal atresia / stenosis | 20 | 1 | 12 | 3.08 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 103 | 0 | 4 | 10.00 |
| Epispadias | 1 | 0 | 0 | 0.09 |
| Indeterminate sex | 9 | 0 | 3 | 1.12 |
| Renal agenesis | 1 | 1 | 5 | 0.65 |
| Cystic kidney | 29 | 0 | 24 | 4.95 |
| Bladder exstrophy | 3 | 0 | 1 | 0.37 |
| Polydactyly, preaxial | 7 | 0 | 3 | 0.93 |
| Total Limb reduction defects (include unspecified) | 33 | 3 | 19 | 5.14 |
| Transverse | 23 | 1 | 5 | 2.71 |
| Preaxial | 4 | 1 | 8 | 1.22 |
| Postaxial | 2 | 1 | 3 | 0.56 |
| Intercalary | 2 | 0 | 1 | 0.28 |
| Mixed | 2 | 0 | 2 | 0.37 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 30 | 1 | 5 | 3.36 |
| Omphalocele | 13 | 0 | 16 | 2.71 |
| Gastroschisis | 20 | 0 | 2 | 2.06 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 2 | 0.19 |
| Trisomy 13 | 5 | 0 | 22 | 2.52 |
| Trisomy 18 | 5 | 0 | 61 | 6.17 |
| Down syndrome, all ages (include age unknown) | 48 | 3 | 203 | 23.74 |
| <20 | 0 | 0 | 3 | 18.30 |
| 20-24 | 6 | 0 | 10 | 11.65 |
| 25-29 | 5 | 1 | 14 | 5.79 |
| 30-34 | 10 | 0 | 49 | 16.15 |
| 35-39 | 8 | 0 | 72 | 47.97 |
| 40-44 | 3 | 2 | 45 | 135.21 |
| 45+ | 0 | 0 | 7 | 351.76 |
| unknown | 16 | 0 | 3 | --- |

nr = not reported

# France-Rhône Alpes: REMERA, Previous years rates 1978 - 2005

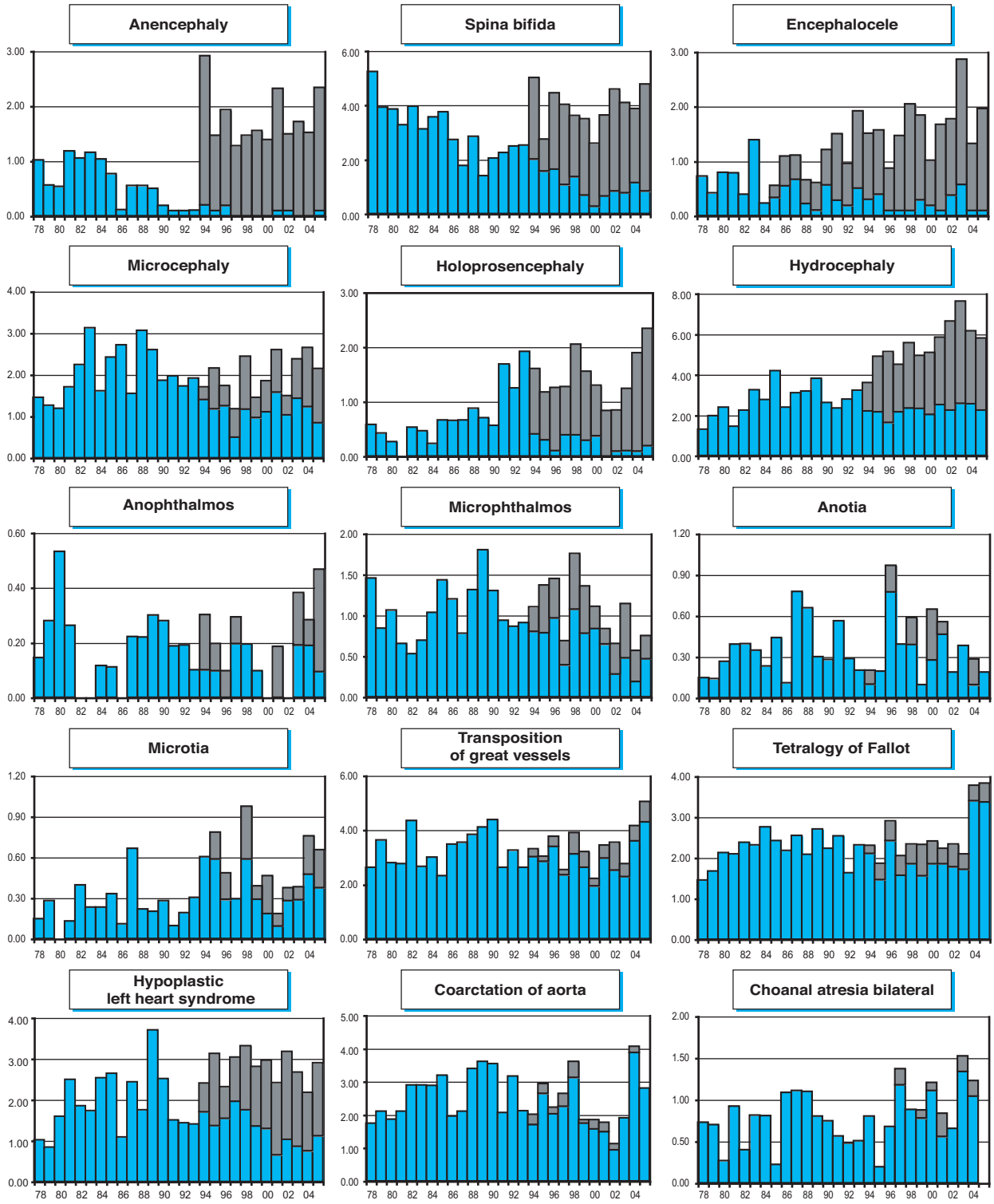
Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1994

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **215,606** | **416,195** | **480,672** | **512,163** | **519,052** | **532,292** |
| Anencephaly | 0.70 | 1.03 | 0.37 | 0.92 | 1.52 | 1.88 |
| Spina bifida | 4.31 | 3.53 | 2.14 | 2.99 | 3.62 | 4.19 |
| Encephalocele | 0.65 | 0.67 | 0.94 | 1.48 | 1.44 | 1.92 |
| Microcephaly | 1.30 | 2.23 | 2.35 | 1.89 | 1.73 | 2.25 |
| Holoprosencephaly | 0.42 | 0.38 | 0.69 | 1.52 | 1.48 | 1.43 |
| Hydrocephaly | 1.90 | 2.84 | 3.02 | 3.36 | 5.05 | 6.41 |
| Anophthalmos | 0.32 | 0.10 | 0.21 | 0.20 | 0.13 | 0.26 |
| Microphthalmos | 1.11 | 0.89 | 1.29 | 1.03 | 1.27 | 0.79 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.19 | 0.36 | 0.42 | 0.29 | 0.54 | 0.32 |
| Microtia | 0.14 | 0.26 | 0.29 | 0.39 | 0.52 | 0.47 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 3.01 | 2.98 | 3.89 | 2.97 | 3.12 | 3.79 |
| Tetralogy of Fallot | 1.76 | 2.40 | 2.35 | 2.13 | 2.41 | 2.86 |
| Hypoplastic left heart syndrome | 1.16 | 2.26 | 2.33 | 1.97 | 2.89 | 2.67 |
| Coarctation of aorta | 1.90 | 2.81 | 2.95 | 2.46 | 2.43 | 2.33 |
| Choanal atresia, bilateral | 0.56 | 0.62 | 0.96 | 0.51 | 1.00 | 0.85 |
| Cleft palate without cleft lip | 4.17 | 5.00 | 4.66 | 6.29 | 6.32 | 5.35 |
| Cleft lip with or without cleft palate | 6.86 | 6.42 | 6.20 | 7.52 | 8.05 | 7.06 |
| Oesophageal atresia / stenosis with or without fistula | 2.13 | 2.40 | 2.60 | 3.22 | 3.08 | 2.69 |
| Small intestine atresia / stenosis | 1.62 | 1.44 | 1.85 | 1.97 | 2.64 | 2.80 |
| Anorectal atresia / stenosis | 2.09 | 2.84 | 3.29 | 3.32 | 3.99 | 3.55 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 6.40 | 6.05 | 9.99 | 9.41 | 12.52 | 11.59 |
| Epispadias | 0.19 | 0.19 | 0.29 | 0.12 | 0.31 | 0.13 |
| Indeterminate sex | 0.60 | 0.74 | 0.81 | 0.72 | 0.58 | 0.68 |
| Renal agenesis | 0.42 | 0.79 | 0.62 | 0.80 | 1.52 | 1.20 |
| Cystic kidney | 0.28 | 1.54 | 2.50 | 3.63 | 4.57 | 4.68 |
| Bladder exstrophy | 0.23 | 0.17 | 0.33 | 0.41 | 0.31 | 0.34 |
| Polydactyly, preaxial | 0.70 | 0.82 | 1.46 | 2.15 | 2.08 | 1.35 |
| Total Limb reduction defects (include unspecified) | 4.64 | 4.16 | 4.33 | 4.53 | 5.18 | 4.73 |
| Transverse | 2.41 | 2.04 | 2.58 | 2.23 | 2.56 | 2.44 |
| Preaxial | 0.65 | 0.70 | 0.64 | 0.57 | 0.87 | 1.13 |
| Postaxial | 0.37 | 0.24 | 0.48 | 0.33 | 0.39 | 0.47 |
| Intercalary | 0.42 | 0.60 | 0.27 | 0.55 | 0.44 | 0.32 |
| Mixed | 0.60 | 0.50 | 0.33 | 0.29 | 0.37 | 0.30 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 1.76 | 2.96 | 2.33 | 2.85 | 2.77 | 3.06 |
| Omphalocele | 1.07 | 1.11 | 1.25 | 1.41 | 2.50 | 2.46 |
| Gastroschisis | 0.42 | 0.84 | 0.92 | 1.23 | 1.21 | 1.62 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.28 | 0.14 | 0.40 | 0.43 | 0.48 | 0.17 |
| Trisomy 13 | 0.28 | 0.58 | 1.00 | 1.13 | 1.81 | 2.10 |
| Trisomy 18 | 0.93 | 0.99 | 1.98 | 3.03 | 4.32 | 4.60 |
| Down syndrome, all ages (include age unknown) | 11.87 | 11.05 | 10.96 | 14.70 | 19.96 | 22.56 |
| <20 | 8.95 | 3.24 | 5.69 | 6.69 | 11.92 | 6.00 |
| 20-24 | 7.09 | 6.69 | 5.38 | 6.39 | 8.33 | 7.97 |
| 25-29 | 5.63 | 5.36 | 7.13 | 6.46 | 7.82 | 7.74 |
| 30-34 | 12.92 | 10.12 | 9.39 | 10.00 | 14.05 | 13.53 |
| 35-39 | 27.15 | 30.92 | 21.90 | 33.54 | 44.07 | 47.83 |
| 40-44 | 115.63 | 59.62 | 54.30 | 101.05 | 143.51 | 144.84 |
| 45+ | 123.46 | 112.36 | 110.38 | 294.12 | 273.75 | 231.71 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 7 years
nr = not reported

## France-Rhône Alpes : (REMERA)

'Time trends 1978-2005 (Birth prevalence rates per 10,000)

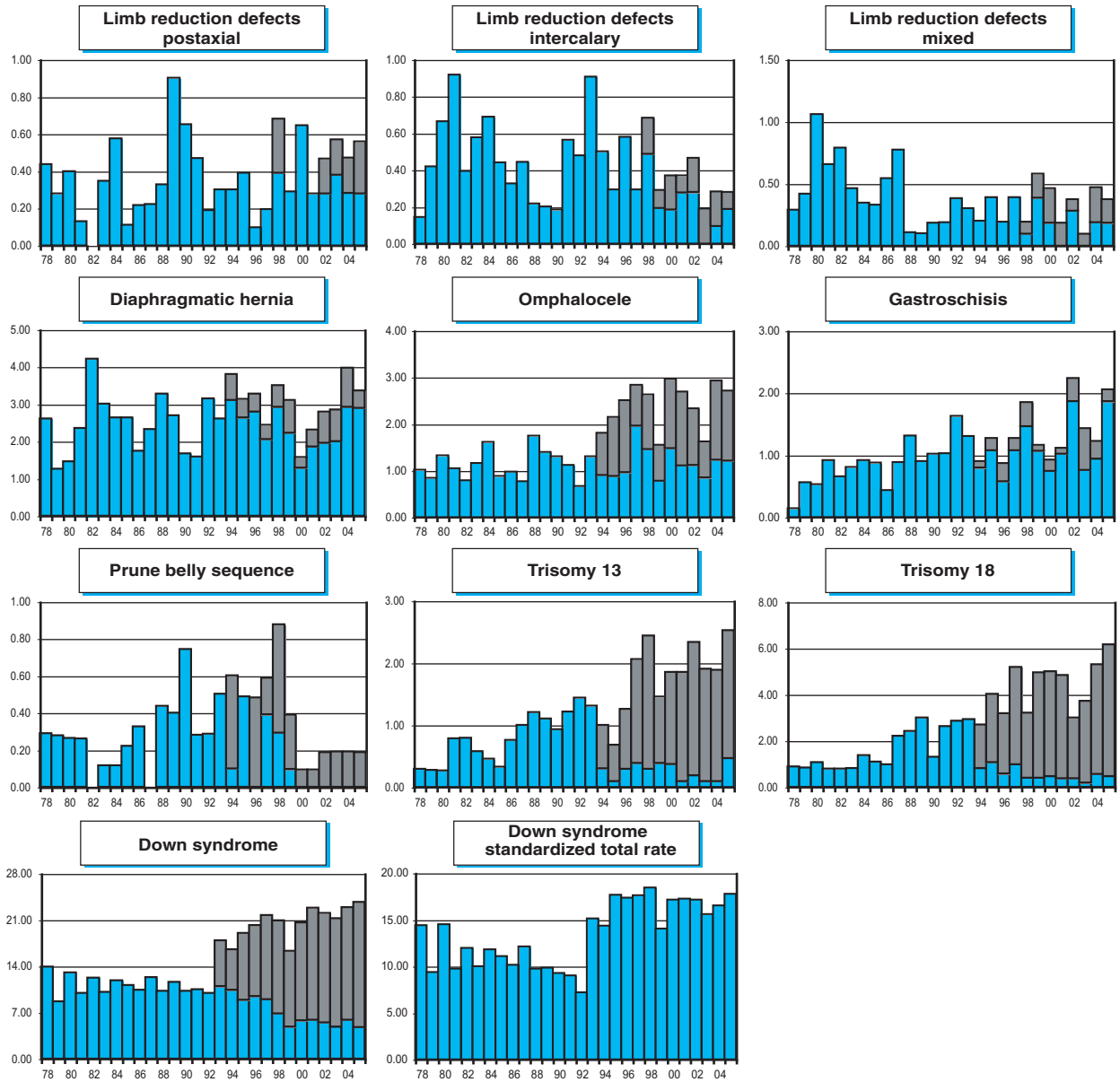



**Note:** L+S rates, ToP rates

## France-Rhône Alpes : (REMERA)


**Note:** L+S rates, ToP rates

## France-Rhône Alpes : (REMERA)




**Note:** L+S rates, ToP rates

## France: Strasbourg

Strasbourg Prospective Study of Congenital Malformations.

**History:**
The registry was started in 1979. The Programme became an associate member of the Clearinghouse in 1982.

**Size and coverage:**
All births in an area including and around Strasbourg and the department of Bas-Rhin are covered -13,000 to 13,500 annually, or 1,8% of all births in France.

**Legislation and funding:**
The Programme is a research Programme, recognised by the local University, the local health authorities and funded by the Institut national de Veille Sanitaire, Ministry of Health, INSERM and CREGEMES (Centre Régional de Génétique Médicale de Strasbourg)

**Sources of ascertainment:**
Reports are obtained from medical files of gynecologists, pediatricians, fetopathologists and radiologists. No control infant is selected.

**Exposure information:**
Information on various exposures is obtained after analysis of hospital files (maternity files or pediatric files). More precise information may be obtained from genetic files (if it exists) after direct interview of the mother

**Background information:**
General demographic information is obtained from the National Institute of Statistics. Further information is obtained from Social Security Records and Health Sheets.

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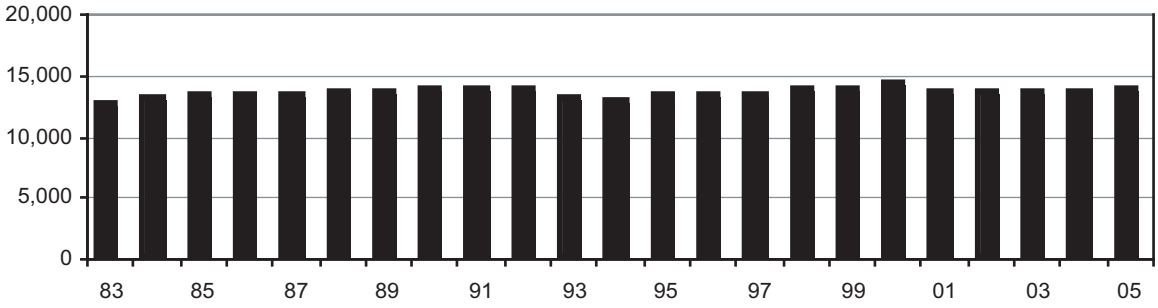
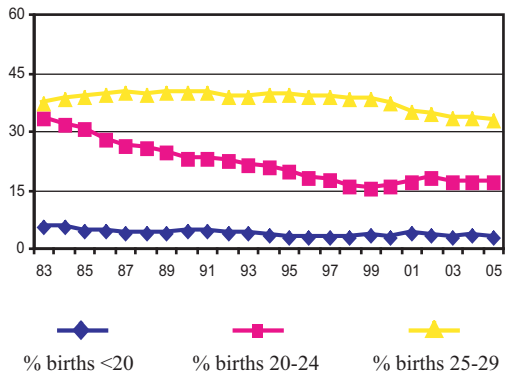
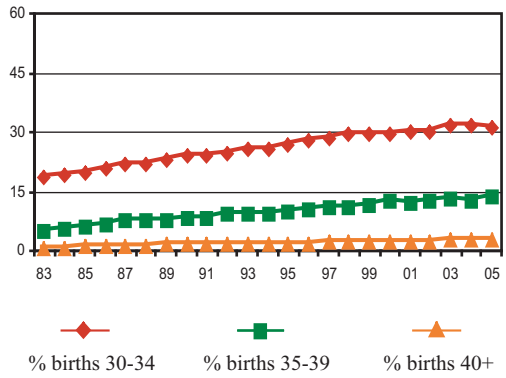
## France: Strasbourg

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 26 | 96.30 | Cystic kidney | 7 | 25.93 |
| Spina bifida | 24 | 92.31 | Limb reduction defects | 13 | 35.14 |
| Encephalocele | 6 | 100.00 | Diaphragmatic hernia | 4 | 21.05 |
| Holoprosencephaly | 15 | 100.00 | Omphalocele | 11 | 84.62 |
| Hydrocephaly | 18 | 90.00 | Gastroschisis | 0 | 0.00 |
| Hypoplastic left heart syndrome | 9 | 90.00 | Trisomy 13 | 15 | 93.75 |
| Cleft palate without cleft lip | 3 | 8.57 | Trisomy 18 | 28 | 90.32 |
| Cleft lip with or without cleft palate | 15 | 26.32 | Down syndrome | 62 | 60.78 |
| Renal agenesis | 10 | 37.04 | | | |

Total ToPs with births defects = 279 (Ratio ToPs/Births: 6.86 per 1,000 births)
*ToPs/ToPs+Births

# France: Strasbourg, 2005

| | |
|---|---|
| Live births (LB) | 13,635 |
| Stillbirths (SB) | 119 |
| Total births | 13,754 |
| Number of terminations of pregnancy (ToP) for birth defects | 89 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | 5 | 4.36 |
| Spina bifida | 1 | 0 | 12 | 9.45 |
| Encephalocele | 0 | 0 | 1 | 0.73 |
| Microcephaly | 2 | 0 | 1 | 2.18 |
| Holoprosencephaly | 0 | 0 | 4 | 2.91 |
| Hydrocephaly | 0 | 0 | 5 | 3.64 |
| Anophthalmos | 1 | 0 | 0 | 0.73 |
| Microphthalmos | 3 | 0 | 2 | 3.64 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 1 | 0 | 1 | 1.45 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 2 | 0 | 0 | 1.45 |
| Tetralogy of Fallot | 3 | 0 | 2 | 3.64 |
| Hypoplastic left heart syndrome | 0 | 1 | 2 | 2.18 |
| Coarctation of aorta | 5 | 0 | 0 | 3.64 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 15 | 0 | 1 | 11.63 |
| Cleft lip with or without cleft palate | 10 | 0 | 6 | 11.63 |
| Oesophageal atresia / stenosis with or without fistula | 3 | 0 | 0 | 2.18 |
| Small intestine atresia / stenosis | 4 | 0 | 0 | 2.91 |
| Anorectal atresia / stenosis | 6 | 0 | 4 | 7.27 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 26 | 0 | 0 | 18.90 |
| Epispadias | 1 | 0 | 0 | 0.73 |
| Indeterminate sex | 0 | 1 | 0 | 0.73 |
| Renal agenesis | 8 | 0 | 1 | 6.54 |
| Cystic kidney | 8 | 0 | 4 | 8.72 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 5 | 0 | 1 | 4.36 |
| Total Limb reduction defects (include unspecified) | 7 | 0 | 7 | 10.18 |
| Transverse | 1 | 0 | 1 | 1.45 |
| Preaxial | 0 | 0 | 2 | 1.45 |
| Postaxial | 0 | 0 | 1 | 0.73 |
| Intercalary | 0 | 0 | 1 | 0.73 |
| Mixed | 6 | 0 | 2 | 5.82 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 2 | 0 | 2 | 2.91 |
| Omphalocele | 0 | 0 | 4 | 2.91 |
| Gastroschisis | 2 | 0 | 0 | 1.45 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 4 | 2.91 |
| Trisomy 18 | 0 | 1 | 8 | 6.54 |
| Down syndrome, all ages (include age unknown) | 7 | 1 | 23 | 22.54 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 0 | 0 | 0 | 0.00 |
| 25-29 | 2 | 0 | 3 | 11.07 |
| 30-34 | 2 | 0 | 7 | 20.89 |
| 35-39 | 1 | 1 | 12 | 75.68 |
| 40-44 | 2 | 0 | 1 | 80.43 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# France: Strasbourg, Previous years rates 1983 - 2005

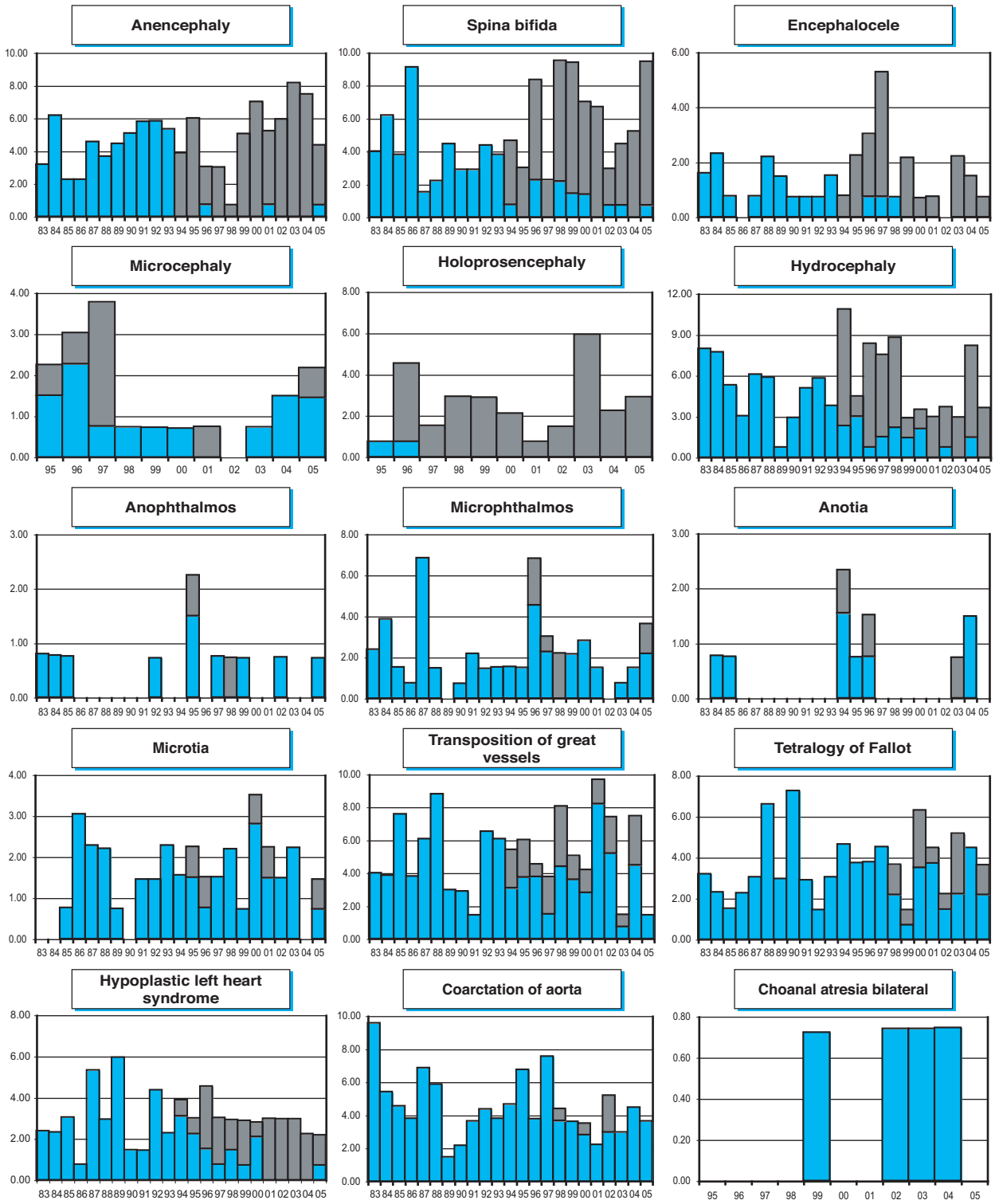
Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1994

| | 1974-1980 | 1981-1985* | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | **38,613** | **67,098** | **66,826** | **68,143** | **67,544** |
| Anencephaly | | 3.88 | 4.02 | 5.39 | 3.82 | 6.22 |
| Spina bifida | | 4.66 | 4.02 | 3.74 | 7.34 | 5.77 |
| Encephalocele | | 1.55 | 1.04 | 1.20 | 2.35 | 1.04 |
| Microcephaly | | nr | nr | 1.53* | 1.76 | 1.04 |
| Holoprosencephaly | | nr | nr | 0.76* | 2.79 | 2.66 |
| Hydrocephaly | | 6.99 | 3.73 | 5.99 | 6.16 | 4.29 |
| Anophthalmos | | 0.78 | 0.00 | 0.60 | 0.44 | 0.30 |
| Microphthalmos | | 2.59 | 1.94 | 1.65 | 3.38 | 1.48 |
| Unspecified Anophthalmos / Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 0.52 | 0.00 | 0.60 | 0.29 | 0.56* |
| Microtia | | 0.26 | 1.64 | 1.80 | 1.91 | 1.48 |
| Unspecified Anotia / Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 5.18 | 4.92 | 5.09 | 5.14 | 5.48 |
| Tetralogy of Fallot | | 2.33 | 4.47 | 3.14 | 3.96 | 4.00 |
| Hypoplastic left heart syndrome | | 2.59 | 3.28 | 2.99 | 3.23 | 2.66 |
| Coarctation of aorta | | 6.47 | 4.02 | 4.64 | 4.55 | 3.70 |
| Choanal atresia, bilateral | | nr | nr | 0.00 | 0.15 | 0.44 |
| Cleft palate without cleft lip | | 9.32 | 8.64 | 9.58 | 6.31 | 8.44 |
| Cleft lip with or without cleft palate | | 8.55 | 9.69 | 13.02 | 13.21 | 13.03 |
| Oesophageal atresia / stenosis with or without fistula | | 2.85 | 2.68 | 2.39 | 2.94 | 3.85 |
| Small intestine atresia / stenosis | | nr | nr | 1.91* | 2.20 | 1.92 |
| Anorectal atresia / stenosis | | 4.40 | 5.07 | 5.09 | 7.34 | 4.74 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | 1.52* | nr |
| Hypospadias | | 16.32 | 25.49 | 26.04 | 20.55 | 22.65 |
| Epispadias | | nr | nr | 0.00* | 0.29 | 0.30 |
| Indeterminate sex | | nr | nr | 0.38* | 0.29 | 1.04 |
| Renal agenesis | | nr | nr | 2.67* | 5.87 | 7.99 |
| Cystic kidney | | nr | nr | 5.35* | 8.81 | 7.40 |
| Bladder exstrophy | | nr | nr | 0.38* | 0.44 | 0.30 |
| Polydactyly, preaxial | | nr | nr | 2.67* | 4.26 | 3.55 |
| Total Limb reduction defects (include unspecified) | | 5.70 | 7.75 | 7.93 | 10.86 | 8.44 |
| Transverse | | 4.14 | 4.62 | 3.29 | 5.87 | 3.70 |
| Preaxial | | 1.55 | 2.09 | 1.65 | 0.88 | 0.89 |
| Postaxial | | 0.00 | 0.60 | 0.30 | 0.44 | 0.59 |
| Intercalary | | 0.00 | 0.00 | 0.90 | 0.73 | 0.15 |
| Mixed | | 0.00 | 0.45 | 0.45 | 0.73 | 2.81 |
| Unspecified | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 3.88 | 5.07 | 4.34 | 4.99 | 3.55 |
| Omphalocele | | 2.07 | 4.17 | 4.19 | 3.67 | 2.96 |
| Gastroschisis | | 1.81 | 1.94 | 2.99 | 1.61 | 1.78 |
| Unspecified Omphalocele / Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | nr | nr | 0.38* | 0.29 | 0.89 |
| Trisomy 13 | | nr | nr | 2.67* | 1.76 | 2.81 |
| Trisomy 18 | | nr | nr | 3.82* | 3.96 | 7.11 |
| Down syndrome, all ages (include age unknown) | | 10.62 | 16.99 | 25.74 | 25.97 | 25.76 |
| <20 | | 5.05 | 14.69 | 17.01 | 15.06 | 4.86 |
| 20-24 | | 8.19 | 7.61 | 11.92 | 9.81 | 10.38 |
| 25-29 | | 4.08 | 8.66 | 13.74 | 10.75 | 5.71 |
| 30-34 | | 12.25 | 18.05 | 15.43 | 23.79 | 16.31 |
| 35-39 | | 43.17 | 52.35 | 97.93 | 81.02 | 82.58 |
| 40-44 | | 209.79 | 261.78 | 268.27 | 192.31 | 152.25 |
| 45+ | | 0.00 | 217.39 | 222.22 | 0.00 | 0.00 |
| unspecified | | --- | --- | --- | --- | --- |

* data include less than 5 years
nr = not reported

## France: Strasbourg

'Time trends 1983-2005 (Birth prevalence rates per 10,000)

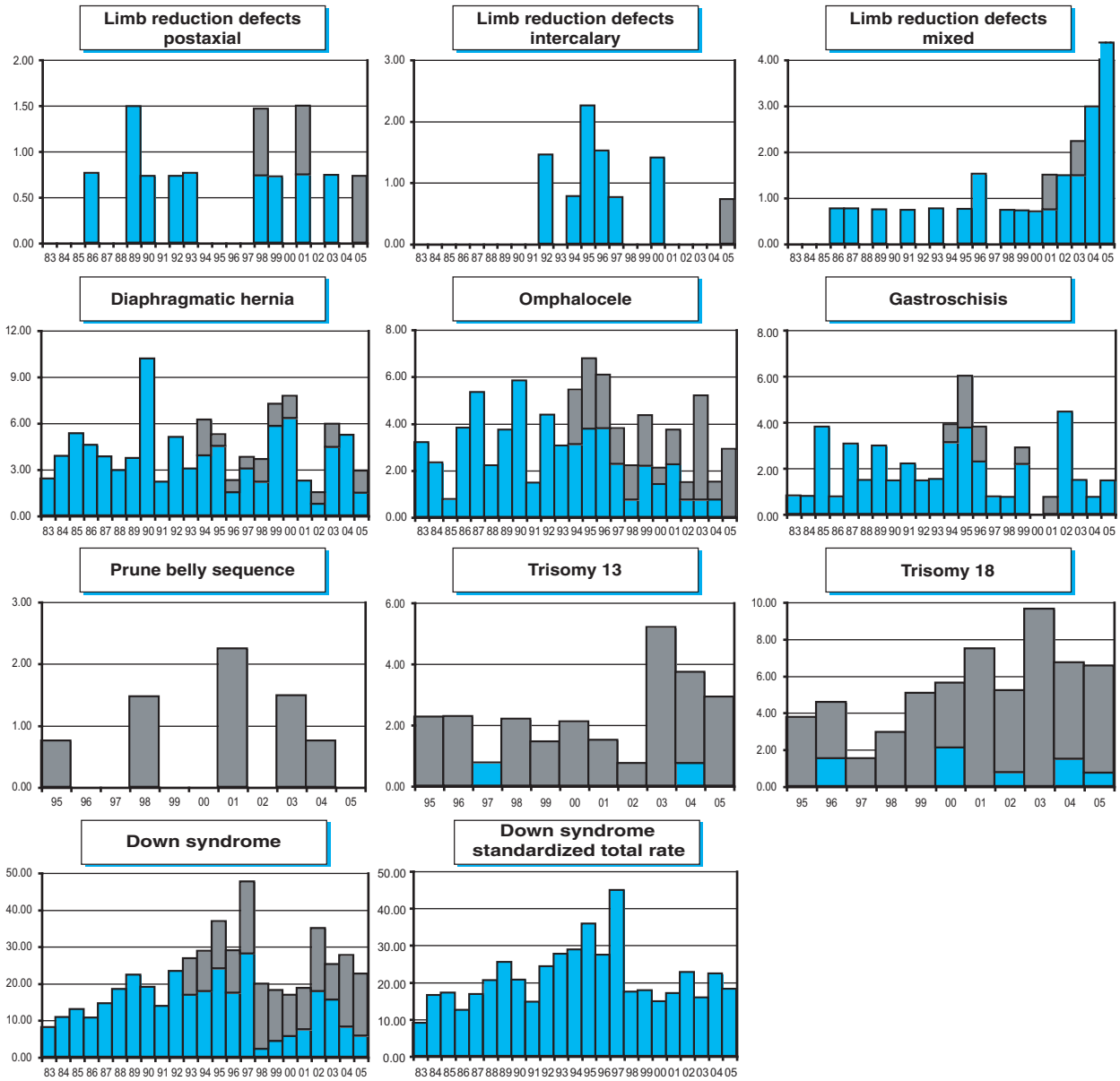



**Note:** L+S rates, ToP rates

## France: Strasbourg


**Note:** L+S rates, ToP rates

## France: Strasbourg




**Note:** L+S rates, ToP rates

## Germany: Saxony-Anhalt
Malformation Monitoring Saxony-Anhalt

**History**:
Since 1980 in the city of Magdeburg all live- and stillbirths, abortions after the 16th week of gestation (spontaneous and induced abortions according to medical evidence based on prenatal diagnoses of congenital defects), and postnatal anomalies or congenital defects have been recorded up to the first week of life. After the reunification of Germany and the creation of the Federal state of Saxony-Anhalt, the survey of congenital defects included approximately two-thirds of all births with postnatal anomalies and congenital defects in the same federal state. Since 1 January 2000 the survey region includes the entire state of Saxony-Anhalt. Saxony-Anhalt has 2.44 million inhabitants (31.12.2006) and annual births at a rate of about 17 000 children (2006). The survey system is multi-centric and based on population.

**Legislation and funding:**
1980 to 1989: Ministry of Health of the former German Democratic Republic
1990 to 1992: Medical Faculty, Magdeburg
1993 to 1995: Ministry of Health, Federal Republic of Germany
since 1995: Ministry of Labour, Women, Health and Social Security of the Federal State of Saxony-Anhalt. The Malformation Monitoring is working in order of Ministry of Labour, Women, Health and Social Security of the Federal State of Saxony-Anhalt.

**Sources of ascertainment:**
The co-operation partner are:
- 29 obstetrics departments
- 27 children hospitals
- 10 institutions of prenatal diagnostic (geneticists, gynecologists)
- 6 departments of pathology

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

**Background information:**
Population based registry (Federal State Saxony-Anhalt); written informed consent of the mother (parents); name and address don't registered; two healthy "controls" per one malformed child; inclusion of terminations of pregnancy, spontaneous abortions after 16th week of gestation, live and stillborn babies; definition of stillbirth: >/= 500 grams; maximum age to include diagnoses: 1 year (almost 1sh week of life); annual report (in German).

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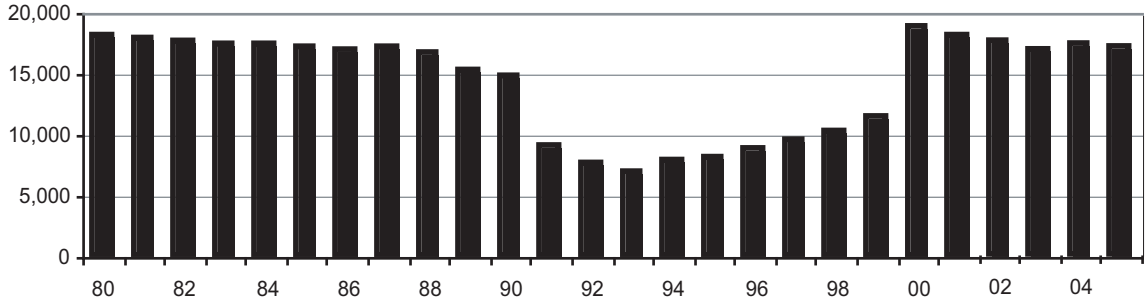
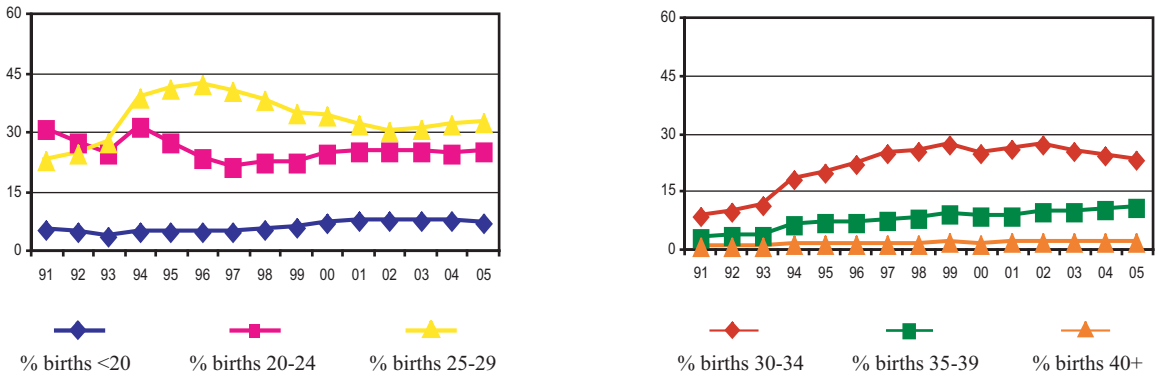
## Germany: Saxony Anhalt

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 18 | 100.0 | Cystic kidney | 9 | 18.8 |
| Spina bifida | 17 | 51.5 | Limb reduction defects | 14 | 35.0 |
| Encephalocele | 6 | 75.0 | Diaphragmatic hernia | 6 | 42.9 |
| Holoprosencephaly | 10 | 100.0 | Omphalocele | 16 | 72.7 |
| Hydrocephaly | 14 | 50.0 | Gastroschisis | 6 | 22.2 |
| Hypoplastic left heart syndrome | 5 | 50.0 | Trisomy 13 | 3 | 100.0 |
| Cleft palate without cleft lip | 6 | 13.6 | Trisomy 18 | 18 | 78.3 |
| Cleft lip with or without cleft palate | 12 | 14.8 | Down syndrome | 42 | 57.5 |
| Renal agenesis | 11 | 91.7 | | | |

Total ToPs with births defects = 239 (Ratio ToPs/Births: 4.63 per 1,000 births)
*ToPs/ToPs+Births

# Germany: Saxony Anhalt, 2005

| | |
|---|---|
| Live births (LB) | 17,166 |
| Stillbirths (SB) | 66 |
| Total births | 17,232 |
| Number of terminations of pregnancy (ToP) for birth defects | 83 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 4 | 2.32 |
| Spina bifida | 3 | 0 | 2 | 2.90 |
| Encephalocele | 0 | 0 | 0 | 0.00 |
| Microcephaly | 40 | 1 | 0 | 23.79 |
| Holoprosencephaly | 0 | 0 | 1 | 0.58 |
| Hydrocephaly | 6 | 0 | 5 | 6.38 |
| Anophthalmos | 0 | 0 | 1 | 0.58 |
| Microphthalmos | 1 | 0 | 0 | 0.58 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 1 | 0.58 |
| Microtia | 1 | 0 | 0 | 0.58 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 9 | 0 | 0 | 5.22 |
| Tetralogy of Fallot | 10 | 1 | 0 | 6.38 |
| Hypoplastic left heart syndrome | 1 | 0 | 3 | 2.32 |
| Coarctation of aorta | 6 | 0 | 0 | 3.48 |
| Choanal atresia, bilateral | 1 | 0 | 0 | 0.58 |
| Cleft palate without cleft lip | 16 | 0 | 2 | 10.45 |
| Cleft lip with or without cleft palate | 21 | 0 | 4 | 14.51 |
| Oesophageal atresia / stenosis with or without fistula | 3 | 0 | 1 | 2.32 |
| Small intestine atresia / stenosis | 3 | 0 | 0 | 1.74 |
| Anorectal atresia / stenosis | 4 | 0 | 2 | 3.48 |
| Undescended testis (36 weeks of gestation or later) | 28 | 0 | 0 | 16.25 |
| Hypospadias | 15 | 0 | 0 | 8.70 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 1 | 0 | 0 | 0.58 |
| Cystic kidney | 14 | 0 | 4 | 10.45 |
| Bladder exstrophy | 0 | 0 | 2 | 1.16 |
| Polydactyly, preaxial | 3 | 0 | 0 | 1.74 |
| Total Limb reduction defects (include unspecified) | 4 | 0 | 4 | 4.64 |
| Transverse | 1 | 0 | 1 | 1.16 |
| Preaxial | 0 | 0 | 0 | 0.00 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 1 | 0 | 1 | 1.16 |
| Mixed | 0 | 0 | 2 | 1.16 |
| Unspecified | 2 | 0 | 0 | 1.16 |
| Diaphragmatic hernia | 1 | 0 | 2 | 1.74 |
| Omphalocele | 3 | 0 | 6 | 5.22 |
| Gastroschisis | 5 | 0 | 3 | 4.64 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 2 | 1.16 |
| Trisomy 13 | 0 | 0 | 1 | 0.58 |
| Trisomy 18 | 1 | 0 | 6 | 4.06 |
| Down syndrome, all ages (include age unknown) | 5 | 0 | 14 | 11.03 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 0 | 2.34 |
| 25-29 | 1 | 0 | 2 | 5.36 |
| 30-34 | 0 | 0 | 2 | 4.99 |
| 35-39 | 3 | 0 | 6 | 50.08 |
| 40-44 | 0 | 0 | 4 | 131.58 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

# Germany: Saxony Anhalt, Previous years rates 1980 - 2005

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

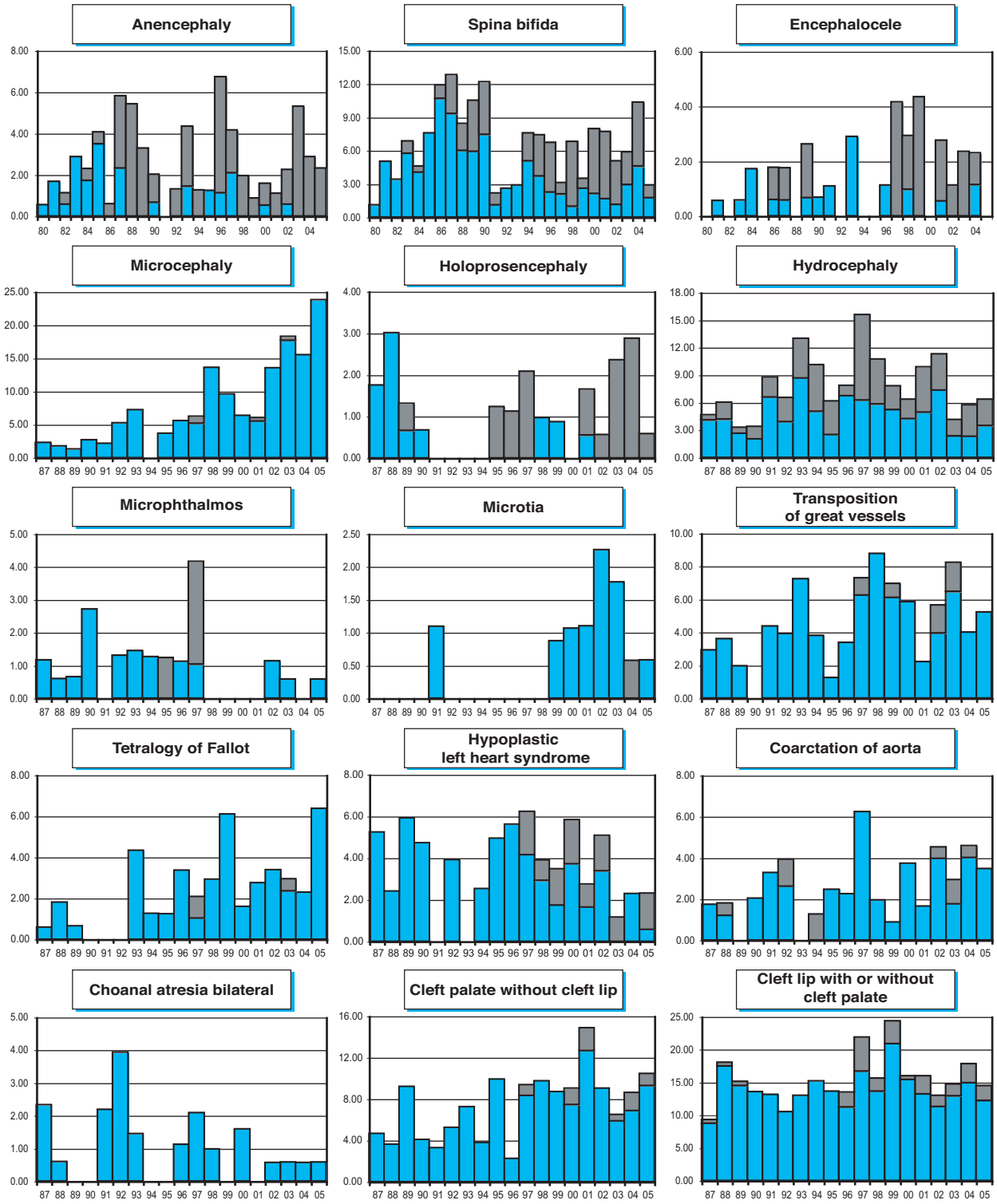
| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **18,113** | **87,531** | **80,602** | **39,659** | **59,053** | **87,458** |
| Anencephaly | 0.55 | 2.40 | 3.47 | 1.51 | 2.71 | 2.74 |
| Spina bifida | 1.10 | 5.48 | 11.17 | 4.54 | 5.93 | 6.40 |
| Encephalocele | 0.00 | 0.57 | 1.36 | 0.76 | 2.20 | 1.72 |
| Microcephaly | nr | nr | 2.04* | 3.53 | 8.13 | 15.32 |
| Holoprosencephaly | nr | nr | 1.73* | 0.25 | 0.85 | 1.60 |
| Hydrocephaly | nr | nr | 4.39* | 8.83 | 9.14 | 7.55 |
| Anophthalmos | nr | nr | 0.00* | 1.01 | 0.00 | 0.23 |
| Microphthalmos | nr | nr | 1.25* | 1.01 | 0.85 | 0.46 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | 0.00* | 0.25 | 0.00 | 0.34 |
| Microtia | nr | nr | 0.00* | 0.25 | 0.51 | 1.26 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | nr | nr | 2.20* | 4.03 | 6.43 | 5.03 |
| Tetralogy of Fallot | nr | nr | 0.78* | 1.26 | 3.05 | 3.54 |
| Hypoplastic left heart syndrome | nr | nr | 4.55* | 2.27 | 5.08 | 2.74 |
| Coarctation of aorta | nr | nr | 1.41* | 2.27 | 3.05 | 3.43 |
| Choanal atresia, bilateral | nr | nr | 0.78* | 1.51 | 1.19 | 0.46 |
| Cleft palate without cleft lip | nr | nr | 5.33* | 5.80 | 8.13 | 9.95 |
| Cleft lip with or without cleft palate | nr | nr | 13.96* | 13.11 | 18.12 | 15.21 |
| Oesophageal atresia / stenosis with or without fistula | nr | nr | 2.82* | 1.51 | 2.88 | 2.97 |
| Small intestine atresia / stenosis | nr | nr | 1.25* | 3.28 | 1.69 | 2.29 |
| Anorectal atresia / stenosis | nr | nr | 3.61* | 3.53 | 2.37 | 3.09 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | 11.45* | 19.67 | 12.02 | 10.75 |
| Hypospadias | nr | nr | 13.02* | 20.93 | 15.41 | 8.35 |
| Epispadias | nr | nr | 0.31* | 0.50 | 0.51 | 0.23 |
| Indeterminate sex | nr | nr | 0.47* | 0.00 | 1.19 | 0.46 |
| Renal agenesis | nr | nr | 1.57* | 1.51 | 2.71 | 2.06 |
| Cystic kidney | nr | nr | 2.04* | 4.79 | 3.05 | 6.97 |
| Bladder exstrophy | nr | nr | 0.78* | 0.25 | 0.34 | 0.23 |
| Polydactyly, preaxial | nr | nr | 0.47* | 3.03 | 3.22 | 3.54 |
| Total Limb reduction defects (include unspecified) | nr | nr | 5.96* | 5.30 | 8.64 | 7.20 |
| Transverse | nr | nr | nr | nr | 4.79* | 2.63 |
| Preaxial | nr | nr | nr | nr | 0.00* | 0.57 |
| Postaxial | nr | nr | nr | nr | 0.00* | 0.00 |
| Intercalary | nr | nr | nr | nr | 1.60* | 1.60 |
| Mixed | nr | nr | nr | nr | 1.60* | 1.72 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | nr | nr | 1.88* | 0.50 | 1.69 | 2.74 |
| Omphalocele | nr | nr | 5.33* | 1.26 | 3.39 | 3.54 |
| Gastroschisis | nr | nr | 1.10* | 2.52 | 3.39 | 4.34 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | 0.16* | 1.01 | 1.02 | 0.91 |
| Trisomy 13 | 0.00 | 0.34 | 0.50* | 0.50 | 2.03 | 0.69 |
| Trisomy 18 | 0.55 | 1.03 | 0.87* | 1.26 | 1.35 | 3.89 |
| Down syndrome, all ages (include age unknown) | 5.52 | 8.80 | 8.81* | 11.60 | 16.93 | 15.44 |
| <20 | nr | nr | nr | nr | 0.00* | 6.27 |
| 20-24 | nr | nr | nr | nr | 4.37* | 5.99 |
| 25-29 | nr | nr | nr | nr | 11.04* | 8.40 |
| 30-34 | nr | nr | nr | nr | 14.94* | 10.94 |
| 35-39 | nr | nr | nr | nr | 71.20* | 49.04 |
| 40-44 | nr | nr | nr | nr | 120.48* | 174.51 |
| 45+ | nr | nr | nr | nr | nr | 392.16 |
| unspecified | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

## Germany: Saxony Anhalt

'Time trends 1980-2005 (Birth prevalence rates per 10,000)

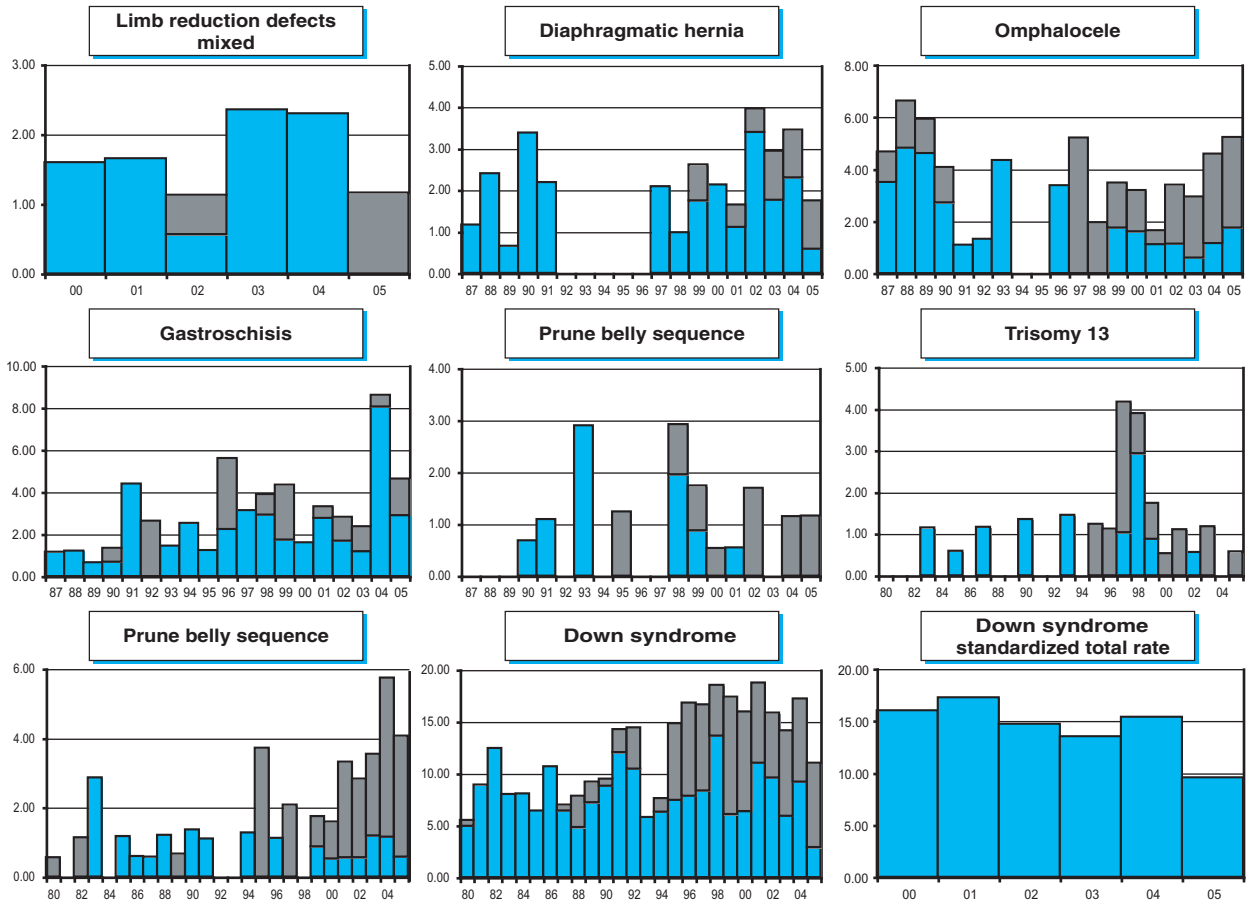



**Note:** L+S rates, ToP rates

## Germany: Saxony Anhalt


**Note:** L+S rates, ToP rates

## Germany: Saxony Anhalt




**Note:** 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 and is a full member.

**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.

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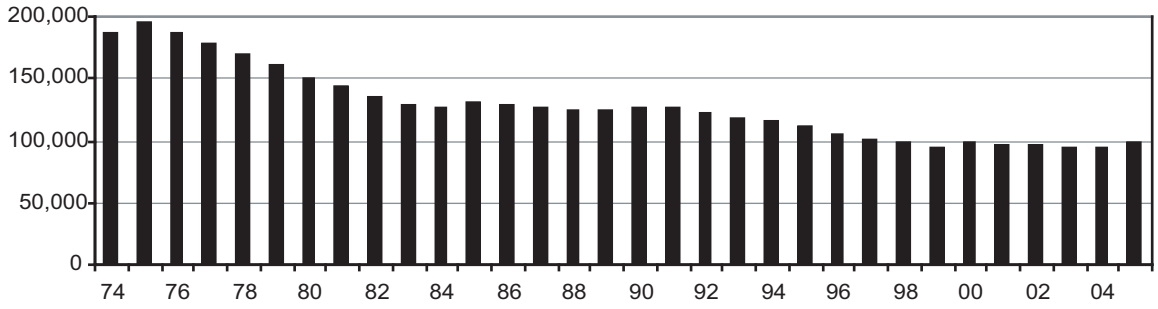
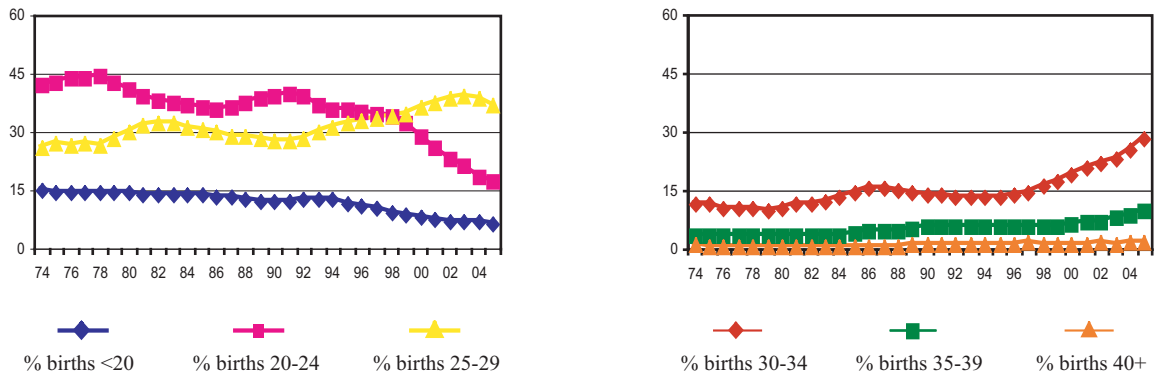
## Hungary

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 37 | 88.1 | Cystic kidney | 9 | 9.5 |
| Spina bifida | 50 | 54.9 | Limb reduction defects | 6 | 6.3 |
| Encephalocele | 4 | 33.3 | Diaphragmatic hernia | 4 | 20.0 |
| Holoprosencephaly | 4 | 20.0 | Omphalocele | 18 | 54.5 |
| Hydrocephaly | 51 | 46.4 | Gastroschisis | 10 | 58.8 |
| Hypoplastic left heart syndrome | 9 | 23.1 | Trisomy 13 | 18 | 72.0 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 37 | 72.5 |
| Cleft lip with or without cleft palate | 4 | 1.9 | Down syndrome | 153 | 38.1 |
| Renal agenesis | 7 | 36.8 | | | |

Total ToPs with births defects = 739 (Ratio ToPs/Births: 25.59 per 1,000 births)
*ToPs/ToPs+Births

# Hungary: 2005

| | |
|---|---|
| Live births (LB) | 97,496 |
| Stillbirths (SB) | 506 |
| Total births | 98,002 |
| Number of terminations of pregnancy (ToP) for birth defects | 257 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 3 | 0 | 10 | 1.33 |
| Spina bifida | 18 | 0 | 16 | 3.47 |
| Encephalocele | 1 | 0 | 0 | 0.10 |
| Microcephaly | 12 | 0 | 0 | 1.22 |
| Holoprosencephaly | 3 | 0 | 0 | 0.31 |
| Hydrocephaly | 19 | 2 | 17 | 3.88 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 3 | 0 | 0 | 0.31 |
| Unspecified Anophthalmos / Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 8 | 0 | 0 | 0.82 |
| Microtia | 1 | 0 | 0 | 0.10 |
| Unspecified Anotia / Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 12 | 0 | 2 | 1.43 |
| Tetralogy of Fallot | 25 | 0 | 0 | 2.55 |
| Hypoplastic left heart syndrome | 11 | 0 | 6 | 1.73 |
| Coarctation of aorta | 20 | 0 | 0 | 2.04 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 30 | 0 | 0 | 3.06 |
| Cleft lip with or without cleft palate | 68 | 0 | 1 | 7.04 |
| Oesophageal atresia / stenosis with or without fistula | 20 | 0 | 1 | 2.14 |
| Small intestine atresia / stenosis | 14 | 0 | 0 | 1.43 |
| Anorectal atresia / stenosis | 15 | 0 | 0 | 1.53 |
| Undescended testis (36 weeks of gestation or later) | 181 | 0 | 0 | 18.47 |
| Hypospadias | 281 | 0 | 0 | 28.67 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | 2 | 0 | 0 | 0.20 |
| Renal agenesis | 4 | 0 | 2 | 0.61 |
| Cystic kidney | 29 | 0 | 4 | 3.37 |
| Bladder exstrophy | 0 | 0 | 1 | 0.10 |
| Polydactyly, preaxial | 79 | 0 | 0 | 8.06 |
| Total Limb reduction defects (include unspecified) | 32 | 0 | 1 | 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 | 7 | 0 | 1 | 0.82 |
| Omphalocele | 6 | 0 | 3 | 0.92 |
| Gastroschisis | 3 | 0 | 3 | 0.61 |
| Unspecified Omphalocele / Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 1 | 5 | 0.71 |
| Trisomy 18 | 3 | 0 | 14 | 1.73 |
| Down syndrome, all ages (include age unknown) | 82 | 0 | 60 | 14.49 |
| <20 | 5 | 0 | 2 | 11.05 |
| 20-24 | 15 | 0 | 1 | 9.52 |
| 25-29 | 18 | 0 | 3 | 5.76 |
| 30-34 | 24 | 0 | 13 | 13.47 |
| 35-39 | 18 | 0 | 28 | 48.99 |
| 40+ | 7 | 0 | 15 | 141.84 |
| unknown | 0 | 0 | 0 | 0.00 |

NOTE1: Epispadias included in Hypospadias
NOTE2: Only isolated birth defects are reported
nr = not reported

# Hungary: Previous years rates 1974 - 2005

Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1994

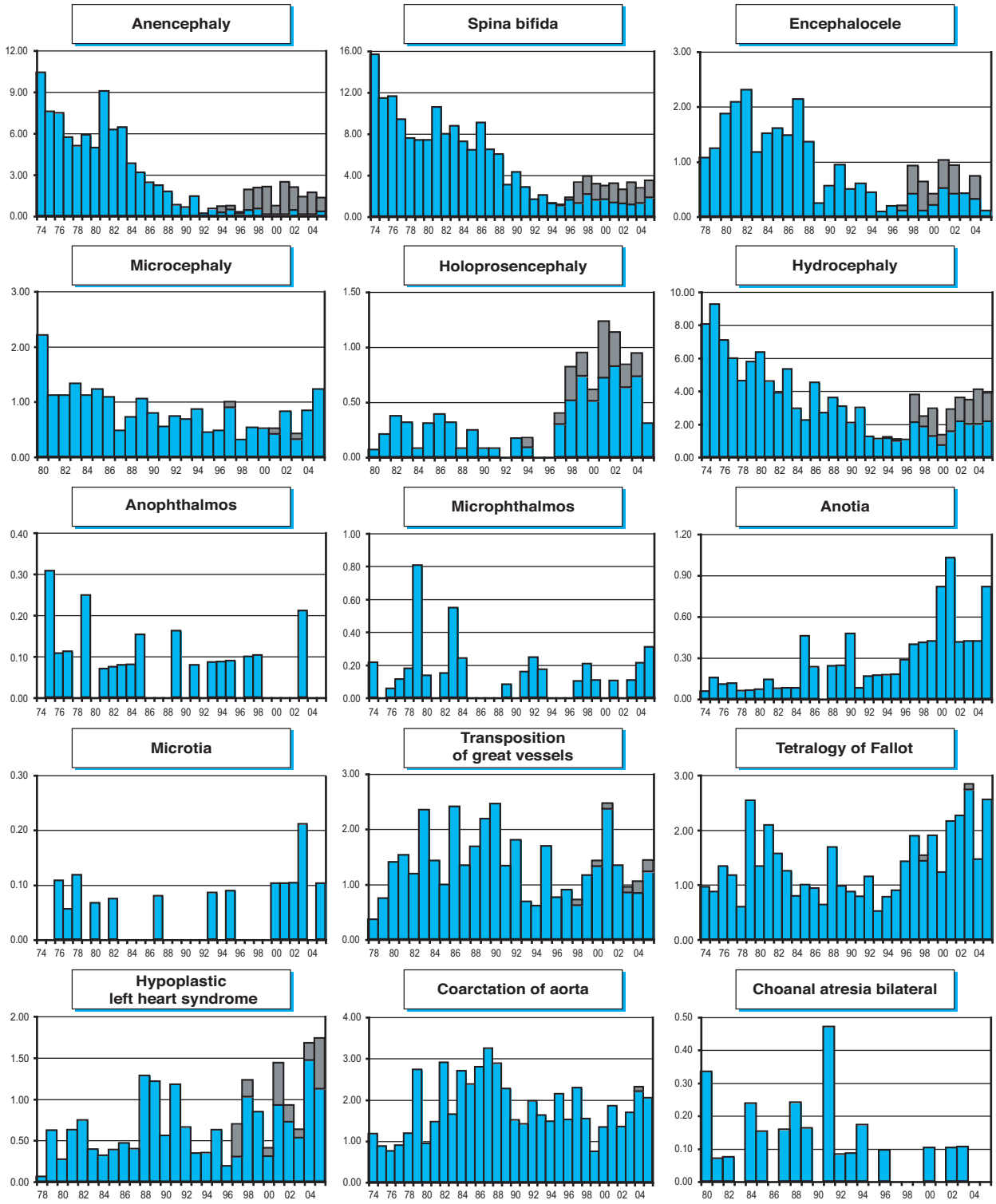
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **1,230,902** | **663,967** | **631,149** | **595,926** | **497,607** | **483,716** |
| Anencephaly | 6.82 | 5.80 | 1.57 | 0.70 | 1.39 | 1.78 |
| Spina bifida | 10.23 | 8.22 | 5.80 | 1.81 | 2.99 | 3.04 |
| Encephalocele | 1.37* | 1.75 | 1.16 | 0.52 | 0.46 | 0.64 |
| Microcephaly | 2.20* | 1.17 | 0.82 | 0.65 | 0.56 | 0.76 |
| Holoprosencephaly | 0.07* | 0.26 | 0.22 | 0.08 | 0.54 | 0.89 |
| Hydrocephaly | 6.80 | 3.80 | 3.18 | 1.54 | 2.29 | 3.58 |
| Anophthalmos | 0.11 | 0.09 | 0.03 | 0.07 | 0.04 | 0.04 |
| Microphthalmos | 0.20 | 0.18 | 0.02 | 0.12 | 0.08 | 0.14 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.09 | 0.17 | 0.24 | 0.15 | 0.46 | 0.62 |
| Microtia | 0.05 | 0.02 | 0.02 | 0.03 | 0.02 | 0.10 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 0.81* | 1.49 | 2.01 | 1.22 | 0.98 | 1.45 |
| Tetralogy of Fallot | 1.23 | 1.36 | 1.01 | 0.82 | 1.59 | 2.25 |
| Hypoplastic left heart syndrome | 0.31* | 0.50 | 0.78 | 0.64 | 0.66 | 1.28 |
| Coarctation of aorta | 1.19 | 2.20 | 2.54 | 1.71 | 1.49 | 1.84 |
| Choanal atresia, bilateral | 0.33* | 0.11 | 0.11 | 0.17 | 0.04 | 0.04 |
| Cleft palate without cleft lip | 3.85 | 4.70 | 3.72 | 3.14 | 2.75 | 3.43 |
| Cleft lip with or without cleft palate | 10.87 | 11.22 | 9.38 | 8.64 | 6.43 | 6.95 |
| Oesophageal atresia / stenosis with or without fistula | 1.96* | 1.64 | 1.93 | 1.11 | 0.96 | 1.24 |
| Small intestine atresia / stenosis | 1.33* | 1.43 | 1.28 | 0.96 | 0.52 | 1.03 |
| Anorectal atresia / stenosis | 2.14* | 2.26 | 1.90 | 1.31 | 0.88 | 1.12 |
| Undescended testis (36 weeks of gestation or later) | 15.68* | 17.65 | 16.43 | 14.83 | 9.45 | 14.86 |
| Hypospadias | 16.28 | 21.19 | 21.22 | 21.13 | 18.99 | 23.84 |
| Epispadias | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | 0.13* | 0.35 | 0.33 | 0.17 | 0.12 | 0.45 |
| Renal agenesis | 1.41* | 0.83 | 1.20 | 0.59 | 0.18 | 0.48 |
| Cystic kidney | 0.00* | 0.05 | 0.24 | 0.44 | 1.63 | 2.85 |
| Bladder exstrophy | 0.13* | 0.42 | 0.35 | 0.05 | 0.08 | 0.10 |
| Polydactyly, preaxial | 0.00* | 1.20 | 2.11 | 1.14 | 6.43 | 8.15 |
| Total Limb reduction defects (include unspecified) | nr | 4.35* | 3.90 | 2.72 | 3.22 | 3.12 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 1.96 | 2.35 | 2.19 | 1.38 | 0.84 | 0.54 |
| Omphalocele | nr | 1.98* | 1.14 | 0.72 | 0.86 | 1.16 |
| Gastroschisis | nr | 0.46* | 0.52 | 0.59 | 0.58 | 0.79 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | nr | 0.00 | 0.10 | 0.00 |
| Trisomy 13 | nr | 0.17* | 0.24 | 0.20 | 0.26 | 0.76 |
| Trisomy 18 | nr | 0.25* | 0.30 | 0.22 | 0.72 | 1.72 |
| Down syndrome, all ages (include age unknown) | 8.98 | 8.03 | 8.32 | 8.16 | 8.32 | 14.06 |
| <20 | nr | 1.80* | 1.97 | 1.64 | 2.77 | 6.52 |
| 20-24 | nr | 2.11* | 2.88 | 2.06 | 3.58 | 7.47 |
| 25-29 | nr | 3.55* | 4.28 | 2.75 | 4.42 | 7.99 |
| 30-34 | nr | 5.02* | 5.50 | 4.52 | 6.71 | 13.93 |
| 35-39 | nr | 11.82* | 17.97 | 18.28 | 21.12 | 41.01 |
| 40-44 | nr | 57.93* | 57.61 | 78.04 | 106.78 | 164.09 |
| 45+ | nr | nr | nr | nr | nr | nr |
| unspecified | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

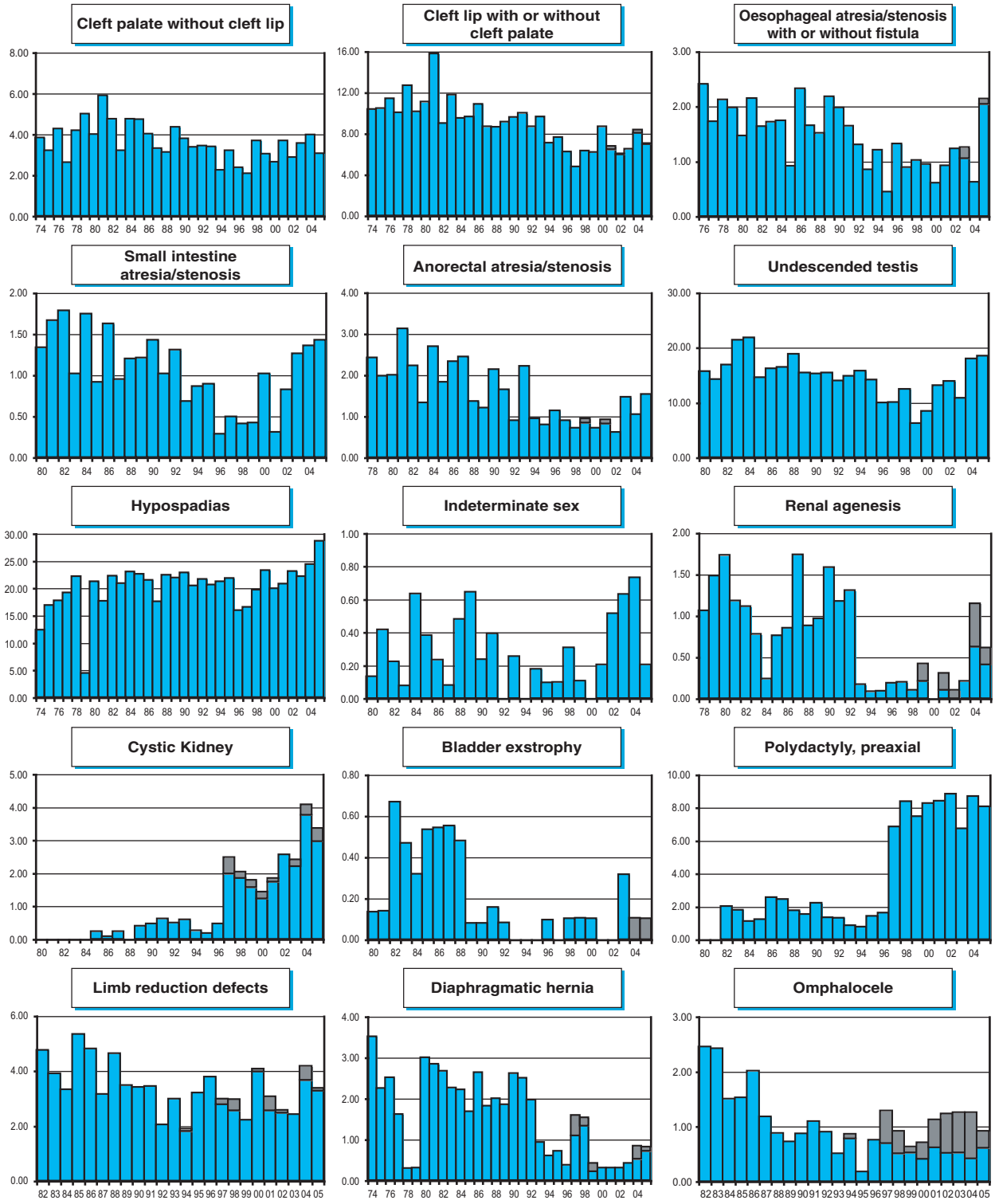
## Hungary

'Time trends 1974-2005 (Birth prevalence rates per 10,000)



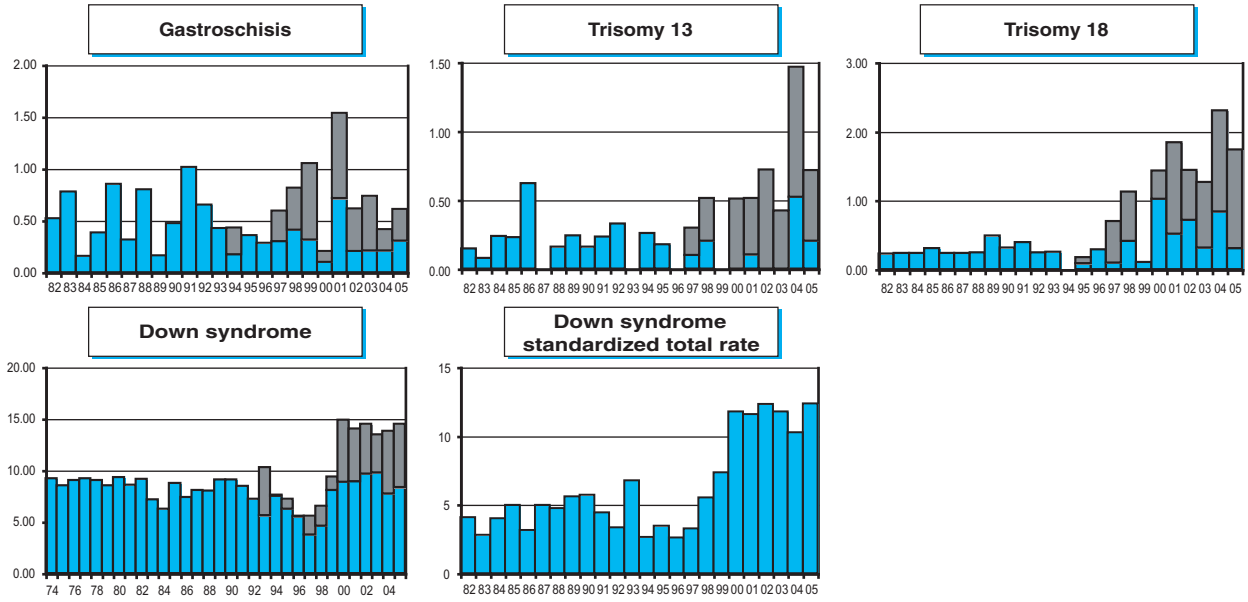

**Note:** L+S rates, ToP rates

## Hungary




**Note:** L+S rates, ToP rates

## Hungary




**Note:** L+S rates, ToP rates

## Iran: TROCA

Tabriz Registry of Congenital Anomalies (TROCA)

**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.

**Size and coverage:**
TROCA is a hospital-based registry and situated in the North-West 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 (15000 births per year) in the area.

**Legislation and funding:**
The programme has been financially supported by the National Public Health Management Centre (NPMC) as a research grant. TROCA is located in the Alzahra University hospital of Tabriz University of Medical Sciences.

**Exposure information:**
Some exposure information are 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.

**Addresses and Staff:**
Saeed Dastgiri, PhD, Programme Director
Department of Community and Family Medicine
School of Medicine
Tabriz University of Medical Sciences
Tabriz, Iran
**Phone:** 98 914 415 7039
**Fax:** 98 411 336 4668
**E-mail:** saeed.dastgiri@gmail.com
**Web:** http://www.tbzmed.ac.ir/troca

# Iran: 2005

| | |
|---|---|
| Live births (LB) | 20,477 |
| Stillbirths (SB) | 225 |
| Total births | 20,702 |
| Number of terminations of pregnancy (ToP) for birth defects | 17 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 13 | 19 | nr | 15.46 |
| Spina bifida | 2 | nr | nr | 0.97 |
| Encephalocele | 1 | nr | nr | 0.48 |
| Microcephaly | 1 | 1 | nr | 0.97 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | 26 | 2 | nr | 13.53 |
| Anophthalmos | nr | nr | nr | nr |
| Microphthalmos | 1 | nr | nr | 0.48 |
| Unspecified Anophthalmos/ Microphthalmos | 1 | nr | nr | 0.48 |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 1 | nr | nr | 0.48 |
| Tetralogy of Fallot | nr | nr | nr | nr |
| Hypoplastic left heart syndrome | nr | nr | nr | nr |
| Coarctation of aorta | nr | nr | nr | nr |
| Choanal atresia, bilateral | nr | nr | nr | nr |
| Cleft palate without cleft lip | 8 | 1 | nr | 4.35 |
| Cleft lip with or without cleft palate | 5 | nr | nr | 2.42 |
| Oesophageal atresia / stenosis with or without fistula | 4 | nr | nr | 1.93 |
| Small intestine atresia / stenosis | nr | nr | nr | nr |
| Anorectal atresia / stenosis | 3 | nr | nr | 1.45 |
| Undescended testis (36 weeks of gestation or later) | 55 | 1 | nr | 27.05 |
| Hypospadias | 14 | nr | nr | 6.76 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 2 | nr | nr | 0.97 |
| Cystic kidney | nr | nr | nr | nr |
| Bladder exstrophy | nr | nr | nr | nr |
| Polydactyly, preaxial | 17 | nr | nr | 8.21 |
| Total Limb reduction defects (include unspecified) | 42 | nr | nr | 20.29 |
| 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 | 42 | nr | nr | 20.29 |
| Diaphragmatic hernia | nr | nr | nr | nr |
| Omphalocele | nr | nr | nr | nr |
| Gastroschisis | nr | nr | nr | nr |
| Unspecified Omphalocele/Gastroschisis | nr | nr | nr | nr |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | nr | nr | nr | nr |
| Trisomy 18 | 2 | nr | nr | 0.97 |
| Down syndrome, all ages (include age unknown) | 7 | nr | nr | 3.38 |
| <20 | nr | nr | nr | nr |
| 20-24 | 1 | nr | nr | 1.75 |
| 25-29 | nr | nr | nr | nr |
| 30-34 | 1 | nr | nr | 3.68 |
| 35-39 | 1 | nr | nr | 10.67 |
| 40-44 | nr | nr | nr | nr |
| 45+ | nr | nr | nr | nr |
| unknown | 4 | nr | nr | 1.93 |

nr = not reported

# Ireland
## Dublin EUROCAT Registry

**Ireland**
**Dublin EUROCAT Registry**

**History:**
Register began in September 1979 and joined EUROCAT at the same time. Joined International Clearinghouse in 1997.

**Size and coverage:**
The Registry is population-based and situated in the East of Ireland covering the counties of Dublin, Wicklow and Kildare. About one third (22,000 births) of all births in Ireland occur in this region.

**Legislation and funding:**
The Registry is located within the Population Health Directorate of the Health Service Executive. Staffing includes a full time Research Nurse and a part time secretary and is led by a Specialist in Public Health Medicine. Funding is provided by the Department of Health through the Health Service Executive. There is a Steering Committee comprised of specialists from each of Maternity and Paediatric Hospitals in the catchment plus a representative from the Department of Health.

**Exposure information**:
For each malformed infant reported, very limited information is given on certain exposures. No information is available on controls.

**Sources of ascertainment:**
All live and still births included. Termination of pregnancy is not legal in Ireland.

**Addresses and Staff:**
Bob McDonnell, MD, Programme Director
Population Health Directorate
Health Service Executive
Dr. Steeven's Hospital
Dublin 8 - Ireland
**Phone:** 353-1-6352752
**Fax:** 353-1-6353745
**E-mail:** bob.mcdonnell@mailf.hse.ie

Virginia Delany, Registry Co-ordinator/Research nurse
Population Health Directorate
Health Service Executive
Dr. Steeven's Hospital
Dublin 8 - Ireland
**Phone:** 353-1-6352751
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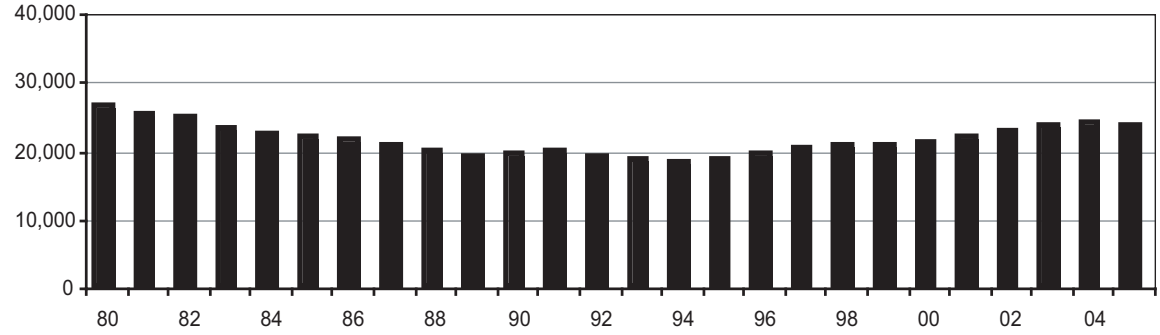
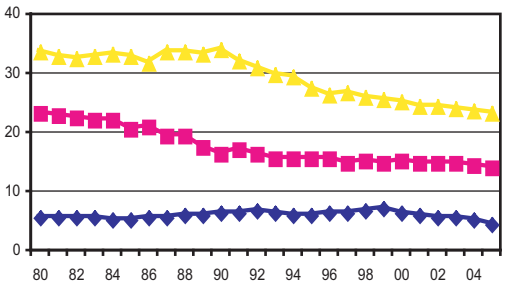
**E-mail:** virginia.delaney@mailf.hse.ie

## Ireland: Dublin

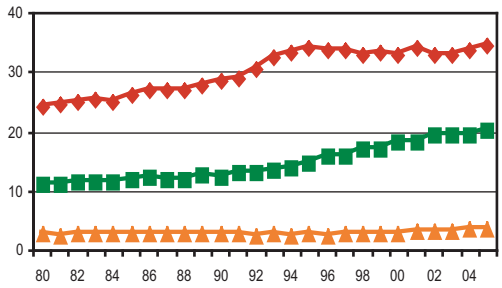
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# Ireland: Dublin, 2005

| | |
|---|---|
| Live births (LB)* | 23,306 |
| Stillbirths (SB)* | 130 |
| Total births* | 23,436 |
| 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 | 2 | 3 | | 2.13 |
| Spina bifida | 4 | 0 | | 1.71 |
| Encephalocele | 1 | 1 | | 0.85 |
| Microcephaly | 3 | 1 | | 1.71 |
| Holoprosencephaly | 2 | 2 | | 1.71 |
| Hydrocephaly | 2 | 1 | | 1.28 |
| Anophthalmos | 0 | 0 | | 0.00 |
| Microphthalmos | 3 | 0 | | 1.28 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 0 | 0 | | 0.00 |
| Microtia | 1 | 0 | | 0.43 |
| Unspecified Anotia/Microtia | 0 | 0 | | 0.00 |
| Transposition of great vessels | 9 | 1 | | 4.27 |
| Tetralogy of Fallot | 12 | 0 | | 5.12 |
| Hypoplastic left heart syndrome | 7 | 2 | | 3.84 |
| Coarctation of aorta | 12 | 0 | | 5.12 |
| Choanal atresia, bilateral | 4 | 0 | | 1.71 |
| Cleft palate without cleft lip | 17 | 0 | | 7.25 |
| Cleft lip with or without cleft palate | 19 | 0 | | 8.11 |
| Oesophageal atresia / stenosis with or without fistula | 5 | 1 | | 2.56 |
| Small intestine atresia / stenosis | 2 | 0 | | 0.85 |
| Anorectal atresia / stenosis | 11 | 0 | | 4.69 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | | nr |
| Hypospadias | 16 | 0 | | 6.83 |
| Epispadias | nr | nr | | nr |
| Indeterminate sex | 0 | 0 | | 0.00 |
| Renal agenesis | 4 | 2 | | 2.56 |
| Cystic kidney | 10 | 1 | | 4.69 |
| Bladder exstrophy | 4 | 0 | | 1.71 |
| Polydactyly, preaxial | 15 | 1 | | 6.83 |
| Total Limb reduction defects (include unspecified) | 5 | 1 | | 2.56 |
| 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 | 5 | 0 | | 2.13 |
| Omphalocele | 6 | 2 | | 3.41 |
| Gastroschisis | 9 | 0 | | 3.84 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | | 0.00 |
| Prune belly sequence | 0 | 0 | | 0.00 |
| Trisomy 13 | 5 | 2 | | 2.99 |
| Trisomy 18 | 5 | 8 | | 5.55 |
| Down syndrome, all ages (include age unknown) | 52 | 1 | | 22.61 |
| <20 | 2 | 0 | | 19.96 |
| 20-24 | 3 | 0 | | 9.30 |
| 25-29 | 2 | 0 | | 3.69 |
| 30-34 | 14 | 0 | | 17.40 |
| 35-39 | 22 | 0 | | 46.89 |
| 40-44 | 8 | 1 | | 106.89 |
| 45+ | 1 | 0 | | 434.78 |
| unknown | 0 | 0 | | 0.00 |

nr = not reported
* estimated

# Ireland: Dublin, Previous years rates 1980 - 2005

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

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **26,202** | **117,167** | **99,902** | **93,658** | **101,861** | **115,776** |
| Anencephaly | 17.94 | 13.23 | 8.21 | 4.59 | 3.24 | 2.85 |
| Spina bifida | 2.67 | 16.64 | 10.01 | 6.19 | 5.20 | 3.80 |
| Encephalocele | 1.53 | 2.73 | 1.30 | 2.78 | 1.47 | 1.12 |
| Microcephaly | 3.43 | 4.18 | 2.70 | 4.06 | 4.32 | 3.37 |
| Holoprosencephaly | 0.00 | 0.34 | 0.40 | 0.64 | 1.28 | 1.21 |
| Hydrocephaly | nr | nr | nr | nr | 2.06 | 2.25 |
| Anophthalmos | 0.00 | 0.34 | 0.00 | 0.43 | 0.59 | 0.17 |
| Microphthalmos | 0.00 | 0.85 | 1.30 | 1.17 | 2.85 | 0.86 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | nr | nr | 0.11* |
| Microtia | nr | nr | nr | nr | nr | 0.11* |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | nr | nr | nr | nr | 5.10 | 3.89 |
| Tetralogy of Fallot | 2.29 | 2.99 | 2.60 | 3.10 | 3.73 | 2.94 |
| Hypoplastic left heart syndrome | 2.29 | 2.05 | 2.10 | 2.24 | 1.96 | 2.59 |
| Coarctation of aorta | 3.82 | 4.78 | 6.81 | 5.87 | 6.09 | 6.82 |
| Choanal atresia, bilateral | 0.38 | 0.34 | 0.70 | 1.17 | 1.87 | 1.04 |
| Cleft palate without cleft lip | 7.25 | 7.00 | 6.71 | 8.54 | 7.66 | 8.72 |
| Cleft lip with or without cleft palate | 9.92 | 9.64 | 7.31 | 9.40 | 8.25 | 7.69 |
| Oesophageal atresia / stenosis with or without fistula | 4.58 | 3.76 | 3.40 | 3.20 | 3.44 | 2.16 |
| Small intestine atresia / stenosis | 2.29 | 2.90 | 2.70 | 2.03 | 2.26 | 0.86 |
| Anorectal atresia / stenosis | 2.67 | 3.41 | 4.30 | 2.88 | 2.06 | 2.94 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 12.21 | 15.28 | 10.61 | 12.71 | 18.85 | 11.83 |
| Epispadias | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | 0.00 | 0.17 | 0.30 | 0.11 | 0.39 | 0.26 |
| Renal agenesis | 5.34 | 5.29 | 3.70 | 4.06 | 4.81 | 1.64 |
| Cystic kidney | 1.14 | 4.01 | 1.80 | 4.59 | 3.34 | 3.37 |
| Bladder exstrophy | nr | nr | nr | nr | 0.49 | 1.04 |
| Polydactyly, preaxial | 8.01 | 6.66 | 4.90 | 5.66 | 7.56 | 8.90 |
| Total Limb reduction defects (include unspecified) | 4.58 | 3.84 | 4.20 | 3.95 | 4.61 | 4.06 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 3.05 | 3.24 | 4.20 | 4.80 | 4.42 | 3.37 |
| Omphalocele | 2.29 | 2.56 | 2.60 | 2.14 | 2.65 | 3.71 |
| Gastroschisis | 0.00 | 0.34 | 0.40 | 1.17 | 2.65 | 3.02 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.00 | 0.09 | 0.50 | 0.53 | 0.39 | 0.69 |
| Trisomy 13 | 1.14 | 1.02 | 1.20 | 0.64 | 3.44 | 2.07 |
| Trisomy 18 | 1.91 | 2.30 | 2.00 | 3.20 | 4.12 | 4.23 |
| Down syndrome, all ages (include age unknown) | 21.75 | 18.44 | 17.92 | 20.93 | 23.27 | 20.64 |
| <20 | nr | nr | nr | 17.96* | 7.57 | 8.38 |
| 20-24 | nr | nr | nr | 7.84* | 7.89 | 7.77 |
| 25-29 | nr | nr | nr | 9.68* | 10.30 | 5.83 |
| 30-34 | nr | nr | nr | 17.85* | 18.25 | 13.40 |
| 35-39 | nr | nr | nr | 42.21* | 51.61 | 45.59 |
| 40-44 | nr | nr | nr | 164.38* | 144.81 | 130.60 |
| 45+ | nr | nr | nr | 869.57* | 560.75 | 192.31 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

## Ireland: Dublin

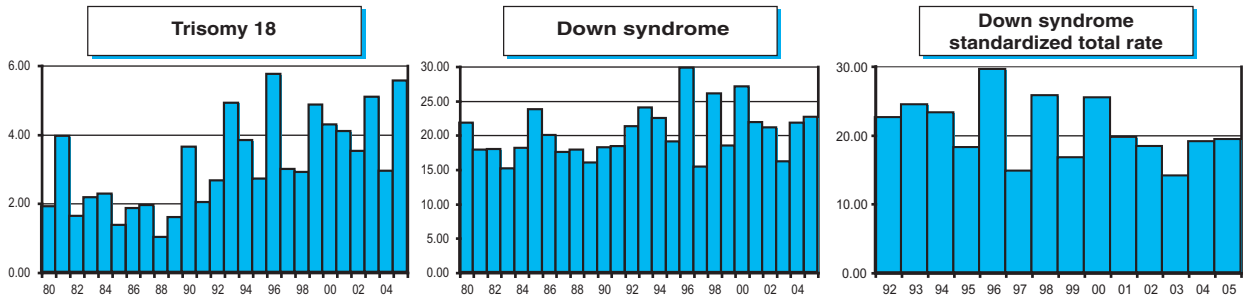
Time trends 1980-2005 (Birth prevalence rates per 10,000)


**Note:** L+S rates

## Ireland: Dublin


**Note:** L+S rates

## Ireland: Dublin




**Note:** L+S rates

## Israel: IBDSP

Israel Birth Defects Surveillance Program

**History :**
The program started in one hospital in 1966 and was a founder member of the Clearinghouse. It was a full member until 1986, when it became an associate member.

**Size and coverage:**
Reports are now obtained from five hospitals: three are located in the central region of the country, one in the south ( with the largest number of births in the country ) and one in the north. The total number of births is now around 40,000 per year ( 27% of all births in Israel).
Stillbirths of 20 weeks gestation or more and 500 grams or more are included.
The registry of termination of pregnancy began in 1995.

**Legislation and funding:**
The Registry is a research program supported by research grants without any governmental support.

**Sources and ascertainment:**
Reporting is voluntary. Reports are obtained from the Departments of Neonatology in the participating hospitals. The included hospitals are:
Rabin Medical Center , Beilinson Hospital , Petah Tikva ( Prof Sirota, Prof Linder )
Lis Medical Center, Tel-Aviv ( Prof Dolberg);
these two hospitals are affiliated to Sackler School of Medicine, Tel-Aviv University.
Kaplan Hospital, Rehovot ( Prof Shinwell) affiliated to Hebrew University,Jerusalem.
The hospital in the south is Soroka Medical Center, Beer Sheva (Dr Landau, Prof Zmora ) affiliated to Ben-Gurion University of the Negev, Beer-Sheva.
The hospital in the north is Bnai Zion Medical Center, Haifa ( Dr Bader), affiliated to Faculty of Medicine, Technion, Haifa.

**Exposure information:**
Complete anamneses are obtained by interviews of mothers of all malformed infants. The socio-demographic data of all the other women with normal newborns are also obtained.

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

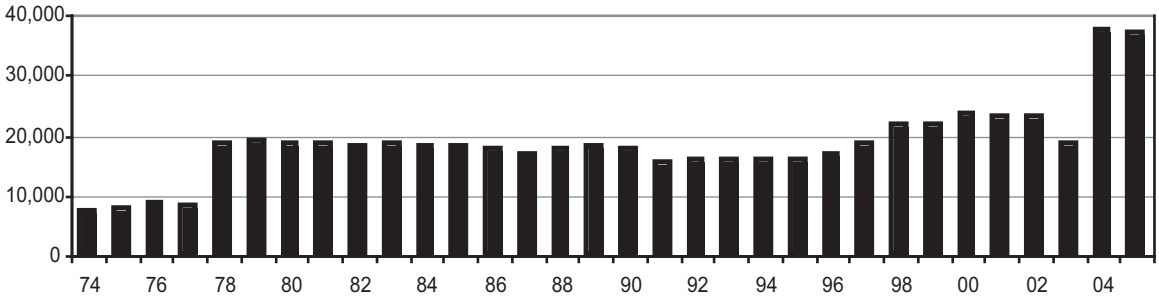
**Addresses and Staff:**
Prof Paul Merlob
Rabin Medical Center and Schneider Children Hospital,
Petah-Tikva, 49100
**Phone:** 972- 052 – 567 22 32
**Fax:** 972 – 3- 922 00 68
**E-mail:** merlobp@post.tau.ac.il

Dr Daniela Landau
Department of Neonatology
Soroka Medical Center
Beer-Sheva
**Phone:** 972 – 8- 6400272
**Fax:** 972 – 8 – 6400545

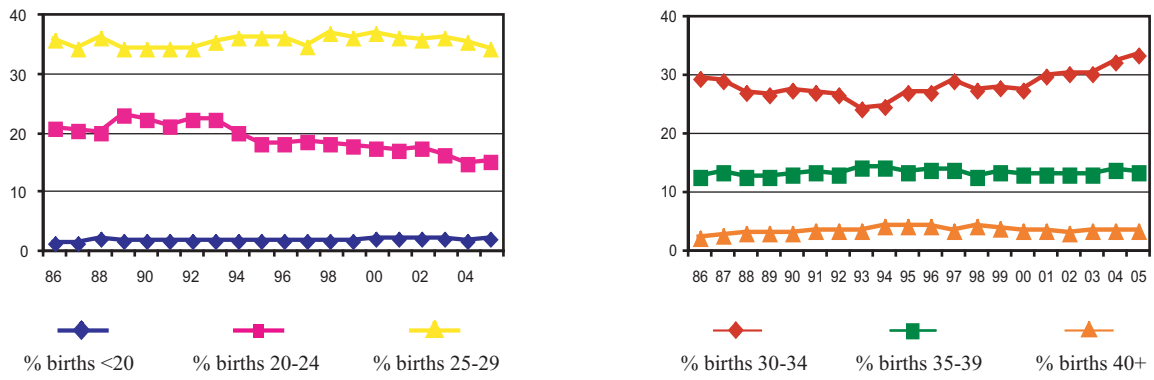
**E-mail:** landaud@bgumail.bgu.ac.il

## Israel: IBDSP

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 6 | 26.1 | Cystic kidney | 1 | 5.3 |
| Spina bifida | 10 | 28.6 | Limb reduction defects | 2 | 7.4 |
| Encephalocele | 2 | 66.7 | Diaphragmatic hernia | 3 | 15.0 |
| Holoprosencephaly | 2 | 50.0 | Omphalocele | 2 | 22.2 |
| Hydrocephaly | 11 | 21.6 | Gastroschisis | 0 | 0.0 |
| Hypoplastic left heart syndrome | 5 | 22.7 | Trisomy 13 | 2 | 40.0 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 2 | 14.3 |
| Cleft lip with or without cleft palate | 4 | 9.1 | Down syndrome | 37 | 38.1 |
| Renal agenesis | 1 | 7.7 | | | |

Total ToPs with births defects = 112 (Ratio ToPs/Births: 1.20 per 1,000)
*ToPs/ToPs+Births

# Israel: IBDMS, 2005

| | |
|---|---|
| Live births (LB) | 36,598 |
| Stillbirths (SB) | 331 |
| Total births | 36,929 |
| Number of terminations of pregnancy (ToP) for birth defects | 37 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 5 | 1 | 0 | 1.62 |
| Spina bifida | 7 | 2 | 5 | 3.79 |
| Encephalocele | 0 | 0 | 0 | 0.00 |
| Microcephaly | 6 | 0 | 1 | 1.90 |
| Holoprosencephaly | 2 | 0 | 1 | 0.81 |
| Hydrocephaly | 14 | 1 | 0 | 4.06 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 1 | 0 | 0 | 0.27 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 1 | 0 | 0 | 0.27 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 12 | 0 | 3 | 4.06 |
| Tetralogy of Fallot | 17 | 0 | 2 | 5.15 |
| Hypoplastic left heart syndrome | 6 | 0 | 4 | 2.71 |
| Coarctation of aorta | 13 | 1 | 2 | 4.33 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 20 | 0 | 0 | 5.42 |
| Cleft lip with or without cleft palate | 17 | 0 | 1 | 4.87 |
| Oesophageal atresia/stenosis with or without fistula | 13 | 0 | 0 | 3.52 |
| Small intestine atresia/stenosis | 1 | 0 | 0 | 0.27 |
| Anorectal atresia/stenosis | 5 | 0 | 0 | 1.35 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 108 | 0 | 0 | 29.25 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 5 | 0 | 0 | 1.35 |
| Cystic kidney | 9 | 0 | 0 | 2.44 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 4 | 0 | 0 | 1.08 |
| Total Limb reduction defects (include unspecified) | 7 | 0 | 1 | 2.17 |
| Transverse | 3 | 0 | 1 | 1.08 |
| Preaxial | 3 | 0 | 0 | 0.81 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 1 | 0 | 0 | 0.27 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 8 | 0 | 0 | 2.17 |
| Omphalocele | 2 | 0 | 2 | 1.08 |
| Gastroschisis | 1 | 0 | 0 | 0.27 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 2 | 1 | 0 | 0.81 |
| Trisomy 13 | 1 | 0 | 1 | 0.54 |
| Trisomy 18 | 5 | 0 | 0 | 1.35 |
| Down syndrome, all ages (include age unknown) | 23 | 0 | 14 | 10.02 |
| <20 | 1 | 0 | 0 | 15.92 |
| 20-24 | 3 | 0 | 0 | 5.49 |
| 25-29 | 4 | 0 | 1 | 3.99 |
| 30-34 | 4 | 0 | 5 | 7.34 |
| 35-39 | 5 | 0 | 5 | 20.36 |
| 40-44 | 6 | 0 | 3 | 90.27 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# Israel: IBDMS, Previous years rates 1974 - 2005

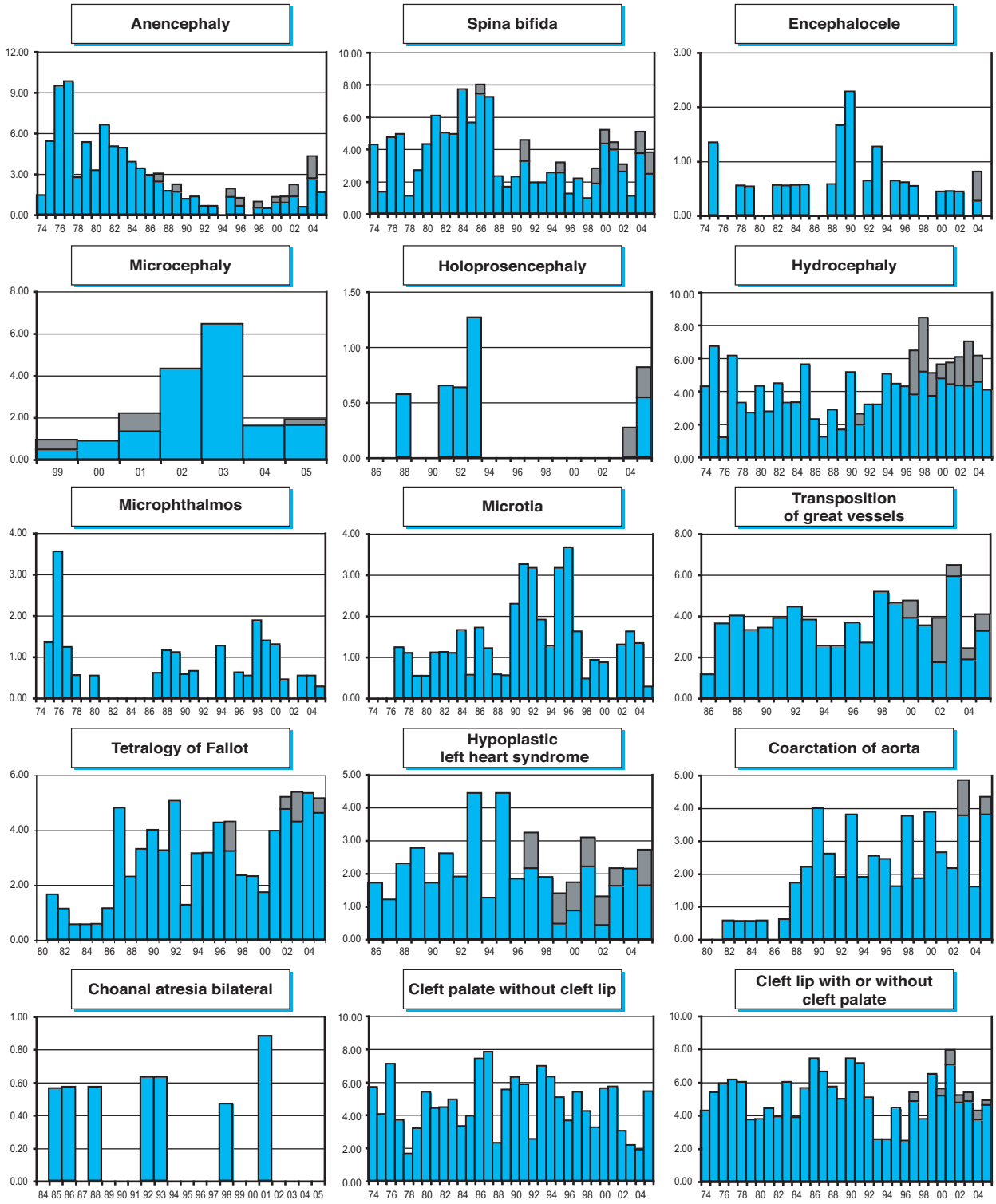
Birth prevalence rates: (LB+SB) * 10,000 until 1985
Birth prevalence rates: (LB+SB+TOP) * 10,000 since 1986

| | 1974-80 | 1981-85 | 1986-90 | 1991-95 | 1996-00 | 2001-05 |
|---|---|---|---|---|---|---|
| **Births** | **86,899** | **90,506** | **87,374** | **78,764** | **101,249** | **138,884** |
| Anencephaly | 4.83 | 4.75 | 2.17 | 0.89 | 0.79 | 2.23 |
| Spina bifida | 3.11 | 5.86 | 4.23 | 2.79 | 2.57 | 3.74 |
| Encephalocele | 0.35 | 0.44 | 0.92 | 0.51 | 0.30 | 0.36 |
| Microcephaly | nr | nr | 0.00 | 0.00 | 0.40 | 2.88 |
| Arhinencephaly/Holoprosencephaly | nr | 0.28 | 0.11 | 0.51 | 0.00 | 0.29 |
| Hydrocephaly | 3.80 | 3.87 | 2.63 | 3.68 | 6.02 | 5.62 |
| Anophthalmos | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 | 0.14 |
| Microphthalmos | 0.81 | 0.00 | 0.69 | 0.38 | 1.19 | 0.36 |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.00 | 0.00 | 0.00 | 0.13 | 0.00 | 0.00 |
| Microtia | 0.58 | 1.10 | 1.26 | 2.54 | 1.38 | 0.86 |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | nr | nr | 3.09 | 3.43 | 4.25 | 3.82 |
| Tetralogy of Fallot | nr | 0.88 | 3.09 | 3.17 | 2.86 | 5.04 |
| Hypoplastic left heart syndrome | nr | nr | 1.95 | 2.92 | 1.98 | 2.30 |
| Coarctation of aorta | nr | 0.44 | 1.72 | 2.54 | 2.77 | 3.02 |
| Choanal atresia, bilateral | nr | 0.28* | 0.23 | 0.25 | 0.10 | 0.14 |
| Cleft palate without cleft lip | 4.03 | 4.20 | 5.84 | 5.33 | 4.44 | 3.67 |
| Cleft lip with or without cleft palate | 4.83 | 4.75 | 6.41 | 4.32 | 4.84 | 5.33 |
| Oesophageal atresia/stenosis with or without fistula | 1.61 | 1.66 | 3.09 | 3.81 | 2.27 | 2.09 |
| Small intestine atresia/stenosis | nr | 0.83* | 1.37 | 1.27 | 0.40 | 1.01 |
| Anorectal atresia/stenosis | 1.84 | 3.09 | 4.23 | 3.30 | 2.37 | 1.58 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 28.42 | 26.52 | 34.68 | 41.77 | 36.44 | 34.27 |
| Epispadias | 0.12 | 0.11 | 0.00 | 0.25 | 0.20 | 0.22 |
| Indeterminate sex | nr | nr | nr | nr | nr | 0.27* |
| Renal agenesis | nr | nr | 0.69 | 0.89 | 0.49 | 0.94 |
| Cystic kidney | 0.46 | 0.99 | 1.26 | 1.14 | 1.78 | 2.02 |
| Bladder exstrophy | 0.12 | 0.22 | 0.92 | 0.25 | 0.30 | 0.50 |
| Polydactyly, preaxial | 0.23 | 0.66 | 0.46 | 0.38 | 1.19 | 1.01 |
| Total Limb reduction defects (include unspecified) | 3.11 | 3.09 | 2.63 | 3.55 | 1.09 | 2.45 |
| Transverse | nr | 0.69* | 1.26 | 1.52 | 0.40 | 1.08 |
| Preaxial | nr | 0.69* | 0.57 | 0.25 | 0.49 | 1.08 |
| Postaxial | nr | 0.42* | 0.11 | 0.76 | 0.00 | 0.07 |
| Intercalary | nr | 0.28* | 0.34 | 0.25 | 0.20 | 0.14 |
| Mixed | nr | 0.69* | 0.34 | 0.76 | 0.00 | 0.07 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 2.15* | 2.65 | 1.83 | 2.67 | 1.58 | 2.02 |
| Omphalocele | 1.61 | 2.54 | 0.80 | 1.27 | 0.40 | 0.94 |
| Gastroschisis | 0.00 | 0.77 | 0.23 | 0.00 | 0.20 | 0.36 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.46 | 0.11 | 0.11 | 0.00 | 0.10 | 0.29 |
| Trisomy 13 | nr | 0.83* | 0.34 | 0.38 | 0.49 | 0.50 |
| Trisomy 18 | nr | 0.56* | 0.69 | 0.63 | 1.28 | 1.30 |
| Down syndrome, all ages (include age unknown) | 10.24 | 11.05 | 11.90 | 6.86 | 9.09 | 10.08 |
| <20 | nr | nr | nr | nr | nr | 16.31 |
| 20-24 | nr | nr | nr | 0.00 | 2.76 | 4.61 |
| 25-29 | nr | nr | nr | 2.90 | 5.49 | 4.93 |
| 30-34 | nr | nr | nr | 6.91 | 8.23 | 6.91 |
| 35-39 | nr | nr | nr | 17.90 | 16.54 | 19.74 |
| 40-44 | nr | nr | nr | 38.74 | 58.34 | 79.98 |
| 45+ | nr | nr | nr | 79.37 | 81.97 | 92.17 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years
nr = not reported

## Israel: IBDSP

'Time trends 1974-2005 (Birth prevalence rates per 10,000)

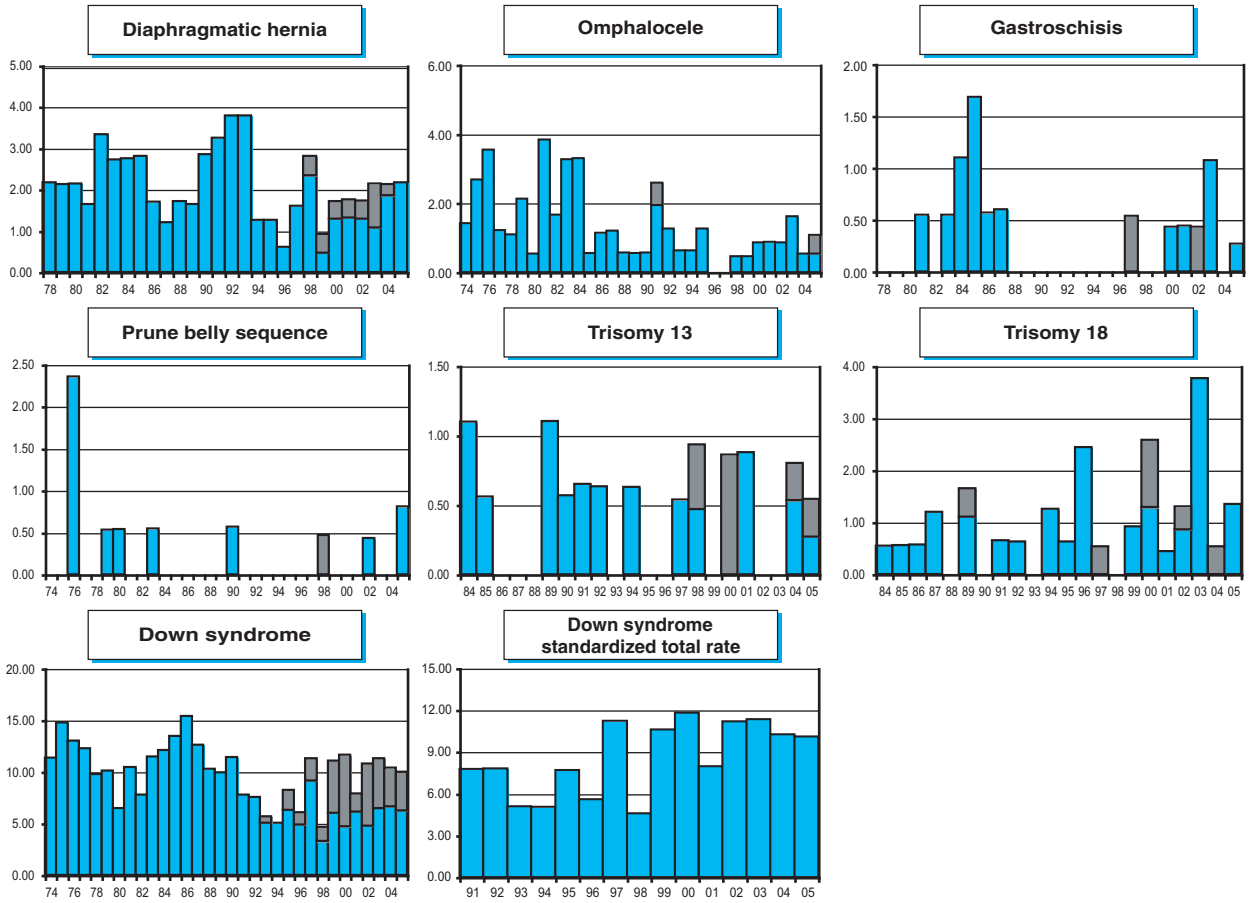



**Note:** L+S rates, ToP rates

## Israel: IBDSP


**Note:** L+S rates, ToP rates

## Israel: IBDSP




**Note:** L+S rates, ToP rates

## Italy: BDRCam

Birth Defects Registry of Campania (BDRCam)

**History**:
The Registry started in 1991and 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.

**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.

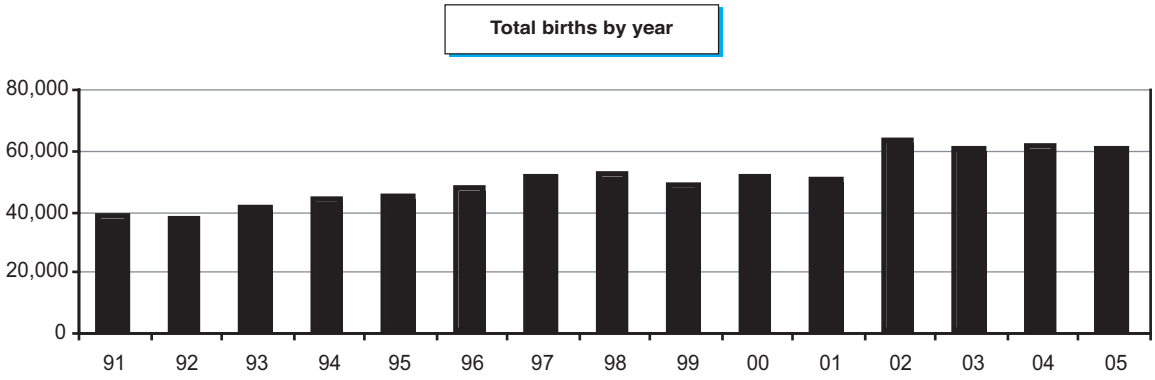
**Sources of ascertainment:**
Reports are obtained from delivery units and pediatric clinics at the participating hospitals. For selected malformations multiple sources are used with follow-up to one year using specific records from pediatric specialties departments dealing with malformed infants.

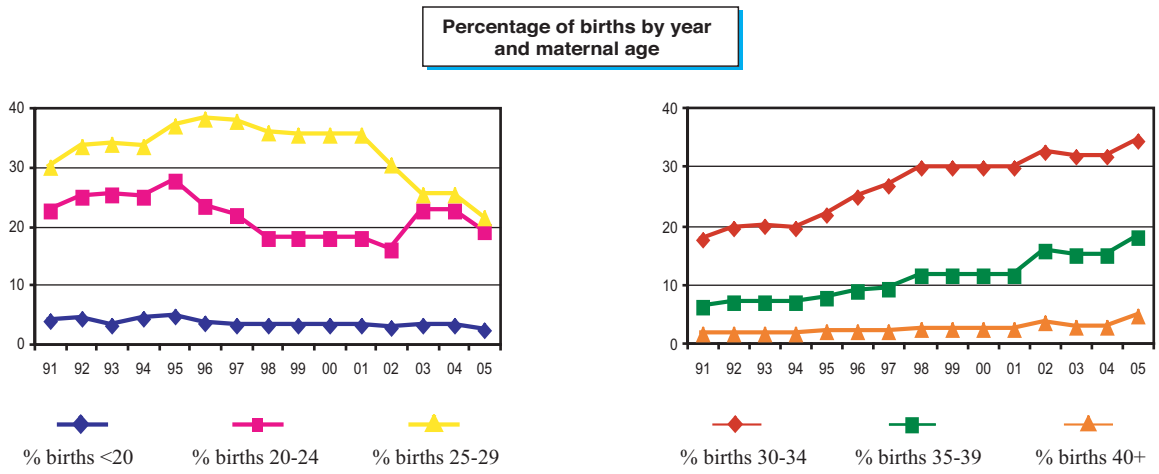
**Exposure information:**
For each malformed infant reported, information is given on certain exposures, including maternal drug usage and parental occupation. Beginning from 2002 informations on controls are available but only partially on induced abortions.

**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),
Medical Genetics Division,
Azienda Ospedaliera "G. Rummo", Via dell'Angelo 1,
82100 Benevento, Italy
**Phone:** 39- 0824-57374
**Fax:** 39-0824-57495
**E-mail:** giorecam@tin.it

## Italy: BDRCam




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 61 | 92.4 | Cystic kidney | 18 | 48.6 |
| Spina bifida | 41 | 80.4 | Limb reduction defects | 21 | 30.9 |
| Encephalocele | 11 | 100.0 | Diaphragmatic hernia | 12 | 36.4 |
| Holoprosencephaly | 13 | 72.2 | Omphalocele | 31 | 77.5 |
| Hydrocephaly | 116 | 92.1 | Gastroschisis | 7 | 100.0 |
| Hypoplastic left heart syndrome | 34 | 89.5 | Trisomy 13 | 11 | 84.6 |
| Cleft palate without cleft lip | 6 | 10.3 | Trisomy 18 | 37 | 84.1 |
| Cleft lip with or without cleft palate | 25 | 25.0 | Down syndrome | 167 | 68.4 |
| Renal agenesis | 21 | 39.6 | | | |

Total ToPs with births defects = 801 (Ratio ToPs/Births: 4.41 per 1,000)
*ToPs/ToPs+Births

# Italy: BDRCam, 2005

| | |
|---|---|
| Live births (LB) | 59,432 |
| Stillbirths (SB) | 114 |
| Total births | 59,546 |
| Number of terminations of pregnancy (ToP) for birth defects | 272 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 23 | 3.86 |
| Spina bifida | 2 | 0 | 8 | 1.68 |
| Encephalocele | 0 | 0 | 4 | 0.67 |
| Microcephaly | 4 | 0 | 1 | 0.84 |
| Holoprosencephaly | 2 | 0 | 3 | 0.84 |
| Hydrocephaly | 6 | 0 | 34 | 6.72 |
| Anophthalmos | 2 | 0 | 0 | 0.34 |
| Microphthalmos | 2 | 0 | 0 | 0.34 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 2 | 0 | 0 | 0.34 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 8 | 0 | 0 | 1.34 |
| Tetralogy of Fallot | 4 | 0 | 4 | 1.34 |
| Hypoplastic left heart syndrome | 1 | 0 | 10 | 1.85 |
| Coarctation of aorta | 5 | 0 | 2 | 1.18 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.34 |
| Cleft palate without cleft lip | 17 | 0 | 1 | 3.02 |
| Cleft lip with or without cleft palate | 23 | 0 | 7 | 5.04 |
| Oesophageal atresia/stenosis with or without fistula | 9 | 0 | 1 | 1.68 |
| Small intestine atresia/stenosis | 6 | 0 | 2 | 1.34 |
| Anorectal atresia/stenosis | 10 | 1 | 3 | 2.35 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 30 | 0 | 0 | 5.04 |
| Epispadias | 3 | 0 | 0 | 0.50 |
| Indeterminate sex | 4 | 0 | 0 | 0.67 |
| Renal agenesis | 9 | 0 | 5 | 2.35 |
| Cystic kidney | 7 | 1 | 5 | 2.18 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 7 | 0 | 0 | 1.18 |
| Total Limb reduction defects (include unspecified) | 13 | 0 | 8 | 3.53 |
| Transverse | 11 | 0 | 5 | 2.69 |
| Preaxial | 1 | 0 | 2 | 0.50 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 0 | 0 | 1 | 0.17 |
| Mixed | 1 | 0 | 0 | 0.17 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 6 | 0 | 7 | 2.18 |
| Omphalocele | 1 | 0 | 8 | 1.51 |
| Gastroschisis | 0 | 0 | 0 | 0.00 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 1 | 0.17 |
| Trisomy 13 | 0 | 0 | 4 | 0.67 |
| Trisomy 18 | 2 | 0 | 18 | 3.36 |
| Down syndrome, all ages (include age unknown) | 16 | 1 | 53 | 11.76 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 2 | 0 | 0 | 1.77 |
| 25-29 | 1 | 0 | 2 | 2.38 |
| 30-34 | 2 | 0 | 9 | 5.42 |
| 35-39 | 7 | 0 | 24 | 24.25 |
| 40-44 | 1 | 1 | 12 | 63.55 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 3 | 0 | 6 | 137.40 |

nr = not reported

# Italy: BDRCam, Previous years rates 1991 - 2005

Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1994

| | 1974-80 | 1981-85 | 1986-90 | 1991-95 | 1996-00 | 2001-05 |
|---|---|---|---|---|---|---|
| **Total births** | | | | **202,482** | **246,252** | **292,752** |
| Anencephaly | | | | 1.83 | 3.05 | 3.25 |
| Spina bifida | | | | 3.16 | 3.33 | 2.56 |
| Encephalocele | | | | 0.64 | 1.02 | 0.79 |
| Microcephaly | | | | 1.04 | 0.77 | 0.79 |
| Holoprosencephaly | | | | 0.44 | 1.22 | 1.02 |
| Hydrocephaly | | | | 4.10 | 5.44 | 6.11 |
| Anophthalmos | | | | 0.64 | 0.37 | 0.20 |
| Microphthalmos | | | | 0.25 | 0.24 | 0.65 |
| Unspecified Anophthalmos / Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | 0.69 | 0.53 | 0.41 |
| Microtia | | | | 0.35 | 0.85 | 0.38 |
| Unspecified Anotia / Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 1.58 | 2.19 | 1.06 |
| Tetralogy of Fallot | | | | 2.72 | 2.40 | 2.22 |
| Hypoplastic left heart syndrome | | | | 0.99 | 1.99 | 1.84 |
| Coarctation of aorta | | | | 1.68 | 1.91 | 1.02 |
| Choanal atresia, bilateral | | | | 0.20 | 0.16 | 0.24 |
| Cleft palate without cleft lip | | | | 4.30 | 4.91 | 3.59 |
| Cleft lip with or without cleft palate | | | | 7.06 | 7.23 | 5.19 |
| Oesophageal atresia / stenosis with or without fistula | | | | 2.27 | 2.03 | 1.74 |
| Small intestine atresia / stenosis | | | | 2.02 | 2.11 | 1.30 |
| Anorectal atresia / stenosis | | | | 3.06 | 3.01 | 2.43 |
| Undescended testis (36 weeks of gestation or later) | | | | nr | nr | nr |
| Hypospadias | | | | 3.51 | 3.53 | 5.47 |
| Epispadias | | | | 0.25 | 0.20 | 0.17 |
| Indeterminate sex | | | | 0.40 | 0.69 | 0.79 |
| Renal agenesis | | | | 1.93 | 3.37 | 3.25 |
| Cystic kidney | | | | 1.78 | 2.60 | 2.08 |
| Bladder exstrophy | | | | 0.35 | 0.16 | 0.00 |
| Polydactyly, preaxial | | | | 1.78 | 1.87 | 1.23 |
| Total Limb reduction defects (include unspecified) | | | | 5.28 | 5.08 | 3.96 |
| Transverse | | | | 3.61 | 2.68 | 2.46 |
| Preaxial | | | | 0.69 | 0.97 | 0.68 |
| Postaxial | | | | 0.25 | 0.49 | 0.51 |
| Intercalary | | | | 0.35 | 0.53 | 0.24 |
| Mixed | | | | 0.15 | 0.12 | 0.07 |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 1.98 | 2.44 | 2.12 |
| Omphalocele | | | | 1.38 | 1.99 | 2.05 |
| Gastroschisis | | | | 0.35 | 0.61 | 0.51 |
| Unspecified Omphalocele / Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.00* | 0.13* | 0.10 |
| Trisomy 13 | | | | 0.84 | 0.61 | 0.72 |
| Trisomy 18 | | | | 1.04 | 1.95 | 1.95 |
| Down syndrome, all ages (include age unknown) | | | | 12.54 | 13.08 | 12.74 |
| <20 | | | | 3.59 | 3.74 | 4.70 |
| 20-24 | | | | 6.51 | 3.51 | 1.92 |
| 25-29 | | | | 7.51 | 5.25 | 4.78 |
| 30-34 | | | | 12.77 | 9.71 | 7.38 |
| 35-39 | | | | 36.46 | 28.39 | 31.26 |
| 40-44 | | | | 102.45 | 125.31 | 89.71 |
| 45+ | | | | 197.37 | 226.24 | 70.30 |
| unknown | | | | --- | --- | --- |

* data include less than 5 years

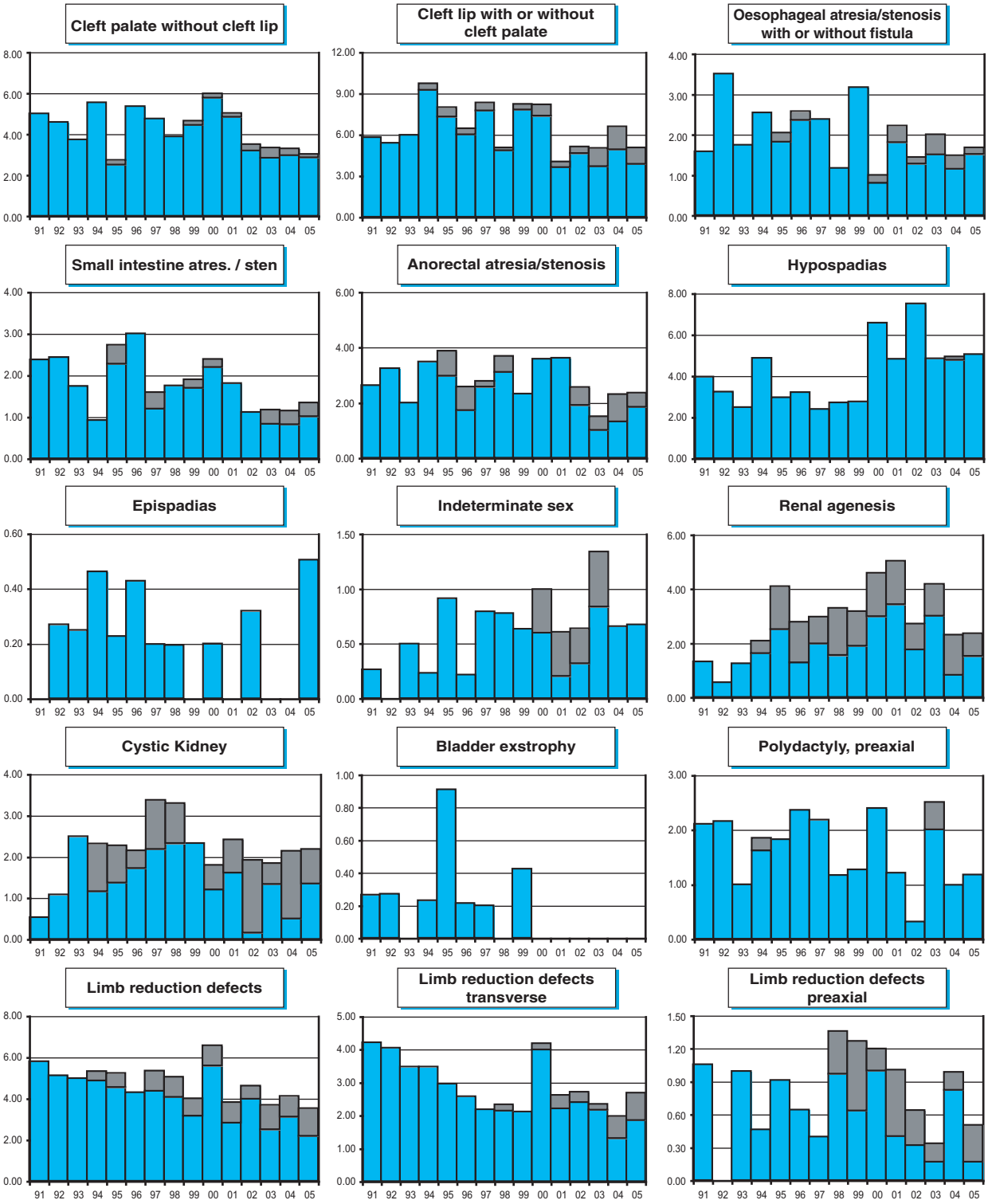
nr = not reported

## Italy: BDRCam

'Time trends 1991-2005 (Birth prevalence rates per 10,000)

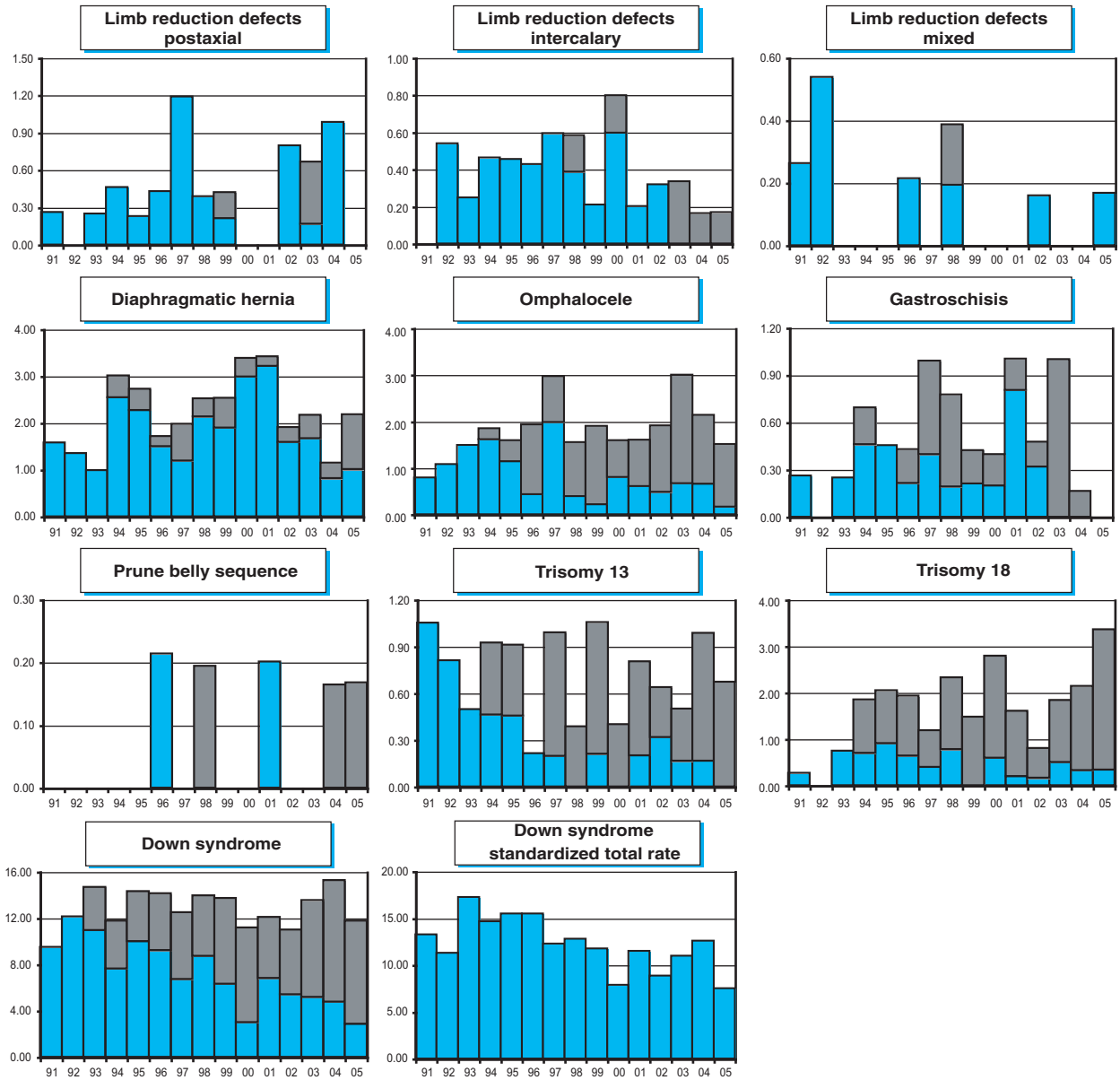

**Note:** L+S rates, ToP rates

## Italy: BDRCam




**Note:** L+S rates, ToP rates

## Italy: BDRCam




**Note:** L+S rates, ToP rates

## Italy: IMER

Emilia-Romagna Registry of Congenital Malformations (IMER)

**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.

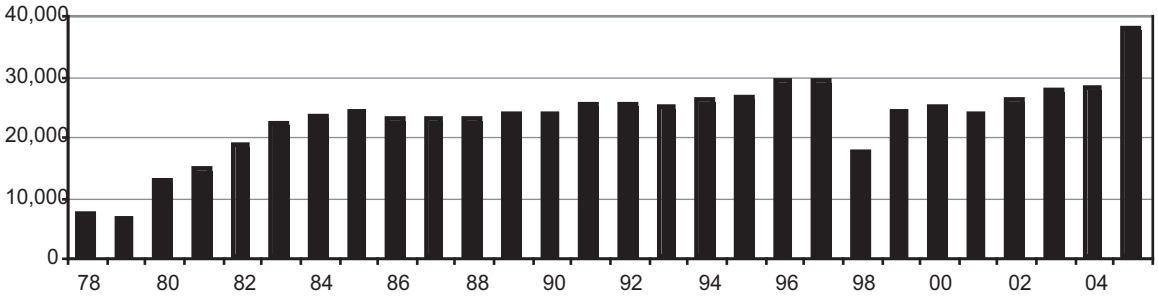
**Background information:**
Some general demographic information is known for all births in the area. For each participating hospital, the number of livebirths and stillbirths are known.

**Addresses and Staff:**
Guido Cocchi, MD, Programme Director
Istituto di Pediatria Preventiva/Neonatologia
Universita' di Bologna
Via Massarenti 11
40138 Bologna, Italy
**Phone:** 39-051-342754 /6364654
**Fax:** 39-051-342754
**E-mail:** guido.cocchi@unibo.it

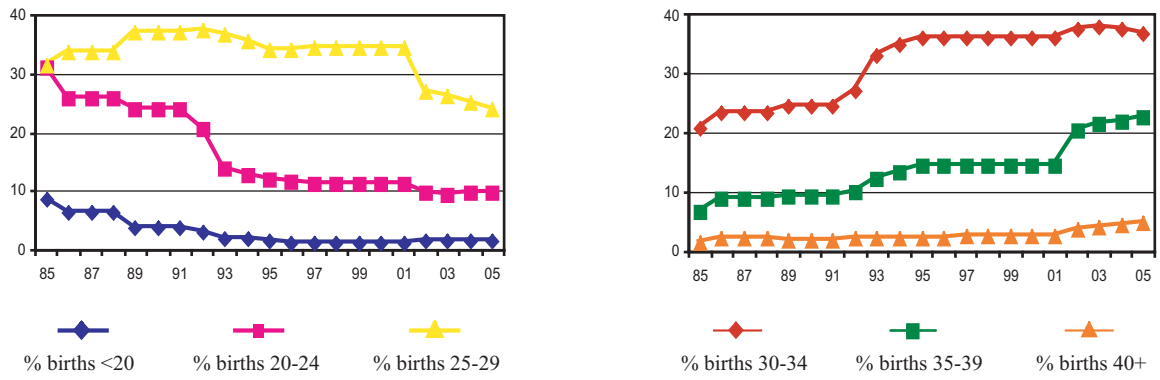
**Website:** www.unife.it/imer

## Italy: IMER

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 21 | 85.7 | Cystic kidney | 48 | 35.4 |
| Spina bifida | 42 | 73.8 | Limb reduction defects | 56 | 33.9 |
| Encephalocele | 9 | 66.7 | Diaphragmatic hernia | 37 | 35.1 |
| Holoprosencephaly | 14 | 92.9 | Omphalocele | 27 | 63.0 |
| Hydrocephaly | 62 | 69.4 | Gastroschisis | 14 | 28.6 |
| Hypoplastic left heart syndrome | 25 | 48.0 | Trisomy 13 | 19 | 89.5 |
| Cleft palate without cleft lip | 38 | 0.0 | Trisomy 18 | 47 | 83.0 |
| Cleft lip with or without cleft palate | 64 | 23.4 | Down syndrome | 186 | 62.9 |
| Renal agenesis | 36 | 38.9 | | | |

Total ToPs with births defects = 561 (Ratio ToPs/Births: 6.03 per 1,000)
*ToPs/ToPs+Births

# Italy: IMER, 2005

| | |
|---|---|
| Live births (LB) | 37,530 |
| Stillbirths (SB) | 75 |
| Total births | 37,605 |
| Number of terminations of pregnancy (ToP) for birth defects | 186 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 2 | 8 | 2.66 |
| Spina bifida | 4 | 0 | 7 | 2.93 |
| Encephalocele | 1 | 1 | 2 | 1.06 |
| Microcephaly | 6 | 0 | 0 | 1.60 |
| Holoprosencephaly | 0 | 0 | 5 | 1.33 |
| Hydrocephaly | 6 | 0 | 9 | 3.99 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 4 | 0 | 2 | 1.60 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 8 | 0 | 0 | 2.13 |
| Microtia | 5 | 0 | 0 | 1.33 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 11 | 0 | 5 | 4.25 |
| Tetralogy of Fallot | 11 | 0 | 3 | 3.72 |
| Hypoplastic left heart syndrome | 4 | 0 | 8 | 3.19 |
| Coarctation of aorta | 14 | 0 | 3 | 4.52 |
| Choanal atresia, bilateral | 3 | 0 | 0 | 0.80 |
| Cleft palate without cleft lip | 15 | 0 | 0 | 3.99 |
| Cleft lip with or without cleft palate | 20 | 0 | 7 | 7.18 |
| Oesophageal atresia / stenosis with or without fistula | 18 | 0 | 1 | 5.05 |
| Small intestine atresia / stenosis | 5 | 0 | 1 | 1.60 |
| Anorectal atresia / stenosis | 9 | 0 | 1 | 2.66 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 55 | 0 | 1 | 14.89 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 4 | 0 | 0 | 1.06 |
| Renal agenesis | 7 | 0 | 7 | 3.72 |
| Cystic kidney | 16 | 0 | 6 | 5.85 |
| Bladder exstrophy | 1 | 0 | 0 | 0.27 |
| Polydactyly, preaxial | 16 | 0 | 1 | 4.52 |
| Total Limb reduction defects (include unspecified) | 18 | 0 | 4 | 5.85 |
| Transverse | 5 | 0 | 2 | 1.86 |
| Preaxial | 4 | 0 | 1 | 1.33 |
| Postaxial | 3 | 0 | 0 | 0.80 |
| Intercalary | 0 | 0 | 0 | 0.00 |
| Mixed | 1 | 0 | 0 | 0.27 |
| Unspecified | 5 | 0 | 1 | 1.60 |
| Diaphragmatic hernia | 8 | 0 | 0 | 2.13 |
| Omphalocele | 1 | 0 | 1 | 0.53 |
| Gastroschisis | 2 | 0 | 1 | 0.80 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 4 | 1.06 |
| Trisomy 18 | 0 | 0 | 13 | 3.46 |
| Down syndrome, all ages (include age unknown) | 31 | 0 | 42 | 19.41 |
| <20 | 2 | 0 | 0 | 34.97 |
| 20-24 | 1 | 0 | 0 | 2.73 |
| 25-29 | 2 | 0 | 2 | 4.48 |
| 30-34 | 5 | 0 | 10 | 10.90 |
| 35-39 | 13 | 0 | 17 | 35.19 |
| 40-44 | 7 | 0 | 12 | 111.24 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 1 | 0 | 1 | 57.47 |

nr = not reported

# Italy: IMER, Previous years rates 1978 - 2005

Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1994

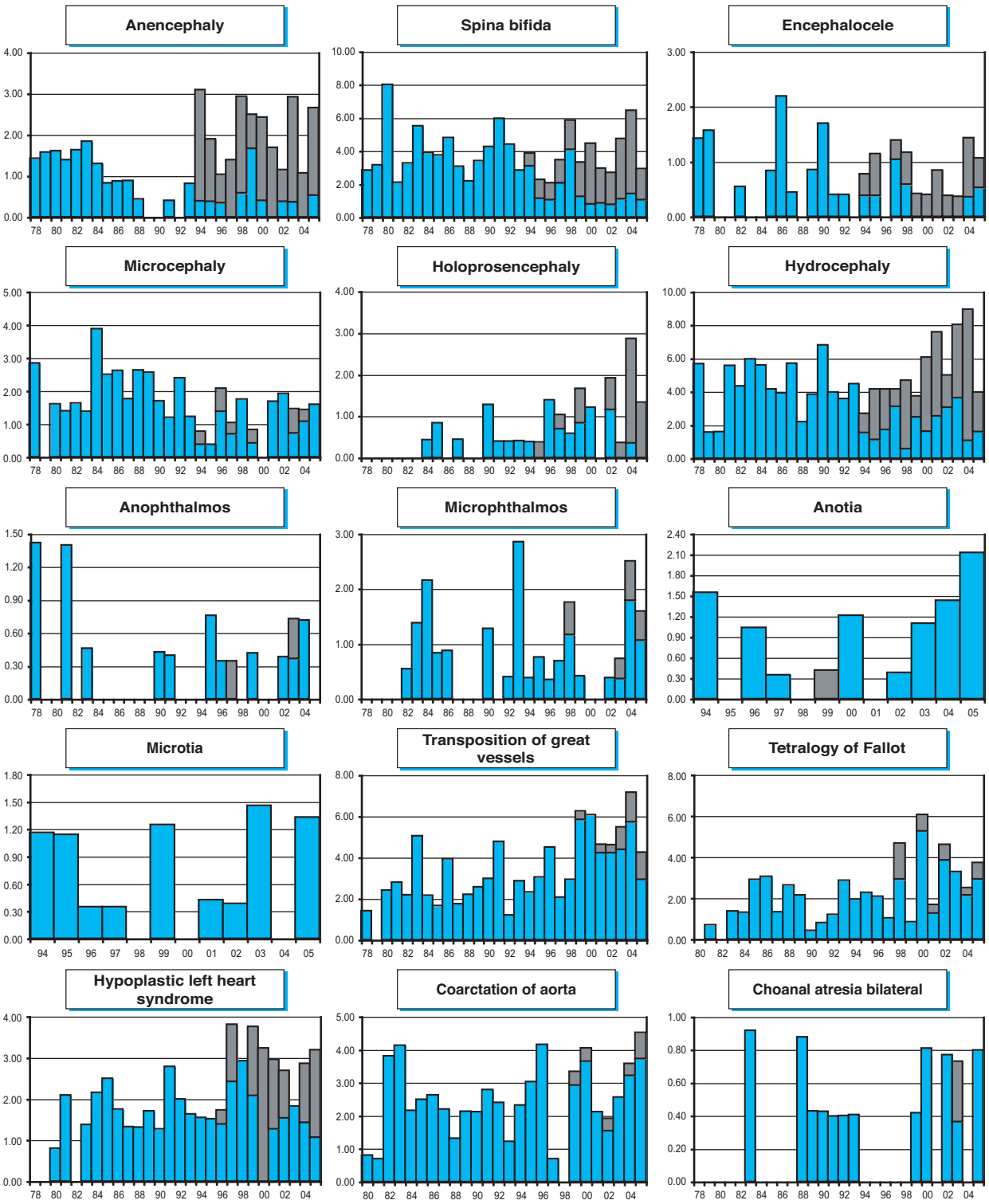
| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **25,878** | **101,622** | **115,334** | **126,943** | **123,541** | **142,659** |
| Anencephaly | 1.55 | 1.38 | 0.43 | 1.26 | 1.94 | 1.96 |
| Spina bifida | 5.41 | 3.84 | 3.55 | 3.86 | 3.64 | 3.93 |
| Encephalocele | 0.77 | 0.30 | 1.04 | 0.55 | 0.65 | 0.84 |
| Microcephaly | 1.55 | 2.26 | 2.25 | 1.18 | 1.13 | 1.61 |
| Holoprosencephaly | 0.00 | 0.30 | 0.35 | 0.39 | 1.21 | 1.33 |
| Hydrocephaly | 2.71 | 5.12 | 4.51 | 3.78 | 4.53 | 6.52 |
| Anophthalmos | 0.39 | 0.30 | 0.09 | 0.24 | 0.24 | 0.35 |
| Microphthalmos | 0.00 | 1.08 | 0.43 | 0.87 | 0.57 | 1.12 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | 0.77* | 0.65 | 1.12 |
| Microtia | nr | nr | nr | 1.15* | 0.40 | 0.77 |
| Unspecified Anotia / Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 1.55 | 2.76 | 2.69 | 2.84 | 4.37 | 5.19 |
| Tetralogy of Fallot | 0.00 | 1.38 | 1.91 | 1.81 | 2.75 | 3.22 |
| Hypoplastic left heart syndrome | 0.39 | 1.67 | 1.47 | 1.89 | 3.08 | 2.73 |
| Coarctation of aorta | 0.80 | 2.76 | 2.08 | 2.36 | 2.59 | 3.08 |
| Choanal atresia, bilateral | 0.00 | 0.20 | 0.35 | 0.24 | 0.24 | 0.49 |
| Cleft palate without cleft lip | 3.48 | 5.12 | 7.20 | 4.88 | 4.05 | 3.93 |
| Cleft lip with or without cleft palate | 6.57 | 8.07 | 6.59 | 6.07 | 5.91 | 6.38 |
| Oesophageal atresia / stenosis with or without fistula | 3.09 | 3.94 | 3.90 | 3.70 | 3.56 | 3.50 |
| Small intestine atresia / stenosis | 2.71 | 2.26 | 3.90 | 3.23 | 2.75 | 2.66 |
| Anorectal atresia / stenosis | 0.39 | 3.35 | 3.12 | 2.99 | 2.67 | 3.72 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 20.87 | 18.89 | 20.64 | 17.27* | 16.59 | 16.12 |
| Epispadias | nr | nr | nr | 0.00* | 0.00 | 0.00 |
| Indeterminate sex | nr | nr | nr | 0.00* | 0.24 | 0.56 |
| Renal agenesis | 3.09 | 0.98 | 1.56 | 1.50 | 3.08 | 4.28 |
| Cystic kidney | 0.39 | 0.59 | 0.78 | 0.39 | 4.21 | 4.49 |
| Bladder exstrophy | 0.77 | 0.30 | 0.69 | 0.08 | 0.24 | 0.21 |
| Polydactyly, preaxial | 9.27 | 9.64 | 7.37 | 6.14 | 3.89 | 2.59 |
| Total Limb reduction defects (include unspecified) | nr | 6.25* | 5.72 | 4.73 | 4.45 | 4.98 |
| Transverse | nr | 4.16* | 3.12 | 2.44 | 2.02 | 1.61 |
| Preaxial | nr | 0.00* | 0.78 | 0.79 | 1.13 | 0.91 |
| Postaxial | nr | 0.42* | 0.61 | 0.47 | 0.49 | 0.77 |
| Intercalary | nr | 0.42* | 0.78 | 0.47 | 0.57 | 0.42 |
| Mixed | nr | 0.42* | 0.35 | 0.32 | 0.08 | 0.21 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 1.16 | 1.67 | 2.08 | 3.15 | 3.24 | 3.50 |
| Omphalocele | 2.32 | 1.57 | 1.82 | 2.05 | 1.70 | 2.52 |
| Gastroschisis | 0.00 | 0.98 | 0.78 | 0.95 | 0.73 | 1.19 |
| Unspecified Omphalocele / Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.39 | 0.49 | 0.26 | 0.24 | 0.40 | 0.14 |
| Trisomy 13 | 1.16 | 1.38 | 0.87 | 0.71 | 0.89 | 1.89 |
| Trisomy 18 | 0.39 | 1.38 | 1.04 | 1.26 | 2.35 | 4.70 |
| Down syndrome, all ages (include age unknown) | 17.78 | 13.48 | 12.40 | 15.83 | 19.43 | 18.65 |
| <20 | nr | 0.00* | 3.36 | 6.68 | 13.25 | 14.49 |
| 20-24 | nr | 4.05* | 5.19 | 5.73 | 9.88 | 5.64 |
| 25-29 | nr | 13.22* | 9.43 | 9.85 | 5.90 | 4.45 |
| 30-34 | nr | 14.01* | 16.42 | 13.67 | 13.02 | 11.37 |
| 35-39 | nr | 62.15* | 27.93 | 34.53 | 46.97 | 36.18 |
| 40-44 | nr | 55.40* | 61.64 | 125.33 | 161.68 | 107.70 |
| 45+ | nr | 333.33* | 0.00 | 165.29 | 198.68 | 86.58 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

## Italy: IMER

Time trends 1978-2003 (Birth prevalence rates per 10,000)

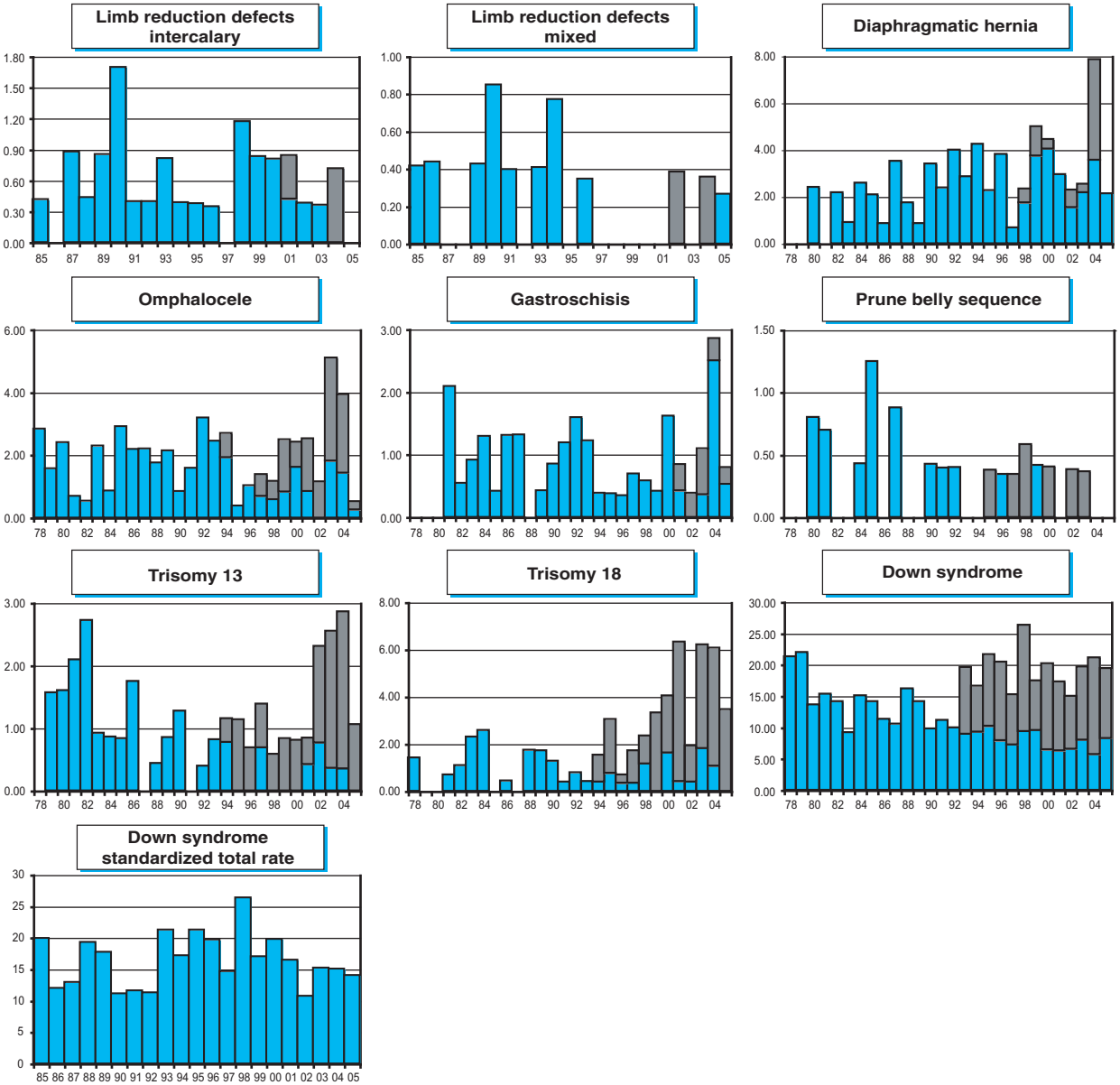



**Note:** L+S rates, ToP rates

## Italy: IMER


**Note:** L+S rates, ToP rates

## Italy: IMER




**Note:** L+S rates, ToP rates

## Italy: ISMAC

Sicilian Registry of Congenital Malformations (ISMAC)

**History:**
The registry started in 1991 and became an associate member of the Clearinghouse in 1996.

**Size and coverage:**
The Programme is hospital based and actually collaborates with four south-east provinces (Catania, Enna, Ragusa and Siracusa) of the nine Sicilian provinces, with a covering rate higher than 75% and with more than 19,000 controlled newborns for year. Stillbirths are included.

**Legislation and funding:**
The Programme is a surveillance Programme with a voluntary participation, supported by ASMAC-Associazione per la Prevenzione Sociale e per il Trattamento delle Malformazioni Congenite and under Osservatorio Epidemiologico Regionale-Sicilia.

**Sources of ascertainment:**
Reports are obtained from delivery units, pediatric units and other specialistic departments.

Exposure information: for each malformed infant reported, information is given on certain exposures, including maternal drug usange and parental occupation. Up to now no information on controls is available.

**Background information:**
Up to now little background information is available and no information on controls is available.

**Addresses and Staff:**
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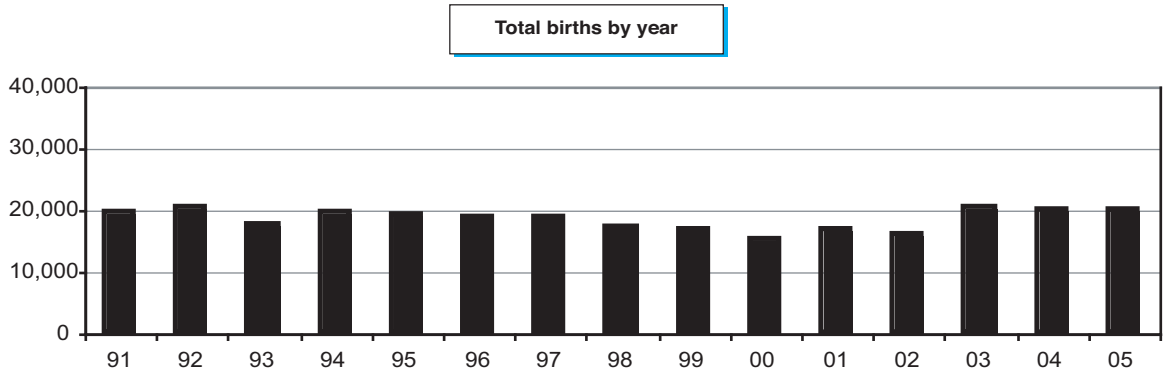
## Italy: ISMAC


Total births by year
40,000
30,000
20,000
10,000
0
91
92
93
94
95
96
97
98
99
00
01
02
03
04
05

# Italy: ISMAC, 2005

| | |
|---|---|
| Live births (LB) | 19,960 |
| Stillbirths (SB) | nr |
| Total births | 19,960 |
| 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 | 0 | nr | 0.00 |
| Spina bifida | 4 | 0 | nr | 2.00 |
| Encephalocele | 1 | 0 | nr | 0.50 |
| Microcephaly | 0 | 0 | nr | 0.00 |
| Holoprosencephaly | 0 | 0 | nr | 0.00 |
| Hydrocephaly | 7 | 0 | nr | 3.51 |
| Anophthalmos | 0 | 0 | nr | 0.00 |
| Microphthalmos | 0 | 0 | nr | 0.00 |
| Unspecified Anophthalmos/ Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 1 | 0 | nr | 0.50 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 3 | 0 | nr | 1.50 |
| Tetralogy of Fallot | 6 | 0 | nr | 3.01 |
| Hypoplastic left heart syndrome | 1 | 0 | nr | 0.50 |
| Coarctation of aorta | 8 | 0 | nr | 4.01 |
| Choanal atresia, bilateral | 1 | 0 | nr | 0.50 |
| Cleft palate without cleft lip | 12 | 0 | nr | 6.01 |
| Cleft lip with or without cleft palate | 7 | 0 | nr | 3.51 |
| Oesophageal atresia / stenosis with or without fistula | 4 | 0 | nr | 2.00 |
| Small intestine atresia / stenosis | 0 | 0 | nr | 0.00 |
| Anorectal atresia / stenosis | 5 | 0 | nr | 2.51 |
| Undescended testis (36 weeks of gestation or later) | 33 | 0 | nr | 16.53 |
| Hypospadias | 49 | 0 | nr | 24.55 |
| Epispadias | 0 | 0 | nr | 0.00 |
| Indeterminate sex | 1 | 0 | nr | 0.50 |
| Renal agenesis | 1 | 0 | nr | 0.50 |
| Cystic kidney | 1 | 0 | nr | 0.50 |
| Bladder exstrophy | 1 | 0 | nr | 0.50 |
| Polydactyly, preaxial | 7 | 0 | nr | 3.51 |
| Total Limb reduction defects (include unspecified) | 5 | 0 | nr | 2.51 |
| Transverse | 1 | 0 | nr | 0.50 |
| Preaxial | 3 | 0 | nr | 1.50 |
| Postaxial | 1 | 0 | nr | 0.50 |
| Intercalary | 0 | 0 | nr | 0.00 |
| Mixed | 0 | 0 | nr | 0.00 |
| Unspecified | 0 | 0 | nr | 0.00 |
| Diaphragmatic hernia | 6 | 0 | nr | 3.01 |
| Omphalocele | 0 | 0 | nr | 0.00 |
| Gastroschisis | 0 | 0 | nr | 0.00 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13 | 1 | 0 | nr | 0.50 |
| Trisomy 18 | 1 | 0 | nr | 0.50 |
| Down syndrome, all ages (include age unknown) | 8 | 0 | nr | 4.01 |
| <20 | 0 | 0 | nr | nr |
| 20-24 | 0 | 0 | nr | nr |
| 25-29 | 1 | 0 | nr | nr |
| 30-34 | 5 | 0 | nr | nr |
| 35-39 | 2 | 0 | nr | nr |
| 40-44 | 0 | 0 | nr | nr |
| 45+ | 0 | 0 | nr | nr |
| unknown | 0 | 0 | nr | nr |

nr = not reported

# Italy: ISMAC, Previous years rates 1991 - 2005

Birth prevalence rates: (LB+SB) * 10,000
Birth prevalence rates: (LB+SB+TOP) * 10,000 only for 2000-2001-2004

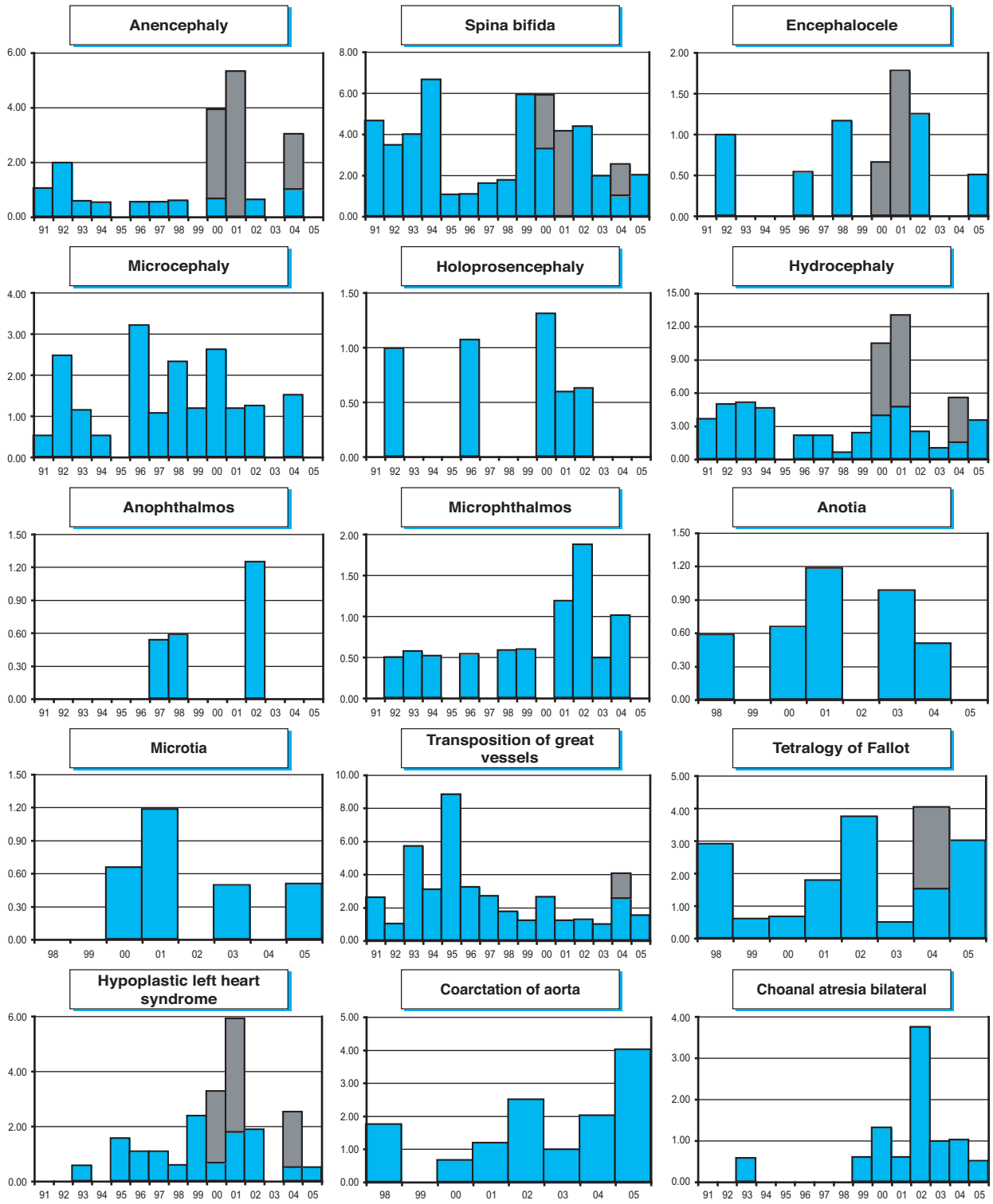
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | | | **96,220** | **87,015** | **93,312** |
| Anencephaly | | | | 0.83 | 1.03 | 1.71 |
| Spina bifida | | | | 3.95 | 3.10 | 2.89 |
| Encephalocele | | | | 0.21 | 0.46 | 0.64 |
| Microcephaly | | | | 0.94 | 2.07 | 0.75 |
| Holoprosencephaly | | | | 0.21 | 0.46 | 0.21 |
| Hydrocephaly | | | | 3.64 | 3.33 | 4.93 |
| Anophthalmos | | | | 0.00 | 0.23 | 0.21 |
| Microphthalmos | | | | 0.31 | 0.34 | 0.86 |
| Unspecified Anophthalmos / Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | nr | 0.40* | 0.54 |
| Microtia | | | | nr | 0.20* | 0.43 |
| Unspecified Anotia / Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 4.16 | 2.30 | 1.82 |
| Tetralogy of Fallot | | | | nr | 1.41* | 2.57 |
| Hypoplastic left heart syndrome | | | | 0.42 | 1.61 | 2.04 |
| Coarctation of aorta | | | | nr | 0.81* | 2.14 |
| Choanal atresia, bilateral | | | | 0.10 | 0.44* | 1.29 |
| Cleft palate without cleft lip | | | | 4.78 | 4.83 | 4.61 |
| Cleft lip with or without cleft palate | | | | 6.55 | 6.90 | 4.07 |
| Oesophageal atresia / stenosis with or without fistula | | | | 3.53 | 2.30 | 2.25 |
| Small intestine atresia / stenosis | | | | 7.38 | 1.72 | 1.50 |
| Anorectal atresia / stenosis | | | | 3.01 | 2.53 | 1.82 |
| Undescended testis (36 weeks of gestation or later) | | | | 5.92 | 11.49 | 15.54 |
| Hypospadias | | | | nr | 17.81 | 21.86 |
| Epispadias | | | | 0.00 | 0.44* | 0.21 |
| Indeterminate sex | | | | 0.42 | 0.23 | 0.64 |
| Renal agenesis | | | | 1.56 | 1.03 | 1.18 |
| Cystic kidney | | | | 0.83 | 1.26 | 2.57 |
| Bladder exstrophy | | | | 0.10 | 0.59* | 0.32 |
| Polydactyly, preaxial | | | | 0.21 | 2.49* | 2.57 |
| Total Limb reduction defects (include unspecified) | | | | 3.43 | 2.87 | 3.11 |
| Transverse | | | | nr | 2.42* | 2.14 |
| Preaxial | | | | nr | 0.00* | 0.32 |
| Postaxial | | | | nr | 0.40* | 0.11 |
| Intercalary | | | | nr | 0.00* | 0.00 |
| Mixed | | | | nr | 0.00* | 0.00 |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 1.87 | 1.61 | 2.79 |
| Omphalocele | | | | 1.77 | 1.38 | 0.75 |
| Gastroschisis | | | | 0.83 | 0.92 | 1.82 |
| Unspecified Omphalocele / Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.00 | 0.15* | 0.00 |
| Trisomy 13 | | | | 0.21 | 1.17* | 1.29 |
| Trisomy 18 | | | | 0.52 | 0.92 | 1.29 |
| Down syndrome, all ages (include age unknown) | | | | 13.20 | 12.53 | 8.57 |
| <20 | | | | nr | nr | nr |
| 20-24 | | | | nr | nr | nr |
| 25-29 | | | | nr | nr | nr |
| 30-34 | | | | nr | nr | nr |
| 35-39 | | | | nr | nr | nr |
| 40-44 | | | | nr | nr | nr |
| 45+ | | | | nr | nr | nr |
| unknown | | | | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

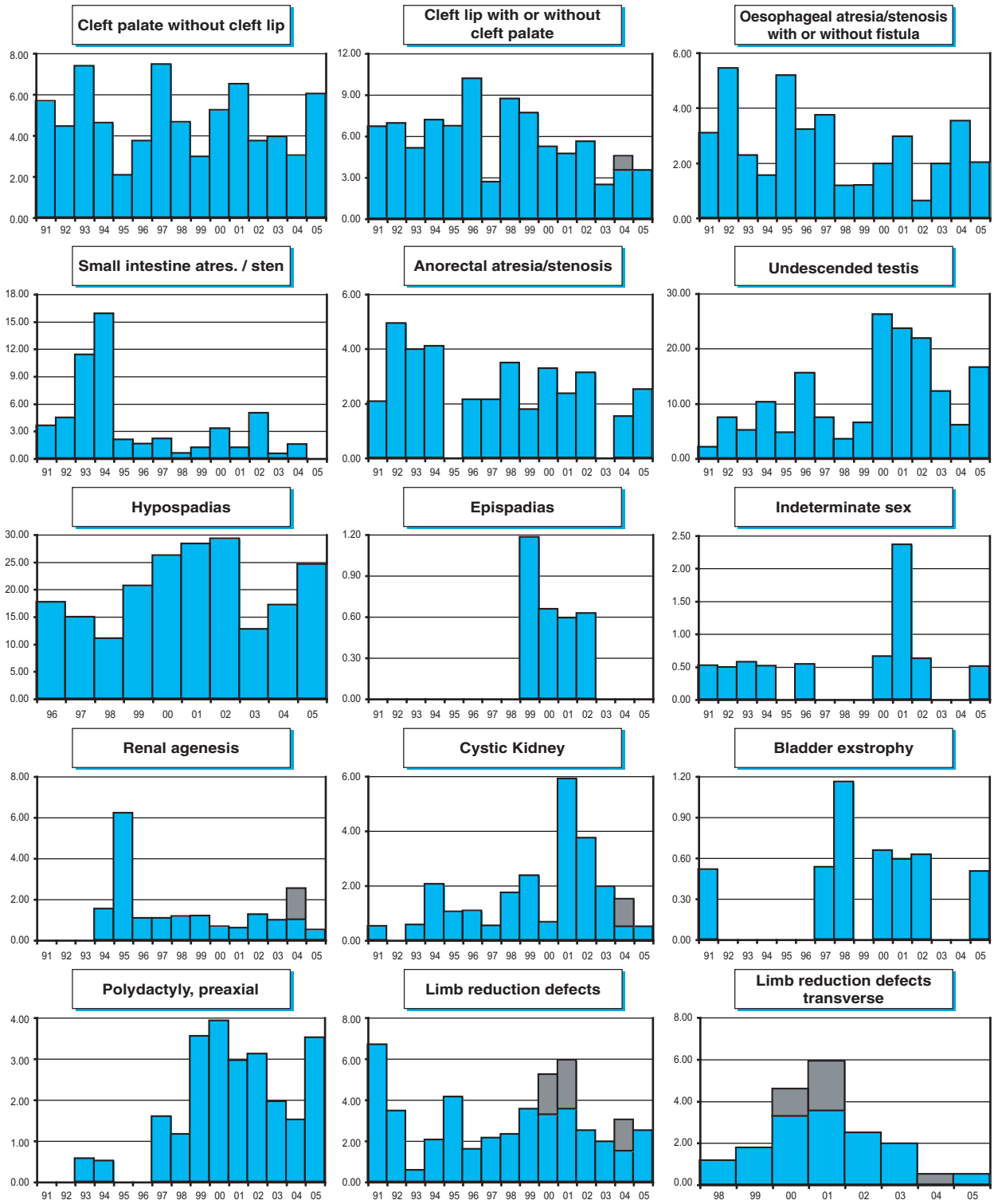
## Italy: ISMAC

'Time trends 1991-2005 (Birth prevalence rates per 10,000)



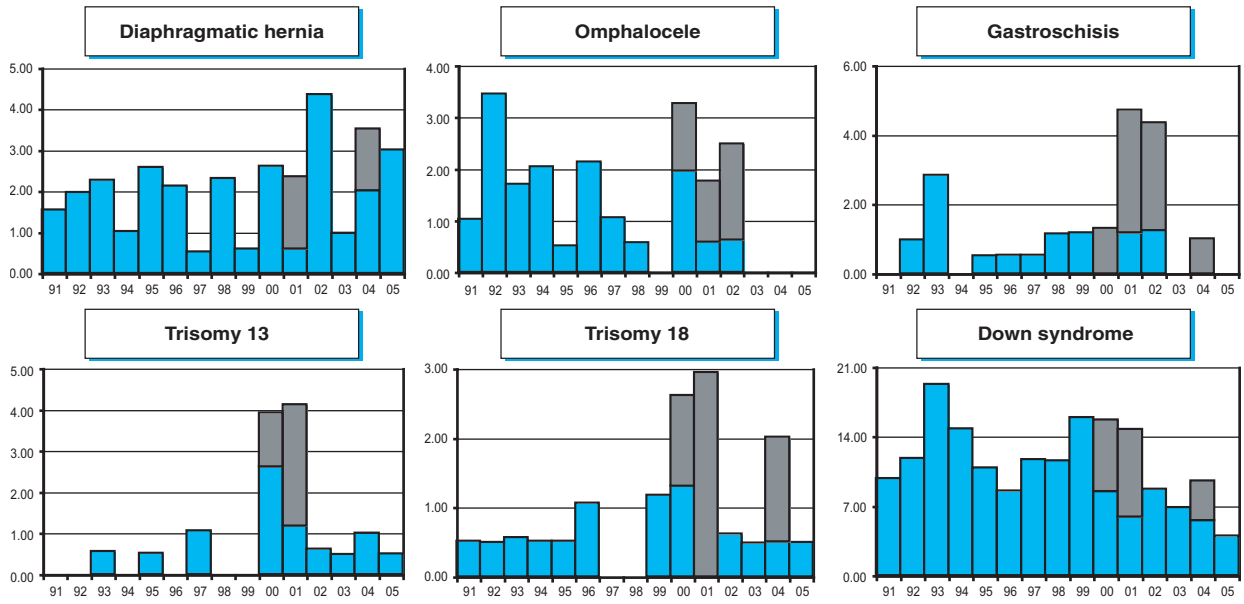

**Note:** L+S rates, ToP rates

## Italy: ISMAC




**Note:** L+S rates, ToP rates

## Italy: ISMAC




**Note:** L+S rates, ToP rates

## 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 a member of Eurocat in 1985, and an associate 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.

**Addresses and Staff:**
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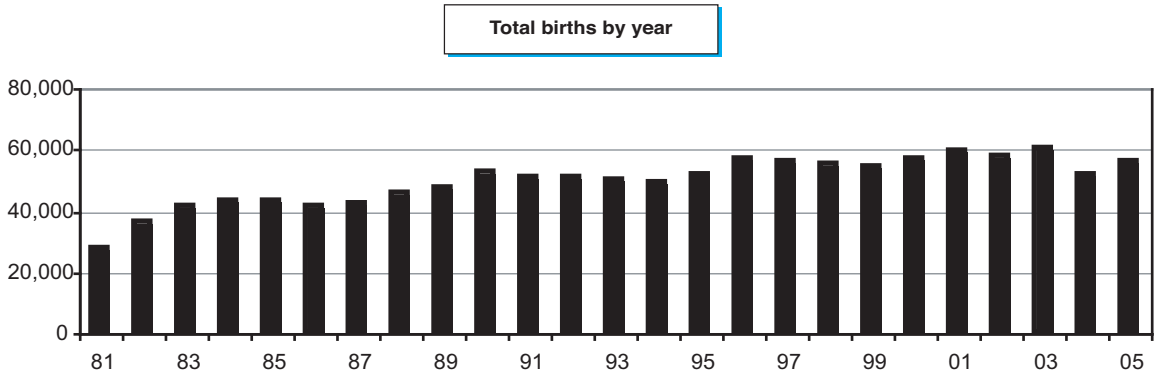
Genetica Medica Dipartimento Pediatria
Via Giustiniani, 3
35128 Padova, Italy
**Phone:** 39-049-8213513
**Fax:** 39-049-8211425
**E-mail:** tenconi@child.pedi.unipd.it

**Website:** www.genetica.pediatria.unipd.it

## Italy: North East




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 16 | 80.0 | Cystic kidney | 5 | 35.7 |
| Spina bifida | 23 | 48.9 | Limb reduction defects | 20 | 30.3 |
| Encephalocele | 3 | 50.0 | Diaphragmatic hernia | 5 | 14.3 |
| Holoprosencephaly | 9 | 69.2 | Omphalocele | 13 | 59.1 |
| Hydrocephaly | 27 | 47.4 | Gastroschisis | 3 | 17.6 |
| Hypoplastic left heart syndrome | 9 | 40.9 | Trisomy 13 | 14 | 82.4 |
| Cleft palate without cleft lip | 8 | 7.1 | Trisomy 18 | 23 | 74.2 |
| Cleft lip with or without cleft palate | 17 | 12.2 | Down syndrome | 106 | 42.2 |
| Renal agenesis | 6 | 75.0 | | | |

Total ToPs with births defects = 401 (Ratio ToPs/Births: 2.45 per 1,000)
*ToPs/ToPs+Births

# Italy: North East, 2005

| | |
|---|---|
| Live births (LB) | 55,683 |
| Stillbirths (SB) | 398 |
| Total births | 56,081 |
| Number of terminations of pregnancy (ToP) for birth defects | 146 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 5 | 0.89 |
| Spina bifida | 8 | 0 | 7 | 2.67 |
| Encephalocele | 1 | 0 | 0 | 0.18 |
| Microcephaly | 4 | 0 | 0 | 0.71 |
| Holoprosencephaly | 3 | 0 | 1 | 0.71 |
| Hydrocephaly | 9 | 0 | 11 | 3.57 |
| Anophthalmos | 1 | 0 | 0 | 0.18 |
| Microphthalmos | 5 | 0 | 1 | 1.07 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 10 | 0 | 1 | 1.96 |
| Unspecified Anotia/Microtia | 1 | 0 | 0 | 0.18 |
| Transposition of great vessels | 8 | 0 | 0 | 1.43 |
| Tetralogy of Fallot | 13 | 0 | 0 | 2.32 |
| Hypoplastic left heart syndrome | 6 | 0 | 2 | 1.43 |
| Coarctation of aorta | 7 | 0 | 0 | 1.25 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.36 |
| Cleft palate without cleft lip | 34 | 0 | 2 | 6.42 |
| Cleft lip with or without cleft palate | 35 | 0 | 7 | 7.49 |
| Oesophageal atresia/stenosis with or without fistula | 7 | 0 | 0 | 1.25 |
| Small intestine atresia/stenosis | 13 | 0 | 1 | 2.50 |
| Anorectal atresia/stenosis | 8 | 0 | 5 | 2.32 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 19 | 0 | 0 | 3.39 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 0 | 0 | 5 | 0.89 |
| Cystic kidney | 3 | 0 | 1 | 0.71 |
| Bladder exstrophy | 1 | 0 | 0 | 0.18 |
| Polydactyly, preaxial | 9 | 0 | 0 | 1.60 |
| Total Limb reduction defects (include unspecified) | 17 | 0 | 12 | 5.17 |
| Transverse | 6 | 0 | 0 | 1.07 |
| Preaxial | 3 | 0 | 1 | 0.71 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 1 | 0 | 1 | 0.36 |
| Mixed | 1 | 0 | 0 | 0.18 |
| Unspecified | 6 | 0 | 10 | 2.85 |
| Diaphragmatic hernia | 8 | 0 | 1 | 1.60 |
| Omphalocele | 1 | 0 | 4 | 0.89 |
| Gastroschisis | 5 | 0 | 0 | 0.89 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | 3 | 0.71 |
| Trisomy 18 | 2 | 0 | 7 | 1.60 |
| Down syndrome, all ages (include age unknown) | 36 | 0 | 33 | 12.30 |
| <20 | 0 | 0 | 0 | nr |
| 20-24 | 0 | 0 | 0 | nr |
| 25-29 | 2 | 0 | 3 | nr |
| 30-34 | 4 | 0 | 3 | nr |
| 35-39 | 13 | 0 | 13 | nr |
| 40-44 | 3 | 0 | 14 | nr |
| 45+ | 1 | 0 | 0 | nr |
| unknown | 13 | 0 | 0 | nr |

nr = not reported

# Italy: North East, Previous years rates 1981 - 2005

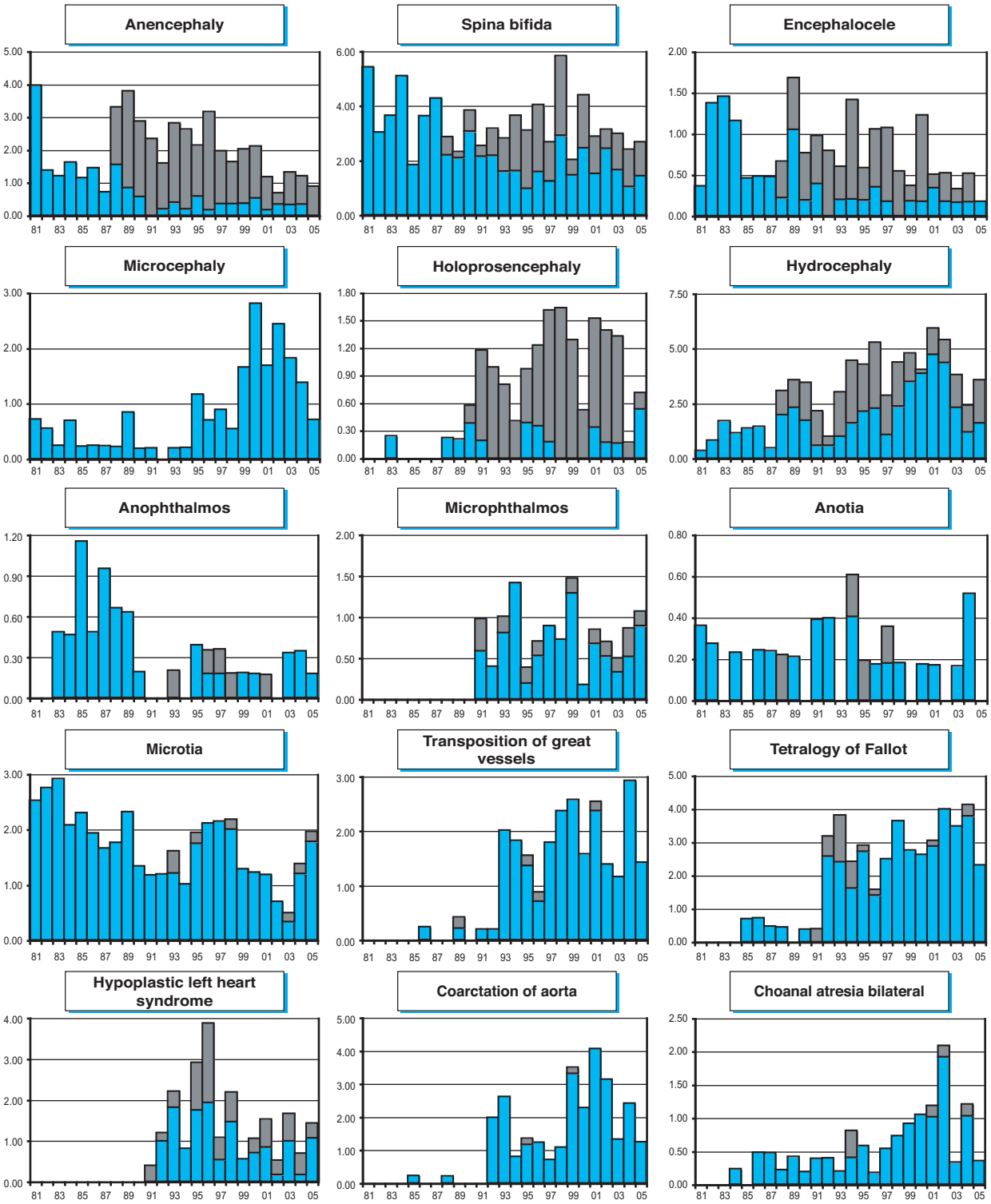
Birth prevalence rates: (LB+SB) * 10,000 until 1995
Birth prevalence rates: (LB+SB+TOP) * 10,000 from 1996

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | **191,827** | **228,340** | **252,097** | **279,021** | **290,902** |
| Anencephaly | | 1.72 | 2.50 | 2.30 | 2.19 | 1.07 |
| Spina bifida | | 3.70 | 3.37 | 3.05 | 3.80 | 2.82 |
| Encephalocele | | 0.99 | 0.83 | 0.87 | 0.86 | 0.41 |
| Microcephaly | | 0.47 | 0.35 | 0.36 | 1.33 | 1.62 |
| Holoprosencephaly | | 0.05 | 0.22 | 0.87 | 1.25 | 1.03 |
| Hydrocephaly | | 1.15 | 2.50 | 2.98 | 4.26 | 4.23 |
| Anophthalmos | | 0.47 | 0.57 | 0.12 | 0.25 | 0.21 |
| Microphthalmos | | 0.00 | 0.00 | 0.83 | 0.79 | 0.79 |
| Unspecified Anophthalmos/Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 0.16 | 0.18 | 0.32 | 0.18 | 0.17 |
| Microtia | | 2.50 | 1.80 | 1.39 | 1.79 | 1.13 |
| Unspecified Anotia/Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 0.00 | 0.13 | 1.15 | 1.83 | 1.89 |
| Tetralogy of Fallot | | 0.16 | 0.39 | 2.54 | 2.62 | 3.40 |
| Hypoplastic left heart syndrome | | 0.00 | 0.00 | 1.51 | 1.76 | 1.17 |
| Coarctation of aorta | | 0.05 | 0.04 | 1.35 | 1.76 | 2.44 |
| Choanal atresia, bilateral | | 0.05 | 0.35 | 0.48 | 0.68 | 1.03 |
| Cleft palate without cleft lip | | 4.27 | 5.96 | 4.68 | 4.80 | 5.95 |
| Cleft lip with or without cleft palate | | 9.12 | 8.23 | 7.81 | 7.74 | 7.49 |
| Oesophageal atresia/stenosis with or without fistula | | 2.50 | 1.97 | 3.05 | 2.51 | 2.78 |
| Small intestine atresia/stenosis | | 0.42 | 0.83 | 1.11 | 0.90 | 2.34 |
| Anorectal atresia/stenosis | | 2.61 | 3.11 | 2.30 | 2.87 | 2.54 |
| Undescended testis (36 weeks of gestation or later) | | nr | nr | nr | nr | nr |
| Hypospadias | | 6.93 | 6.57 | 6.47 | 10.14 | 12.10 |
| Epispadias | | 0.10 | 0.13 | 0.12 | 0.18 | 0.03 |
| Indeterminate sex | | nr | nr | nr | nr | nr |
| Renal agenesis | | 0.73 | 0.88 | 0.44 | 0.72 | 0.41 |
| Cystic kidney | | 0.00 | 0.00 | 0.32 | 0.68 | 1.13 |
| Bladder exstrophy | | 0.26 | 0.39 | 0.20 | 0.18 | 0.21 |
| Polydactyly, preaxial | | 1.62 | 2.63 | 2.30 | 1.86 | 1.72 |
| Total Limb reduction defects (include unspecified) | | 5.68 | 6.44 | 5.47 | 4.69 | 4.30 |
| Transverse | | 3.18 | 3.50 | 2.82 | 2.69 | 1.44 |
| Preaxial | | 0.00 | 0.18 | 0.87 | 0.57 | 0.45 |
| Postaxial | | 0.00 | 0.18 | 0.16 | 0.14 | 0.28 |
| Intercalary | | 0.63 | 0.88 | 0.87 | 0.39 | 0.24 |
| Mixed | | 1.88 | 0.74 | 0.16 | 0.18 | 0.03 |
| Unspecified | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 0.31 | 0.66 | 0.67 | 1.04 | 1.96 |
| Omphalocele | | 1.36 | 1.40 | 1.27 | 1.18 | 1.27 |
| Gastroschisis | | 0.78 | 0.83 | 0.56 | 0.50 | 0.89 |
| Unspecified Omphalocele/Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | 0.05 | 0.04 | 0.36 | 0.14 | 0.07 |
| Trisomy 13 | | 0.83 | 0.57 | 0.79 | 1.11 | 1.07 |
| Trisomy 18 | | 0.94 | 1.80 | 2.26 | 2.72 | 1.99 |
| Down syndrome, all ages (include age unknown) | | 14.34 | 15.94 | 16.62 | 16.09 | 16.02 |
| <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 = not reported

## Italy: North East

'Time trends 1981-2005 (Birth prevalence rates per 10,000)

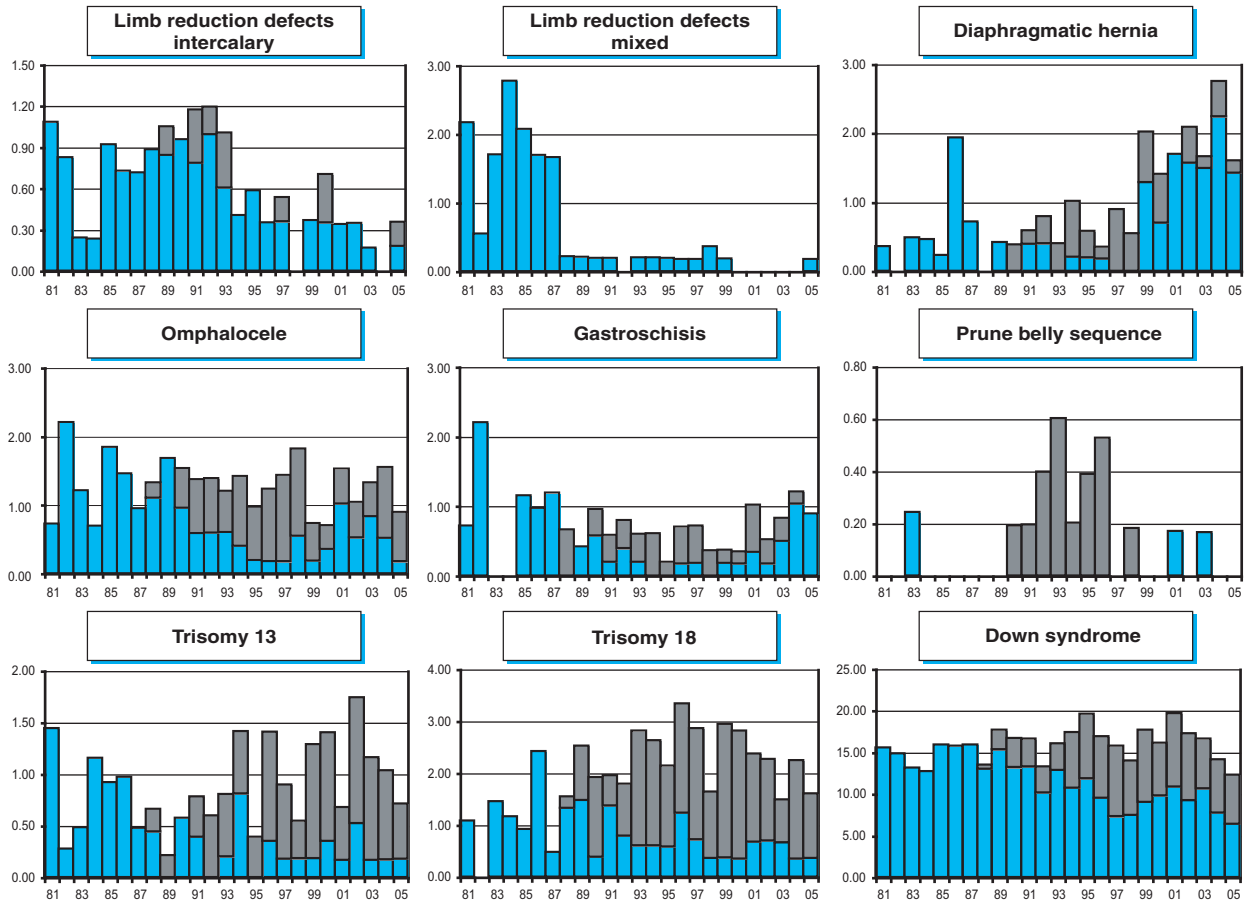



**Note:** L+S rates, ToP rates

## Italy: North East


**Note:** L+S rates, ToP rates

## Italy: North East




**Note:** L+S rates, ToP rates

## Italy: Tuscany
Tuscany Registry of Congenital Defects (RTDC)

**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 millions inhabitants and 25,000 births/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, neonatal and pediatric surgery units, prenatal diagnostic centers and pathology services. Mothers are interviewed by using a standardized questionnaire.

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

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

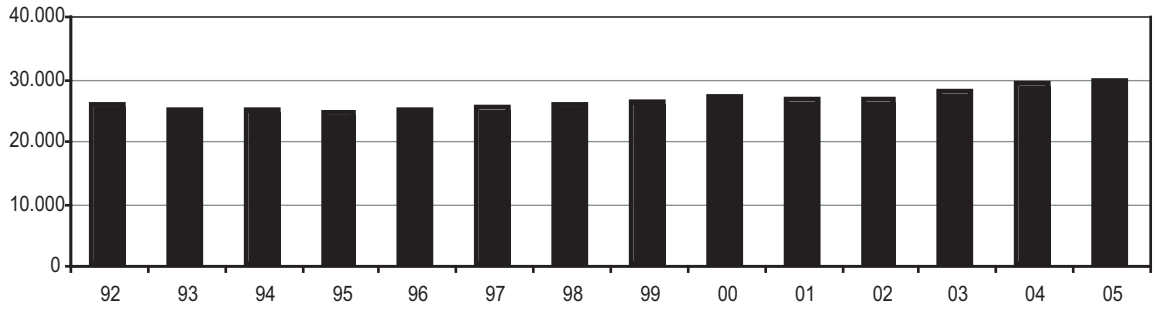
**Addresses and Staff:**
IFC-Unit of Epidemiology
CNR Area di Ricerca di San Cataldo
Via Moruzzi 1 – 56124 Pisa
Fabrizio Bianchi, BSc, Programme Director
**Phone:** 39-050-3152100
**Fax:** 39-050-3152095
**E-mail:** fabriepi@ifc.cnr.it

Anna Pierini, BSc, Project Manager
**Phone:** 39-050-3152102
**Fax:** 39-050-3152095
**E-mail:** apier@ifc.cnr.it

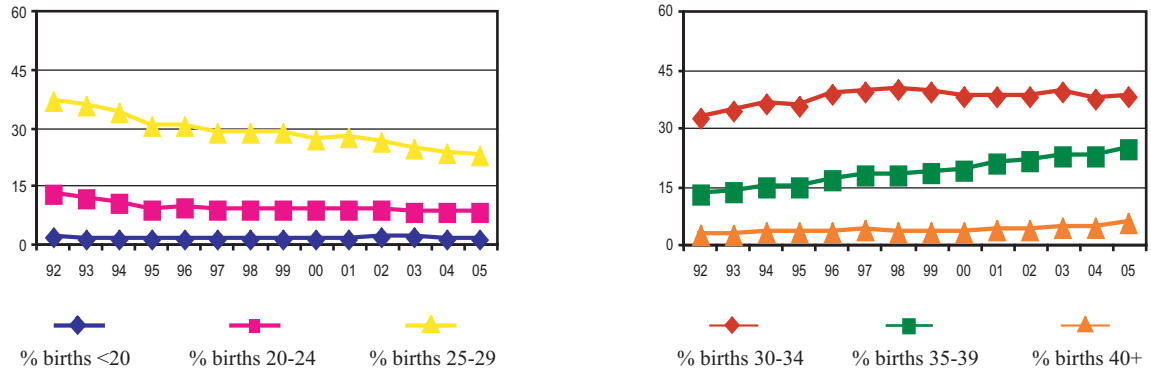
**Web:** www.rtdc.it

## Italy: Tuscany

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 9 | 81.8 | Cystic kidney | 9 | 24.3 |
| Spina bifida | 18 | 64.3 | Limb reduction defects | 8 | 17.0 |
| Encephalocele | 3 | 75.0 | Diaphragmatic hernia | 2 | 14.3 |
| Holoprosencephaly | 9 | 81.8 | Omphalocele | 7 | 50.0 |
| Hydrocephaly | 22 | 51.2 | Gastroschisis | 5 | 71.4 |
| Hypoplastic left heart syndrome | 8 | 44.4 | Trisomy 13 | 4 | 40.0 |
| Cleft palate without cleft lip | 2 | 4.7 | Trisomy 18 | 22 | 84.6 |
| Cleft lip with or without cleft palate | 6 | 12.0 | Down syndrome | 105 | 75.0 |
| Renal agenesis | 5 | 71.4 | | | |

Total ToPs with births defects = 292 (Ratio ToPs/Births: 3.40 per 1,000)
*ToPs/ToPs+Births

# Italy: Tuscany, 2005

| | |
|---|---|
| Live births (LB) | 29,322 |
| Stillbirths (SB) | 89 |
| Total births | 29,411 |
| Number of terminations of pregnancy (ToP) for birth defects | 76 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 2 | 0 | 4 | 2.04 |
| Spina bifida | 1 | 0 | 5 | 2.04 |
| Encephalocele | 1 | 0 | 2 | 1.02 |
| Microcephaly | 0 | 0 | 1 | 0.34 |
| Holoprosencephaly | 0 | 0 | 3 | 1.02 |
| Hydrocephaly | 5 | 0 | 5 | 3.40 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 1 | 0 | 0 | 0.34 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.34 |
| Microtia | 3 | 0 | 0 | 1.02 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 4 | 0 | 0 | 1.36 |
| Tetralogy of Fallot | 7 | 0 | 1 | 2.72 |
| Hypoplastic left heart syndrome | 3 | 0 | 3 | 2.04 |
| Coarctation of aorta | 5 | 0 | 0 | 1.70 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 11 | 0 | 0 | 3.74 |
| Cleft lip with or without cleft palate | 18 | 0 | 2 | 6.80 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | 0 | 2.72 |
| Small intestine atresia/stenosis | 2 | 0 | 0 | 0.68 |
| Anorectal atresia/stenosis | 4 | 0 | 0 | 1.36 |
| Undescended testis (36 weeks of gestation or later) | 6 | 0 | 0 | 2.04 |
| Hypospadias | 21 | 0 | 0 | 7.14 |
| Epispadias | 1 | 0 | 0 | 0.34 |
| Indeterminate sex | 0 | 0 | 0 | 0.00 |
| Renal agenesis | 1 | 0 | 1 | 0.68 |
| Cystic kidney | 8 | 0 | 0 | 2.72 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 0 | 0 | 0 | 0.00 |
| Total Limb reduction defects (include unspecified) | 9 | 0 | 3 | 4.08 |
| Transverse | 3 | 0 | 0 | 1.02 |
| Preaxial | 1 | 0 | 0 | 0.34 |
| Postaxial | 0 | 0 | 2 | 0.68 |
| Intercalary | 0 | 0 | 0 | 0.00 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 5 | 0 | 1 | 2.04 |
| Diaphragmatic hernia | 0 | 0 | 0 | 0.00 |
| Omphalocele | 0 | 0 | 4 | 1.36 |
| Gastroschisis | 0 | 0 | 1 | 0.34 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | 0 | 0.34 |
| Trisomy 18 | 1 | 0 | 6 | 2.38 |
| Down syndrome, all ages (include age unknown) | 12 | 0 | 27 | 13.26 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 0 | 0 | 0 | 0.00 |
| 25-29 | 2 | 0 | 1 | 4.55 |
| 30-34 | 2 | 0 | 3 | 4.45 |
| 35-39 | 5 | 0 | 14 | 26.48 |
| 40-44 | 2 | 0 | 8 | 67.07 |
| 45+ | 0 | 0 | 1 | 212.77 |
| unknown | 1 | 0 | 0 | 833.33 |

# Italy: Tuscany, Previous years rates 1992 - 2005

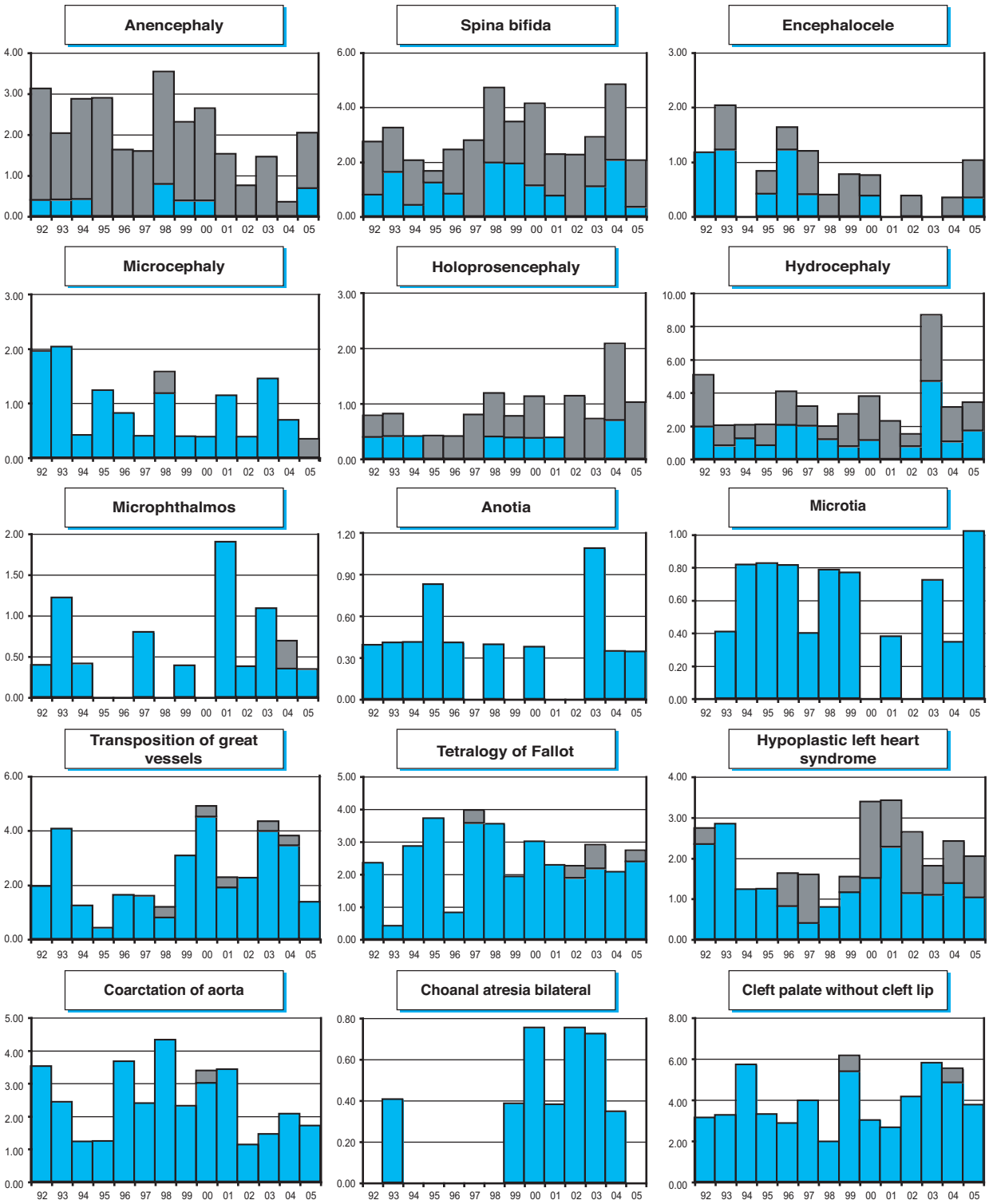
Birth prevalence rates: (LB+SB+TOP) * 10,000 since 1992 except for LRD subcategories (ToPs reported since 1998)

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995* | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | | | | **99,132** | **127,968** | **139,034** |
| Anencephaly | | | | 2.72 | 2.34 | 1.22 |
| Spina bifida | | | | 2.42 | 3.52 | 2.88 |
| Encephalocele | | | | 1.01 | 0.94 | 0.36 |
| Microcephaly | | | | 1.41 | 0.70 | 0.79 |
| Holoprosencephaly | | | | 0.61 | 0.86 | 1.08 |
| Hydrocephaly | | | | 2.82 | 3.13 | 3.81 |
| Anophthalmos | | | | 0.00 | 0.23 | 0.07 |
| Microphthalmos | | | | 0.50 | 0.23 | 0.86 |
| Unspecified Anophthalmos/Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | 0.50 | 0.23 | 0.36 |
| Microtia | | | | 0.50 | 0.55 | 0.50 |
| Unspecified Anotia/Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 1.92 | 2.50 | 2.81 |
| Tetralogy of Fallot | | | | 2.32 | 2.66 | 2.45 |
| Hypoplastic left heart syndrome | | | | 2.02 | 1.80 | 2.45 |
| Coarctation of aorta | | | | 2.12 | 3.20 | 1.94 |
| Choanal atresia, bilateral | | | | 0.10 | 0.23 | 0.43 |
| Cleft palate without cleft lip | | | | 3.83 | 3.59 | 4.39 |
| Cleft lip with or without cleft palate | | | | 7.46 | 6.72 | 5.90 |
| Oesophageal atresia/stenosis with or without fistula | | | | 2.32 | 2.66 | 2.23 |
| Small intestine atresia/stenosis | | | | 1.01 | 0.55 | 1.22 |
| Anorectal atresia/stenosis | | | | 1.61 | 2.34 | 2.73 |
| Undescended testis (36 weeks of gestation or later) | | | | 4.44 | 7.19 | 8.42 |
| Hypospadias | | | | 5.55 | 3.05 | 7.34 |
| Epispadias | | | | 0.30 | 0.16 | 0.29 |
| Indeterminate sex | | | | 1.01 | 0.55 | 0.36 |
| Renal agenesis | | | | 1.61 | 1.41 | 0.86 |
| Cystic kidney | | | | 3.13 | 3.59 | 4.10 |
| Bladder exstrophy | | | | 0.30 | 0.23 | 0.14 |
| Polydactyly, preaxial | | | | 0.91 | 1.25 | 0.86 |
| Total Limb reduction defects (include unspecified) | | | | 5.04 | 5.47 | 5.68 |
| Transverse | | | | 3.63 | 3.28 | 3.60 |
| Preaxial | | | | 0.20 | 0.39 | 0.58 |
| Postaxial | | | | 0.10 | 0.47 | 0.14 |
| Intercalary | | | | 0.20 | 0.70 | 0.43 |
| Mixed | | | | 0.50 | 0.47 | 0.07 |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 1.41 | 1.41 | 2.16 |
| Omphalocele | | | | 2.12 | 1.48 | 1.58 |
| Gastroschisis | | | | 0.40 | 0.55 | 0.50 |
| Unspecified Omphalocele/Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.10 | 0.16 | 0.00 |
| Trisomy 13 | | | | 0.50 | 0.94 | 1.08 |
| Trisomy 18 | | | | 2.32 | 3.75 | 2.66 |
| Down syndrome, all ages (include age unknown) | | | | 14.22 | 16.25 | 16.40 |
| <20 | | | | 0.00 | 0.00 | 0.00 |
| 20-24 | | | | 8.35 | 2.64 | 4.31 |
| 25-29 | | | | 8.31 | 8.22 | 2.93 |
| 30-34 | | | | 12.21 | 11.83 | 8.30 |
| 35-39 | | | | 28.51 | 28.99 | 27.13 |
| 40-44 | | | | 64.03 | 119.47 | 136.47 |
| 45+ | | | | 238.10 | 0.00 | 197.04 |
| uknown | | | | --- | --- | --- |

* data include less than 5 years

## Italy: Tuscany

Time trends 1992-2005 (Birth prevalence rates per 10,000)

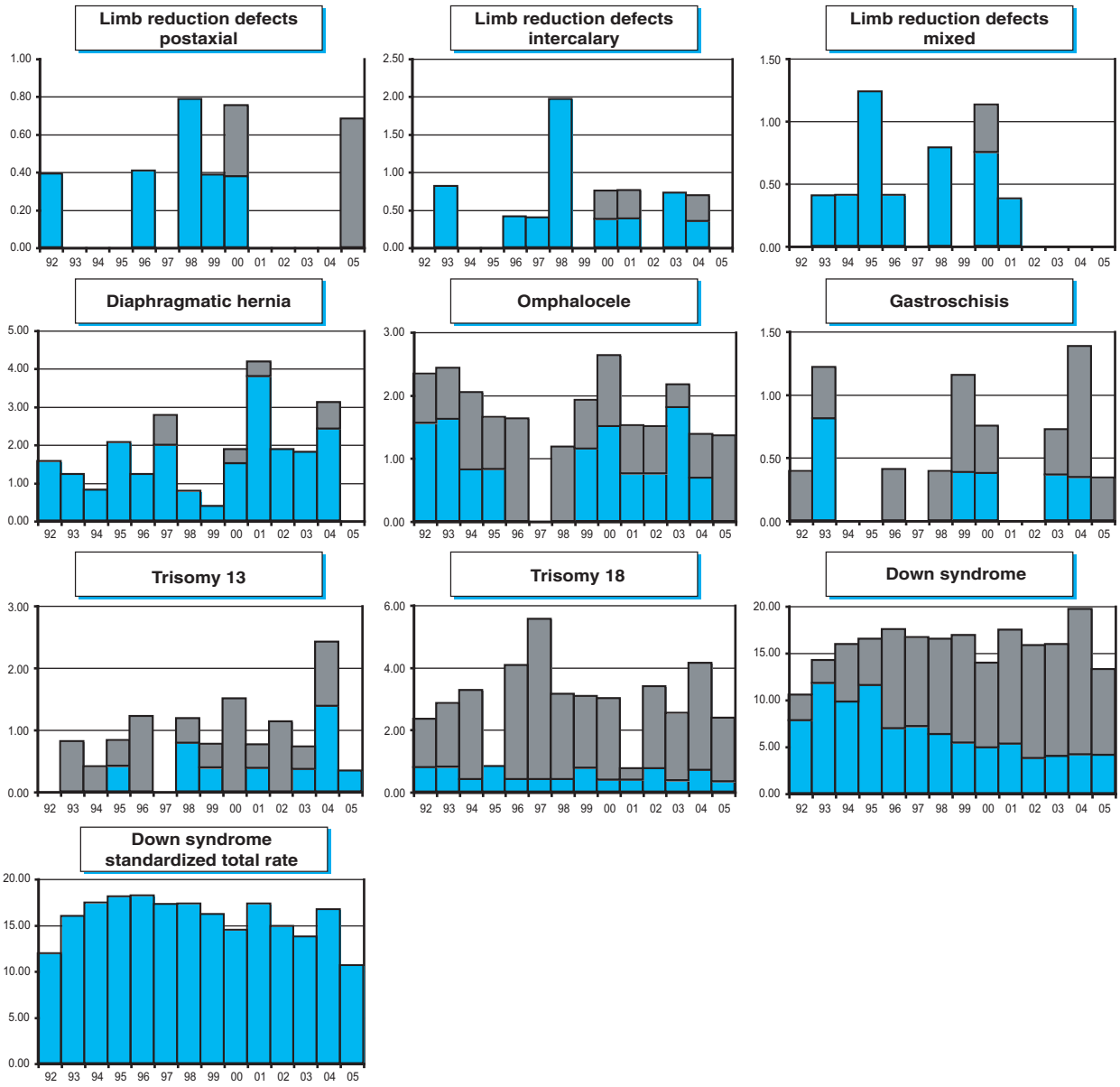



**Note:** L+S rates, ToP rates

## Italy: Tuscany


**Note:** L+S rates, ToP rates

## Italy: Tuscany




**Note:** L+S rates, ToP rates

## Japan: JAOG

Japan Association of Obstetricians and Gynaecologists (JAOG)

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

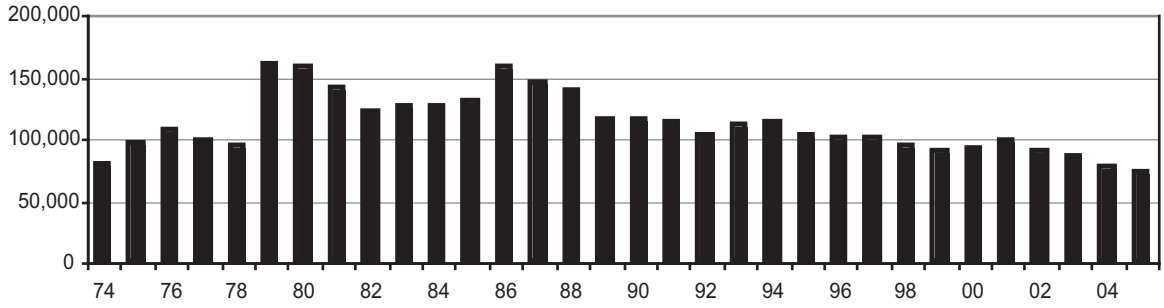
**Addresses and Staff:**
Fumiki Hirahara, MD
Yokohama City University Hospital
Dept. OB V GYN
3-9 Fukuura, Kanazawaku,
Yokohama, 236-0004, Japan
**Phone:** 81-45-787-2689
**Fax:** 81-45-787-2689

**E-mail:** hirafu@med.yokohama-cu.ac.jp

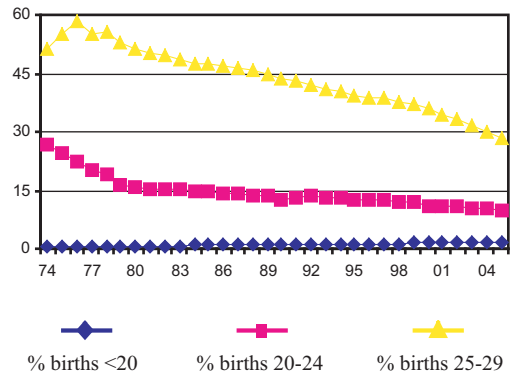
Michiko Yamanaka
**Phone:** 81-45-716-5366
**E-mail:** michikoy@yha.att.ne.jp

## Japan: JAOG

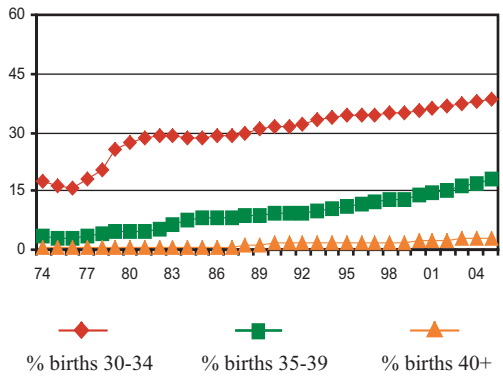
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# Japan: JAOG, 2005

| | |
|---|---|
| Live births (LB) | 71,765 |
| Stillbirths (SB) | 464 |
| Total births | 72,229 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 5 | 5 | nr | 1.38 |
| Spina bifida | 30 | 4 | nr | 4.71 |
| Encephalocele | 8 | 0 | nr | 1.11 |
| Microcephaly | 5 | 0 | nr | 0.69 |
| Holoprosencephaly | 8 | 10 | nr | 2.49 |
| Hydrocephaly | 46 | 6 | nr | 7.20 |
| Anophthalmos | 1 | 2 | nr | 0.42 |
| Microphthalmos | 2 | 0 | nr | 0.28 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | nr | nr | nr | nr |
| Microtia | 7 | 2 | nr | 1.25 |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 30 | 1 | nr | 4.29 |
| Tetralogy of Fallot | 41 | 0 | nr | 5.68 |
| Hypoplastic left heart syndrome | 35 | 4 | nr | 5.40 |
| Coarctation of aorta | 26 | 1 | nr | 3.74 |
| Choanal atresia, bilateral | nr | nr | nr | nr |
| Cleft palate without cleft lip | 46 | 2 | nr | 6.65 |
| Cleft lip with or without cleft palate | 117 | 13 | nr | 18.00 |
| Oesophageal atresia/stenosis with or without fistula | 41 | 3 | nr | 6.09 |
| Small intestine atresia/stenosis | 66 | 4 | nr | 9.69 |
| Anorectal atresia/stenosis | 43 | 3 | nr | 6.37 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 33 | 0 | nr | 4.57 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 11 | 3 | nr | 1.94 |
| Cystic kidney | 29 | 9 | nr | 5.26 |
| Bladder exstrophy | 2 | 0 | nr | 0.28 |
| Polydactyly, preaxial | 46 | 0 | nr | 6.37 |
| Total Limb reduction defects (include unspecified) | 25 | 2 | nr | 3.74 |
| Transverse | 3 | 1 | nr | 0.55 |
| Preaxial | 6 | 0 | nr | 0.83 |
| Postaxial | 4 | 1 | nr | 0.69 |
| Intercalary | 3 | 0 | nr | 0.42 |
| Mixed | 8 | 0 | nr | 1.11 |
| Unspecified | 1 | 0 | nr | 0.14 |
| Diaphragmatic hernia | 37 | 3 | nr | 5.54 |
| Omphalocele | 21 | 3 | nr | 3.32 |
| Gastroschisis | 17 | 4 | nr | 2.91 |
| Unspecified Omphalocele/Gastroschisis | 2 | 0 | nr | 0.28 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13 | 12 | 5 | nr | 2.35 |
| Trisomy 18 | 34 | 16 | nr | 6.92 |
| Down syndrome, all ages (include age unknown) | 87 | 4 | nr | 12.60 |
| <20 | 0 | 0 | nr | 0.00 |
| 20-24 | 4 | 0 | nr | 0.55 |
| 25-29 | 13 | 2 | nr | 2.08 |
| 30-34 | 26 | 0 | nr | 3.60 |
| 35-39 | 34 | 1 | nr | 4.85 |
| 40-44 | 10 | 1 | nr | 1.52 |
| 45+ | 0 | 0 | nr | 0.00 |
| unknown | 0 | 0 | nr | 0.00 |

nr = not reported

# Japan: JAOG, Previous years rates 1974 - 2005

Prevalence rates: (LB+SB) * 10,000

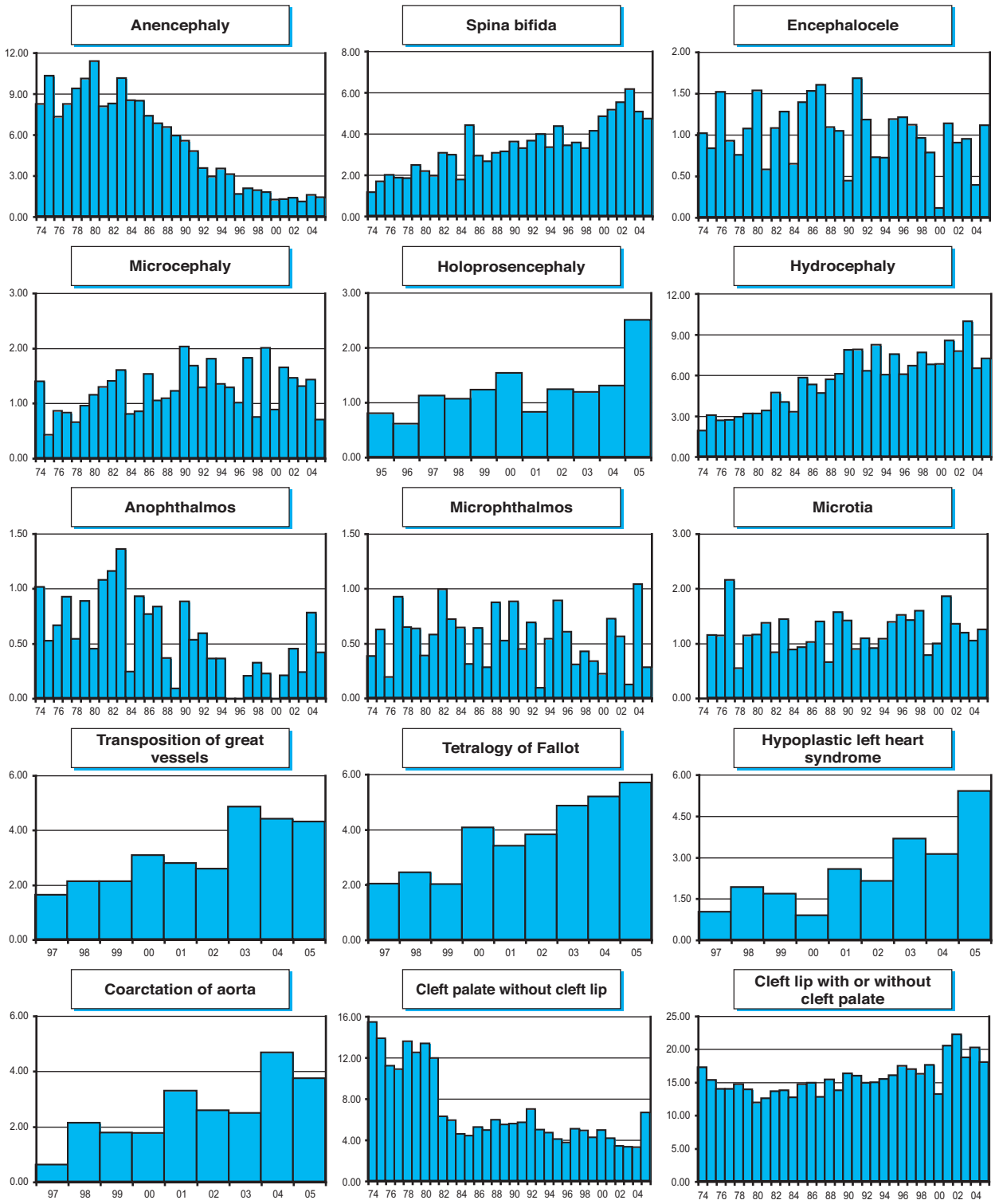
| | 1974-1980 | 1981-85 | 1986-90 | 1991-95 | 1996-00 | 2001-05 |
|---|---|---|---|---|---|---|
| **Births** | **787,267** | **641,607** | **669,652** | **539,382** | **474,407** | **420,750** |
| Anencephaly | 9.48 | 8.65 | 6.50 | 3.56 | 1.71 | 1.31 |
| Spina bifida | 1.96 | 2.81 | 3.03 | 3.69 | 3.82 | 5.32 |
| Encephalocele | 1.13 | 0.98 | 1.18 | 1.09 | 0.84 | 0.90 |
| Microcephaly | 0.90 | 1.18 | 1.36 | 1.48 | 1.29 | 1.33 |
| Holoprosencephaly | nr | nr | nr | 0.79* | 1.10 | 1.35 |
| Hydrocephaly | 2.85 | 4.21 | 5.78 | 7.17 | 6.77 | 8.03 |
| Anophthalmos | 0.70 | 0.95 | 0.60 | 0.37 | 0.15 | 0.40 |
| Microphthalmos | 0.53 | 0.64 | 0.63 | 0.52 | 0.38 | 0.55 |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | nr | nr | nr |
| Microtia | 1.08 | 1.09 | 1.18 | 1.06 | 1.26 | 1.35 |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | nr | nr | nr | nr | 2.21* | 3.71 |
| Tetralogy of Fallot | nr | nr | nr | nr | 2.61* | 4.49 |
| Hypoplastic left heart syndrome | nr | nr | nr | nr | 1.36* | 3.28 |
| Coarctation of aorta | nr | nr | nr | nr | 1.54* | 3.30 |
| Choanal atresia, bilateral | nr | nr | nr | nr | nr | nr |
| Cleft palate without cleft lip | 12.85 | 6.70 | 5.39 | 5.25 | 4.55 | 4.06 |
| Cleft lip with or without cleft palate | 14.07 | 13.39 | 14.52 | 15.41 | 16.25 | 19.99 |
| Oesophageal atresia/stenosis with or without fistula | 1.05* | 1.22 | 1.64 | 2.35 | 2.87 | 5.04 |
| Small intestine atresia/stenosis | nr | nr | nr | nr | 4.42* | 6.63 |
| Anorectal atresia/stenosis | 3.96 | 3.58 | 4.42 | 4.17 | 4.38 | 5.70 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 1.83 | 2.35 | 2.42 | 3.02 | 2.95 | 4.52 |
| Epispadias | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr | nr | nr |
| Renal agenesis | nr | nr | 1.09 | 1.56 | 1.67 | 2.14 |
| Cystic kidney | nr | nr | nr | nr | 2.77* | 4.80 |
| Bladder exstrophy | 0.17* | 0.12 | 0.16 | 0.09 | 0.21 | 0.31 |
| Polydactyly, preaxial | nr | nr | 5.88 | 6.64 | 6.09 | 6.44 |
| Total Limb reduction defects (include unspecified) | nr | nr | nr | 3.29* | 3.18 | 3.64 |
| Transverse | nr | nr | nr | 0.30* | 0.36 | 0.40 |
| Preaxial | nr | nr | nr | 0.52* | 0.57 | 0.67 |
| Postaxial | nr | nr | nr | 0.21* | 0.34 | 0.33 |
| Intercalary | nr | nr | nr | 1.41* | 0.95 | 0.81 |
| Mixed | nr | nr | nr | 0.52* | 0.59 | 0.93 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | nr | nr | 2.05 | 3.08 | 4.43 | 6.01 |
| Omphalocele | 1.09 | 1.37 | 3.03 | 2.98 | 3.56 | 3.45 |
| Gastroschisis | 1.05 | 0.86 | 1.33 | 1.45 | 2.11 | 2.66 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | nr | 0.19* | 0.00 | 0.02 |
| Trisomy 13 | nr | nr | nr | 0.46* | 0.95 | 1.50 |
| Trisomy 18 | nr | nr | nr | 2.20* | 4.01 | 8.03 |
| Down syndrome, all ages (include age unknown) | 4.25* | 4.72 | 6.17 | 6.27 | 8.43 | 10.53 |
| <20 | nr | nr | nr | 5.81* | 1.64 | 6.13 |
| 20-24 | nr | nr | nr | 2.15* | 2.98 | 4.05 |
| 25-29 | nr | nr | nr | 3.98* | 5.39 | 5.45 |
| 30-34 | nr | nr | nr | 5.67* | 8.02 | 8.87 |
| 35-39 | nr | nr | nr | 16.38* | 18.78 | 21.37 |
| 40+ | nr | nr | nr | 66.60* | 47.53 | 56.67 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

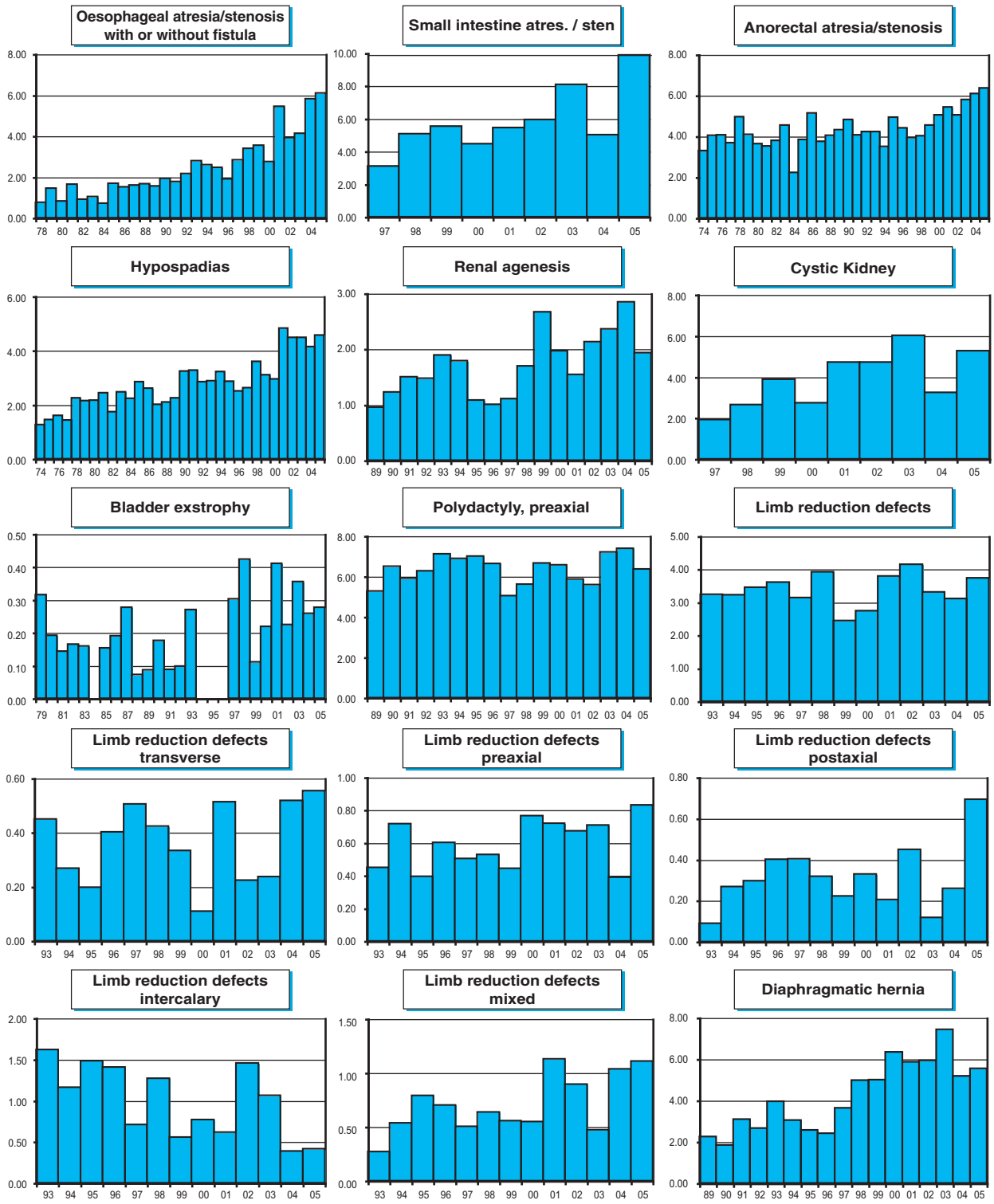
## Japan: JAOG

Time trends 1974-2005 (Birth prevalence rates per 10,000)



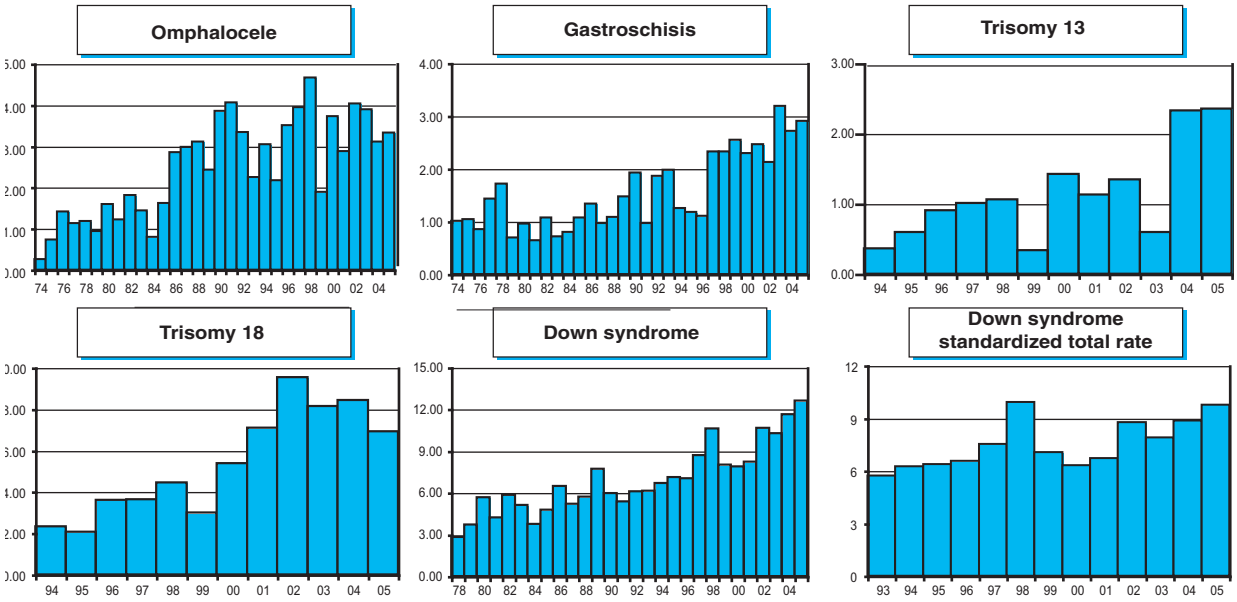

**Note:** L+S rates

## Japan: JAOG




**Note:** L+S rates

## Japan: JAOG




**Note:** L+S rates

## Malta

Malta Congenital Anomalies Register (MCAR)

**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 4000 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.

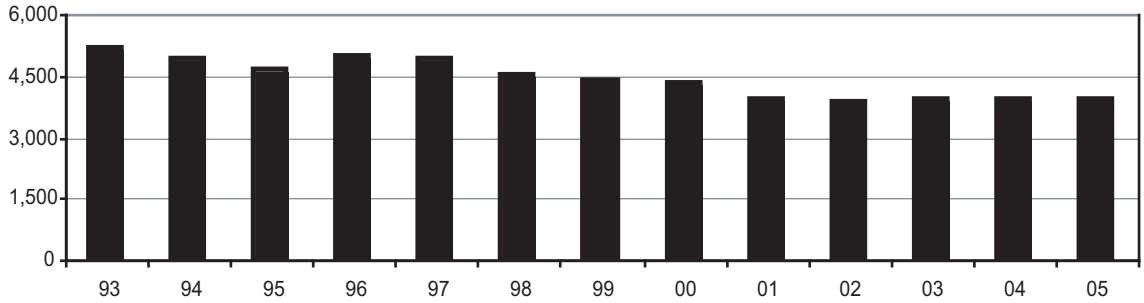
**Exposure information:**
Information regarding maternal exposure to medicinal drugs, smoking, alcohol and drug abuse as well as parental occupation are collected for all malformed infants and fetuses.

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

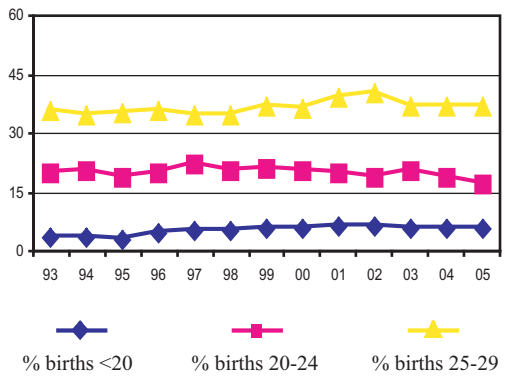
**Addresses and Staff:**
Miriam Gatt, MD, Programme Director
Malta Congenital Anomalies Registry
Department of Health Information and Research
95, Guardamangia Hill
Guardamangia PTA 1313
Malta
**Phone:** 356 25599000
**Fax:** 356 25599385
**E-mail:** miriam.gatt@gov.mt

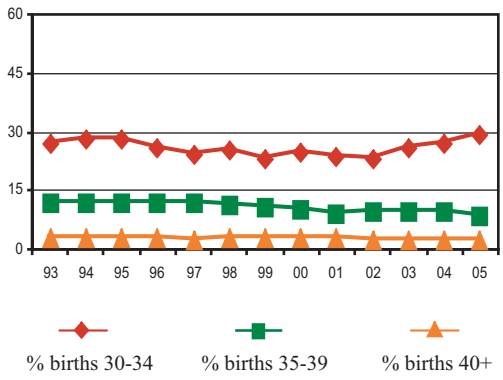
## Malta

**Total births by year**




**Percentage of births by year and maternal age**



# Malta: MCAR, 2005

| | |
|---|---|
| Live births (LB) | 2,357 |
| Stillbirths (SB) | 8 |
| Total births | 3,865 |
| 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 | 1 | 0 | | 2.59 |
| Spina bifida | 1 | 0 | | 2.59 |
| Encephalocele | 2 | 0 | | 5.17 |
| Microcephaly | 0 | 0 | | 0.00 |
| Holoprosencephaly | 0 | 0 | | 0.00 |
| Hydrocephaly | 2 | 0 | | 5.17 |
| Anophthalmos | 0 | 0 | | 0.00 |
| Microphthalmos | 0 | 0 | | 0.00 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 0 | 0 | | 0.00 |
| Microtia | 0 | 0 | | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | | 0.00 |
| Transposition of great vessels | 1 | 0 | | 2.59 |
| Tetralogy of Fallot | 3 | 0 | | 7.76 |
| Hypoplastic left heart syndrome | 0 | 0 | | 0.00 |
| Coarctation of aorta | 2 | 0 | | 5.17 |
| Choanal atresia, bilateral | 1 | 0 | | 2.59 |
| Cleft palate without cleft lip | 7 | 0 | | 18.11 |
| Cleft lip with or without cleft palate | 2 | 0 | | 5.17 |
| Oesophageal atresia/stenosis with or without fistula | 1 | 0 | | 2.59 |
| Small intestine atresia/stenosis | 0 | 0 | | 0.00 |
| Anorectal atresia/stenosis | 1 | 0 | | 2.59 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | | nr |
| Hypospadias | 9 | 0 | | 23.29 |
| Epispadias | 0 | 0 | | 0.00 |
| Indeterminate sex | 0 | 0 | | 0.00 |
| Renal agenesis | 0 | 0 | | 0.00 |
| Cystic kidney | 1 | 0 | | 2.59 |
| Bladder exstrophy | 0 | 0 | | 0.00 |
| Polydactyly, total | 3 | 0 | | 7.76 |
| Total Limb reduction defects (include unspecified) | 0 | 0 | | 0.00 |
| 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 | 0 | 0 | | 0.00 |
| Omphalocele | 1 | 0 | | 2.59 |
| Gastroschisis | 0 | 0 | | 0.00 |
| Unspecified Omphalocele/Gastroschisis | 1 | 0 | | 2.59 |
| Prune belly sequence | 0 | 0 | | 0.00 |
| Trisomy 13 | 0 | 0 | | 0.00 |
| Trisomy 18 | 1 | 0 | | 2.59 |
| Down syndrome, all ages (include age unknown) | 11 | 0 | | 28.46 |
| <20 | 0 | 0 | | 0.00 |
| 20-24 | 1 | 0 | | 15.22 |
| 25-29 | 0 | 0 | | 0.00 |
| 30-34 | 2 | 0 | | 17.70 |
| 35-39 | 4 | 0 | | 121.95 |
| 40-44 | 4 | 0 | | 500.00 |
| 45+ | 0 | 0 | | 0.00 |
| unknown | 0 | 0 | | 0.00 |

nr = not reported

# Malta: MCAR, Previous years rates 1993 - 2005

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

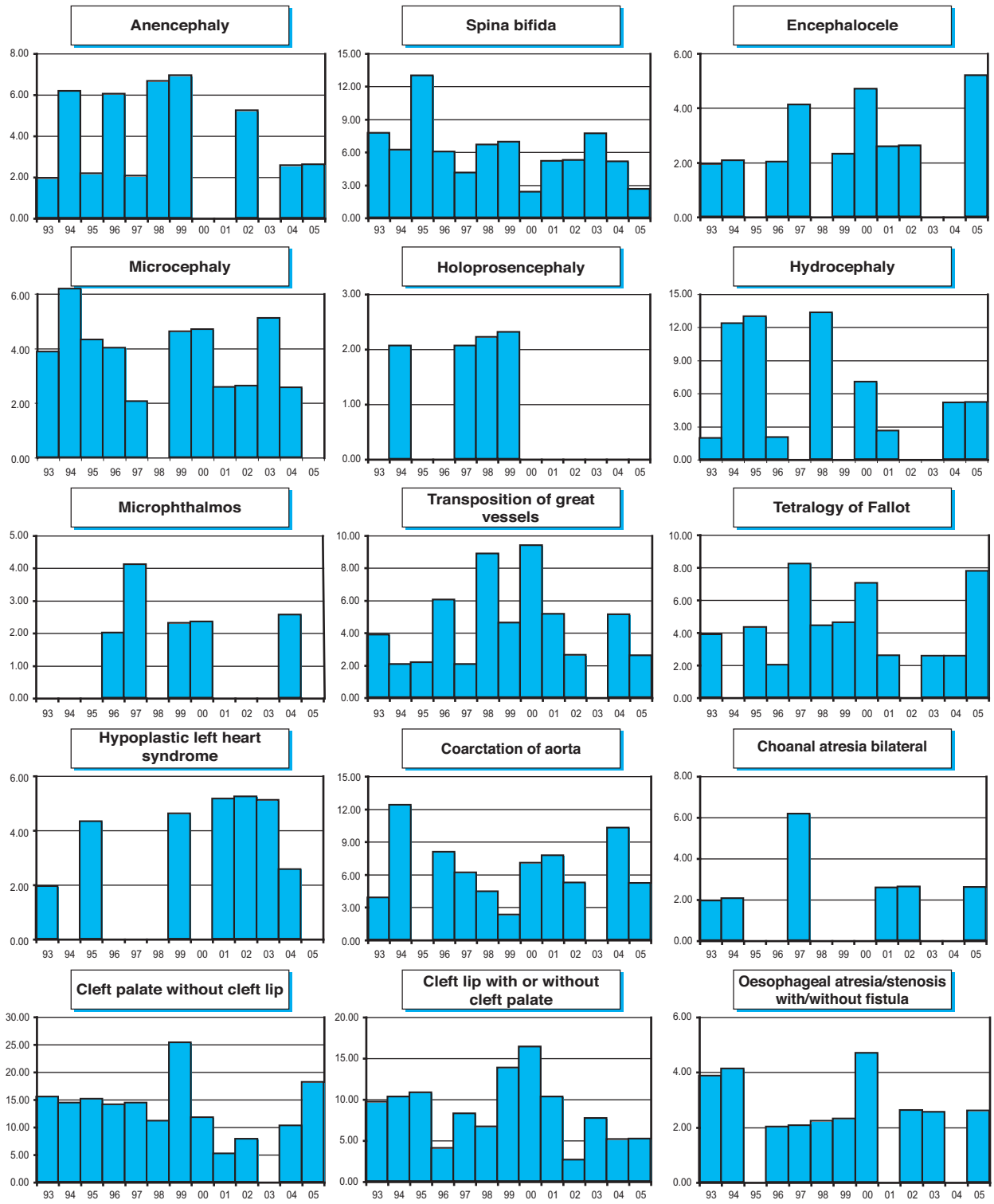
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995* | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | **14,668** | **22,964** | **19,396** |
| Anencephaly | | | | 3.41 | 4.35 | 2.06 |
| Spina bifida | | | | 8.86 | 5.23 | 5.16 |
| Encephalocele | | | | 1.36 | 2.61 | 2.06 |
| Microcephaly | | | | 4.77 | 3.05 | 2.58 |
| Holoprosencephaly | | | | 0.68 | 1.31 | 0.00 |
| Hydrocephaly | | | | 8.86 | 4.35 | 2.58 |
| Anophthalmos | | | | 0.68 | 0.00 | 0.00 |
| Microphthalmos | | | | 0.00 | 2.18 | 0.52 |
| Unspecified Anophthalmos/Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | 0.00 | 0.00 | 0.00 |
| Microtia | | | | 0.00 | 0.00 | 0.00 |
| Unspecified Anotia/Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 2.73 | 6.10 | 3.09 |
| Tetralogy of Fallot | | | | 2.73 | 5.23 | 3.09 |
| Hypoplastic left heart syndrome | | | | 2.05 | 0.87 | 3.61 |
| Coarctation of aorta | | | | 5.45 | 5.66 | 5.67 |
| Choanal atresia, bilateral | | | | 1.36 | 1.31 | 1.55 |
| Cleft palate without cleft lip | | | | 15.00 | 15.24 | 8.25 |
| Cleft lip with or without cleft palate | | | | 10.23 | 9.58 | 6.19 |
| Oesophageal atresia/stenosis with or without fistula | | | | 2.73 | 2.61 | 1.55 |
| Small intestine atresia/stenosis | | | | 0.68 | 1.74 | 2.58 |
| Anorectal atresia/stenosis | | | | 3.41 | 3.92 | 5.67 |
| Undescended testis (36 weeks of gestation or later) | | | | nr | nr | nr |
| Hypospadias | | | | 10.91 | 18.72 | 19.08 |
| Epispadias | | | | 2.05 | 0.44 | 0.00 |
| Indeterminate sex | | | | 1.36 | 1.31 | 0.52 |
| Renal agenesis | | | | 1.36 | 0.87 | 2.06 |
| Cystic kidney | | | | 4.77 | 3.92 | 1.55 |
| Bladder exstrophy | | | | 0.00 | 0.00 | 0.00 |
| Polydactyly, preaxial | | | | 14.32 | 16.11 | 16.50 |
| Total Limb reduction defects (include unspecified) | | | | 7.50 | 5.23 | 4.64 |
| Transverse | | | | nr | nr | nr |
| Preaxial | | | | nr | nr | nr |
| Postaxial | | | | nr | nr | nr |
| Intercalary | | | | nr | nr | nr |
| Mixed | | | | nr | nr | nr |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 4.77 | 6.53 | 2.58 |
| Omphalocele | | | | 2.73 | 1.74 | 2.06 |
| Gastroschisis | | | | 1.36 | 0.87 | 1.03 |
| Unspecified Omphalocele/Gastroschisis | | | | --- | --- | --- |
| Prune belly sequence | | | | 0.68 | 0.44 | 0.00 |
| Trisomy 13 | | | | 0.00 | 0.44 | 0.52 |
| Trisomy 18 | | | | 2.05 | 3.48 | 5.16 |
| Down syndrome, all ages (include age unknown) | | | | 20.45 | 14.81 | 23.20 |
| <20 | | | | 0.00 | 16.75 | 0.00 |
| 20-24 | | | | 0.00 | 0.00 | 2.72 |
| 25-29 | | | | 5.83 | 4.92 | 8.14 |
| 30-34 | | | | 22.24 | 14.17 | 26.03 |
| 35-39 | | | | 68.81 | 42.60 | 84.70 |
| 40-44 | | | | 166.21 | 132.01 | 238.10 |
| 45+ | | | | 0.00 | 416.67 | 0.00 |
| unknown | | | | --- | --- | --- |

* data include less than 5 years

nr = not reported

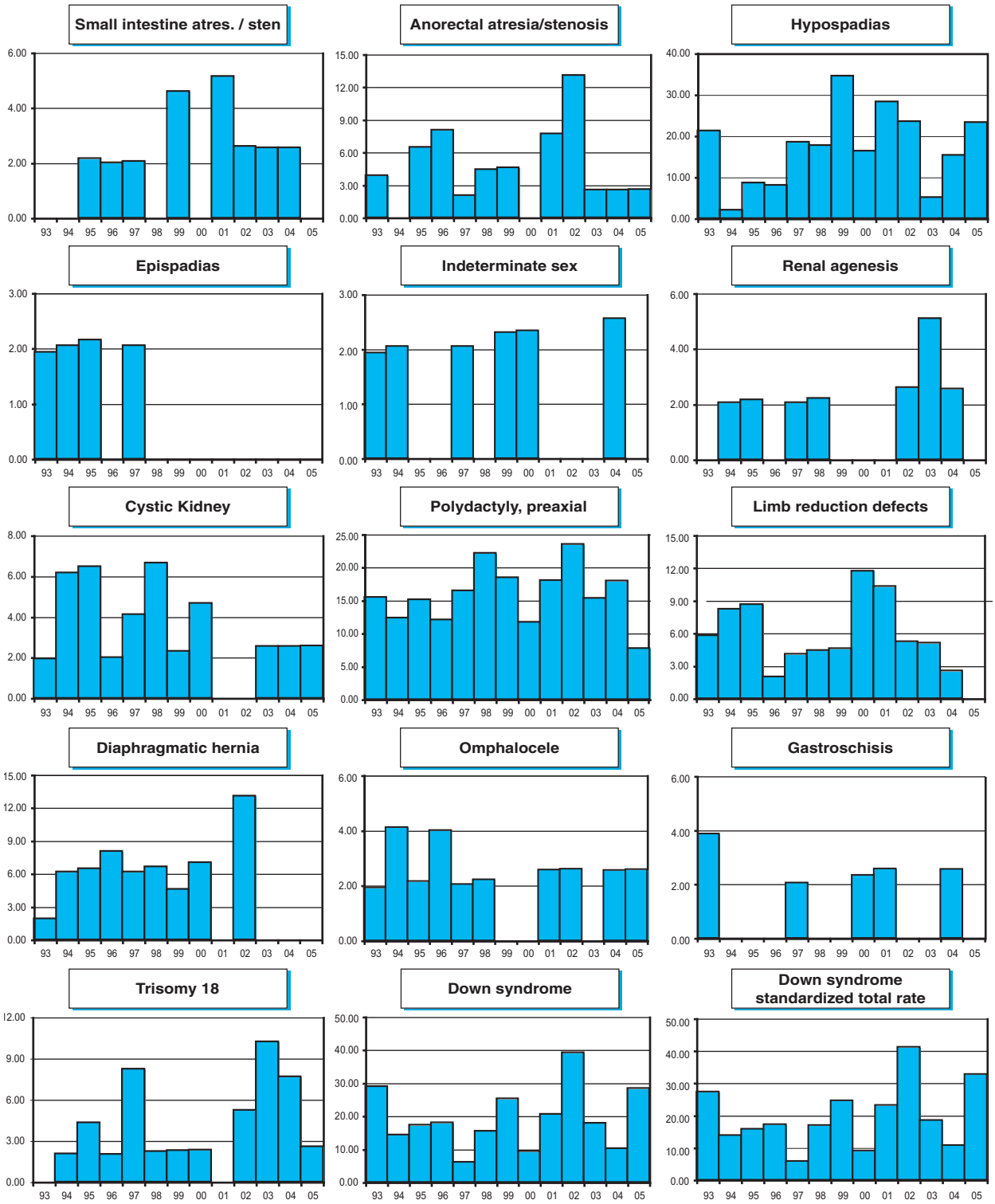
## Malta

'Time trends 1993-2005 (Birth prevalence rates per 10,000)




**Note:** L+S rates

## Malta




**Note:** L+S rates

## Mexico: RYVEMCE

Mexican Registry and Epidemiological Surveillance of External Congenital Malformations (RYVEMCE)

**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 15 hospitals in 11 cities in Mexico. Participation is voluntary. The annual number of births is approximately 40.000, 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.

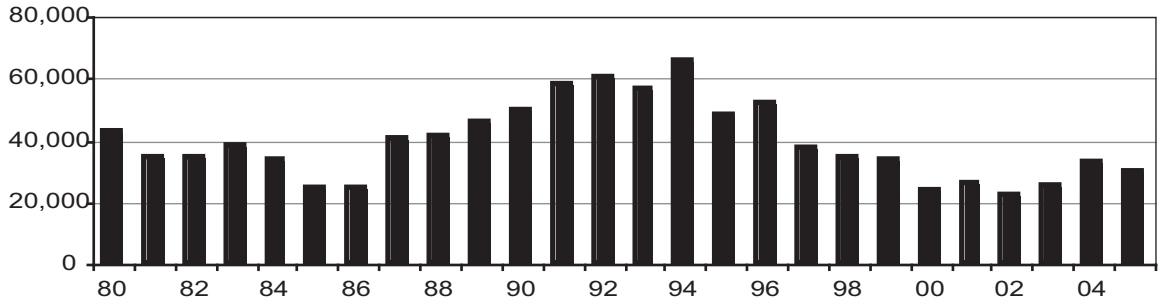
**Exposure information:**
The mother of each reported infant and the mother of a control infant-the next non-malformed infant born at that hospital with the same sex as the proband - are interviewed on various exposures, including drug usage and parental occupation.

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

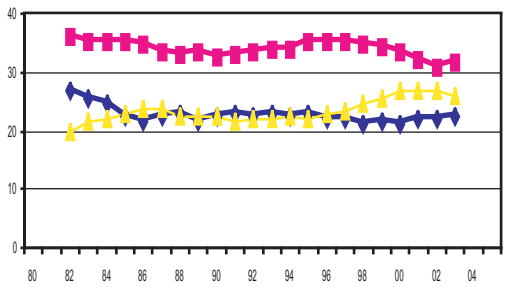
**Addresses and Staff:**
Osvaldo Mutchinick, MD, Programme Director
RYVEMCE Departamento de Genética, Inst. Nacional de Ciencias Médicas y Nutrición
Vasco de Quiroga 15,Tlalpan, C.P.14000
Mexico DF, Mexico
**Phone:** 52-55-54870900 (ext 2514 and 2515)
**Fax:** 52-55-56556138
**E-mail:** osvaldo@servidor.unam.mx

## Mexico: RYVEMCE

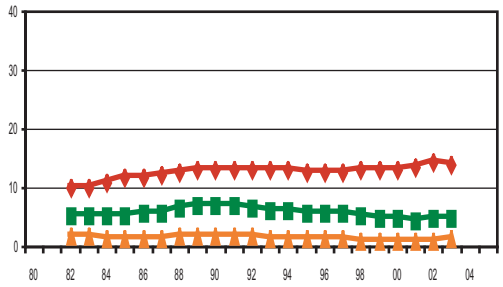
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# Mexico: RYVEMCE, 2005

| | |
|---|---|
| Live births (LB) | 29,089 |
| Stillbirths (SB) | 374 |
| Total births | 29,463 |
| 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 | 0 | 8 | | 2.72 |
| Spina bifida | 14 | 0 | | 4.75 |
| Encephalocele | 3 | 0 | | 1.02 |
| Microcephaly | 7 | 2 | | 3.05 |
| Holoprosencephaly | 6 | 2 | | 2.72 |
| Hydrocephaly | 19 | 3 | | 7.47 |
| Anophthalmos* | 2 | 2 | | 1.36 |
| Microphthalmos | nr | nr | | nr |
| Unspecified Anophthalmos/Microphthalmos | nr | nr | | nr |
| Anotia** | 25 | 2 | | 9.16 |
| Microtia | nr | nr | | nr |
| Unspecified Anotia/Microtia | nr | nr | | nr |
| Transposition of great vessels | 2 | 0 | | 0.68 |
| Tetralogy of Fallot | 0 | 0 | | 0.00 |
| Hypoplastic left heart syndrome | 0 | 0 | | 0.00 |
| Coarctation of aorta | 0 | 0 | | 0.00 |
| Choanal atresia, bilateral | 0 | 0 | | 0.00 |
| Cleft palate without cleft lip | 8 | 1 | | 3.05 |
| Cleft lip with or without cleft palate | 34 | 3 | | 12.56 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | | 2.72 |
| Small intestine atresia/stenosis | 7 | 0 | | 2.38 |
| Anorectal atresia/stenosis | 12 | 1 | | 4.41 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | | nr |
| Hypospadias | 17 | 0 | | 5.77 |
| Epispadias | 1 | 0 | | 0.34 |
| Indeterminate sex | 4 | 4 | | 2.72 |
| Renal agenesis | 2 | 1 | | 1.02 |
| Cystic kidney | 2 | 0 | | 0.68 |
| Bladder exstrophy | 1 | 0 | | 0.34 |
| Polydactyly, preaxial | 32 | 1 | | 11.20 |
| Total Limb reduction defects (include unspecified) | 12 | 7 | | 6.45 |
| Transverse | 8 | 2 | | 3.39 |
| Preaxial | 1 | 1 | | 0.68 |
| Postaxial | 3 | 1 | | 1.36 |
| Intercalary | 0 | 0 | | 0.00 |
| Mixed | 0 | 0 | | 0.00 |
| Unspecified | 0 | 3 | | 1.02 |
| Diaphragmatic hernia | 3 | 1 | | 1.36 |
| Omphalocele | 4 | 2 | | 2.04 |
| Gastroschisis | 17 | 0 | | 5.77 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | | 0.00 |
| Prune belly sequence | 2 | 0 | | 0.68 |
| Trisomy 13 | 0 | 0 | | 0.00 |
| Trisomy 18 | 0 | 0 | | 0.00 |
| Down syndrome, all ages (include age unknown) | 32 | 1 | | 11.20 |
| <20 | 6 | 0 | | 8.82 |
| 20-24 | 8 | 0 | | 7.97 |
| 25-29 | 6 | 0 | | 9.01 |
| 30-34 | 6 | 0 | | 16.60 |
| 35-39 | 4 | 0 | | 23.53 |
| 40-44 | 2 | 1 | | 77.12 |
| 45+ | 0 | 0 | | 0.00 |
| unknown | 0 | 0 | | 0.00 |

* = include Microphthalmos and Unspecified Anophthalmos/ Microphthalmos
** = include Microtia and Unspecified Anotia/Microtia
nr = not reported

# Mexico: RYVEMCE, Previous years rates 1980 - 2005

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

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **42,344** | **164,972** | **201,504** | **288,684** | **180,775** | **135,139** |
| Anencephaly | 17.71 | 18.18 | 20.25 | 15.93 | 12.94 | 5.77 |
| Spina bifida | 13.46 | 11.76 | 16.62 | 14.86 | 14.11 | 7.47 |
| Encephalocele | 4.25 | 2.91 | 3.18 | 2.32 | 2.43 | 1.41 |
| Microcephaly | 2.13 | 2.55 | 2.58 | 1.91 | 1.88 | 2.00 |
| Holoprosencephaly | 0.00 | 0.18 | 0.40 | 0.87 | 0.61 | 1.85 |
| Hydrocephaly | 6.14 | 5.64 | 4.76 | 5.99 | 6.20 | 7.03 |
| Anophthalmos** | 2.13 | 2.67 | 1.69 | 1.84 | 1.05 | 2.00 |
| Microphthalmos | nr | nr | nr | nr | nr | nr |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia*** | 7.79 | 6.61 | 6.30 | 6.58 | 6.47 | 9.40 |
| Microtia | nr | nr | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 0.00 | 0.06 | 0.10 | 0.17 | 0.28 | 0.30 |
| Tetralogy of Fallot | 0.00 | 0.00 | 0.00 | 0.28 | 0.11 | 0.15 |
| Hypoplastic left heart syndrome | 0.00 | 0.00 | 0.05 | 0.00 | 0.00 | 0.22 |
| Coarctation of aorta | 0.24 | 0.00 | 0.05 | 0.10 | 0.00 | 0.00 |
| Choanal atresia, bilateral | 0.24 | 0.24 | 0.40 | 0.52 | 0.17 | 0.15 |
| Cleft palate without cleft lip | 3.78 | 2.79 | 3.77 | 3.95 | 2.38 | 2.81 |
| Cleft lip with or without cleft palate | 14.17 | 12.67 | 12.56 | 12.51 | 12.11 | 15.61 |
| Oesophageal atresia/stenosis with or without fistula | 1.65 | 1.09 | 2.28 | 1.91 | 2.38 | 2.96 |
| Small intestine atresia/stenosis | 1.42 | 0.48 | 0.99 | 1.25 | 1.44 | 2.22 |
| Anorectal atresia/stenosis | 4.72 | 3.76 | 4.71 | 4.99 | 4.48 | 5.03 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | nr | nr |
| Hypospadias | 4.49 | 3.94 | 4.22 | 5.23 | 3.32 | 4.22 |
| Epispadias | nr | nr | nr | nr | nr | 0.16 |
| Indeterminate sex | 1.89 | 1.64 | 2.18 | 2.53 | 1.99 | 2.89 |
| Renal agenesis | 0.71 | 0.24 | 0.50 | 0.62 | 0.50 | 0.52 |
| Cystic kidney | 0.24 | 0.30 | 0.55 | 0.87 | 1.00 | 1.70 |
| Bladder exstrophy | 0.47 | 0.48 | 0.40 | 0.38 | 0.61 | 0.15 |
| Polydactyly, preaxial | 10.39 | 11.88 | 14.14 | 12.68 | 11.40 | 13.54 |
| Total Limb reduction defects (include unspecified) | 5.20 | 6.12 | 6.90 | 5.96 | 5.09 | 6.96 |
| Transverse | nr | nr | nr | nr | nr | 3.85 |
| Preaxial | nr | nr | nr | nr | nr | 1.26 |
| Postaxial | nr | nr | nr | nr | nr | 0.44 |
| Intercalary | nr | nr | nr | nr | nr | 0.44 |
| Mixed | nr | nr | nr | nr | nr | 0.67 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 0.94 | 0.42 | 1.09 | 0.97 | 0.89 | 1.41 |
| Omphalocele | 2.60 | 1.39 | 1.44 | 1.73 | 1.49 | 2.52 |
| Gastroschisis | 0.47 | 0.79 | 1.49 | 2.01 | 3.10 | 5.33 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.94 | 1.21 | 1.29 | 0.80 | 0.94 | 0.59 |
| Trisomy 13 | 0.71 | 0.18 | 0.30 | 0.21 | 0.11 | 0.22 |
| Trisomy 18 | 1.18 | 0.61 | 0.55 | 0.31 | 0.06 | 0.30 |
| Down syndrome, all ages (include age unknown) | 14.41 | 11.88 | 14.24 | 14.06 | 11.56 | 11.03 |
| <20 | | 6.13* | 10.76 | 7.95 | 6.65 | 6.64 |
| 20-24 | | 5.63* | 6.27 | 8.21 | 4.61 | 7.45 |
| 25-29 | | 4.64* | 9.57 | 12.56 | 6.40 | 6.85 |
| 30-34 | | 14.72* | 17.32 | 14.25 | 13.79 | 10.70 |
| 35-39 | | 45.92* | 43.52 | 41.83 | 45.11 | 53.13 |
| 40-44 | | 155.72* | 131.49 | 146.58 | 266.27 | 106.46 |
| 45+ | | 466.67* | 262.01 | 173.91 | 389.61 | 0.00* |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

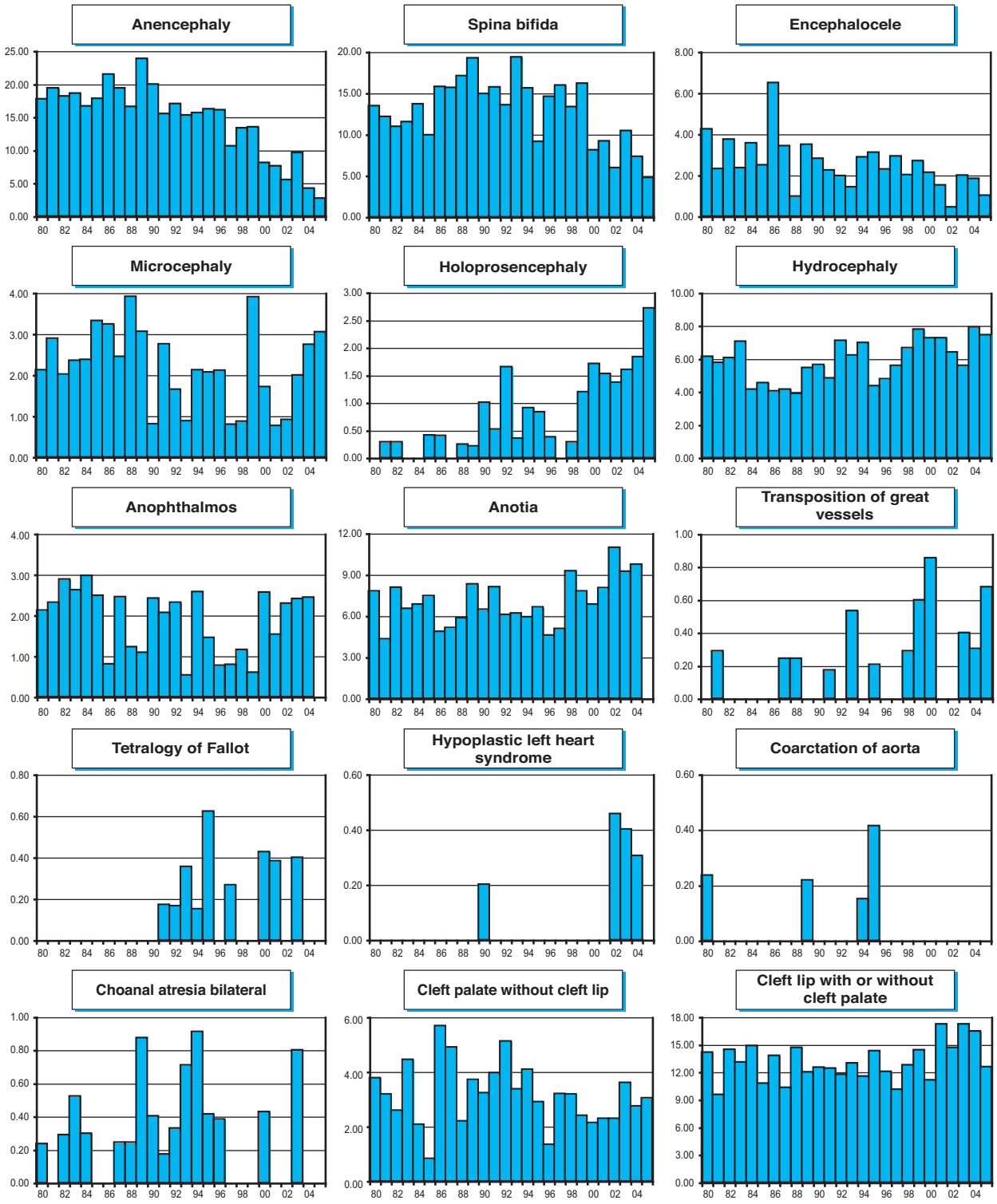
** = include Microphthalmos and Unspecified Anophthalmos/ Microphthalmos

*** = include Microtia and Unspecified Anotia/Microtia

nr = not reported

## Mexico: RYVEMCE

'Time trends 1980-2005 (Birth prevalence rates per 10,000)

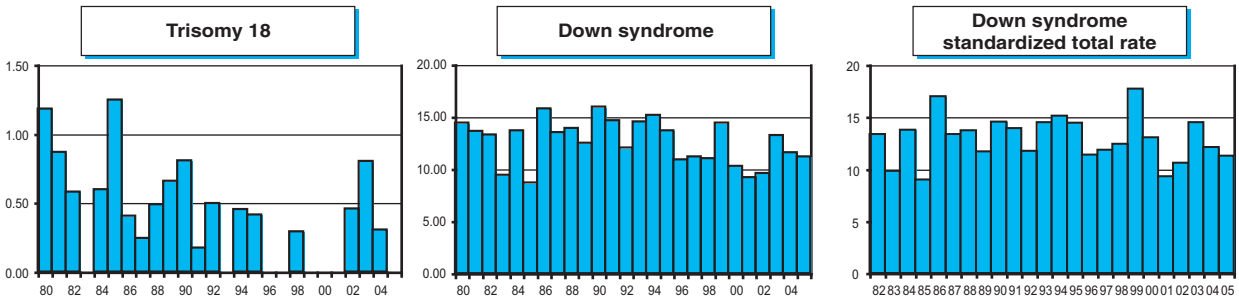



**Note:** L+S rates

## Mexico: RYVEMCE


**Note:** L+S rates

## Mexico: RYVEMCE




**Note:** L+S rates

## New Zealand

New Zealand Congenital Anomalies Monitoring Programme

**History:**
The Programme began in 1975 and became a full member of the ICBDSR in 1979.

**Size and coverage:**
The Programme covers all livebirths (approximately 56,000 per year) delivered or treated in a New Zealand publicly funded hospital. Only these data are included in the quarterly and annual reports to the ICBDSR. Data on stillbirths are retrospectively added to the database together with additional cases derived from the national perinatal and mortality databases. In late 1995 the definition of stillbirth was changed from 28 weeks completed gestation to 20 weeks or more gestation and/or 400g birthweight.

**Legislation and funding:**
The Programme is run and funded by the Public Health Directorate, Ministry of Health.

**Sources of ascertainment:**
Ascertainment is from discharge records of publicly funded hospitals and stillbirth notification forms. Data on voluntary terminations of pregnancy are being added to the database.

**Exposure information:**
No exposure information are currently available, but attempts are being made to obtain such data as well as increase the level of ascertainment.

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

**Addresses and Staff:**
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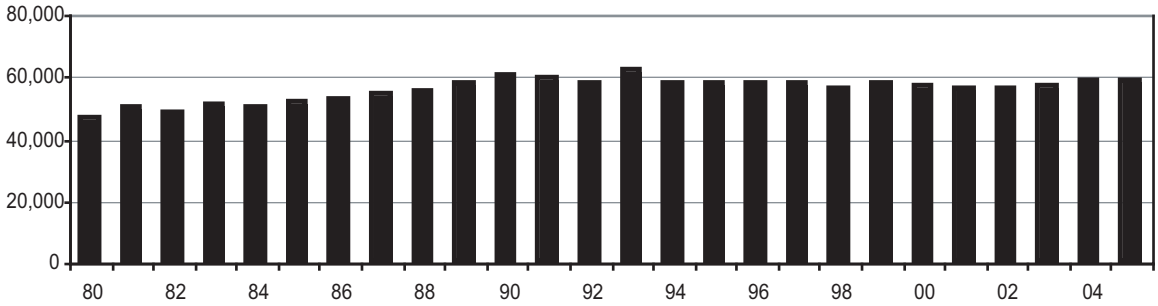
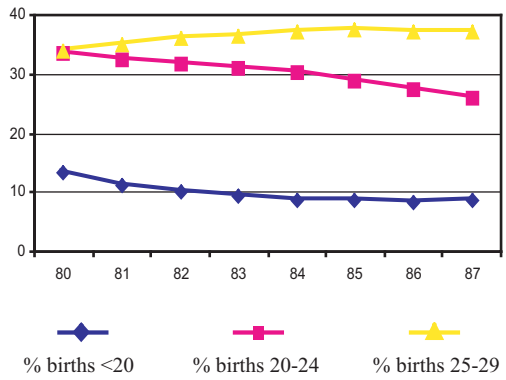
Public Health Intelligence
Public Health Directorate
Ministry of Health
PO Box 5013
Wellington, New Zealand
**Phone:** 64-4-496-2445
**Fax:** 64-4-496-2340
**E-mail:** barry_borman@moh.govt.nz

## New Zealand

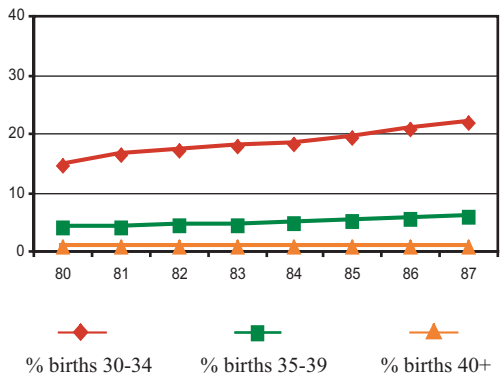
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# New Zealand: 2005

| | |
|---|---|
| Live births (LB) | 57,745 |
| Stillbirths (SB) | 360 |
| Total births | 58,105 |
| 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 | nr | nr | 0.00 |
| Spina bifida | 15 | nr | nr | 2.58 |
| Encephalocele | 5 | nr | nr | 0.86 |
| Microcephaly | 18 | nr | nr | 3.10 |
| Holoprosencephaly | nr | nr | nr | nr |
| Hydrocephaly | 18 | nr | nr | 3.10 |
| Anophthalmos | 1 | nr | nr | 0.17 |
| Microphthalmos | 5 | nr | nr | 0.86 |
| Unspecified Anophthalmos/Microphthalmos | 0 | nr | nr | 0.00 |
| Anotia | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | nr | nr | nr | nr |
| Transposition of great vessels | 25 | nr | nr | 4.30 |
| Tetralogy of Fallot | 36 | nr | nr | 6.20 |
| Hypoplastic left heart syndrome | 9 | nr | nr | 1.55 |
| Coarctation of aorta | 13 | nr | nr | 2.24 |
| Choanal atresia, bilateral | 6 | nr | nr | 1.03 |
| Cleft palate without cleft lip | 54 | nr | nr | 9.29 |
| Cleft lip with or without cleft palate | 36 | nr | nr | 6.20 |
| Oesophageal atresia/stenosis with or without fistula | 13 | nr | nr | 2.24 |
| Small intestine atresia/stenosis | 17 | nr | nr | 2.93 |
| Anorectal atresia/stenosis | 17 | nr | nr | 2.93 |
| Undescended testis (36 weeks of gestation or later) | 342 | nr | nr | 58.86 |
| Hypospadias + epispadias | 139 | nr | nr | 23.92 |
| Epispadias | nr | nr | nr | nr |
| Indeterminate sex | 8 | nr | nr | 1.38 |
| Renal agenesis | 19 | nr | nr | 3.27 |
| Cystic kidney | 37 | nr | nr | 6.37 |
| Bladder exstrophy | 2 | nr | nr | 0.34 |
| Polydactyly, preaxial | 49 | nr | nr | 8.43 |
| Total Limb reduction defects (include unspecified) | 15 | nr | nr | 2.58 |
| 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 | 14 | nr | nr | 2.41 |
| Omphalocele | nr | nr | nr | nr |
| Gastroschisis | nr | nr | nr | nr |
| Unspecified Omphalocele/Gastroschisis | 32 | nr | nr | 5.51 |
| Prune belly sequence | nr | nr | nr | nr |
| Trisomy 13 | 2 | nr | nr | 0.34 |
| Trisomy 18 | 4 | nr | nr | 0.69 |
| Down syndrome, all ages (include age unknown) | 62 | nr | nr | 10.67 |
| <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 = not reported

# New Zealand: Previous years rates 1980 - 2005

Birth prevalence rates: (LB) * 10,000

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **46,180** | **249,914** | **279,760** | **293,819** | **284,949** | **284,803** |
| Anencephaly | 6.93 | 4.56 | 2.14 | 0.75 | 0.49 | 0.25 |
| Spina bifida | 12.56 | 10.08 | 5.72 | 3.98 | 3.16 | 1.97 |
| Encephalocele | nr | 0.66* | 0.74* | 0.00* | 0.39 | 0.53 |
| Microcephaly | nr | nr | nr | nr | 2.77 | 2.91 |
| Holoprosencephaly | nr | nr | nr | nr | nr | nr |
| Hydrocephaly | 7.36 | 3.48 | 3.36 | 2.76 | 3.65 | 3.55 |
| Anophthalmos | nr | nr | nr | nr | 0.00 | 0.07 |
| Microphthalmos | nr | nr | nr | nr | 0.67 | 0.81 |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | nr | nr | nr |
| Microtia | nr | nr | nr | nr | nr | nr |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | nr | nr | 0.55* | nr | 5.12 | 4.78 |
| Tetralogy of Fallot | nr | nr | nr | nr | 4.65* | 4.35 |
| Hypoplastic left heart syndrome | nr | nr | 0.82* | 1.90* | 1.40 | 1.09 |
| Coarctation of aorta | nr | nr | nr | nr | 2.00* | 3.44 |
| Choanal atresia, bilateral | nr | nr | nr | nr | 1.02 | 1.02 |
| Cleft palate without cleft lip | 6.06 | 6.44 | 7.11 | 5.04 | 8.81 | 9.55 |
| Cleft lip with or without cleft palate | 10.83 | 8.56 | 7.58 | 3.57 | 5.62 | 5.93 |
| Oesophageal atresia/stenosis with or without fistula | 1.73 | 1.88 | 1.75 | 2.42 | 2.11 | 1.47 |
| Small intestine atresia/stenosis | nr | nr | nr | nr | 1.79 | 2.27* |
| Anorectal atresia/stenosis | 1.73 | 2.52 | 2.36 | 2.89 | 2.28 | 2.42 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | 67.12* | 73.24 |
| Hypospadias | 12.13 | 13.64 | 12.47 | 12.15* | 22.53 | 28.69 |
| Epispadias | nr | nr | nr | nr | nr | nr |
| Indeterminate sex | nr | nr | nr | nr | 0.48* | 0.81 |
| Renal agenesis | nr | 0.20* | 0.43* | nr | 3.52* | 3.05 |
| Cystic kidney | nr | nr | nr | nr | 5.83 | 5.97 |
| Bladder exstrophy | nr | nr | nr | nr | 0.39* | 0.25 |
| Polydactyly, preaxial | nr | nr | nr | nr | 5.99* | 9.73 |
| Total Limb reduction defects (include unspecified) | 3.03 | 4.00 | 2.97 | 2.04 | 2.63 | 2.88 |
| Transverse | nr | nr | nr | nr | nr | nr |
| Preaxial | nr | nr | nr | nr | nr | nr |
| Postaxial | nr | nr | nr | nr | nr | nr |
| Intercalary | nr | nr | nr | nr | nr | nr |
| Mixed | nr | nr | nr | nr | nr | nr |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | nr | 1.51* | 1.48* | nr | 2.41* | 2.46 |
| Omphalocele | nr | 2.28 | 1.57 | 2.58* | nr | nr |
| Gastroschisis | nr | 0.28 | 0.62* | nr | nr | nr |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | nr | nr | nr | nr |
| Trisomy 13 | nr | nr | nr | nr | 0.42 | 0.42 |
| Trisomy 18 | nr | nr | nr | nr | 0.95 | 1.19 |
| Down syndrome, all ages (include age unknown) | 8.01 | 9.32 | 9.29 | 9.38* | 11.02 | 11.31 |
| <20 | 1.63 | 6.22 | 6.64* | nr | nr | nr |
| 20-24 | 4.52 | 4.55 | 3.14* | nr | nr | nr |
| 25-29 | 8.92 | 8.36 | 8.83* | nr | nr | nr |
| 30-34 | 10.46 | 9.92 | 8.38* | nr | nr | nr |
| 35-39 | 22.27 | 37.78 | 26.30* | nr | nr | nr |
| 40-44 | 82.42 | 97.04 | 274.46* | nr | nr | nr |
| 45+ | 0.00 | 144.93 | 217.39* | nr | nr | nr |
| unspecified | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

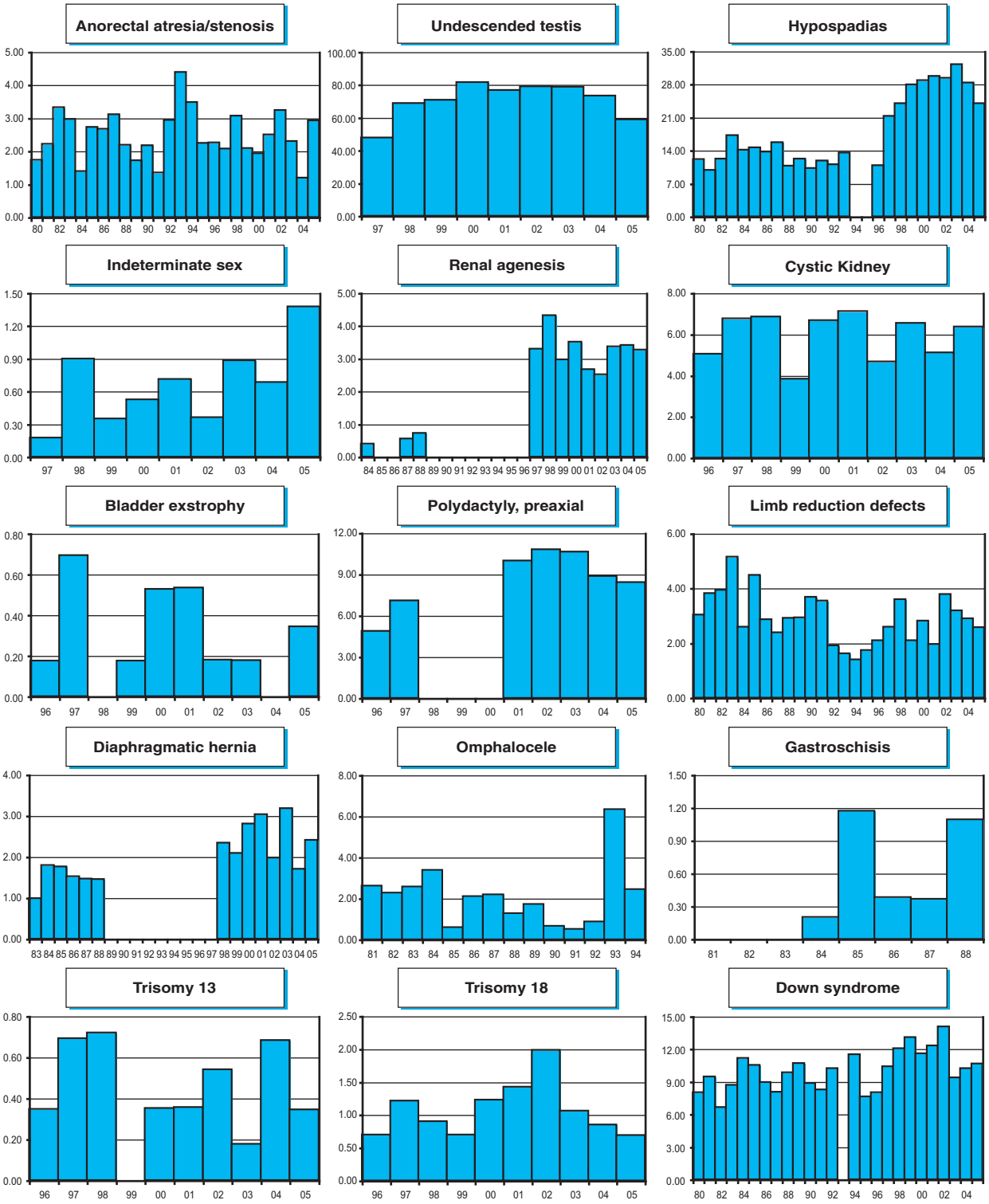
nr = not reported

## New Zealand

Time trends 1980-2005 (Birth prevalence rates per 10,000)

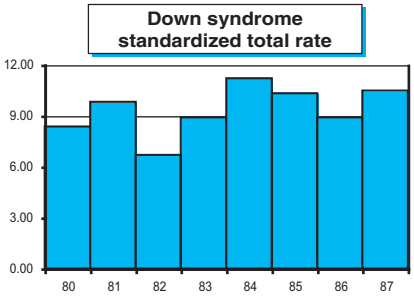

**Note:** L+S rates

## New Zealand




**Note:** L+S rates

## New Zealand




**Note:** L+S rates

## Northern Netherlands

EUROCAT registration 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. 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 16 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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9700 RB Groningen, The Netherlands
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**Fax:** 31-50-3617232
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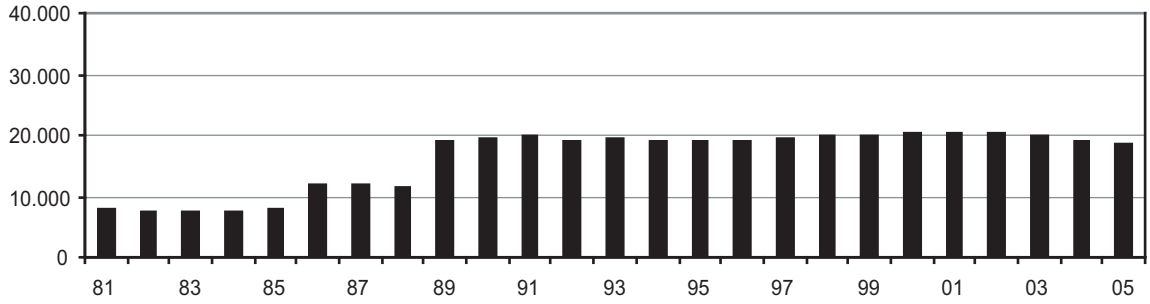
**E-mail:** h.e.k.de.Walle@medgen.umcg.nl

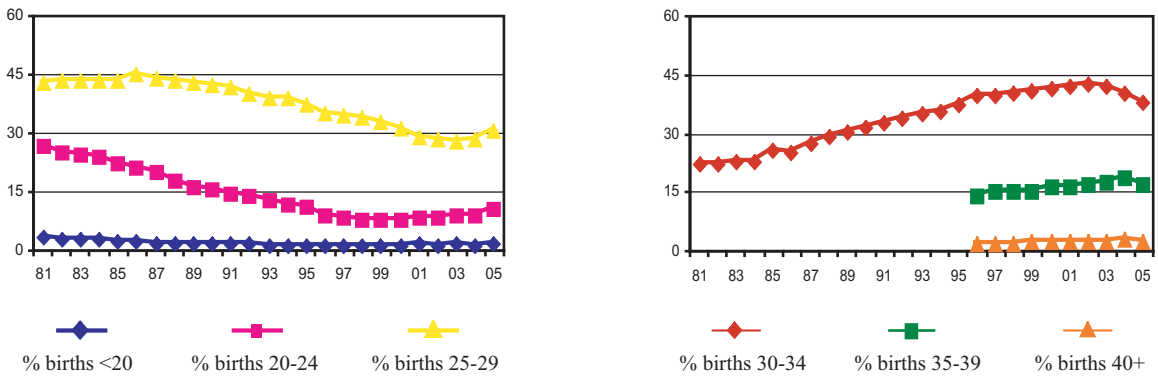
**Web:** www.eurocatnederland.nl

## Northern Netherlands

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 4 | 50.0 | Cystic kidney | 1 | 3.7 |
| Spina bifida | 8 | 29.6 | Limb reduction defects | 2 | 5.1 |
| Encephalocele | 1 | 50.0 | Diaphragmatic hernia | 0 | 0.0 |
| Holoprosencephaly | 2 | 33.3 | Omphalocele | 5 | 38.5 |
| Hydrocephaly | 4 | 23.5 | Gastroschisis | 1 | 20.0 |
| Hypoplastic left heart syndrome | 2 | 16.7 | Trisomy 13 | 3 | 37.5 |
| Cleft palate without cleft lip | 3 | 9.7 | Trisomy 18 | 18 | 47.4 |
| Cleft lip with or without cleft palate | 4 | 4.8 | Down syndrome | 33 | 37.5 |
| Renal agenesis | 2 | 15.4 | | | |

Total ToPs with births defects = 82 (Ratio ToPs/Births: 1.42 per 1,000)
*ToPs/ToPs+Births

# Northern Netherlands: 2005

| | |
|---|---|
| Live births (LB) | 18,419 |
| Stillbirths (SB) | 103 |
| Total births | 18,522 |
| Number of terminations of pregnancy (ToP) for birth defects | 29 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 0 | 2 | 1.08 |
| Spina bifida | 6 | 1 | 2 | 4.86 |
| Encephalocele | 0 | 0 | 0 | 0.00 |
| Microcephaly | 0 | 0 | 0 | 0.00 |
| Holoprosencephaly | 1 | 0 | 1 | 1.08 |
| Hydrocephaly | 4 | 1 | 1 | 3.24 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 1 | 0 | 0 | 0.54 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 1 | 0.54 |
| Microtia | 0 | 0 | 0 | 0.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 7 | 0 | 0 | 3.78 |
| Tetralogy of Fallot | 2 | 1 | 1 | 2.16 |
| Hypoplastic left heart syndrome | 3 | 0 | 0 | 1.62 |
| Coarctation of aorta | 2 | 0 | 0 | 1.08 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 3 | 0 | 1 | 2.16 |
| Cleft lip with or without cleft palate | 24 | 1 | 1 | 14.04 |
| Oesophageal atresia/stenosis with or without fistula | 3 | 0 | 0 | 1.62 |
| Small intestine atresia/stenosis | 3 | 0 | 0 | 1.62 |
| Anorectal atresia/stenosis | 3 | 0 | 0 | 1.62 |
| Undescended testis (36 weeks of gestation or later) | 0 | 0 | 0 | 0.00 |
| Hypospadias | 34 | 0 | 1 | 18.90 |
| Epispadias | 1 | 0 | 0 | 0.54 |
| Indeterminate sex | 0 | 1 | 0 | 0.54 |
| Renal agenesis | 4 | 1 | 0 | 2.70 |
| Cystic kidney | 9 | 0 | 0 | 4.86 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 0 | 0 | 0 | 0.00 |
| Total Limb reduction defects (include unspecified) | 12 | 1 | 0 | 7.02 |
| Transverse | 9 | 1 | 0 | 5.40 |
| Preaxial | 5 | 0 | 0 | 2.70 |
| Postaxial | 3 | 1 | 0 | 2.16 |
| Intercalary | 1 | 0 | 0 | 0.54 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 2 | 0 | 0 | 1.08 |
| Omphalocele | 2 | 0 | 2 | 2.16 |
| Gastroschisis | 1 | 0 | 0 | 0.54 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | 2 | 1.62 |
| Trisomy 18 | 4 | 5 | 9 | 9.72 |
| Down syndrome, all ages (include age unknown) | 22 | 0 | 6 | 15.12 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 1 | 0 | 0 | 51.12 |
| 25-29 | 2 | 0 | 1 | 52.79 |
| 30-34 | 8 | 0 | 0 | 114.78 |
| 35-39 | 8 | 0 | 3 | 348.21 |
| 40-44 | 2 | 0 | 1 | 696.06 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 1 | 0 | 1 | --- |

# Northern Netherlands: Previous years rates 1981 - 2005

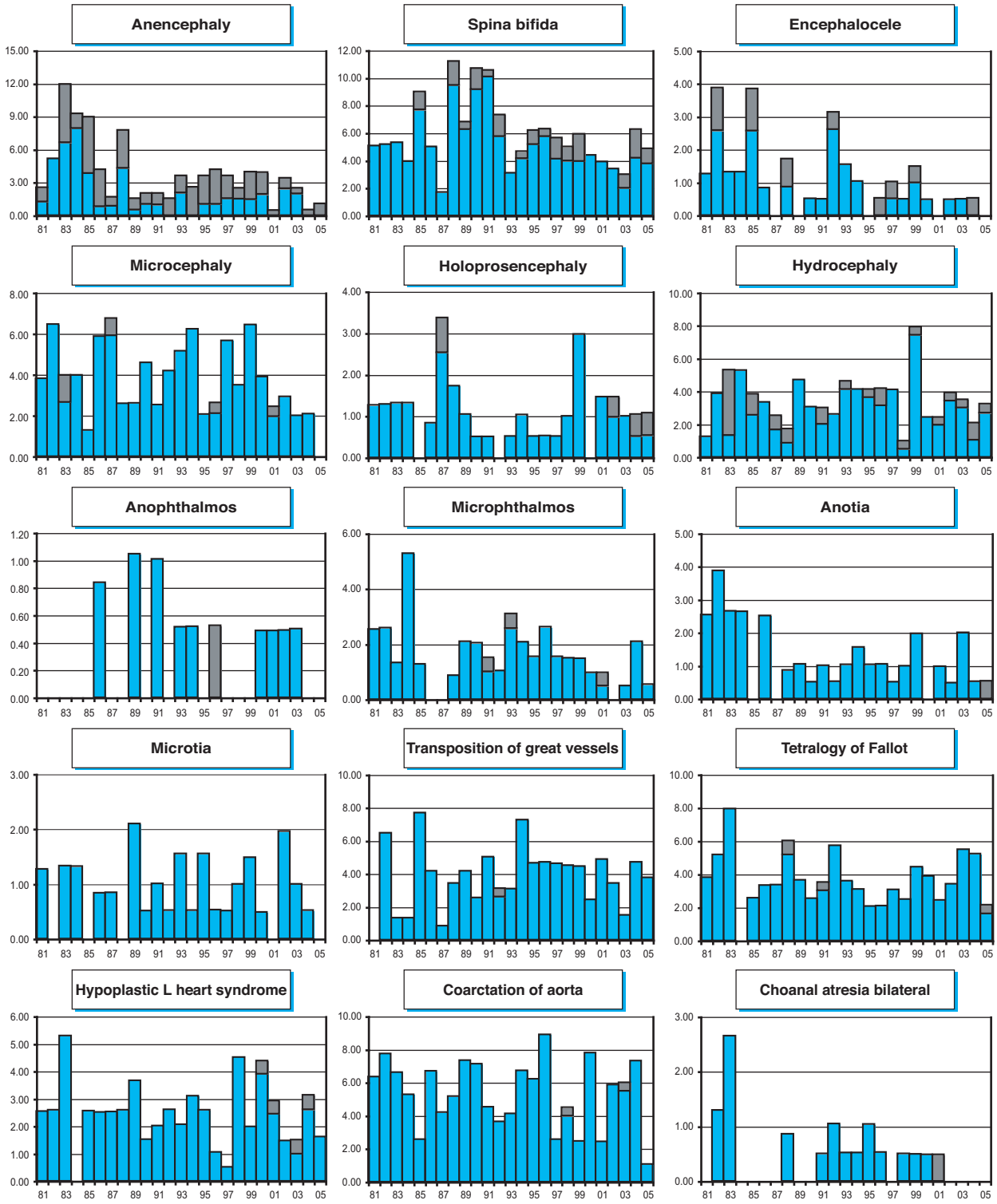
Prevalence rates: (LB+SB+TOP) * 10,000

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | **38,501** | **74,106** | **96,959** | **99,168** | **98,579** |
| Anencephaly | | 7.53 | 3.10 | 2.68 | 3.63 | 1.62 |
| Spina bifida | | 5.71 | 7.42 | 6.39 | 5.45 | 4.26 |
| Encephalocele | | 2.34 | 0.54 | 1.24 | 0.81 | 0.30 |
| Microcephaly | | 3.90 | 4.32 | 4.02 | 4.44 | 1.93 |
| Holoprosencephaly | | 1.04 | 1.35 | 0.52 | 1.01 | 1.22 |
| Hydrocephaly | | 3.90 | 3.24 | 3.71 | 3.93 | 3.04 |
| Anophthalmos | | 0.00 | 0.40 | 0.41 | 0.20 | 0.30 |
| Microphthalmos | | 2.60 | 1.21 | 1.86 | 1.61 | 0.81 |
| Unspecified Anophthalmos/Microphthalmos | | --- | --- | --- | --- | --- |
| Anotia | | 2.34 | 0.94 | 1.03 | 0.91 | 0.91 |
| Microtia | | 0.78 | 0.94 | 1.03 | 0.81 | 0.71 |
| Unspecified Anotia/Microtia | | --- | --- | --- | --- | --- |
| Transposition of great vessels | | 3.38 | 3.10 | 4.64 | 4.13 | 3.65 |
| Tetralogy of Fallot | | 3.90 | 3.64 | 3.61 | 3.23 | 3.75 |
| Hypoplastic left heart syndrome | | 2.60 | 2.56 | 2.48 | 2.52 | 2.13 |
| Coarctation of aorta | | 5.71 | 6.34 | 5.05 | 5.24 | 4.56 |
| Choanal atresia, bilateral | | 0.78 | 0.13 | 0.72 | 0.40 | 0.10 |
| Cleft palate without cleft lip | | 7.01 | 6.48 | 8.46 | 6.86 | 6.19 |
| Cleft lip with or without cleft palate | | 16.62 | 15.25 | 15.47 | 13.41 | 14.51 |
| Oesophageal atresia/stenosis with or without fistula | | 2.60 | 2.70 | 2.99 | 3.93 | 3.55 |
| Small intestine atresia/stenosis | | 2.60 | 2.97 | 2.68 | 2.12 | 1.52 |
| Anorectal atresia/stenosis | | 2.34 | 3.78 | 2.78 | 4.13 | 2.94 |
| Undescended testis (36 weeks of gestation or later) | | 2.08 | 2.29 | 1.44 | 1.31 | 0.91 |
| Hypospadias | | 16.62 | 9.58 | 9.69 | 15.13 | 18.46 |
| Epispadias | | 0.26 | 0.67 | 0.52 | 0.40 | 0.51 |
| Indeterminate sex | | 0.00 | 0.27 | 0.21 | 0.50 | 0.51 |
| Renal agenesis | | 3.64 | 4.45 | 4.43 | 4.74 | 3.85 |
| Cystic kidney | | 1.56 | 5.53 | 4.95 | 4.03 | 4.06 |
| Bladder exstrophy | | 0.26 | 0.27 | 0.10 | 0.20 | 0.20 |
| Polydactyly, preaxial | | 2.08 | 1.62 | 2.06 | 2.42 | 0.51 |
| Total Limb reduction defects (include unspecified) | | 8.05 | 4.72 | 7.22 | 5.65 | 6.59 |
| Transverse | | 5.19 | 2.70 | 3.92 | 3.33 | 4.16 |
| Preaxial | | 1.30 | 0.54 | 0.93 | 0.71 | 1.22 |
| Postaxial | | 0.52 | 0.81 | 1.86 | 1.01 | 1.83 |
| Intercalary | | 0.00 | 0.00 | 0.31 | 0.10 | 0.20 |
| Mixed | | 0.00 | 0.40 | 0.41 | 0.20 | 0.10 |
| Unspecified | | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | | 2.34 | 2.97 | 2.17 | 3.23 | 2.03 |
| Omphalocele | | 1.56 | 2.29 | 3.30 | 2.32 | 1.93 |
| Gastroschisis | | 0.78 | 0.81 | 0.52 | 1.01 | 0.91 |
| Unspecified Omphalocele/Gastroschisis | | --- | --- | --- | --- | --- |
| Prune belly sequence | | 0.26 | 0.27 | 0.52 | 0.30 | 0.20 |
| Trisomy 13 | | 0.52 | 1.21 | 1.34 | 0.91 | 1.32 |
| Trisomy 18 | | 2.08 | 2.56 | 1.65 | 3.63 | 5.38 |
| Down syndrome, all ages (include age unknown) | | 12.21 | 13.90 | 14.23 | 15.63 | 16.33 |
| <20 | | 0.00 | 0.00 | 0.00 | 0.00 | 0.00 |
| 20-24 | | 10.62 | 4.56 | 10.59 | 3.76 | 5.61 |
| 25-29 | | 6.60 | 13.14 | 4.72 | 9.09 | 8.11 |
| 30-34 | | 14.65 | 7.44 | 15.97 | 10.73 | 10.41 |
| 35-39 | | 40.21 | 47.07 | 33.26 | 39.28 | 31.10 |
| 40-44 | | nr | nr | nr | 98.60 | 135.08 |
| 45+ | | nr | nr | nr | 153.85 | 129.87 |
| unknown | | --- | --- | --- | --- | --- |

nr = not reported

## Northern Netherlands

'Time trends 1981-2005 (Birth prevalence rates per 10,000)

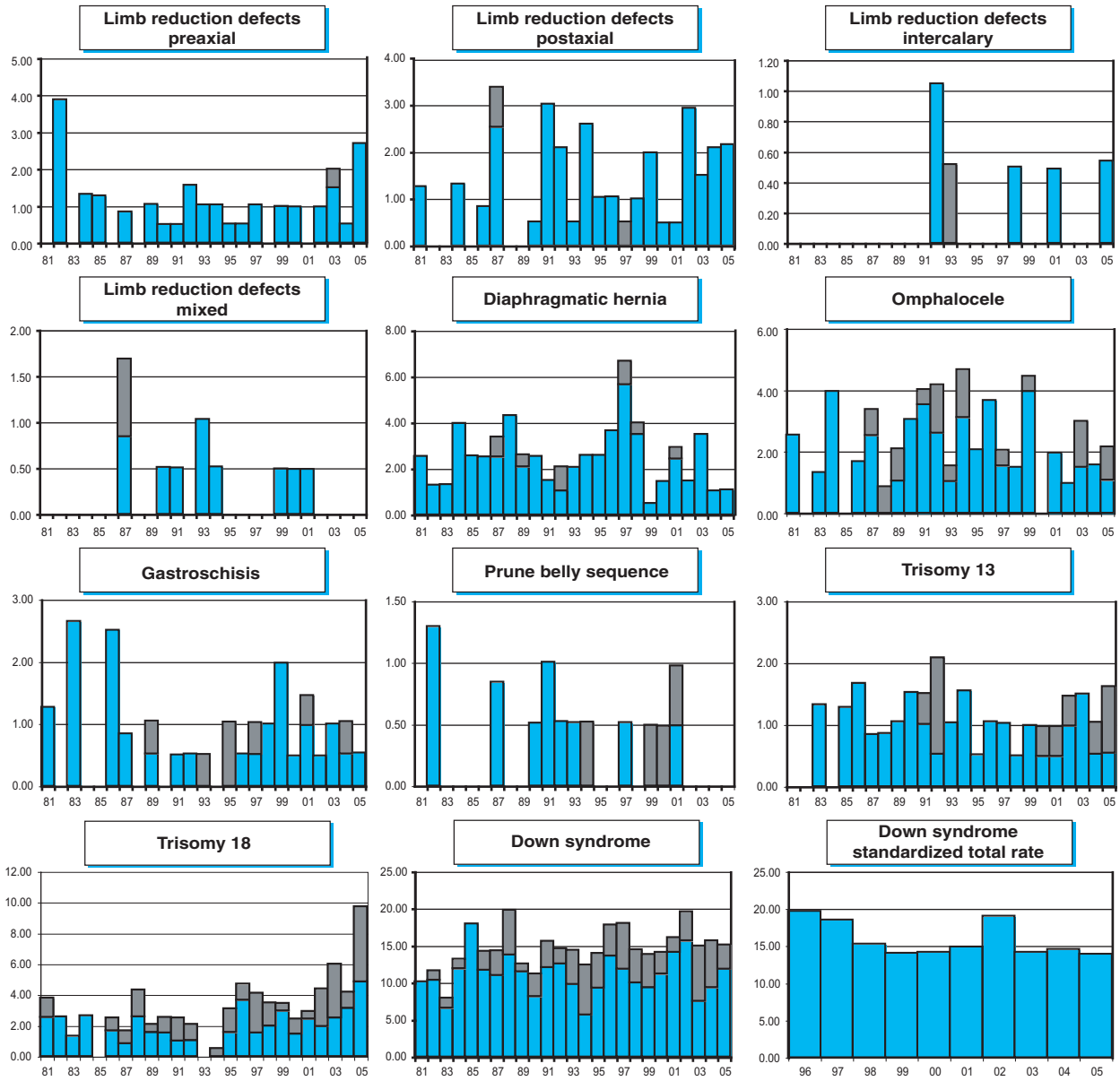



**Note:** L+S rates, ToP rates

## Northern Netherlands


**Note:** L+S rates, ToP rates

## Northern Netherlands




**Note:** L+S rates, ToP rates

## Norway
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.
Stillbirths of 16 weeks or more gestation are included.

**Legislation and funding:**
The Programme part of the Norwegian Institute of Public Health. Reporting is compulsory (Personal Health Data Filing System Act, 2002).

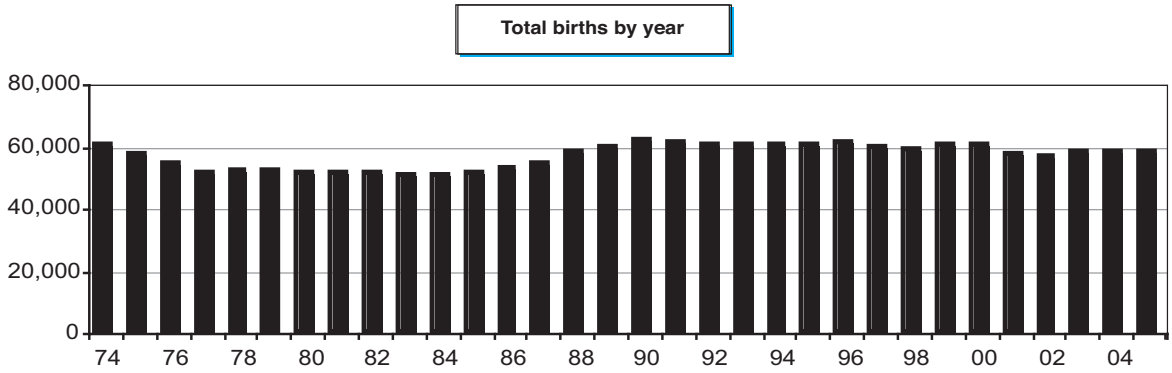
**Sources of ascertainment:**
The registry is based on the notification of births from the delivery units and since 1999 also from the neonatal units.

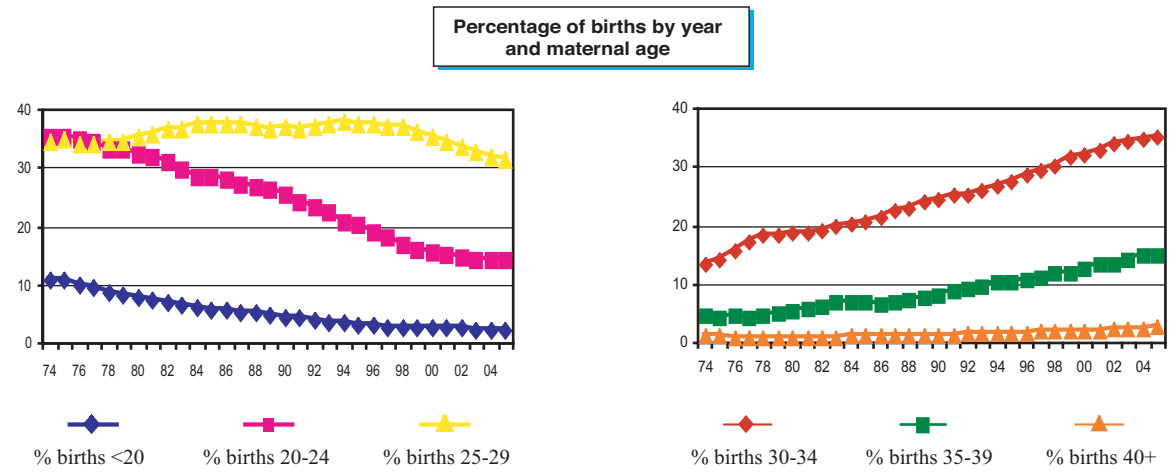
**Exposure information:**
Some basic information, such as maternal disease and since 1999, smoking and occupation, is collected on all infants, malformed or not.

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

**Addresses and Staff:**
Stein Emil Vollset, MD, Programme Director
Medical Birth Registry of Norway
Norwegian Institute of Public Health
Kalfarveien 31
N-5018 Bergen - Norway
**Phone:** 47- 53 20 4002
**Fax:** 47 - 53 20 4001
**E-mail:** stvo@fhi.no

## Norway




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 74 | 82.2 | Cystic kidney | 24 | 27.3 |
| Spina bifida | 41 | 41.0 | Limb reduction defects | 9 | 11.4 |
| Encephalocele | 11 | 52.4 | Diaphragmatic hernia | 11 | 25.0 |
| Holoprosencephaly | 13 | 59.1 | Omphalocele | 21 | 48.8 |
| Hydrocephaly | 42 | 37.5 | Gastroschisis | 5 | 10.0 |
| Hypoplastic left heart syndrome | 19 | 33.9 | Trisomy 13 | 7 | 41.2 |
| Cleft palate without cleft lip | 1 | 0.8 | Trisomy 18 | 34 | 54.0 |
| Cleft lip with or without cleft palate | 7 | 3.0 | Down syndrome | 102 | 29.9 |
| Renal agenesis | 9 | 69.2 | | | |

Total ToPs with births defects = 618 (Ratio ToPs/Births: 3.56 per 1,000)
*ToPs/ToPs+Births

# Norway: MBRN, 2005

| | |
|---|---|
| Live births (LB) | 57,209 |
| Stillbirths (SB) | 459 |
| Total births | 57,916 |
| Number of terminations of pregnancy (ToP) for birth defects | 248 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 3 | 23 | 4.49 |
| Spina bifida | 15 | 4 | 23 | 7.25 |
| Encephalocele | 0 | 0 | 4 | 0.69 |
| Microcephaly | 1 | 0 | 2 | 0.52 |
| Holoprosencephaly | 2 | 1 | 5 | 1.38 |
| Hydrocephaly | 18 | 5 | 16 | 6.73 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 2 | 0 | 0 | 0.35 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.17 |
| Microtia | 2 | 0 | 0 | 0.35 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 21 | 1 | 0 | 3.80 |
| Tetralogy of Fallot | 13 | 0 | 1 | 2.42 |
| Hypoplastic left heart syndrome | 8 | 0 | 6 | 2.42 |
| Coarctation of aorta | 19 | 0 | 1 | 3.45 |
| Choanal atresia, bilateral | 5 | 0 | 0 | 0.86 |
| Cleft palate without cleft lip | 48 | 1 | 0 | 8.46 |
| Cleft lip with or without cleft palate | 77 | 1 | 2 | 13.81 |
| Oesophageal atresia/stenosis with or without fistula | 13 | 0 | 0 | 2.24 |
| Small intestine atresia/stenosis | 6 | 0 | 0 | 1.04 |
| Anorectal atresia/stenosis | 16 | 0 | 1 | 2.94 |
| Undescended testis (36 weeks of gestation or later) | 151 | 0 | 0 | 26.07 |
| Hypospadias | 85 | 0 | 0 | 14.68 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 0 | 2 | 0 | 0.35 |
| Renal agenesis | 1 | 1 | 4 | 1.04 |
| Cystic kidney | 20 | 0 | 10 | 5.18 |
| Bladder exstrophy | 2 | 0 | 0 | 0.35 |
| Polydactyly, preaxial | 64 | 1 | 3 | 11.74 |
| Total Limb reduction defects (include unspecified) | 23 | 0 | 4 | 4.66 |
| Transverse | 17 | 0 | 2 | 3.28 |
| Preaxial | 1 | 0 | 0 | 0.17 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 2 | 0 | 0 | 0.35 |
| Mixed | 4 | 0 | 3 | 1.21 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 8 | 1 | 3 | 2.07 |
| Omphalocele | 4 | 0 | 13 | 2.94 |
| Gastroschisis | 18 | 0 | 4 | 3.80 |
| Unspecified Omphalocele/Gastroschisis | 1 | 0 | 3 | 0.70 |
| Prune belly sequence | 2 | 0 | 5 | 1.21 |
| Trisomy 13 | 3 | 1 | 5 | 1.55 |
| Trisomy 18 | 8 | 4 | 18 | 5.18 |
| Down syndrome, all ages (include age unknown) | 60 | 3 | 44 | 18.48 |
| <20 | 1 | 0 | 0 | 8.35 |
| 20-24 | 3 | 0 | 1 | 4.93 |
| 25-29 | 9 | 1 | 4 | 7.69 |
| 30-34 | 20 | 1 | 6 | 13.37 |
| 35-39 | 21 | 0 | 14 | 40.32 |
| 40-44 | 6 | 1 | 19 | 178.20 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

# Norway: MBRN, Previous years rates 1974 - 2005

Birth prevalence rates: (LB+SB) * 10,000 until 1993
Birth prevalence rates: (LB+SB+TOP) * 10,000 since 1994

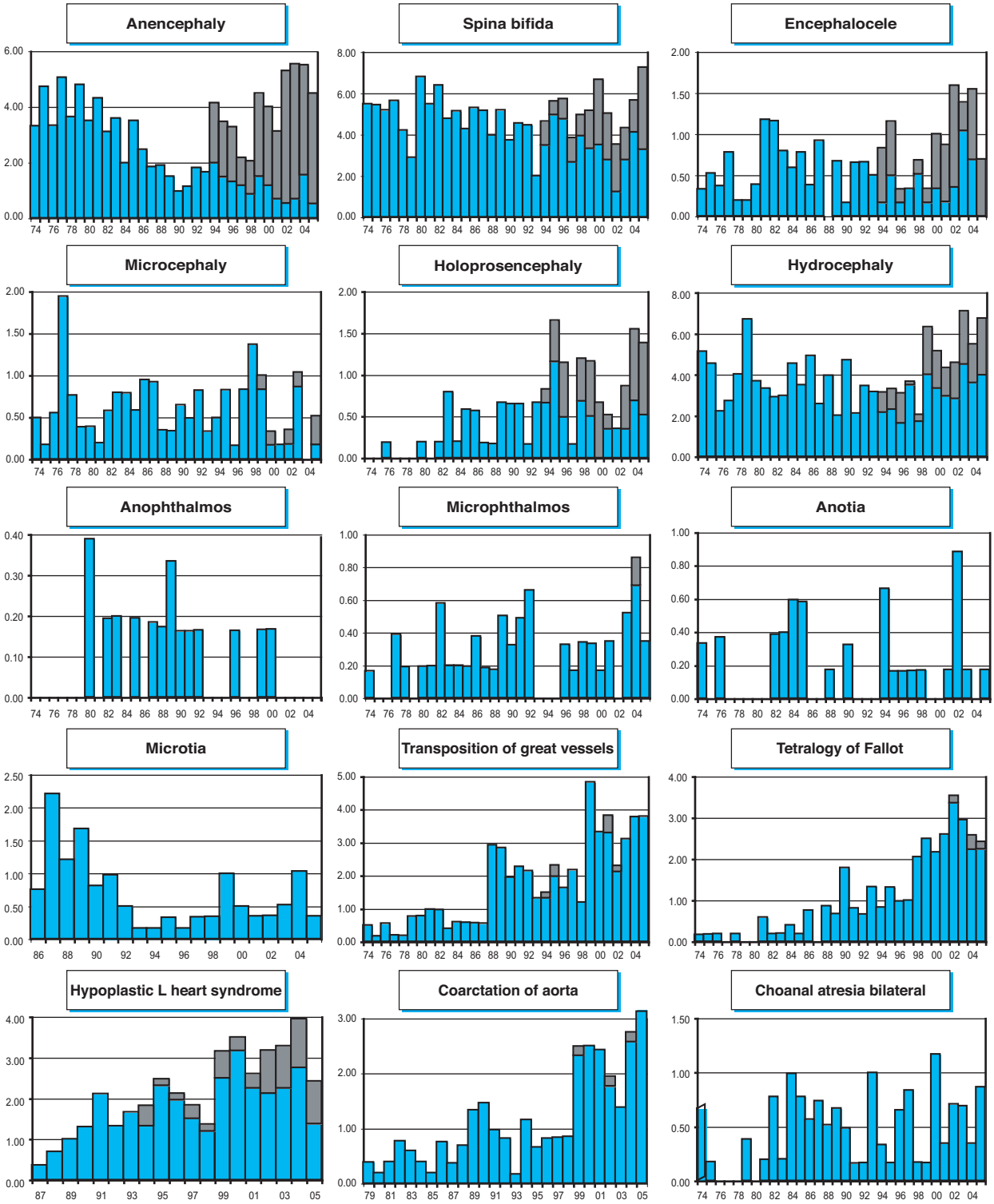
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Total births** | **378,981** | **255,083** | **286,635** | **303,414** | **299,859** | **288,165** |
| Anencephaly | 4.04 | 3.29 | 1.71 | 2.44 | 3.20 | 4.79 |
| Spina bifida | 5.09 | 5.21 | 4.64 | 4.25 | 5.27 | 5.17 |
| Encephalocele | 0.40 | 0.90 | 0.42 | 0.76 | 0.53 | 1.21 |
| Microcephaly | 0.66 | 0.59 | 0.63 | 0.59 | 0.73 | 0.42 |
| Holoprosencephaly | 0.05 | 0.35 | 0.45 | 0.79 | 0.87 | 0.94 |
| Hydrocephaly | 4.17 | 3.45 | 3.63 | 3.03 | 4.07 | 5.66 |
| Anophthalmos | 0.05 | 0.12 | 0.17 | 0.07 | 0.10 | 0.00 |
| Microphthalmos | 0.13 | 0.27 | 0.31 | 0.23 | 0.27 | 0.42 |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.11 | 0.39 | 0.10 | 0.16 | 0.10 | 0.28 |
| Microtia | nr | nr | 1.33 | 0.43 | 0.47 | 0.52 |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 0.45 | 0.71 | 1.81 | 1.91 | 2.63 | 3.37 |
| Tetralogy of Fallot | 0.11 | 0.31 | 0.84 | 0.99 | 1.73 | 2.81 |
| Hypoplastic left heart syndrome | nr | nr | 0.86* | 1.88 | 2.40 | 3.09 |
| Coarctation of aorta | 0.29* | 0.47 | 0.94 | 0.76 | 1.50 | 2.39 |
| Choanal atresia, bilateral | 0.18 | 0.59 | 0.59 | 0.36 | 0.60 | 0.59 |
| Cleft palate without cleft lip | 4.75 | 5.21 | 5.62 | 5.17 | 6.07 | 6.59 |
| Cleft lip with or without cleft palate | 14.41 | 13.56 | 14.13 | 13.32 | 13.21 | 12.94 |
| Oesophageal atresia/stenosis with or without fistula | 1.98 | 1.80 | 2.37 | 2.04 | 2.27 | 2.60 |
| Small intestine atresia/stenosis | 0.95 | 1.02 | 1.26 | 1.55 | 1.40 | 0.90 |
| Anorectal atresia/stenosis | 1.61 | 2.20 | 2.13 | 2.31 | 2.47 | 2.67 |
| Undescended testis (36 weeks of gestation or later) | 17.81 | 14.07 | 16.29 | 17.20 | 18.14 | 28.25 |
| Hypospadias | 13.09 | 14.00 | 16.33 | 15.39 | 14.57 | 16.41 |
| Epispadias | 0.26 | 0.47 | 0.38 | 0.16 | 0.37 | 0.14 |
| Indeterminate sex | 2.51 | 4.08 | 3.98 | 7.05 | 4.84 | 0.52 |
| Renal agenesis | 0.13 | 1.06 | 1.29 | 1.52 | 1.50 | 0.87 |
| Cystic kidney | 0.47 | 1.18 | 1.60 | 2.47 | 3.67 | 5.31 |
| Bladder exstrophy | 0.24 | 0.55 | 0.24 | 0.26 | 0.37 | 0.28 |
| Polydactyly, preaxial | nr | nr | nr | nr | 8.07* | 8.99 |
| Total Limb reduction defects (include unspecified) | 8.39 | 6.82 | 7.19 | 6.86 | 5.77 | 4.20 |
| Transverse | nr | nr | 2.97* | 4.05 | 2.27 | 2.39 |
| Preaxial | nr | nr | 0.82* | 0.56 | 0.50 | 0.45 |
| Postaxial | nr | nr | 0.82* | 0.49 | 0.33 | 0.07 |
| Intercalary | nr | nr | 0.25* | 0.36 | 0.43 | 0.10 |
| Mixed | nr | nr | 0.41* | 0.66 | 1.40 | 1.39 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 2.01 | 2.55 | 2.34 | 2.47 | 2.73 | 2.57 |
| Omphalocele | 2.32 | 1.88 | 2.13 | 1.98 | 2.30 | 2.43 |
| Gastroschisis | 1.32 | 1.49 | 1.92 | 2.24 | 2.90 | 2.85 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | nr | nr | nr | nr | 1.33* | 1.35 |
| Trisomy 13 | nr | nr | nr | nr | 1.08* | 0.76 |
| Trisomy 18 | nr | nr | nr | nr | 2.41* | 2.91 |
| Down syndrome, all ages (include age unknown) | 9.79 | 10.55 | 10.92 | 10.28 | 13.44 | 18.05 |
| <20 | 2.25 | 5.45 | 3.47 | 2.79 | 2.46 | 7.65 |
| 20-24 | 6.27 | 7.11 | 6.58 | 5.64 | 2.74 | 6.27 |
| 25-29 | 7.80 | 6.83 | 6.01 | 7.18 | 6.84 | 7.42 |
| 30-34 | 10.78 | 14.15 | 13.36 | 11.15 | 11.23 | 13.92 |
| 35-39 | 37.22 | 33.97 | 37.42 | 20.98 | 38.93 | 43.66 |
| 40-44 | 127.11 | 63.72 | 80.22 | 84.51 | 130.35 | 152.74 |
| 45+ | 197.04 | 93.46 | 267.86 | 342.47 | 250.00 | 237.15 |
| unknown | --- | --- | --- | --- | --- | --- |

* data include less than 5 and 7 years

nr = not reported

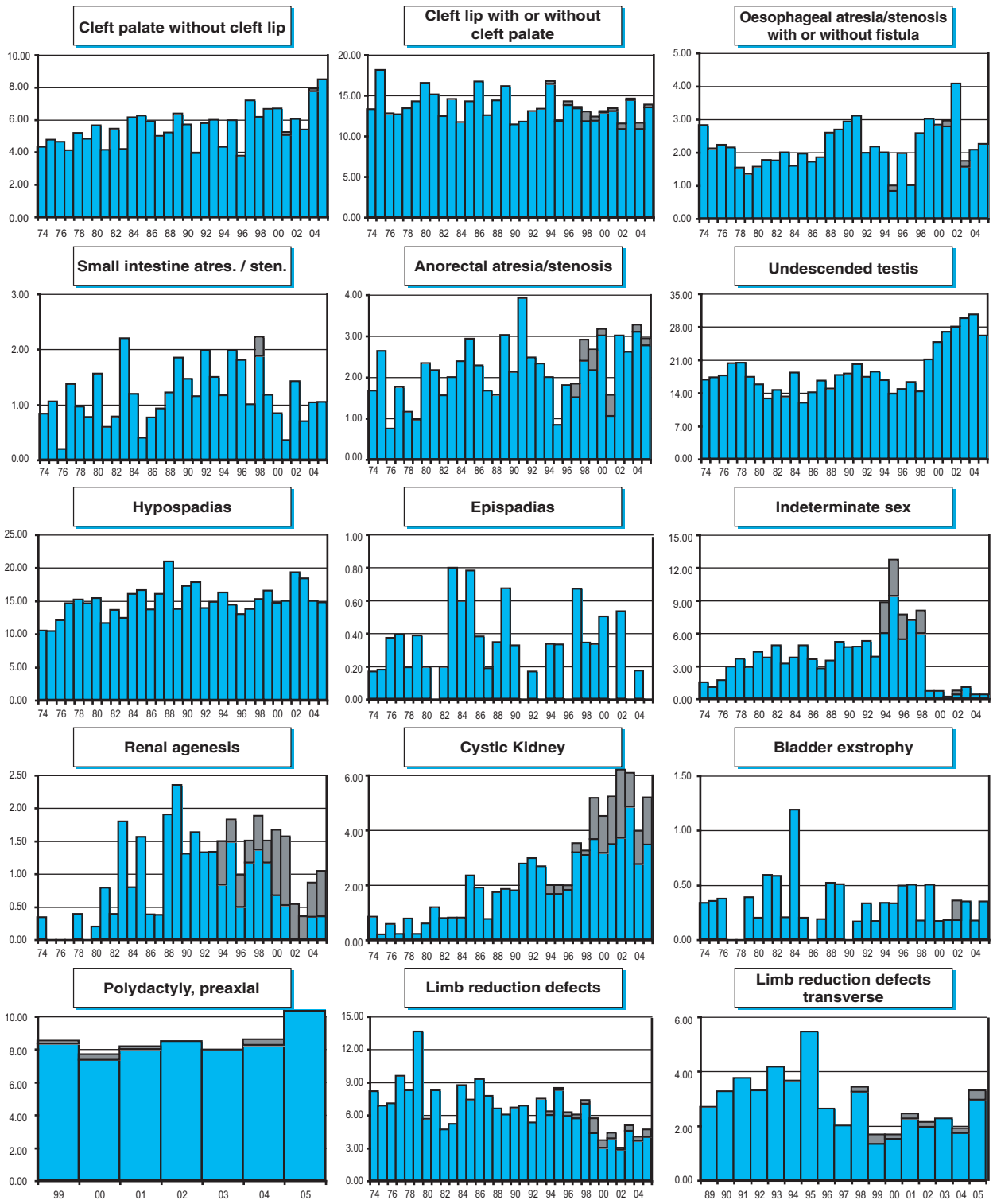
## Norway

'Time trends 1974-2005 (Birth prevalence rates per 10,000)




**Note:** L+S rates, ToP rates

## Norway




**Note:** L+S rates, ToP rates

## Norway


**Note:** L+S rates, ToP rates

## Russia: MRRCM

Moscow Regional Registry of Congenital malformation (MRRCM)

**History:**
Moscow Regional Registry of Congenital malformation started the activity in 1999 and legally defined by the order of the Ministry of Health Care of Russian Federation. MRRCM became a Member of ICBDSR in 2001.

**Size and coverage:**
MRRCM is located as a section of Moscow Regional Medical genetic consultation by The Moscow Regional Research institute of Obstetrics and Gynecology (MONIIAG). Director of the MONIIAG is Professor Vladislav Krasnopolsky. The Head of the Moscow Regional Medical genetic consultation and Director of the Programme of MRRCM is Ludmila Jouthenko. The Programme of Monitoring of Birth defects covers all births in Moscow Region. In 1999 MRRCM observed 45 000 birth. There are about 64 000 births today (2007). The information about babies and fetuses with Birth defects is collected from 53 maternity hospitals among all women consultations and clinics, children clinics. Prenatal diagnosed and terminated fetuses are registered too.

**Legislation and funding:**
Monitoring of the birth of fetuses and babies with congenital malformations is legally defined by the Order of the Ministry of Health Care of Russian Federation in 1999.

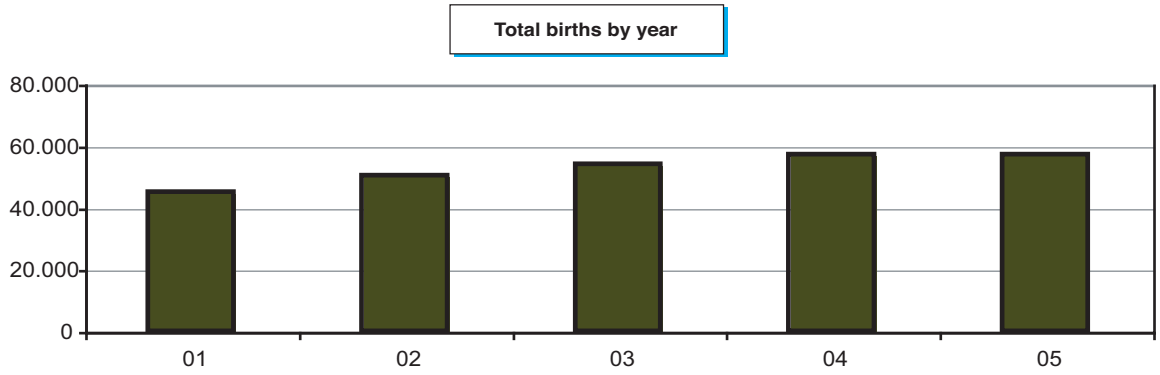
**Sources of ascertainment:**
Reporting is made by neonatologist during the first week of the infant's life in maternity hospitals and by pediatricians during the first year – in pediatric clinics and departments. Reports are collected from cytogenetic laboratories, pathology departments.

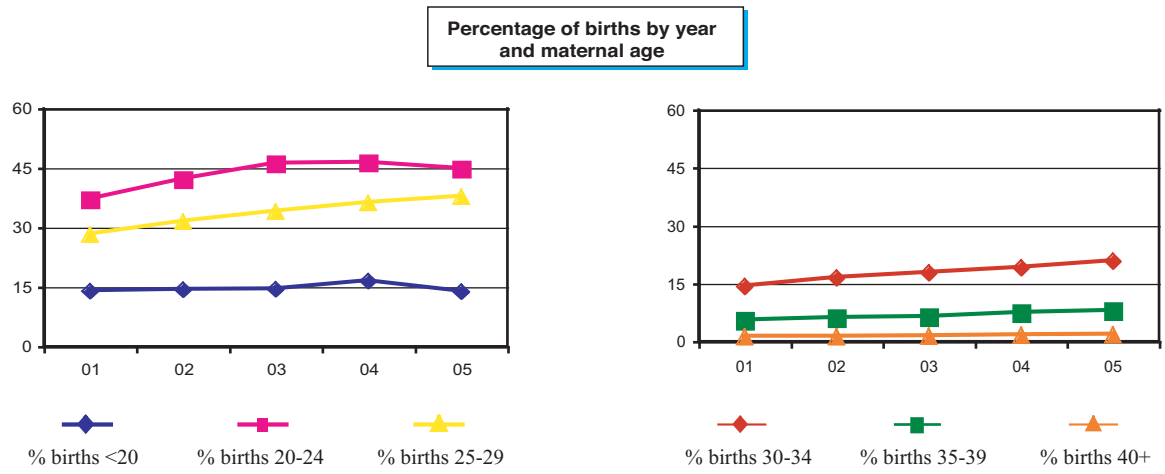
**Exposure information:**
No exposure information is routinely collected in the registry.

**Background information:**
Background information on all births is available from statistics department.

**Addresses and Staff:**
Ludmila Joutchenko, MD, Programme Director
Moscow Regional Research Scientific Institute of Obstetrics and Gynecology
22a, Pokrovka St.
101000 Moscow Russia
**Phone:** 007-4956215398
**Fax:** 007-4956215398
**E-mail:** mrrcm@mail.ru

## Russia: MRRCM




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 46 | 95.8 | Cystic kidney | 12 | 27.3 |
| Spina bifida | 13 | 23.2 | Limb reduction defects | 2 | 5.6 |
| Encephalocele | 5 | 55.6 | Diaphragmatic hernia | 2 | 6.3 |
| Holoprosencephaly | 4 | 100.0 | Omphalocele | 11 | 37.9 |
| Hydrocephaly | 24 | 32.0 | Gastroschisis | 10 | 22.7 |
| Hypoplastic left heart syndrome | 7 | 53.8 | Trisomy 13 | 3 | 100.0 |
| Cleft palate without cleft lip | 0 | 0.0 | Trisomy 18 | 3 | 75.0 |
| Cleft lip with or without cleft palate | 6 | 5.6 | Down syndrome | 16 | 7.4 |
| Renal agenesis | 13 | 76.5 | | | |

Total ToPs with births defects = 440 (Ratio ToPs/Births: 2.61 per 1,000)
*ToPs/ToPs+Births

# Russia: Moscow Region, 2005

| | |
|---|---|
| Live births (LB) | 56,896 |
| Stillbirths (SB) | 252 |
| Total births | 57,148 |
| Number of terminations of pregnancy (ToP) for birth defects | 151 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 0 | 1 | 18 | 3.32 |
| Spina bifida | 6 | 1 | 5 | 2.10 |
| Encephalocele | 1 | 1 | 4 | 1.05 |
| Microcephaly | 2 | 0 | 0 | 0.35 |
| Holoprosencephaly | 0 | 0 | 3 | 0.52 |
| Hydrocephaly | 15 | 0 | 9 | 4.20 |
| Anophthalmos | 1 | 0 | 0 | 0.17 |
| Microphthalmos | 1 | 0 | 0 | 0.17 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 1 | 0 | 0 | 0.17 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 7 | 0 | 2 | 1.57 |
| Tetralogy of Fallot | 7 | 0 | 0 | 1.22 |
| Hypoplastic left heart syndrome | 2 | 0 | 3 | 0.87 |
| Coarctation of aorta | 2 | 0 | 0 | 0.35 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.35 |
| Cleft palate without cleft lip | 26 | 0 | 0 | 4.55 |
| Cleft lip with or without cleft palate | 24 | 0 | 4 | 4.90 |
| Oesophageal atresia/stenosis with or without fistula | 9 | 1 | 0 | 1.75 |
| Small intestine atresia/stenosis | 4 | 0 | 3 | 1.22 |
| Anorectal atresia/stenosis | 14 | 0 | 0 | 2.45 |
| Undescended testis (36 weeks of gestation or later) | 77 | 0 | 0 | 13.47 |
| Hypospadias | 83 | 0 | 0 | 14.52 |
| Epispadias | 0 | 0 | 0 | 0.00 |
| Indeterminate sex | 3 | 0 | 0 | 0.52 |
| Renal agenesis | 1 | 0 | 4 | 0.87 |
| Cystic kidney | 13 | 0 | 2 | 2.62 |
| Bladder exstrophy | 1 | 0 | 0 | 0.17 |
| Polydactyly, preaxial | 17 | 0 | 0 | 2.97 |
| Total Limb reduction defects (include unspecified) | 5 | 1 | 1 | 1.22 |
| Transverse | 3 | 0 | 1 | 0.70 |
| Preaxial | 2 | 1 | 0 | 0.52 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 0 | 0 | 0 | 0.00 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 9 | 0 | 0 | 1.57 |
| Omphalocele | 6 | 0 | 4 | 1.75 |
| Gastroschisis | 11 | 1 | 2 | 2.45 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 0 | 0 | 1 | 0.17 |
| Trisomy 18 | 0 | 0 | 2 | 0.35 |
| Down syndrome, all ages (include age unknown) | 78 | 0 | 6 | 14.70 |
| <20 | 2 | 0 | 0 | 3.22 |
| 20-24 | 14 | 0 | 1 | 7.44 |
| 25-29 | 14 | 0 | 1 | 8.80 |
| 30-34 | 12 | 0 | 2 | 14.93 |
| 35-39 | 23 | 0 | 2 | 69.81 |
| 40-44 | 12 | 0 | 0 | 167.36 |
| 45+ | 1 | 0 | 0 | 243.90 |
| unknown | 0 | 0 | 0 | 0.00 |

## Slovak Republic

Congenital Malformations Monitoring Program of the Slovak Republic

**History:**
In Slovakia the collection of reports from delivery units and processing of data is performed by the National Health Information Center SR, formerly the Institution of Health Information and Statistics (IHIS). The obligation of reporting all groups of congenital malformations results from valid legislation norms. Reporting of congenital malformations began in 1964.
The programme of Slovak Teratological Information Center (STIC) was established in 2003 and consists it cooperates with Slovak Medical University, National Health Information Center, and the Center of Medical Genetics. Research collaboration began in 1995 under the responsibility of Elena Szabova.

**Size and coverage:**
The registry covers all births in the area approximately 55.000 births annually according to the announcements of birth defects from delivery units. Detailed information about cases of CM are collected at the Center of Medical Genetics, Bratislava from western regions of Slovakia (cca 15.000 births ) by Eva Veghova, or under the running research projects at the Slovak Medical University.

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

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

**Exposure Information:**
Detail information on maternal and paternal occupation, drug use, etc. are collected by interviews of case´s and control´s mothers only according to research projects.

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

**Addresses and Staff:**
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Limbova 12
833 03 Bratislava, Slovak Republic
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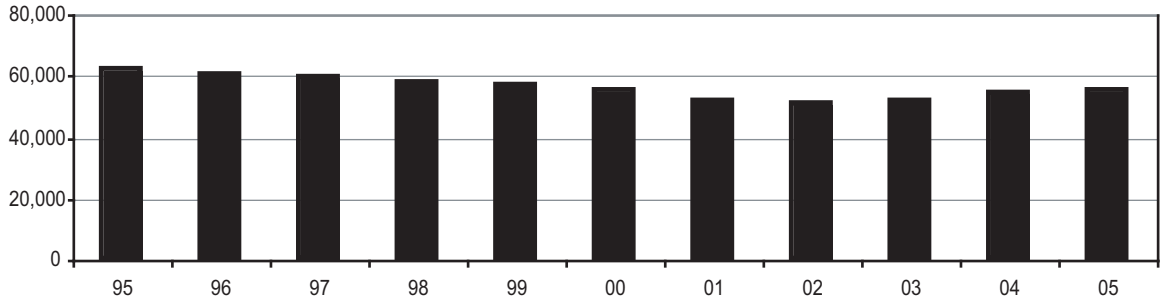
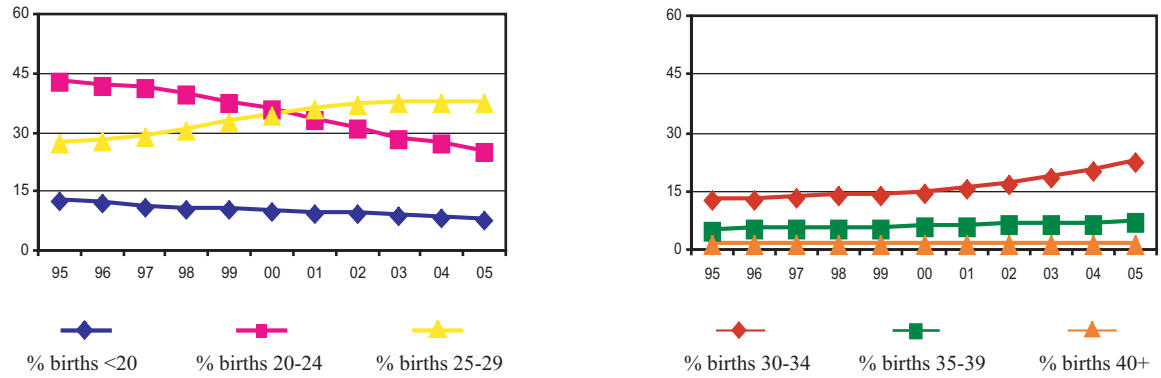
## Slovak Republic

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 4 | 50.0 | Cystic kidney | 0 | 0.0 |
| Spina bifida | 2 | 4.5 | Limb reduction defects | 0 | 0.0 |
| Encephalocele | 7 | 35.0 | Diaphragmatic hernia | 0 | 0.0 |
| Holoprosencephaly | 3 | 30.0 | Omphalocele | 0 | 0.0 |
| Hydrocephaly | 16 | 23.5 | Gastroschisis | 1 | 6.7 |
| Hypoplastic left heart syndrome | 0 | 0.0 | Trisomy 13 | 0 | 0.0 |
| Cleft palate without cleft lip | 3 | 3.7 | Trisomy 18 | 1 | 8.3 |
| Cleft lip with or without cleft palate | 0 | 0.0 | Down syndrome | 16 | 9.2 |
| Renal agenesis | 1 | 1.0 | | | |

Total ToPs with births defects = 107 (Ratio ToPs/Births: 0.67 per 1,000)
*ToPs/ToPs+Births

# Slovak Republic: 2005

| | |
|---|---|
| Live births (LB) | 54,430 |
| Stillbirths (SB) | 195 |
| Total births | 54,625 |
| Number of terminations of pregnancy (ToP) for birth defects | 33 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 2 | 0 | 3 | 0.92 |
| Spina bifida | 13 | 1 | 0 | 2.56 |
| Encephalocele | 3 | 0 | 3 | 1.10 |
| Microcephaly | 3 | 0 | 0 | 0.55 |
| Holoprosencephaly | 2 | 0 | 1 | 0.55 |
| Hydrocephaly | 15 | 0 | 6 | 3.84 |
| Anophthalmos | 0 | 0 | 0 | 0.00 |
| Microphthalmos | 1 | 0 | 0 | 0.18 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 0 | 0 | 0 | 0.00 |
| Microtia | 2 | 0 | 0 | 0.37 |
| Unspecified Anotia/Microtia | 3 | 0 | 0 | 0.55 |
| Transposition of great vessels | 7 | 0 | 0 | 1.28 |
| Tetralogy of Fallot | 5 | 0 | 0 | 0.92 |
| Hypoplastic left heart syndrome | 14 | 0 | 0 | 2.56 |
| Coarctation of aorta | 6 | 0 | 0 | 1.10 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.37 |
| Cleft palate without cleft lip | 29 | 0 | 0 | 5.31 |
| Cleft lip with or without cleft palate | 51 | 0 | 0 | 9.34 |
| Oesophageal atresia/stenosis with or without fistula | 8 | 0 | 0 | 1.46 |
| Small intestine atresia/stenosis | 11 | 1 | 0 | 2.20 |
| Anorectal atresia/stenosis | 14 | 0 | 0 | 2.56 |
| Undescended testis (36 weeks of gestation or later) | 41 | 1 | 0 | 7.69 |
| Hypospadias | 114 | 0 | 0 | 20.87 |
| Epispadias | 1 | 0 | 0 | 0.18 |
| Indeterminate sex | 1 | 0 | 0 | 0.18 |
| Renal agenesis | 36 | 0 | 0 | 6.59 |
| Cystic kidney | 5 | 0 | 0 | 0.92 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 15 | 0 | 0 | 2.75 |
| Total Limb reduction defects (include unspecified) | 28 | 0 | 0 | 5.13 |
| 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 | 1 | 0 | 1.46 |
| Omphalocele | 4 | 0 | 0 | 0.73 |
| Gastroschisis | 2 | 0 | 0 | 0.37 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 1 | 0 | 0 | 0.18 |
| Trisomy 13 | 2 | 0 | 0 | 0.37 |
| Trisomy 18 | 5 | 0 | 0 | 0.92 |
| Down syndrome, all ages (include age unknown) | 48 | 1 | 4 | 9.70 |
| <20 | 0 | 0 | 0 | 0.00 |
| 20-24 | 5 | 0 | 1 | 4.46 |
| 25-29 | 11 | 1 | 0 | 5.88 |
| 30-34 | 14 | 0 | 1 | 12.31 |
| 35-39 | 12 | 0 | 2 | 37.82 |
| 40-44 | 6 | 0 | 0 | 85.71 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# Slovak Republic: Previous years rates 1995 - 2005

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

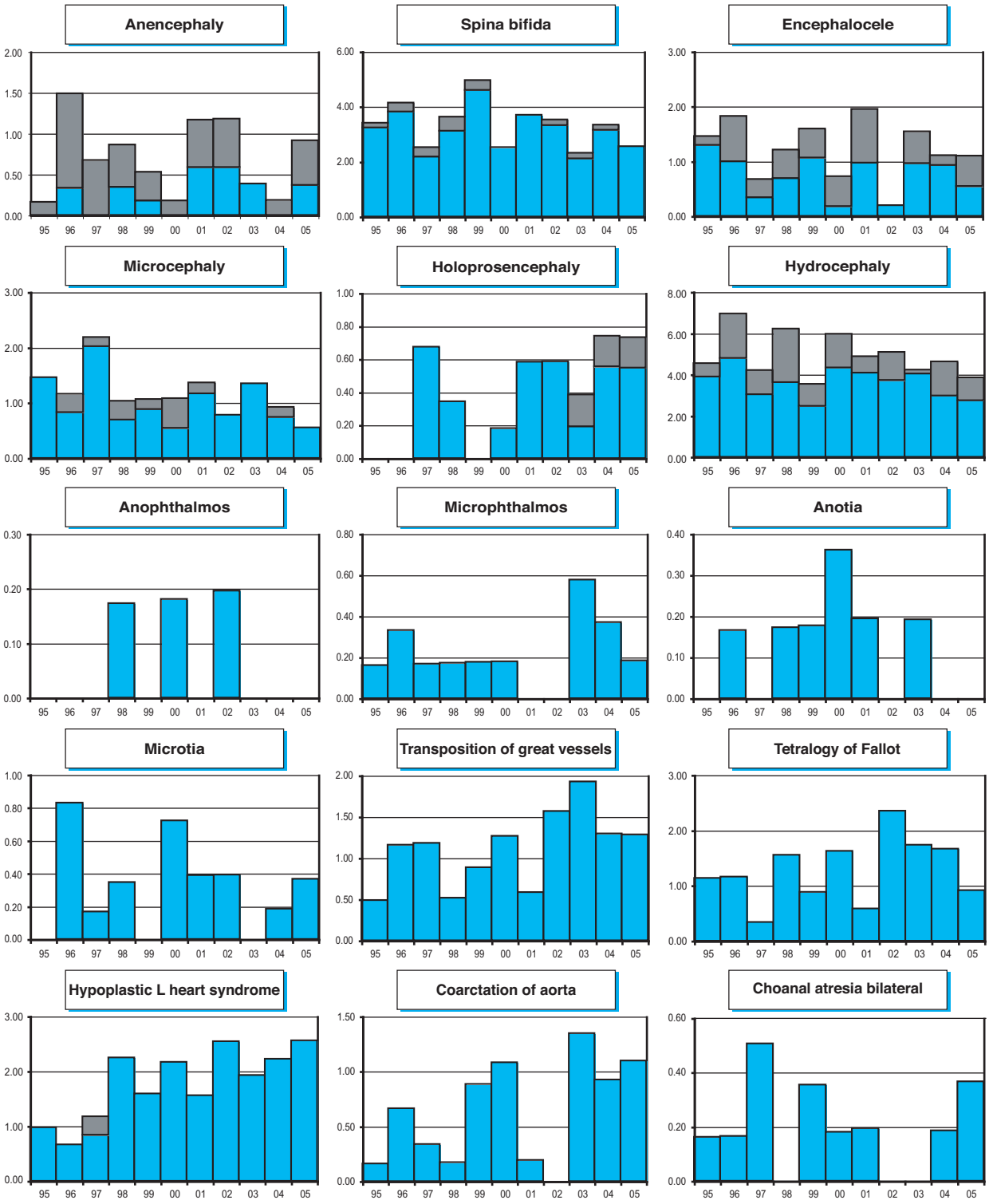
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995* | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | **61,668** | **289,430** | **262,891** |
| Anencephaly | | | | 0.16 | 0.76 | 0.76 |
| Spina bifida | | | | 3.41 | 3.56 | 3.08 |
| Encephalocele | | | | 1.46 | 1.21 | 1.18 |
| Microcephaly | | | | 1.46 | 1.31 | 0.99 |
| Holoprosencephaly | | | | 0.00 | 0.24 | 0.61 |
| Hydrocephaly | | | | 4.54 | 5.39 | 4.53 |
| Anophthalmos | | | | 0.00 | 0.07 | 0.04 |
| Microphthalmos | | | | 0.16 | 0.21 | 0.23 |
| Unspecified Anophthalmos/Microphthalmos | | | | --- | --- | --- |
| Anotia | | | | 0.00 | 0.17 | 0.08 |
| Microtia | | | | 0.00 | 0.41 | 0.27 |
| Unspecified Anotia/Microtia | | | | --- | --- | --- |
| Transposition of great vessels | | | | 0.49 | 1.00 | 1.33 |
| Tetralogy of Fallot | | | | 1.14 | 1.11 | 1.45 |
| Hypoplastic left heart syndrome | | | | 0.97 | 1.55 | 2.17 |
| Coarctation of aorta | | | | 0.16 | 0.62 | 0.72 |
| Choanal atresia, bilateral | | | | 0.16 | 0.24 | 0.15 |
| Cleft palate without cleft lip | | | | 6.81 | 5.08 | 5.59 |
| Cleft lip with or without cleft palate | | | | 8.11 | 10.71 | 10.00 |
| Oesophageal atresia/stenosis with or without fistula | | | | 0.81 | 1.11 | 1.56 |
| Small intestine atresia/stenosis | | | | 1.62 | 1.38 | 2.02 |
| Anorectal atresia/stenosis | | | | 0.65 | 1.76 | 3.16 |
| Undescended testis (36 weeks of gestation or later) | | | | 3.73 | 7.19 | 8.06 |
| Hypospadias | | | | 25.30 | 22.56 | 23.43 |
| Epispadias | | | | 0.16 | 0.17 | 0.15 |
| Indeterminate sex | | | | 0.00 | 0.59 | 0.30 |
| Renal agenesis | | | | 1.14 | 3.18 | 5.90 |
| Cystic kidney | | | | 0.16 | 1.04 | 1.60 |
| Bladder exstrophy | | | | 0.00 | 0.21 | 0.15 |
| Polydactyly, preaxial | | | | 1.30 | 1.80 | 3.65 |
| Total Limb reduction defects (include unspecified) | | | | 3.41 | 3.63 | 3.88 |
| Transverse | | | | nr | nr | nr |
| Preaxial | | | | nr | nr | nr |
| Postaxial | | | | nr | nr | nr |
| Intercalary | | | | nr | nr | nr |
| Mixed | | | | nr | nr | nr |
| Unspecified | | | | --- | --- | --- |
| Diaphragmatic hernia | | | | 0.81 | 1.38 | 1.52 |
| Omphalocele | | | | 0.49 | 0.52 | 0.80 |
| Gastroschisis | | | | 0.32 | 1.07 | 0.91 |
| Unspecified Omphalocele/Gastroschisis | | | | 0.00 | 0.00 | 0.00 |
| Prune belly sequence | | | | 0.00 | 0.03 | 0.23 |
| Trisomy 13 | | | | 0.16 | 0.24 | 0.42 |
| Trisomy 18 | | | | 0.16 | 0.31 | 0.57 |
| Down syndrome, all ages (include age unknown) | | | | 7.95 | 9.57 | 10.46 |
| <20 | | | | 7.91 | 6.53 | 3.63 |
| 20-24 | | | | 6.09 | 6.44 | 3.85 |
| 25-29 | | | | 3.03 | 6.81 | 7.02 |
| 30-34 | | | | 16.68 | 11.70 | 11.83 |
| 35-39 | | | | 27.93 | 34.02 | 45.98 |
| 40-44 | | | | 16.69 | 81.58 | 106.02 |
| 45+ | | | | 0.00 | 243.90 | 230.77 |
| unspecified | | | | --- | --- | --- |

* data include less than 5 years

nr = not reported

## Slovak Republic

'Time trends 1995-2005 (Birth prevalence rates per 10,000)

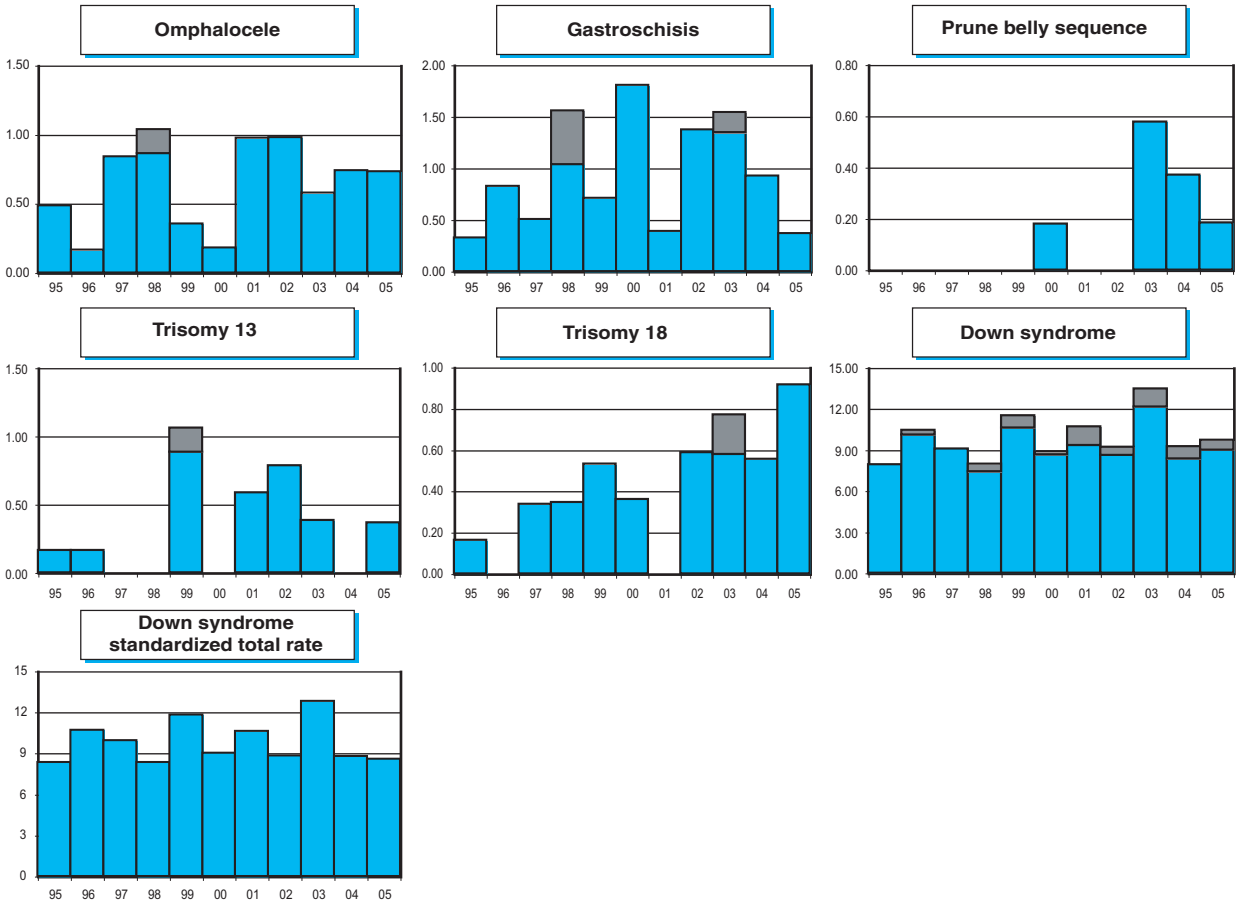



**Note:** L+S rates, ToP rates

## Slovak Republic


**Note:** L+S rates, ToP rates

## Slovak Republic




**Note:** L+S rates, ToP rates

## South America: ECLAMC

Latin American Collaborative Study of Congenital Malformations (ECLAMC)

**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.

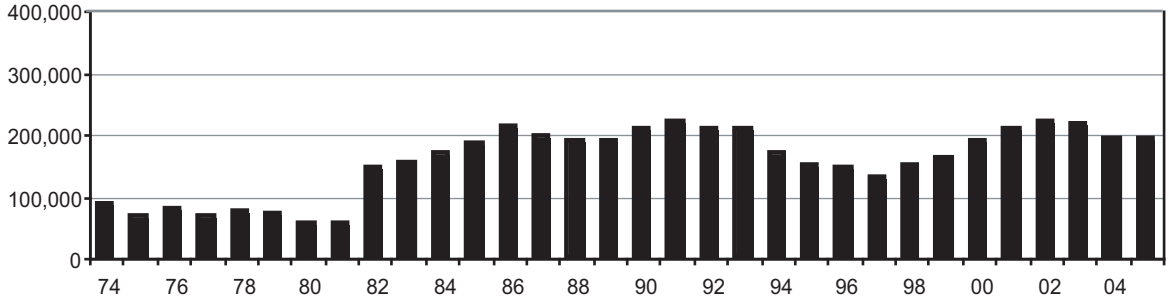
**Exposure information:**
The mother of each reported infant and the mother of a control infant - the next non-malformed infant born at that hospital with the same sex as the proband - are interviewed on various exposures, including drug usage and parental occupation.

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

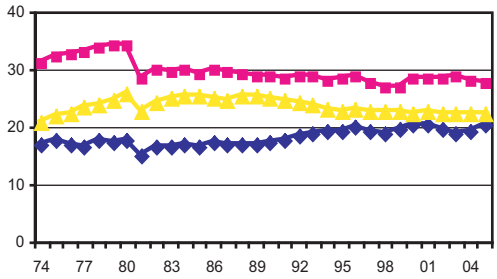
**Addresses and Staff:**
Eduardo Castilla, MD, Programme Director
ECLAMC/Dept.Genetica/FIOCRUZ
C.P. 926
20010-970 Rio de Janeiro, Brazil
**Phone:** 55-21-25528952
**Fax:** 55-21-22604282(5521)
**E-mail:** castilla@centroin.com.br

## South America: ECLAMC

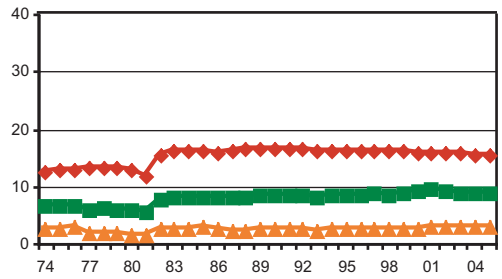
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# South America: 2005

| | |
|---|---|
| Live births (LB) | 190,454 |
| Stillbirths (SB) | 2,428 |
| Total births | 192,882 |
| 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 | 40 | 58 | | 5.08 |
| Spina bifida | 163 | 5 | | 8.71 |
| Encephalocele | 42 | 9 | | 2.64 |
| Microcephaly | 71 | 5 | | 3.94 |
| Holoprosencephaly | 28 | 2 | | 1.56 |
| Hydrocephaly | 256 | 29 | | 14.78 |
| Anophthalmos | 46 | 6 | | 2.70 |
| Microphthalmos | 25 | 0 | | 1.30 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | | 0.00 |
| Anotia | 8 | 0 | | 0.41 |
| Microtia | 129 | 5 | | 6.95 |
| Unspecified Anotia/Microtia | 2 | 0 | | 0.10 |
| Transposition of great vessels | 12 | 0 | | 0.62 |
| Tetralogy of Fallot | 23 | 1 | | 1.24 |
| Hypoplastic left heart syndrome | 20 | 0 | | 1.04 |
| Coarctation of aorta | 6 | 0 | | 0.31 |
| Choanal atresia, bilateral | 3 | 1 | | 0.21 |
| Cleft palate without cleft lip | 101 | 12 | | 5.86 |
| Cleft lip with or without cleft palate | 256 | 24 | | 14.52 |
| Oesophageal atresia/stenosis with or without fistula | 64 | 1 | | 3.37 |
| Small intestine atresia/stenosis | 76 | 1 | | 3.99 |
| Anorectal atresia/stenosis | 102 | 10 | | 5.81 |
| Undescended testis (36 weeks of gestation or later) | 136 | 0 | | 7.05 |
| Hypospadias | 78 | 2 | | 4.15 |
| Epispadias | 1 | 0 | | 0.05 |
| Indeterminate sex | 44 | 4 | | 2.49 |
| Renal agenesis | 45 | 6 | | 2.64 |
| Cystic kidney | 56 | 7 | | 3.27 |
| Bladder exstrophy | 3 | 1 | | 0.21 |
| Polydactyly, preaxial | 82 | 0 | | 4.25 |
| Total Limb reduction defects (include unspecified) | 143 | 27 | | 8.81 |
| Transverse | 56 | 10 | | 3.42 |
| Preaxial | 20 | 6 | | 1.35 |
| Postaxial | 5 | 1 | | 0.31 |
| Intercalary | 12 | 3 | | 0.78 |
| Mixed | 49 | 7 | | 2.90 |
| Unspecified | 1 | 0 | | 0.05 |
| Diaphragmatic hernia | 66 | 4 | | 3.63 |
| Omphalocele | 73 | 14 | | 4.51 |
| Gastroschisis | 93 | 7 | | 5.18 |
| Unspecified Omphalocele/Gastroschisis | 9 | 1 | | 0.52 |
| Prune belly sequence | 12 | 2 | | 0.73 |
| Trisomy 13 | 8 | 5 | | 0.67 |
| Trisomy 18 | 23 | 10 | | 1.71 |
| Down syndrome, all ages (include age unknown) | 352 | 5 | | 18.51 |
| <20 | 36 | 1 | | 9.49 |
| 20-24 | 42 | 1 | | 8.08 |
| 25-29 | 33 | 0 | | 7.71 |
| 30-34 | 49 | 0 | | 16.51 |
| 35-39 | 84 | 2 | | 51.45 |
| 40-44 | 90 | 0 | | 177.03 |
| 45+ | 17 | 1 | | 501.39 |
| unknown | 1 | 0 | | 1.64 |

# South America: Previous years rates 1974 - 2005

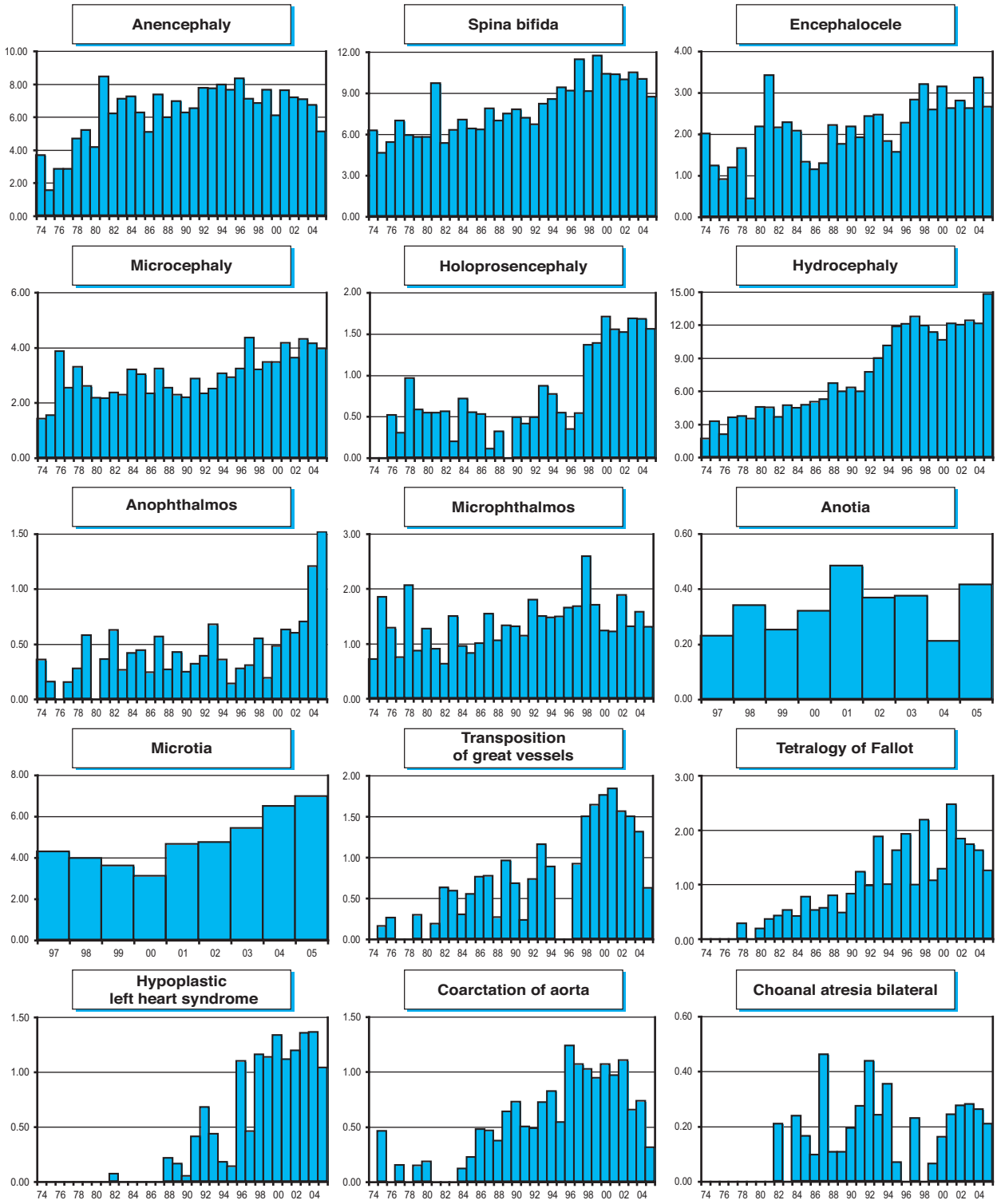
Prevalence rates: (LB+SB) * 10,000

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **493,467** | **706,137** | **992,563** | **953,726** | **771,185** | **1,023,502** |
| Anencephaly | 3.55 | 6.81 | 6.28 | 7.45 | 7.12 | 6.74 |
| Spina bifida | 5.82 | 6.56 | 7.25 | 7.86 | 10.35 | 9.91 |
| Encephalocele | 1.36 | 2.04 | 1.70 | 2.07 | 2.81 | 2.79 |
| Microcephaly | 2.49 | 2.69 | 2.50 | 2.71 | 3.51 | 4.03 |
| Holoprosencephaly | 0.41 | 0.51 | 0.29 | 0.61 | 1.12 | 1.59 |
| Hydrocephaly | 3.06 | 4.39 | 5.80 | 8.63 | 11.63 | 12.62 |
| Anophthalmos | 0.22 | 0.42 | 0.34 | 0.39 | 0.36 | 1.13 |
| Microphthalmos | 1.24 | 0.96 | 1.24 | 1.47 | 1.74 | 1.46 |
| Unspecified Anophthalmos/Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | nr | nr | nr | nr | 0.29* | 0.37 |
| Microtia | nr | nr | nr | nr | 3.66* | 5.60 |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 0.10 | 0.48 | 0.69 | 0.73 | 1.21 | 1.38 |
| Tetralogy of Fallot | 0.06 | 0.52 | 0.63 | 1.33 | 1.48 | 1.79 |
| Hypoplastic left heart syndrome | 0.00 | 0.01 | 0.08 | 0.39 | 1.06 | 1.21 |
| Coarctation of aorta | 0.12 | 0.08 | 0.53 | 0.61 | 1.06 | 0.76 |
| Choanal atresia, bilateral | 0.00 | 0.14 | 0.19 | 0.28 | 0.09 | 0.25 |
| Cleft palate without cleft lip | 3.14 | 3.48 | 3.33 | 3.92 | 3.89 | 5.26 |
| Cleft lip with or without cleft palate | 10.98 | 10.22 | 10.72 | 10.84 | 12.77 | 13.94 |
| Oesophageal atresia/stenosis with or without fistula | 1.97 | 2.51 | 2.66 | 2.95 | 3.50 | 3.70 |
| Small intestine atresia/stenosis | 0.57 | 1.67 | 1.42 | 1.82 | 2.41 | 3.20 |
| Anorectal atresia/stenosis | 2.76 | 3.84 | 3.84 | 4.48 | 5.15 | 5.69 |
| Undescended testis (36 weeks of gestation or later) | 1.54 | 3.94 | 4.53 | 4.83 | 5.55 | 6.86 |
| Hypospadias | 3.53 | 4.87 | 3.79 | 4.59 | 5.36 | 4.63 |
| Epispadias | 0.12 | 0.41 | 0.27 | 0.31 | 0.18 | 0.18 |
| Indeterminate sex | 1.07 | 2.24 | 1.89 | 1.80 | 1.88 | 2.38 |
| Renal agenesis | 0.43 | 0.69 | 1.02 | 1.67 | 2.40 | 2.51 |
| Cystic kidney | 0.57 | 1.10 | 1.74 | 2.09 | 3.99 | 4.13 |
| Bladder exstrophy | 0.12 | 0.28 | 0.28 | 0.22 | 0.39 | 0.32 |
| Polydactyly, preaxial | 2.76 | 2.46 | 2.47 | 2.75 | 2.98 | 4.13 |
| Total Limb reduction defects (include unspecified) | 4.17 | 5.57 | 4.76 | 5.55 | 6.44 | 7.18 |
| Transverse | 2.27 | 2.69 | 2.56 | 2.80 | 3.13 | 3.62 |
| Preaxial | 0.63 | 1.13 | 0.94 | 1.15 | 1.66 | 1.38 |
| Postaxial | 0.26 | 0.50 | 0.27 | 0.44 | 0.45 | 0.38 |
| Intercalary | 0.45 | 0.55 | 0.35 | 0.49 | 0.58 | 0.53 |
| Mixed | 0.45 | 0.62 | 0.50 | 0.50 | 0.52 | 1.09 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 0.81 | 1.26 | 1.83 | 2.40 | 3.64 | 3.78 |
| Omphalocele | 1.09 | 2.20 | 2.24 | 2.61 | 3.18 | 3.70 |
| Gastroschisis | 0.08 | 0.47 | 0.62 | 1.51 | 2.88 | 3.61 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.02 | 0.64 | 0.75 | 0.81 | 1.18 | 0.94 |
| Trisomy 13 | 0.18 | 0.59 | 0.36 | 0.57 | 0.80 | 0.96 |
| Trisomy 18 | 0.24 | 0.86 | 0.88 | 1.20 | 1.88 | 1.91 |
| Down syndrome, all ages (include age unknown) | 14.63 | 15.05 | 15.04 | 15.86 | 18.79 | 19.46 |
| <20 | 7.62 | 6.36 | 7.08 | 6.87 | 7.98 | 7.67 |
| 20-24 | 7.12 | 6.75 | 7.27 | 7.67 | 9.68 | 9.29 |
| 25-29 | 8.14 | 8.02 | 7.14 | 8.45 | 10.21 | 9.60 |
| 30-34 | 13.99 | 15.12 | 16.20 | 15.16 | 17.44 | 16.51 |
| 35-39 | 54.11 | 43.66 | 45.57 | 47.05 | 52.49 | 57.19 |
| 40-44 | 163.64 | 158.38 | 134.57 | 156.76 | 181.76 | 176.99 |
| 45+ | 295.12 | 248.45 | 278.51 | 281.23 | 315.96 | 422.94 |
| unspecified | --- | --- | --- | --- | --- | --- |

* data include less than 5 years

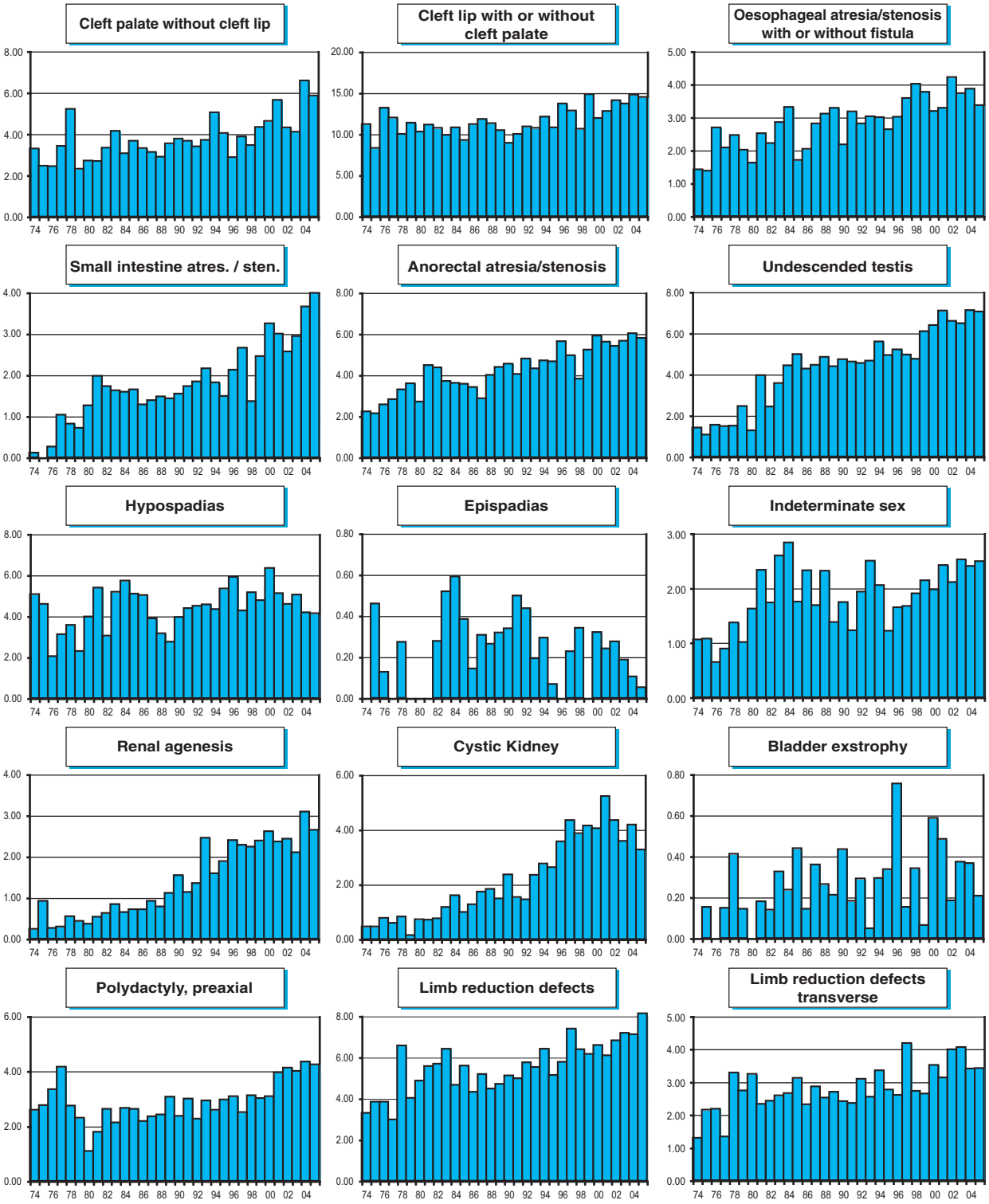
## South America: ECLAMC

'Time trends 1974-2005 (Birth prevalence rates per 10,000)



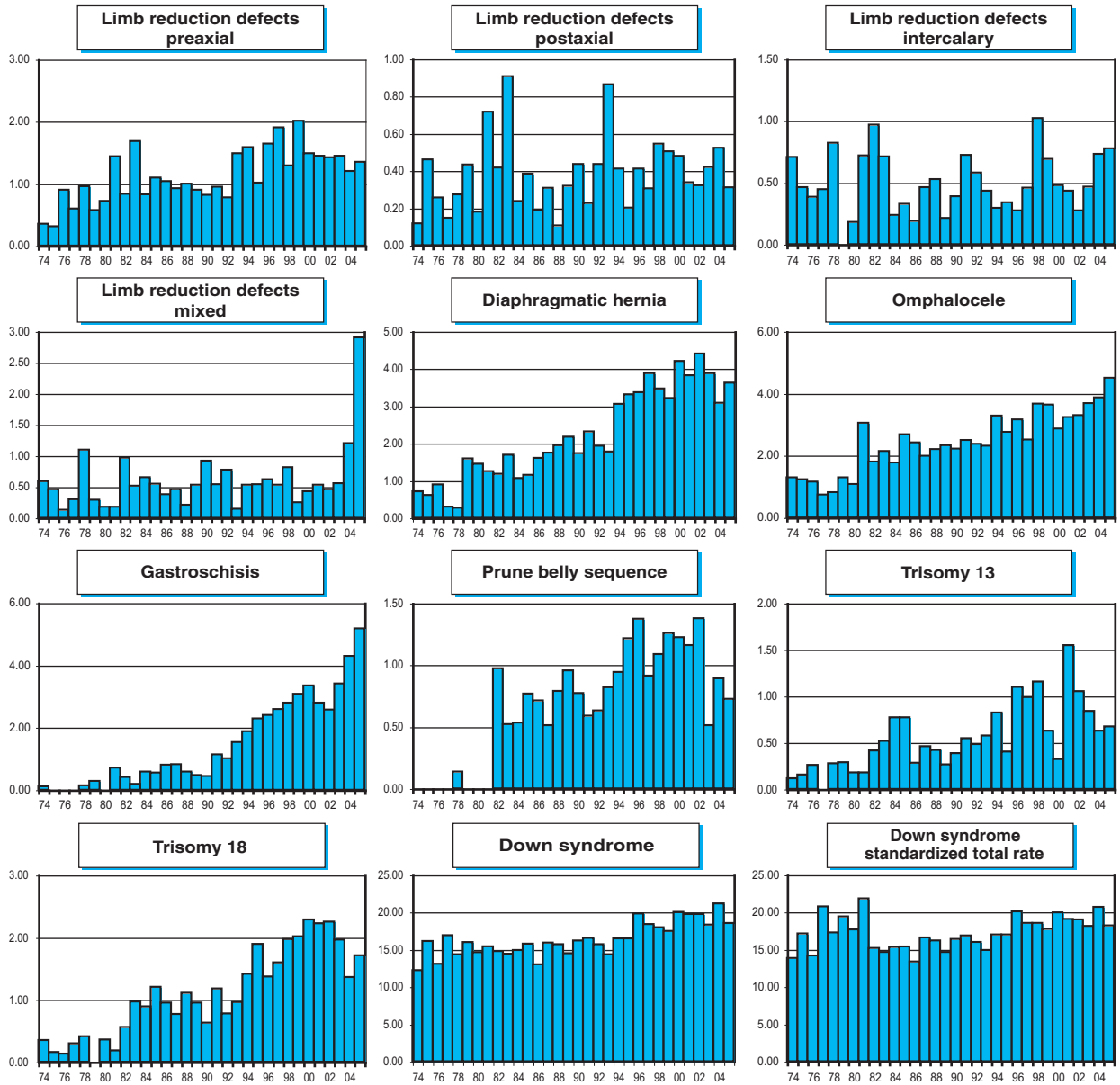

**Note:** L+S rates

## South America: ECLAMC




**Note:** L+S rates

## South America: ECLAMC




**Note:** 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. It became a full member of the ICBDMS in 1979. In January 2002 the ECEMC Programme became integrated into the CIAC (Research Center on Congenital Anomalies), of the Instituto de Salud Carlos III (ISCIII) from the Ministerio de Sanidad y Consumo of Spain, and is also directed by Prof. Martínez-Frías. Activity of the CIAC is coordinated in agreement with the IIER (Institute of Research on Rare Diseases), of the ISCIII too. In 2006 the ECEMC was recognized as an excellence Research programme to be integrated into the CIBERER (Centre for Biomedical Research on Rare Diseases). The ECEMC has 2 Teratogen Information Services since 1991, one for the general population and another one for physicians.

**Size and coverage:**
Data are obtained from hospitals distributed all over Spain. The annual number of births surpasses 100,000, representing about 23% of all Spanish births. Stillbirths of at least 24 weeks or 500 g. have been included since 1980.

**Legislation and funding:**
It is a research programme with voluntary participation of hospitals, and is financed mainly by the Spanish Administration and, partially, by non-governmental organisations.

**Sources of ascertainment:**
The detection period is the first 3 days of life, including major and/or minor/mild defects. The information comes from delivery units and paediatric departments of the participating hospitals. Mothers are interviewed directly to fill in the ECEMC standard protocols, which include more than 300 data for each child (family history, demographic and obstetrical data, prenatal exposures, etc), whether case or control. Controls are defined as the next non-malformed infant born at the same hospital that the case with the same sex as the malformed infant. In many instances, photographs, imaging studies, high-resolution bands karyotypes and molecular analysis when needed (which are performed at the central group of the ECEMC), and other complementary studies are available.

**Exposure information:**
The mother of each reported infant (case or control) is interviewed within the first three days after delivery on several exposures (parental occupation, maternal acute or chronic diseases, drug usage, exposure to other chemical or physical factors).

**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.

**Addresses and Staff:**
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Instituto de Salud Carlos III, C/Sinesio Delgado nº 6
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**Phone:** 34-91-3877538
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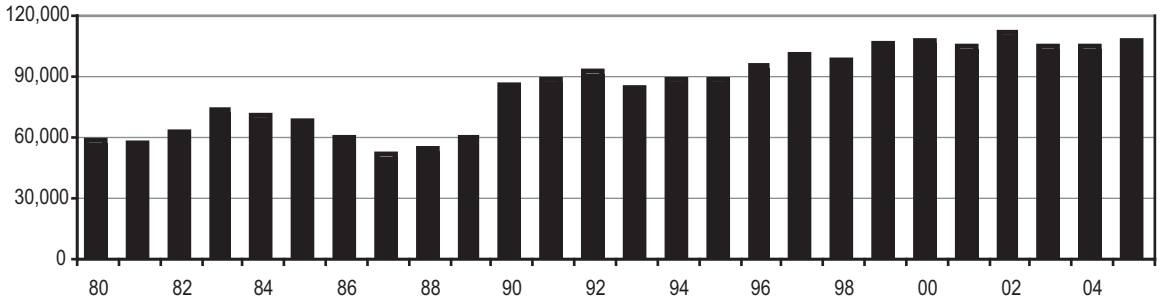
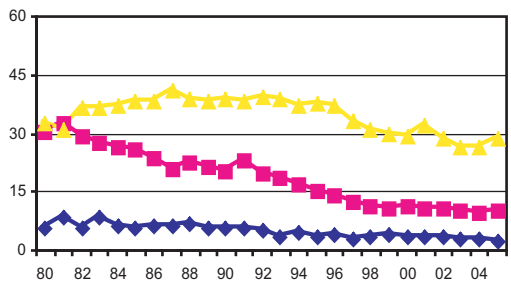
**E-mail:** mlmartinez.frias@isciii.es

## Spain: ECEMC

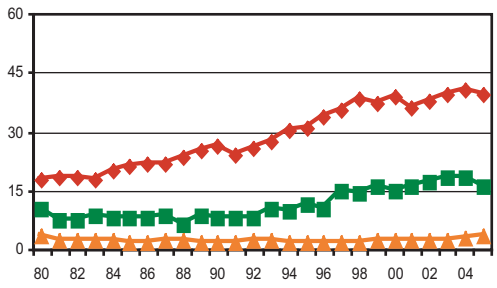
**Total births by year**




**Percentage of births by year and maternal age**




% births <20 % births 20-24 % births 25-29




% births 30-34 % births 35-39 % births 40+

# Spain: ECEMC, 2005

| | |
|---|---|
| Live births (LB) | 106,287 |
| Stillbirths (SB) | 441 |
| Total births | 106,728 |
| 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 | 0 | nr | 0.19 |
| Spina bifida | 16 | 0 | nr | 1.50 |
| Encephalocele | 3 | 0 | nr | 0.28 |
| Microcephaly | 11 | 0 | nr | 1.03 |
| Holoprosencephaly | 5 | 0 | nr | 0.47 |
| Hydrocephaly | 19 | 2 | nr | 1.97 |
| Anophthalmos | 1 | 0 | nr | 0.09 |
| Microphthalmos | 15 | 0 | nr | 1.41 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 0 | 0 | nr | 0.00 |
| Microtia | 14 | 0 | nr | 1.31 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 11 | 2 | nr | 1.22 |
| Tetralogy of Fallot | 8 | 0 | nr | 0.75 |
| Hypoplastic left heart syndrome | 3 | 0 | nr | 0.28 |
| Coarctation of aorta | 8 | 0 | nr | 0.75 |
| Choanal atresia, bilateral | 1 | 0 | nr | 0.09 |
| Cleft palate without cleft lip | 43 | 1 | nr | 4.12 |
| Cleft lip with or without cleft palate | 33 | 0 | nr | 3.09 |
| Oesophageal atresia/stenosis with or without fistula | 20 | 0 | nr | 1.87 |
| Small intestine atresia/stenosis | 4 | 0 | nr | 0.37 |
| Anorectal atresia/stenosis | 21 | 0 | nr | 1.97 |
| Undescended testis (36 weeks of gestation or later) | 26 | 0 | nr | 2.44 |
| Hypospadias | 14 | 0 | nr | 1.31 |
| Epispadias | 2 | 0 | nr | 0.19 |
| Indeterminate sex | 7 | 0 | nr | 0.66 |
| Renal agenesis | 1 | 1 | nr | 0.19 |
| Cystic kidney | 19 | 0 | nr | 1.78 |
| Bladder exstrophy | 1 | 0 | nr | 0.09 |
| Polydactyly, preaxial | 34 | 0 | nr | 3.19 |
| Total Limb reduction defects (include unspecified) | 43 | 0 | nr | 4.03 |
| Transverse | 19 | 0 | nr | 1.78 |
| Preaxial | 6 | 0 | nr | 0.56 |
| Postaxial | 1 | 0 | nr | 0.09 |
| Intercalary | 2 | 0 | nr | 0.19 |
| Mixed | 11 | 0 | nr | 1.03 |
| Unspecified | 4 | 0 | nr | 0.37 |
| Diaphragmatic hernia | 11 | 0 | nr | 1.03 |
| Omphalocele | 9 | 0 | nr | 0.84 |
| Gastroschisis | 1 | 0 | nr | 0.09 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 1 | 0 | nr | 0.09 |
| Trisomy 13 | 4 | 1 | nr | 0.47 |
| Trisomy 18 | 4 | 1 | nr | 0.47 |
| Down syndrome, all ages (include age unknown) | 79 | 0 | nr | 7.40 |
| <20 | 1 | 0 | nr | 3.82 |
| 20-24 | 5 | 0 | nr | 4.62 |
| 25-29 | 12 | 0 | nr | 3.97 |
| 30-34 | 27 | 0 | nr | 6.42 |
| 35-39 | 24 | 0 | nr | 13.83 |
| 40-44 | 8 | 0 | nr | 24.24 |
| 45+ | 1 | 0 | nr | 34.48 |
| unknown | 1 | 0 | nr | 526.32 |

nr = not reported

# Spain: ECEMC, Previous years rates 1980 - 2005

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

| | 1974-1980* | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **56,910** | **325,480** | **305,407** | **436,851** | **499,648** | **528,184** |
| Anencephaly | 6.15 | 4.39 | 2.06 | 1.01 | 0.44 | 0.19 |
| Spina bifida | 4.22 | 4.70 | 4.39 | 3.20 | 2.12 | 1.25 |
| Encephalocele | 1.05 | 1.17 | 0.65 | 0.82 | 0.20 | 0.21 |
| Microcephaly | 1.93 | 2.15 | 2.03 | 2.29 | 1.54 | 1.02 |
| Holoprosencephaly | 0.53 | 0.46 | 0.52 | 0.57 | 0.46 | 0.27 |
| Hydrocephaly | 3.34 | 2.40 | 2.82 | 2.95 | 2.26 | 1.78 |
| Anophthalmos | 0.70 | 0.74 | 0.33 | 0.21 | 0.20 | 0.11 |
| Microphthalmos | 1.76 | 1.87 | 1.80 | 1.69 | 1.30 | 0.97 |
| Unspecified Anophthalmos/Microphthalmos | ---- | ---- | ---- | ---- | ---- | ---- |
| Anotia | 0.00 | 0.03 | 0.00 | 0.21 | 0.12 | 0.08 |
| Microtia | 2.64 | 2.06 | 1.64 | 1.21 | 1.52 | 1.55 |
| Unspecified Anotia/Microtia | ---- | ---- | ---- | ---- | ---- | ---- |
| Transposition of great vessels | 0.88 | 0.61 | 1.11 | 1.51 | 1.32 | 1.25 |
| Tetralogy of Fallot | 0.18 | 0.25 | 0.62 | 1.03 | 1.18 | 0.89 |
| Hypoplastic left heart syndrome | 0.18 | 0.49 | 0.59 | 1.10 | 0.70 | 0.27 |
| Coarctation of aorta | 0.18 | 0.37 | 0.56 | 0.78 | 0.96 | 0.66 |
| Choanal atresia, bilateral | 0.00 | 0.12 | 0.36 | 0.30 | 0.22 | 0.11 |
| Cleft palate without cleft lip | 4.74 | 5.19 | 4.35 | 4.81 | 3.94 | 4.05 |
| Cleft lip with or without cleft palate | 5.27 | 5.87 | 5.17 | 5.91 | 4.14 | 3.64 |
| Oesophageal atresia/stenosis with or without fistula | 1.41 | 2.46 | 1.64 | 2.31 | 1.50 | 2.04 |
| Small intestine atresia/stenosis | 0.70 | 0.49 | 0.52 | 0.53 | 0.40 | 0.57 |
| Anorectal atresia/stenosis | 1.93 | 2.64 | 2.29 | 2.01 | 2.10 | 2.01 |
| Undescended testis (36 weeks of gestation or later) | 1.23 | 1.90 | 2.55 | 2.84 | 2.74 | 2.37 |
| Hypospadias | 2.81 | 2.70 | 2.13 | 2.04 | 1.70 | 2.12 |
| Epispadias | 0.18 | 0.22 | 0.23 | 0.16 | 0.08 | 0.09 |
| Indeterminate sex | 0.35 | 1.20 | 1.11 | 0.66 | 0.58 | 0.55 |
| Renal agenesis | 0.53 | 0.71 | 0.92 | 0.60 | 0.38 | 0.08 |
| Cystic kidney | 1.76 | 1.08 | 1.64 | 1.76 | 1.72 | 1.42 |
| Bladder exstrophy | 0.35 | 0.28 | 0.33 | 0.23 | 0.24 | 0.19 |
| Polydactyly, preaxial | 1.76 | 2.49 | 2.46 | 3.41 | 2.52 | 2.08 |
| Total Limb reduction defects (include unspecified) | 6.15 | 7.28 | 6.88 | 6.80 | 5.50 | 4.45 |
| Transverse | 2.46 | 3.01 | 3.05 | 2.40 | 2.38 | 1.74 |
| Preaxial | 0.53 | 1.26 | 1.05 | 0.89 | 0.66 | 0.59 |
| Postaxial | 0.00 | 0.18 | 0.16 | 0.23 | 0.20 | 0.09 |
| Intercalary | 0.18 | 0.61 | 0.16 | 0.64 | 0.20 | 0.30 |
| Mixed | 1.58 | 1.08 | 1.15 | 1.03 | 1.10 | 0.89 |
| Unspecified | ---- | ---- | ---- | ---- | ---- | ---- |
| Diaphragmatic hernia | 1.93 | 2.86 | 2.03 | 2.22 | 1.22 | 0.74 |
| Omphalocele | 2.46 | 1.57 | 1.34 | 1.19 | 0.70 | 0.59 |
| Gastroschisis | 0.53 | 0.55 | 0.43 | 0.30 | 0.50 | 0.28 |
| Unspecified Omphalocele/Gastroschisis | ---- | ---- | ---- | ---- | ---- | ---- |
| Prune belly sequence | 0.18 | 0.65 | 0.52 | 0.48 | 0.26 | 0.15 |
| Trisomy 13 | 0.35 | 0.34 | 0.46 | 0.46 | 0.50 | 0.40 |
| Trisomy 18 | 0.35 | 1.20 | 0.88 | 0.96 | 0.66 | 0.61 |
| Down syndrome, all ages (include age unknown) | 13.71 | 14.96 | 14.11 | 12.59 | 10.27 | 7.69 |
| <20 | 3.06 | 7.65 | 9.16 | 6.84 | 1.73 | 2.56 |
| 20-24 | 8.15 | 6.22 | 6.37 | 5.32 | 3.72 | 5.34 |
| 25-29 | 5.43 | 6.60 | 7.93 | 7.55 | 6.05 | 4.28 |
| 30-34 | 9.97 | 12.36 | 13.16 | 13.58 | 9.86 | 7.15 |
| 35-39 | 36.66 | 48.39 | 39.05 | 35.42 | 20.85 | 12.28 |
| 40-44 | 96.48 | 175.25 | 154.51 | 60.08 | 53.42 | 35.03 |
| 45+ | 83.33 | 246.91 | 188.09 | 264.15 | 531.91 | 26.46 |
| unknown | ---- | ---- | ---- | ---- | ---- | ---- |

* data include less than 5 and 7 years

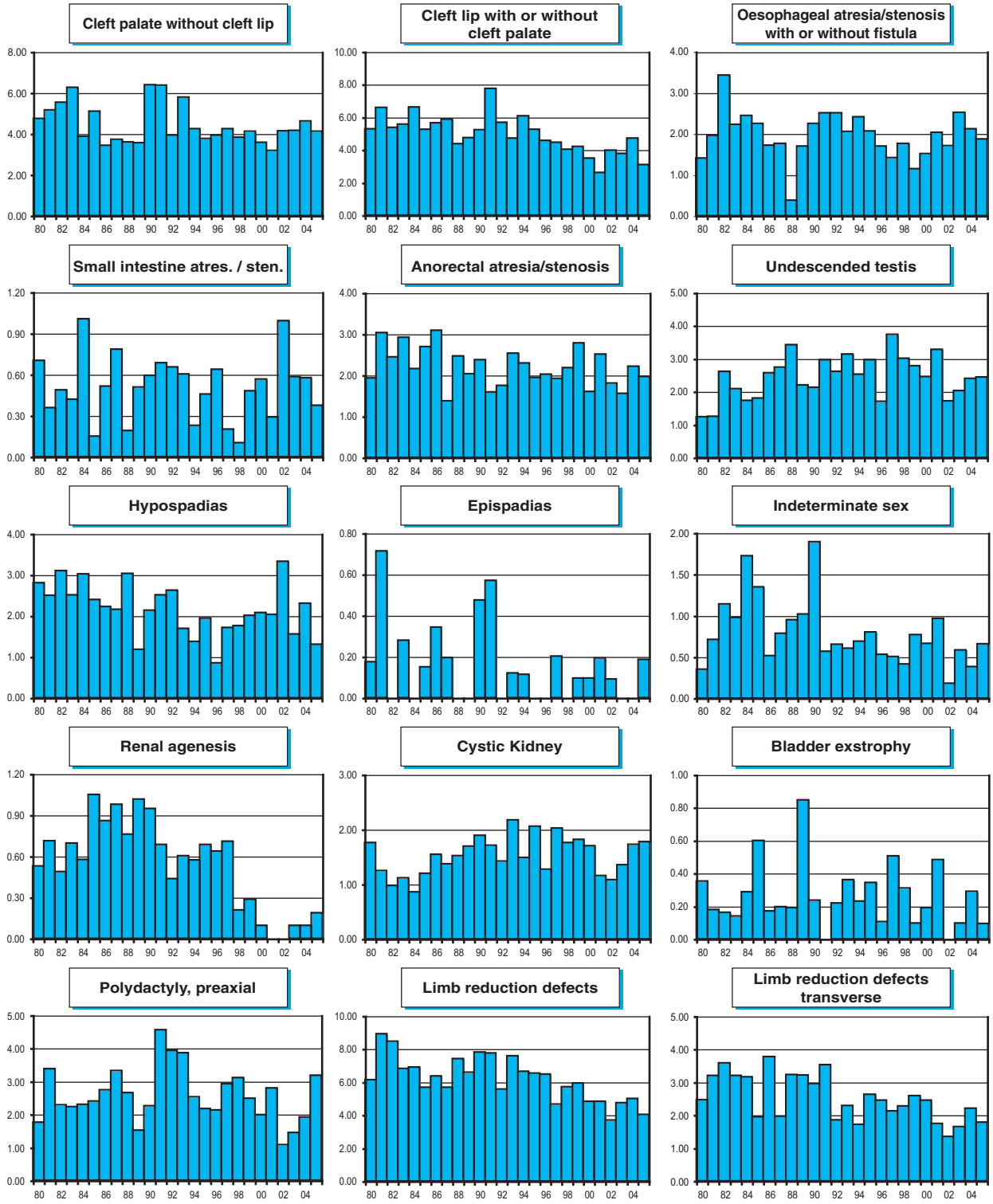
nr = not reported

## Spain: ECEMC

'Time trends 1980-2005 (Birth prevalence rates per 10,000)


**Note:** L+S rates

## Spain: ECEMC




**Note:** L+S rates

## Spain: ECEMC


**Note:** L+S rates

## Sweden
### The Swedish Birth Defects Registry

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

**Size and coverage:**
All births in Sweden are included, approximately 100,000-120,000 annual births. The definition of stillbirth in Sweden is more than 28 weeks. Since 1999 all fetal deaths with congenital malformations more than 22 weeks are reported to the Swedish Birth Defects Registry. In 1999 a special fetal congenital anomalies surveillance system was started to include those fetuses with congenital malformations who were terminated as a result of prenatal diagnosis.

**Legislation and funding:**
Reporting is compulsory for children with malformations, but not for terminated pregnancies with fetuses with congenital malformations.

**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, socio-economic factors, maternal smoking, drug use during pregnancy, contraceptive usage, maternal diseases.

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

**Addresses and Staff:**
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**Phone:** 46-75-2473123
**Fax:** 46-75-2473327
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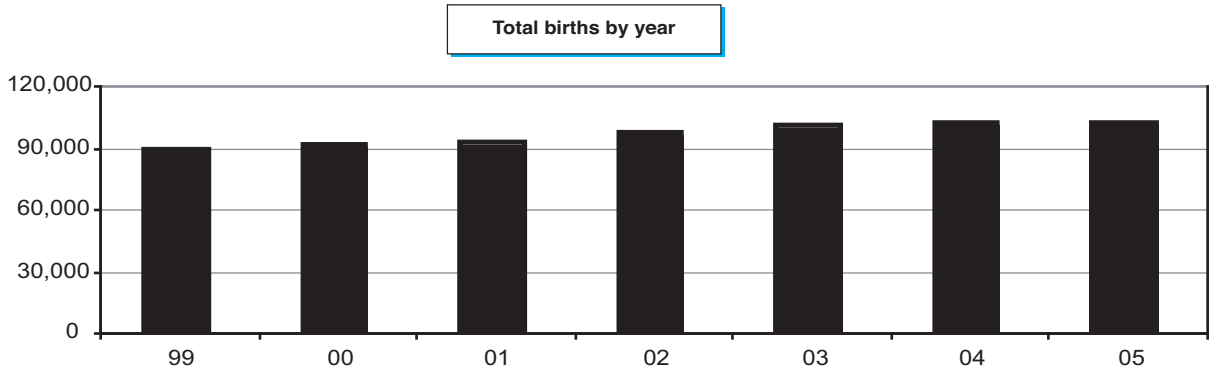
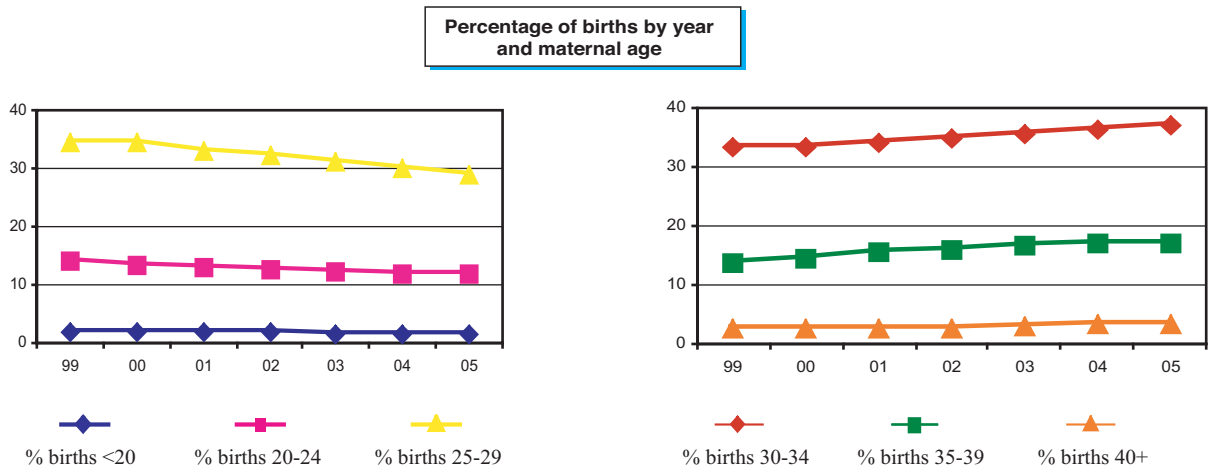
## Sweden




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 100 | 94.3 | Cystic kidney | 54 | 47.4 |
| Spina bifida | 68 | 57.6 | Limb reduction defects | 30 | 20.0 |
| Encephalocele | 25 | 75.8 | Diaphragmatic hernia | 31 | 32.6 |
| Holoprosencephaly | 21 | 65.6 | Omphalocele | 56 | 59.6 |
| Hydrocephaly | 71 | 68.9 | Gastroschisis | 13 | 22.8 |
| Hypoplastic left heart syndrome | 35 | 42.2 | Trisomy 13 | 59 | 64.8 |
| Cleft palate without cleft lip | 9 | 6.0 | Trisomy 18 | 181 | 80.4 |
| Cleft lip with or without cleft palate | 23 | 7.5 | Down syndrome | 408 | 51.1 |
| Renal agenesis | 23 | 71.9 | | | |

Total ToPs with births defects = 1,387 (Ratio ToPs/Births: 4.59 per 1,000)
*ToPs/ToPs+Births

# Sweden: 2005

| | |
|---|---|
| Live births (LB) | 101,346 |
| Stillbirths (SB) | 301 |
| Total births | 101,647 |
| Number of terminations of pregnancy (ToP) for birth defects | 513 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 3 | 0 | 34 | 3.64 |
| Spina bifida | 12 | 0 | 34 | 4.53 |
| Encephalocele | 2 | 0 | 7 | 0.89 |
| Microcephaly | 1 | 0 | 0 | 0.10 |
| Holoprosencephaly | 1 | 2 | 12 | 1.48 |
| Hydrocephaly | 7 | 1 | 20 | 2.75 |
| Anophthalmos | 1 | 0 | 0 | 0.10 |
| Microphthalmos | 7 | 0 | 0 | 0.69 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 11 | 2 | 1 | 1.38 |
| Microtia | 3 | 0 | 0 | 0.30 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 37 | 0 | 1 | 3.74 |
| Tetralogy of Fallot | 26 | 1 | 1 | 2.75 |
| Hypoplastic left heart syndrome | 13 | 0 | 17 | 2.95 |
| Coarctation of aorta | 52 | 0 | 6 | 5.71 |
| Choanal atresia, bilateral | 0 | 0 | 0 | 0.00 |
| Cleft palate without cleft lip | 58 | 0 | 2 | 5.90 |
| Cleft lip with or without cleft palate | 94 | 2 | 9 | 10.33 |
| Oesophageal atresia/stenosis with or without fistula | 19 | 2 | 3 | 2.36 |
| Small intestine atresia/stenosis | 27 | 1 | 3 | 3.05 |
| Anorectal atresia/stenosis | 19 | 0 | 4 | 2.26 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 197 | 0 | 2 | 19.58 |
| Epispadias | 1 | 0 | 0 | 0.10 |
| Indeterminate sex | 1 | 0 | 2 | 0.30 |
| Renal agenesis | 3 | 0 | 7 | 0.98 |
| Cystic kidney | 23 | 0 | 25 | 4.72 |
| Bladder exstrophy | 1 | 0 | 1 | 0.20 |
| Polydactyly, preaxial | 40 | 1 | 5 | 4.53 |
| Total Limb reduction defects (include unspecified) | 30 | 0 | 11 | 4.03 |
| Transverse | 4 | 0 | 6 | 0.98 |
| Preaxial | 2 | 0 | 4 | 0.59 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 2 | 0 | 0 | 0.20 |
| Mixed | 2 | 0 | 1 | 0.30 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 19 | 0 | 13 | 3.15 |
| Omphalocele | 14 | 0 | 24 | 3.74 |
| Gastroschisis | 14 | 0 | 5 | 1.87 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 1 | 0.10 |
| Trisomy 13 | 10 | 1 | 22 | 3.25 |
| Trisomy 18 | 9 | 3 | 75 | 8.56 |
| Down syndrome, all ages (include age unknown) | 129 | 0 | 153 | 27.74 |
| <20 | 1 | 0 | 0 | 5.91 |
| 20-24 | 7 | 0 | 3 | 8.32 |
| 25-29 | 21 | 0 | 4 | 8.49 |
| 30-34 | 50 | 0 | 30 | 21.27 |
| 35-39 | 32 | 0 | 56 | 50.44 |
| 40-44 | 13 | 0 | 55 | 207.06 |
| 45+ | 4 | 0 | 5 | nr |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# Sweden: Previous years rates 1999 - 2005

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

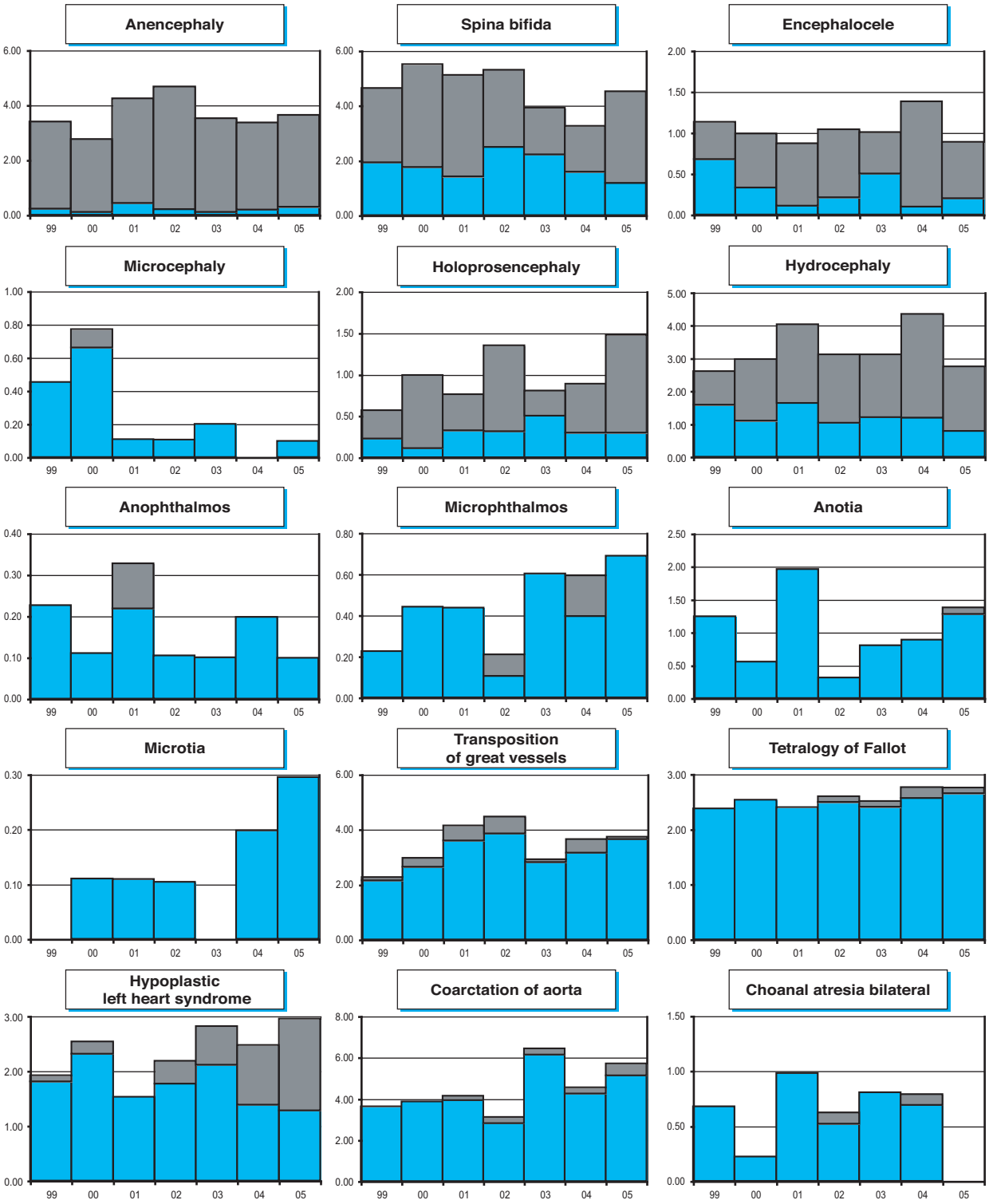
| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000* | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | | **179,308** | **490,406** |
| Anencephaly | | | | | 3.07 | 3.87 |
| Spina bifida | | | | | 5.08 | 4.40 |
| Encephalocele | | | | | 1.06 | 1.04 |
| Microcephaly | | | | | 0.61 | 0.10 |
| Holoprosencephaly | | | | | 0.78 | 1.06 |
| Hydrocephaly | | | | | 2.79 | 3.47 |
| Anophthalmos | | | | | 0.17 | 0.16 |
| Microphthalmos | | | | | 0.33 | 0.51 |
| Unspecified Anophthalmos/Microphthalmos | | | | | --- | --- |
| Anotia | | | | | 0.89 | 1.06 |
| Microtia | | | | | 0.06 | 0.14 |
| Unspecified Anotia/Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | 2.62 | 3.77 |
| Tetralogy of Fallot | | | | | 2.45 | 2.61 |
| Hypoplastic left heart syndrome | | | | | 2.23 | 2.41 |
| Coarctation of aorta | | | | | 3.74 | 4.81 |
| Choanal atresia, bilateral | | | | | 0.45 | 0.63 |
| Cleft palate without cleft lip | | | | | 6.08 | 5.24 |
| Cleft lip with or without cleft palate | | | | | 9.54 | 10.32 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 1.84 | 2.69 |
| Small intestine atresia/stenosis | | | | | 2.12 | 2.71 |
| Anorectal atresia/stenosis | | | | | 2.84 | 3.04 |
| Undescended testis (36 weeks of gestation or later) | | | | | nr | nr |
| Hypospadias | | | | | 19.46 | 20.60 |
| Epispadias | | | | | 0.06 | 0.22 |
| Indeterminate sex | | | | | 0.28 | 0.24 |
| Renal agenesis | | | | | 2.45 | 1.39 |
| Cystic kidney | | | | | 2.51 | 3.63 |
| Bladder exstrophy | | | | | 0.22 | 0.27 |
| Polydactyly, preaxial | | | | | 3.51 | 4.89 |
| Total Limb reduction defects (include unspecified) | | | | | 4.35 | 5.14 |
| Transverse | | | | | 2.45 | 3.57 |
| Preaxial | | | | | 0.33 | 0.29 |
| Postaxial | | | | | 0.22 | 0.12 |
| Intercalary | | | | | 0.11 | 0.24 |
| Mixed | | | | | 1.23 | 0.51 |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 2.79 | 2.81 |
| Omphalocele | | | | | 2.23 | 2.92 |
| Gastroschisis | | | | | 2.01 | 1.81 |
| Unspecified Omphalocele/Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | 0.06 | 0.12 |
| Trisomy 13 | | | | | 2.01 | 2.57 |
| Trisomy 18 | | | | | 5.47 | 6.75 |
| Down syndrome, all ages (include age unknown) | | | | | 21.81 | 25.77 |
| <20 | | | | | 8.76 | 8.47 |
| 20-24 | | | | | 9.37 | 8.66 |
| 25-29 | | | | | 6.34 | 9.56 |
| 30-34 | | | | | 14.71 | 18.35 |
| 35-39 | | | | | 51.25 | 56.13 |
| 40-44 | | | | | 161.92 | 177.40 |
| 45+ | | | | | 421.05 | 477.88 |
| unspecified | | | | | --- | --- |

* data include less than 5 years

nr = not reported

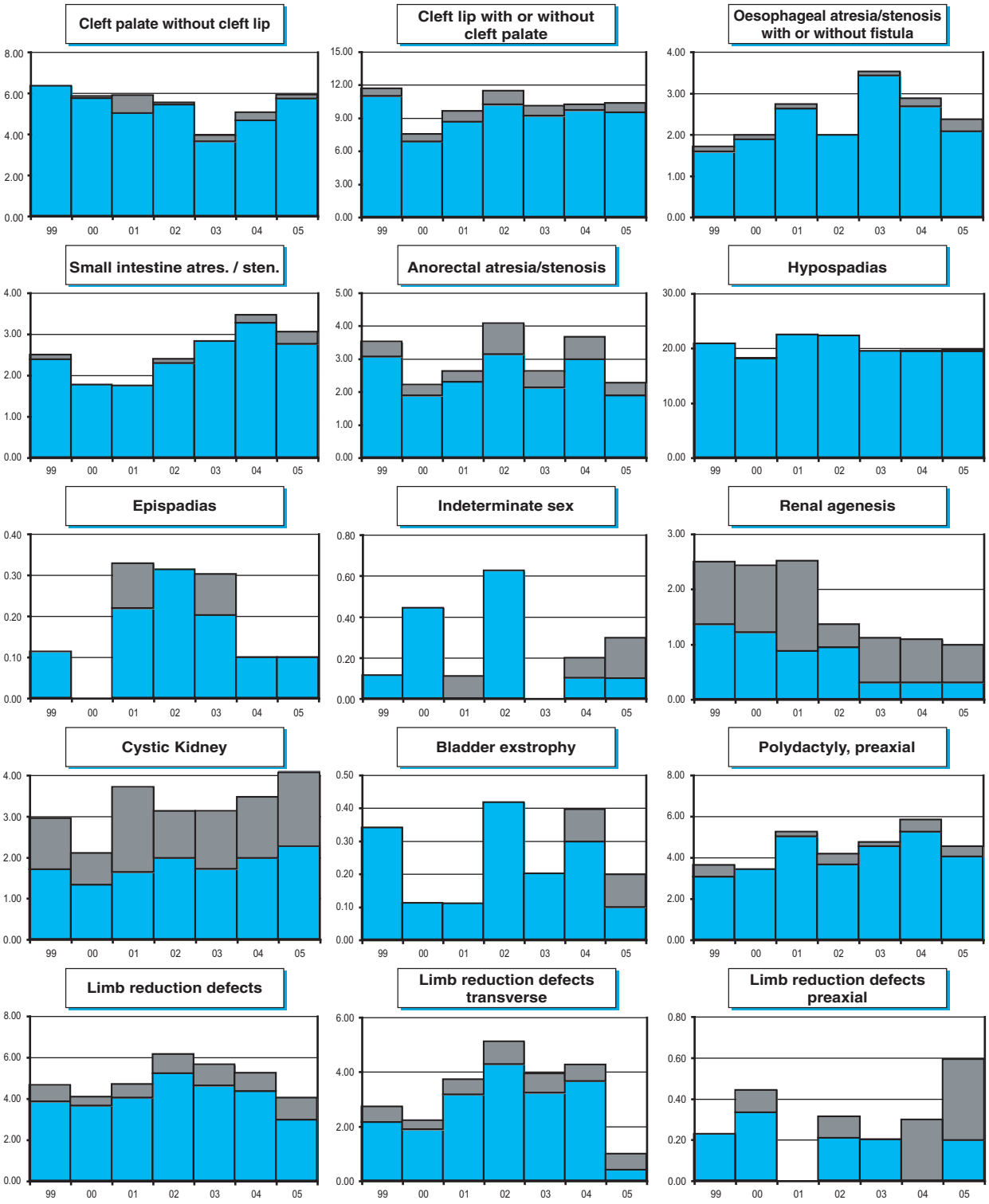
## Sweden

'Time trends 1999-2005 (Birth prevalence rates per 10,000)



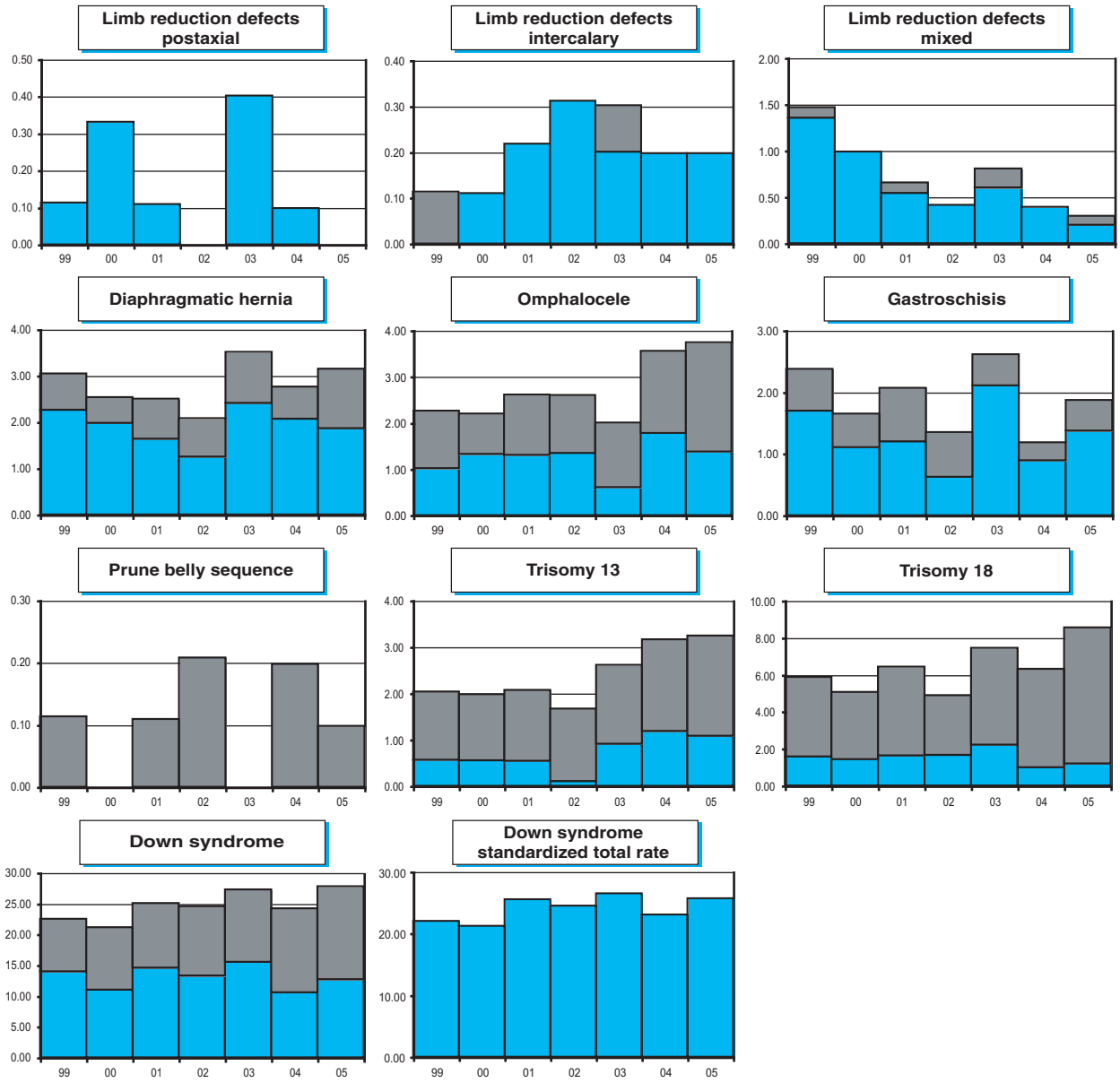

**Note:** L+S rates, ToP rates

## Sweden




**Note:** L+S rates, ToP rates

## Sweden




**Note:** L+S rates, ToP 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 financing of surveillance finally ended and the program was assumed by OMNI-Net, a not-for-profit international organization incorporated in Ukraine. OMNI-Net represents five resource OMNI-centers conducting birth defects surveillance, providing care for children and promoting prevention programs with participation of parental organizations, 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.

**Size and coverage:**
BD surveillance concerns 25000 births in two oblasts (provinces) of Northwest of Ukraine, representing approximately 5% of births in Ukraine. The population is relatively homogeneous, stable (data is pooled from two oblasts). The northern counties (rayons) of both oblasts are contaminated from Chornobyl disaster.

**Legislation and funding:**
OMNI-Net personnel are financed by the Ukrainian Ministry of Health and oblast authorities. 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.

**Sources of ascertainment:**
Relevant hospital admission/discharge summaries are systematically reviewed. Data from specialty clinics, laboratories and other services are explored. Pregnancy, obstetrics, delivery, neonatal and pediatrics records are considered.

**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:**
*International Coordinator:*
Dr. Wladimir Wertelecki,
Department of Medical Genetics,
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307 University Blvd., CCCB, 274,
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*Medical coordinator:*
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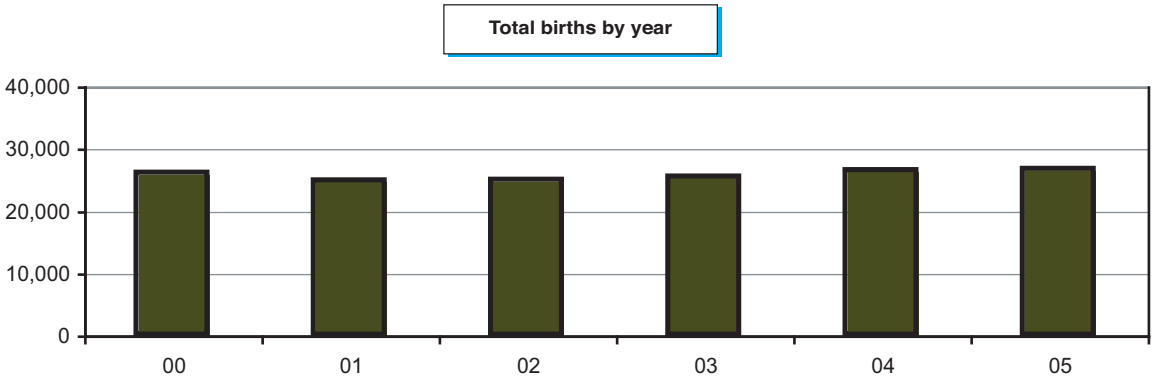
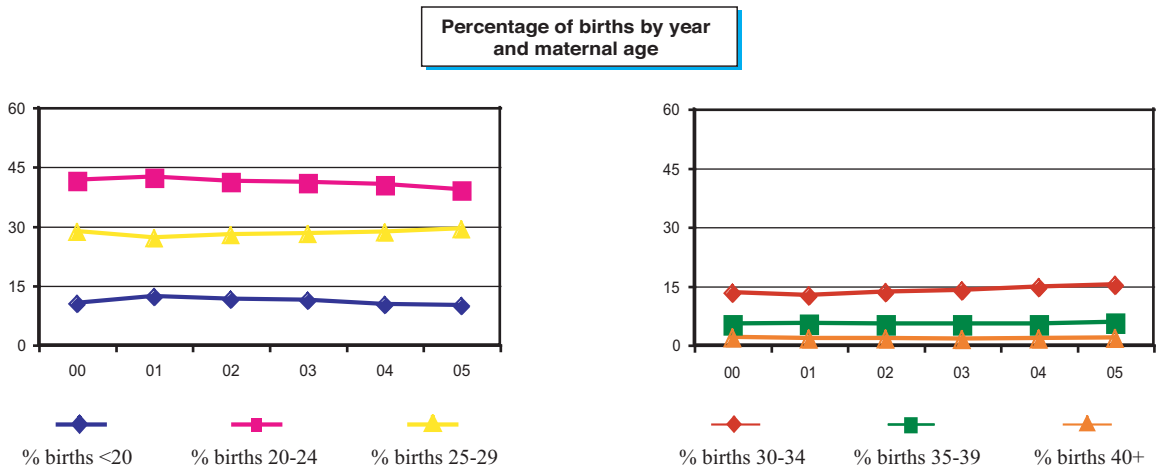
Rivne, Ukraine 33028
**Phone/Fax:** 38 036 262 3447
**E-mail:** bdrivne@bdp.rovno.ua
lyevtushok@bdp.rovno.ua

## Ukraine: OMNI-Net



# Ukraine: OMNI-Net, 2005

| | |
|---|---|
| Live births (LB) | 26,492 |
| Stillbirths (SB) | 121 |
| Total births | 26,613 |
| 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 | 7 | 25 | 12.02 |
| Spina bifida | 6 | 2 | 19 | 10.15 |
| Encephalocele | 1 | 1 | 4 | 2.25 |
| Microcephaly (1) | 8 | 0 | nr | 3.01 |
| Holoprosencephaly | 2 | 0 | nr | 0.75 |
| Hydrocephaly | 14 | 4 | nr | 6.76 |
| Anophthalmos (1) | 0 | 0 | nr | 0.00 |
| Microphthalmos (1) | 4 | 0 | nr | 1.50 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | nr | 0.00 |
| Anotia | 2 | 0 | nr | 0.75 |
| Microtia | 5 | 0 | nr | 1.88 |
| Unspecified Anotia/Microtia | 0 | 0 | nr | 0.00 |
| Transposition of great vessels | 8 | 0 | nr | 3.01 |
| Tetralogy of Fallot | 3 | 0 | nr | 1.13 |
| Hypoplastic left heart syndrome | 6 | 0 | nr | 2.25 |
| Coarctation of aorta | 4 | 0 | nr | 1.50 |
| Choanal atresia, bilateral | 0 | 0 | nr | 0.00 |
| Cleft palate without cleft lip | 13 | 1 | nr | 5.26 |
| Cleft lip with or without cleft palate | 25 | 0 | nr | 9.39 |
| Oesophageal atresia/stenosis with or without fistula | 6 | 2 | nr | 3.01 |
| Small intestine atresia/stenosis | 5 | 0 | nr | 1.88 |
| Anorectal atresia/stenosis | 4 | 0 | nr | 1.50 |
| Undescended testis (36 weeks of gestation or later) | 128 | 0 | nr | 48.10 |
| Hypospadias (2) | 10 | 0 | nr | 3.76 |
| Epispadias | 0 | 0 | nr | 0.00 |
| Indeterminate sex | 0 | 0 | nr | 0.00 |
| Renal agenesis | 0 | 3 | nr | 1.13 |
| Cystic kidney | 7 | 2 | nr | 3.38 |
| Bladder exstrophy | 1 | 0 | nr | 0.38 |
| Polydactyly, preaxial | 11 | 0 | nr | 4.13 |
| Total Limb reduction defects (include unspecified) | 4 | 0 | nr | 1.50 |
| Transverse | 1 | 0 | nr | 0.38 |
| Preaxial | 2 | 0 | nr | 0.75 |
| Postaxial | 1 | 0 | nr | 0.38 |
| Intercalary | 0 | 0 | nr | 0.00 |
| Mixed | 0 | 0 | nr | 0.00 |
| Unspecified | 0 | 0 | nr | 0.00 |
| Diaphragmatic hernia | 3 | 1 | nr | 1.50 |
| Omphalocele | 5 | 0 | nr | 1.88 |
| Gastroschisis | 7 | 0 | nr | 2.63 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | nr | 0.00 |
| Prune belly sequence | 0 | 0 | nr | 0.00 |
| Trisomy 13 (1) | 3 | 0 | nr | 1.13 |
| Trisomy 18 (1) | 2 | 0 | nr | 0.75 |
| Down syndrome, all ages (include age unknown) (1) | 31 | 2 | nr | 12.40 |
| <20 | 7 | 0 | nr | 26.84 |
| 20-24 | 1 | 0 | nr | 0.96 |
| 25-29 | 6 | 0 | nr | 7.74 |
| 30-34 | 7 | 0 | nr | 17.53 |
| 35-39 | 5 | 2 | nr | 47.75 |
| 40-44 | 5 | 0 | nr | 130.21 |
| 45+ | 0 | 0 | nr | 0.00 |
| unknown | 0 | 0 | nr | 0.00 |

(1) Clinical diagnosis only; with photodocumentation or measurements documented
(2) Includes penile, scrotal, and perineal hypospadias only
nr = not reported

# Ukraine: OMNI-Net, Previous years rates 2000 - 2005

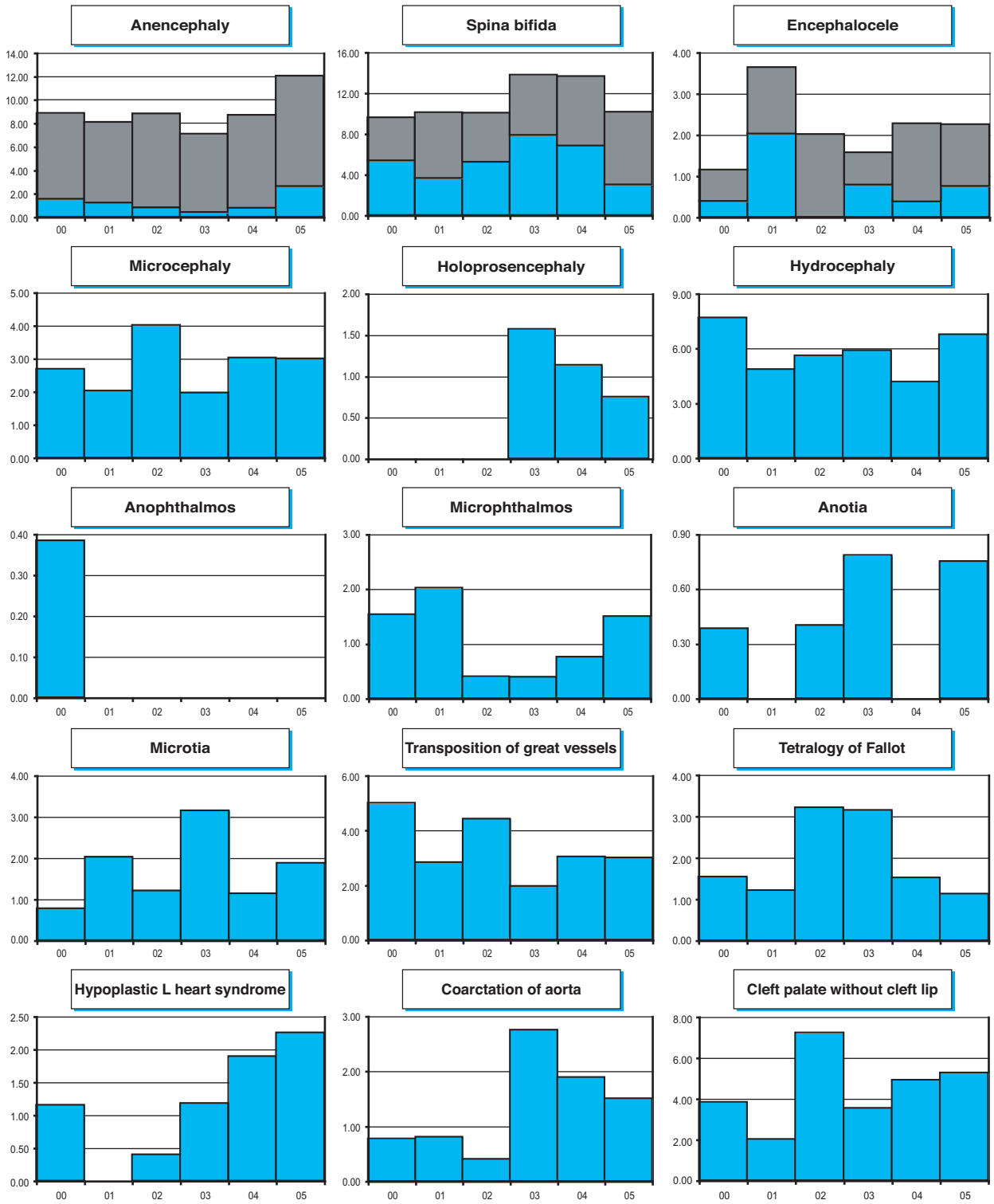
Birth prevalence rates: (LB+SB+TOP) * 10,000 for Anencephaly, Spina bifida and Encephalocele
Birth prevalence rates: (LB+SB) * 10,000 for all other malformations

| | 1974-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000* | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | | **26,025** | **128,135** |
| Anencephaly | | | | | 8.84 | 8.97 |
| Spina bifida | | | | | 9.61 | 11.55 |
| Encephalocele | | | | | 1.15 | 2.34 |
| Microcephaly | | | | | 2.69 | 2.81 |
| Holoprosencephaly | | | | | 0.00 | 0.70 |
| Hydrocephaly | | | | | 7.68 | 5.46 |
| Anophthalmos | | | | | 0.38 | 0.00 |
| Microphthalmos | | | | | 1.54 | 1.01 |
| Unspecified Anophthalmos/Microphthalmos | | | | | --- | --- |
| Anotia | | | | | 0.38 | 0.39 |
| Microtia | | | | | 0.77 | 1.87 |
| Unspecified Anotia/Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | 5.00 | 3.04 |
| Tetralogy of Fallot | | | | | 1.54 | 2.03 |
| Hypoplastic left heart syndrome | | | | | 1.15 | 1.17 |
| Coarctation of aorta | | | | | 0.77 | 1.48 |
| Choanal atresia, bilateral | | | | | 0.00 | 0.00 |
| Cleft palate without cleft lip | | | | | 3.84 | 4.60 |
| Cleft lip with or without cleft palate | | | | | 9.61 | 8.82 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 2.31 | 1.64 |
| Small intestine atresia/stenosis | | | | | 1.54 | 1.56 |
| Anorectal atresia/stenosis | | | | | 1.92 | 2.73 |
| Undescended testis (36 weeks of gestation or later) | | | | | 34.58 | 41.60 |
| Hypospadias | | | | | 3.07 | 3.28 |
| Epispadias | | | | | 0.77 | 0.16 |
| Indeterminate sex | | | | | 0.77 | 0.39 |
| Renal agenesis | | | | | 0.77 | 0.86 |
| Cystic kidney | | | | | 1.54 | 2.42 |
| Bladder exstrophy | | | | | 1.15 | 0.70 |
| Polydactyly, preaxial | | | | | 3.07 | 3.04 |
| Total Limb reduction defects (include unspecified) | | | | | 4.23 | 3.28 |
| Transverse | | | | | 1.54 | 2.03 |
| Preaxial | | | | | 0.77 | 0.47 |
| Postaxial | | | | | 0.77 | 0.16 |
| Intercalary | | | | | 0.38 | 0.23 |
| Mixed | | | | | 0.38 | 0.16 |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 2.31 | 1.72 |
| Omphalocele | | | | | 1.15 | 1.40 |
| Gastroschisis | | | | | 1.15 | 1.33 |
| Unspecified Omphalocele/Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | 0.00 | 0.00 |
| Trisomy 13 | | | | | 0.00 | 0.39 |
| Trisomy 18 | | | | | 1.15 | 0.23 |
| Down syndrome, all ages (include age unknown) | | | | | 10.76 | 12.64 |
| <20 | | | | | 11.23 | 10.09 |
| 20-24 | | | | | 4.64 | 6.90 |
| 25-29 | | | | | 6.76 | 9.47 |
| 30-34 | | | | | 17.60 | 16.41 |
| 35-39 | | | | | 22.34 | 35.80 |
| 40-44 | | | | | 147.42 | 98.21 |
| 45+ | | | | | 0.00 | 851.06 |
| unknown | | | | | --- | --- |

* data include less than 5 years

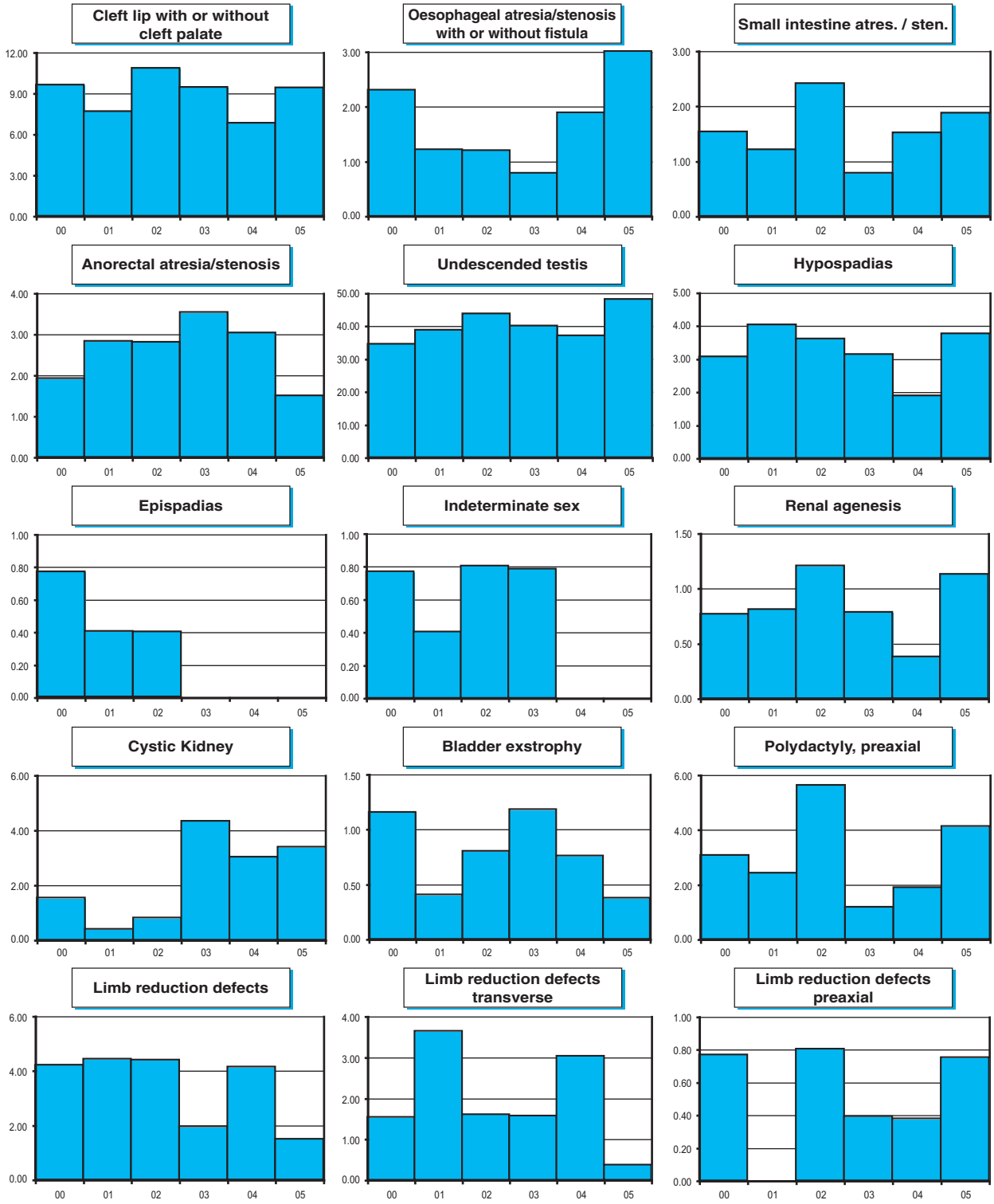
## Ukraine: OMNI-Net

'Time trends 2000-2005 (Birth prevalence rates per 10,000)



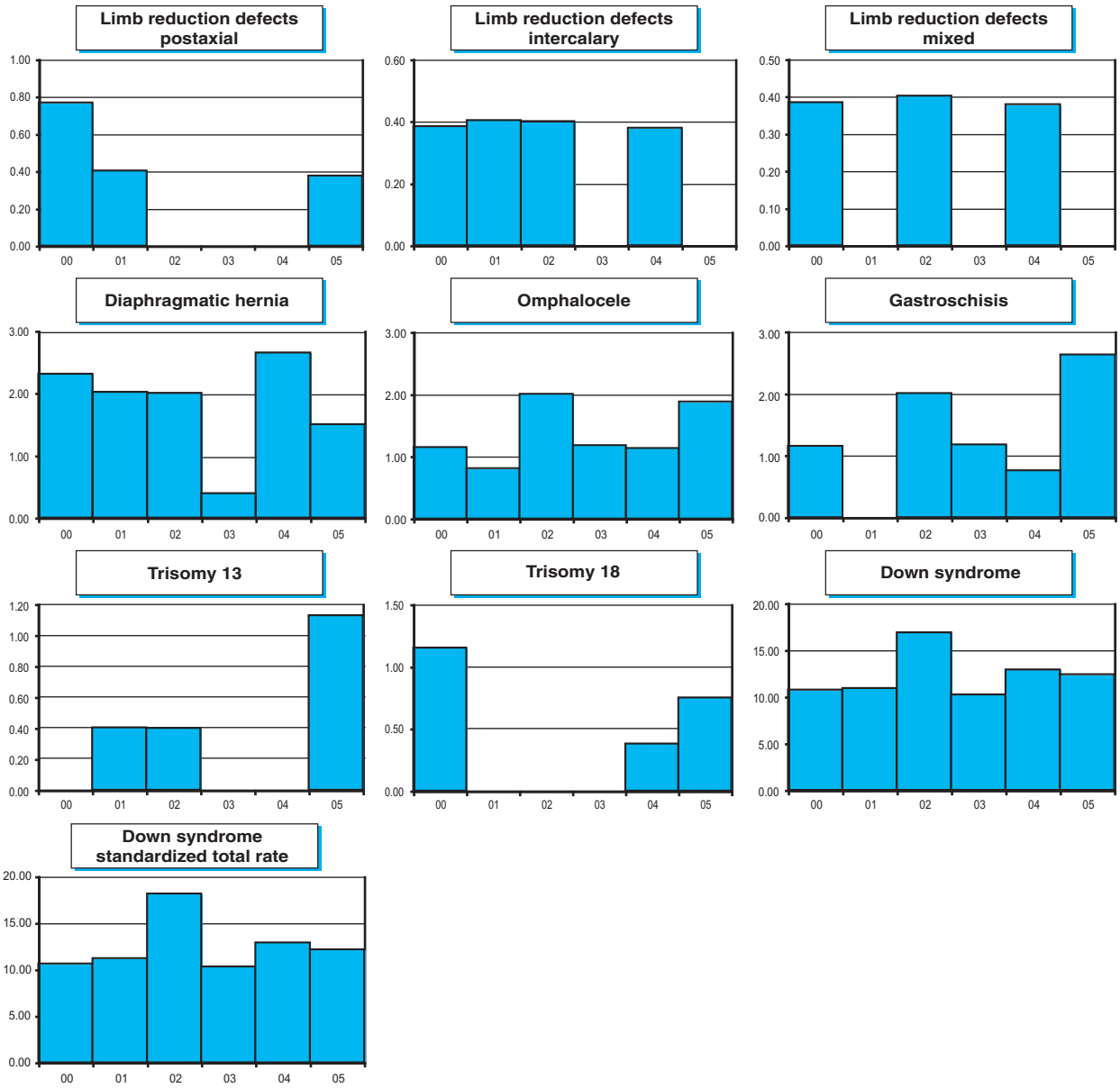

**Note:** L+S rates, ToP rates

## Ukraine: OMNI-Net




**Note:** L+S rates, ToP rates

## Ukraine: OMNI-Net




**Note:** L+S rates, ToP rates

## USA-Atlanta: MACDP

Metropolitan Atlanta Congenital Defects Program

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

**Size and coverage:**
The Programme covers all births within a five-county area in metropolitan Atlanta, Georgia. The annual number of births in this area is approximately 50,000. Stillbirths and terminations of at least 20 weeks gestations are included. Elective terminations at any gestational age are included.

**Legislation and funding:**
In 1994 the Georgia Department of Human Resources (GDHR) added birth defects to the list of legally reportable conditions in Georgia. In 1997 the GDHR authorized the Birth Defects Branch at the Centers for Disease Control and Prevention (CDC) to act with and on its behalf to collect health information on children with birth defects. The Programme 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, and tertiary care centers are used to ascertained malformed infants born in the defined area with a follow-up to age six years.

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

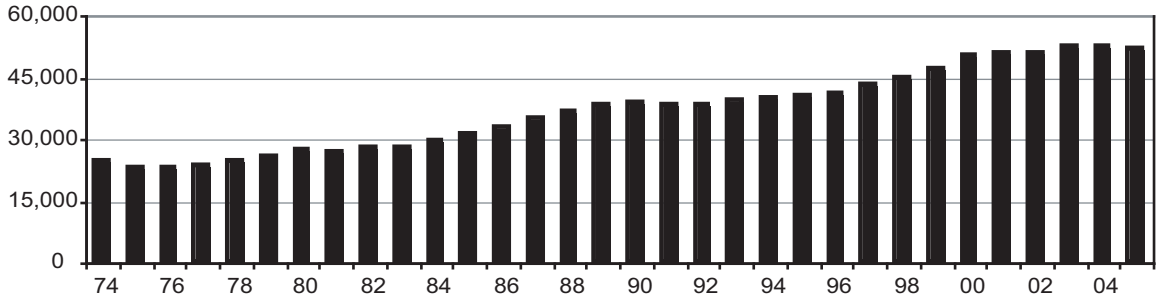
**Background information:**
Demographic information, reproductive history, gestational age, birth weight, and pregnancy outcome information available from vital records for cases and live births and stillbirths.

**Addresses and Staff:**
Adolfo Correa, MD, PhD
Csaba Siffel, MD, PhD
National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention
Mailstop E-86, 1600 Clifton Road
Atlanta, GA 30333, USA
**Phone:** 1-404-498-4090
**Fax:** 1-404-498 3040
**E-mail:** ACorrea@cdc.gov;
CSiffel@cdc.gov

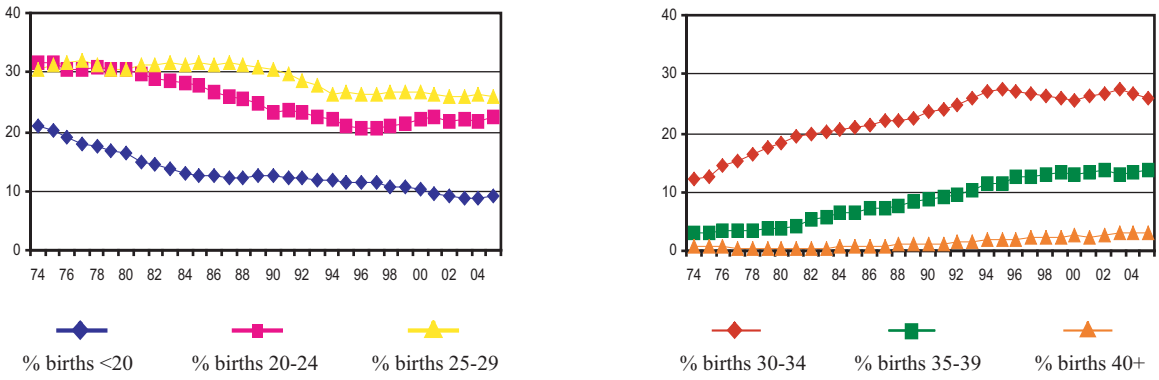
**Web:** http://www.cdc.gov/ncbddd/bd/macdp.htm

## USA-Atlanta: MACDP

**Total births by year**




**Percentage of births by year and maternal age**




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 16 | 57.1 | Cystic kidney | 3 | 3.8 |
| Spina bifida | 18 | 32.1 | Limb reduction defects | 2 | 4.2 |
| Encephalocele | 5 | 23.8 | Diaphragmatic hernia | 2 | 4.3 |
| Holoprosencephaly | 2 | 15.4 | Omphalocele | 3 | 11.5 |
| Hydrocephaly | 2 | 2.4 | Gastroschisis | 0 | 0.0 |
| Hypoplastic left heart syndrome | 0 | 0.0 | Trisomy 13 | 7 | 30.4 |
| Cleft palate without cleft lip | 1 | 1.4 | Trisomy 18 | 28 | 42.4 |
| Cleft lip with or without cleft palate | 5 | 3.5 | Down syndrome | 57 | 21.4 |
| Renal agenesis | 4 | 36.4 | | | |

Total ToPs with births defects = Not reported
*ToPs/ToPs+Births

# USA-Atlanta: MACDP, 2005

| | |
|---|---|
| Live births (LB) | 51,514 |
| Stillbirths (SB) | 546 |
| Total births | 52,060 |
| Number of terminations of pregnancy (ToP) for birth defects | nr |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 3 | 2 | 6 | 2.11 |
| Spina bifida | 16 | 0 | 4 | 3.84 |
| Encephalocele | 7 | 0 | 1 | 1.54 |
| Microcephaly | 37 | 1 | 0 | 7.30 |
| Holoprosencephaly | 5 | 1 | 0 | 1.15 |
| Hydrocephaly | 25 | 4 | 0 | 5.57 |
| Anophthalmos | 5 | 1 | 0 | 1.15 |
| Microphthalmos | 5 | 0 | 0 | 0.96 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.19 |
| Microtia | 5 | 0 | 0 | 0.96 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 23 | 1 | 0 | 4.61 |
| Tetralogy of Fallot | 18 | 0 | 0 | 3.46 |
| Hypoplastic left heart syndrome | 8 | 1 | 0 | 1.73 |
| Coarctation of aorta | 28 | 0 | 0 | 5.38 |
| Choanal atresia, bilateral | 2 | 0 | 0 | 0.38 |
| Cleft palate without cleft lip | 25 | 0 | 0 | 4.80 |
| Cleft lip with or without cleft palate | 42 | 1 | 1 | 8.45 |
| Oesophageal atresia/stenosis with or without fistula | 15 | 0 | 0 | 2.88 |
| Small intestine atresia/stenosis | 9 | 0 | 0 | 1.73 |
| Anorectal atresia/stenosis | 11 | 0 | 3 | 2.69 |
| Undescended testis (36 weeks of gestation or later) | 17 | 0 | 0 | 3.27 |
| Hypospadias | 19 | 0 | 0 | 3.65 |
| Epispadias | 2 | 0 | 0 | 0.38 |
| Indeterminate sex | 5 | 1 | 0 | 1.15 |
| Renal agenesis | 1 | 0 | 1 | 0.38 |
| Cystic kidney | 22 | 0 | 1 | 4.42 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 12 | 0 | 0 | 2.31 |
| Total Limb reduction defects (include unspecified) | 13 | 0 | 1 | 2.69 |
| Transverse | 3 | 0 | 0 | 0.58 |
| Preaxial | 3 | 0 | 1 | 0.77 |
| Postaxial | 2 | 0 | 0 | 0.38 |
| Intercalary | 2 | 0 | 0 | 0.38 |
| Mixed | 0 | 0 | 0 | 0.00 |
| Unspecified | 2 | 0 | 0 | 0.38 |
| Diaphragmatic hernia | 16 | 0 | 1 | 3.27 |
| Omphalocele | 9 | 2 | 1 | 2.31 |
| Gastroschisis | 13 | 2 | 0 | 2.88 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 1 | 1 | 0 | 0.38 |
| Trisomy 13 | 4 | 2 | 0 | 1.15 |
| Trisomy 18 | 8 | 1 | 5 | 2.69 |
| Down syndrome, all ages (include age unknown) | 67 | 0 | 13 | 15.37 |
| <20 | 1 | 0 | 0 | 2.13 |
| 20-24 | 9 | 0 | 1 | 8.57 |
| 25-29 | 4 | 0 | 0 | 2.96 |
| 30-34 | 18 | 0 | 2 | 14.82 |
| 35-39 | 23 | 0 | 5 | 39.11 |
| 40-44 | 12 | 0 | 5 | 119.30 |
| 45+ | 0 | 0 | 0 | 0.00 |
| unknown | 0 | 0 | 0 | 0.00 |

# USA-Atlanta: MACDP, Previous years rates 1974 - 2005

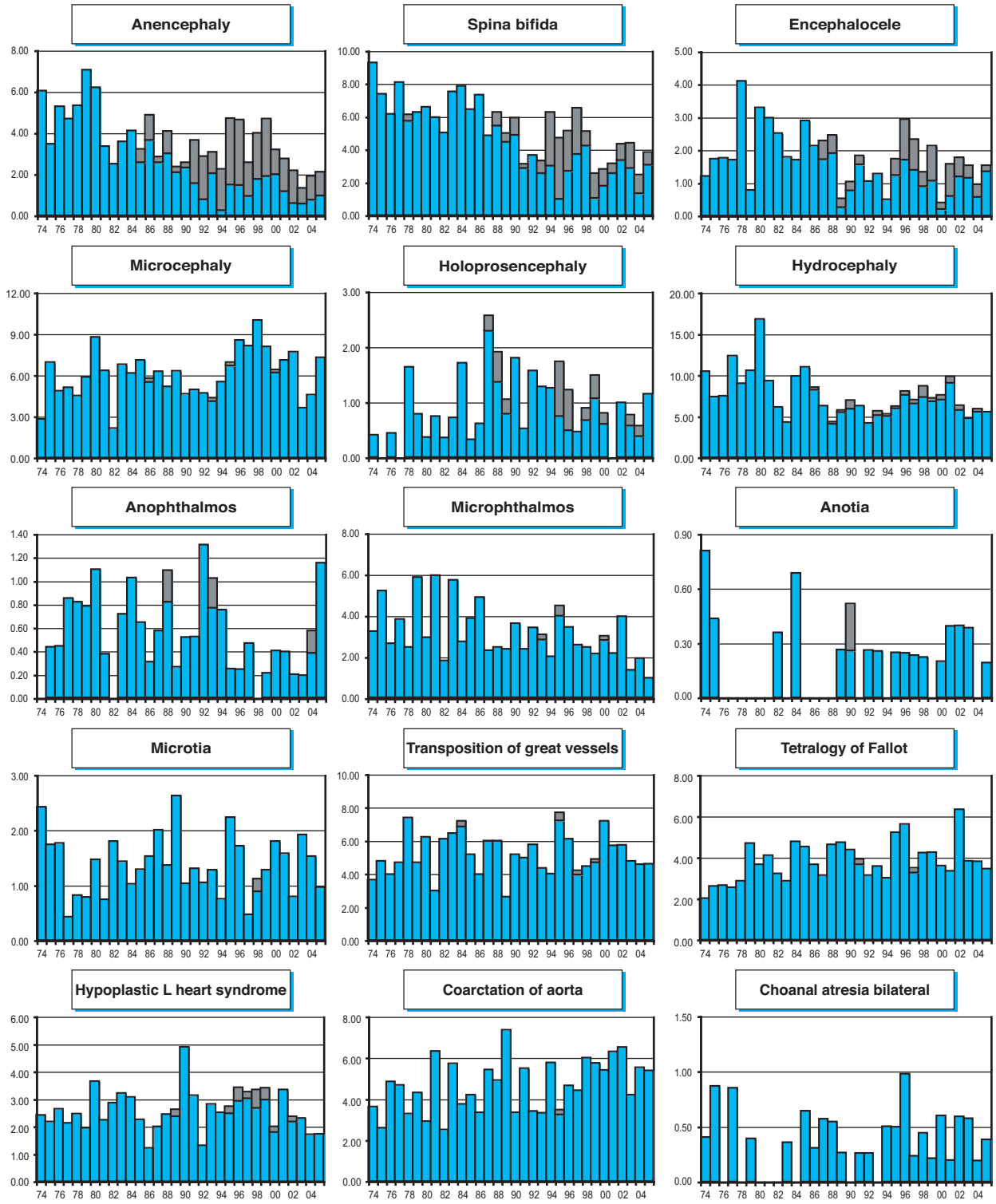
Birth prevalence rates: (LB+SB+TOP) * 10,000

| | 1975-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | **171,284** | **142,873** | **181,304** | **195,642** | **225,770** | **257,942** |
| Anencephaly | 5.49 | 3.36 | 3.31 | 3.32 | 3.81 | 2.05 |
| Spina bifida | 7.12 | 6.58 | 5.85 | 4.24 | 4.34 | 3.64 |
| Encephalocele | 2.10 | 2.38 | 1.65 | 1.28 | 1.77 | 1.47 |
| Microcephaly | 5.60 | 5.74 | 5.63 | 5.32 | 8.19 | 6.05 |
| Holoprosencephaly | 0.53 | 0.77 | 1.60 | 1.28 | 0.97 | 0.70 |
| Hydrocephaly | 10.74 | 8.19 | 6.34 | 5.52 | 7.71 | 6.47 |
| Anophthalmos | 0.64 | 0.56 | 0.55 | 0.77 | 0.27 | 0.50 |
| Microphthalmos | 3.74 | 3.99 | 3.09 | 3.07 | 2.70 | 2.05 |
| Unspecified Anophthalmos / Microphthalmos | --- | --- | --- | --- | --- | --- |
| Anotia | 0.18 | 0.21 | 0.17 | 0.15 | 0.18 | 0.27 |
| Microtia | 1.34 | 1.26 | 1.71 | 1.33 | 1.28 | 1.36 |
| Unspecified Anotia/Microtia | --- | --- | --- | --- | --- | --- |
| Transposition of great vessels | 5.08 | 5.60 | 4.74 | 5.37 | 5.40 | 5.08 |
| Tetralogy of Fallot | 3.04 | 3.92 | 4.14 | 3.78 | 4.21 | 4.15 |
| Hypoplastic left heart syndrome | 2.51 | 2.73 | 2.70 | 2.50 | 3.06 | 2.29 |
| Coarctation of aorta | 3.74 | 4.48 | 4.91 | 4.29 | 5.27 | 5.58 |
| Choanal atresia, bilateral | 0.35 | 0.21 | 0.33 | 0.31 | 0.49 | 0.39 |
| Cleft palate without cleft lip | 7.01 | 4.06 | 5.46 | 4.91 | 6.20 | 5.23 |
| Cleft lip with or without cleft palate | 11.56 | 11.13 | 9.49 | 9.05 | 9.08 | 8.65 |
| Oesophageal atresia/stenosis with or without fistula | 2.34 | 2.80 | 2.04 | 2.35 | 1.99 | 2.05 |
| Small intestine atresia/stenosis | 1.69 | 1.40 | 1.82 | 1.64 | 1.95 | 1.74 |
| Anorectal atresia/stenosis | 4.55 | 3.78 | 4.08 | 3.48 | 3.63 | 2.99 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr | 12.58 | 12.72 |
| Hypospadias | 1.11 | 2.17 | 4.91 | 4.80 | 8.95 | 7.33 |
| Epispadias | 0.99 | 0.91 | 0.55 | 0.61 | 0.35 | 0.43 |
| Indeterminate sex | 2.39 | 1.26 | 1.38 | 1.18 | 1.33 | 1.51 |
| Renal agenesis | 2.10 | 1.82 | 1.16 | 1.33 | 1.11 | 0.89 |
| Cystic kidney | 2.34 | 3.43 | 4.14 | 5.21 | 5.94 | 5.78 |
| Bladder exstrophy | 0.53 | 0.14 | 0.22 | 0.31 | 0.09 | 0.12 |
| Polydactyly, preaxial | 1.93 | 1.68 | 3.31 | 2.96 | 2.35 | 2.33 |
| Total Limb reduction defects (include unspecified) | 6.01 | 4.20 | 4.41 | 5.93 | 6.42 | 4.23 |
| Transverse | 3.68 | 3.01 | 2.65 | 3.99 | 3.28 | 2.44 |
| Preaxial | 1.11 | 0.49 | 0.72 | 1.02 | 1.33 | 0.78 |
| Postaxial | 0.23 | 0.14 | 0.33 | 0.36 | 0.27 | 0.19 |
| Intercalary | 0.53 | 0.21 | 0.33 | 0.10 | 0.31 | 0.16 |
| Mixed | 0.12 | 0.28 | 0.28 | 0.26 | 0.97 | 0.47 |
| Unspecified | --- | --- | --- | --- | --- | --- |
| Diaphragmatic hernia | 2.57 | 1.96 | 3.09 | 2.10 | 2.44 | 2.99 |
| Omphalocele | 3.85 | 3.29 | 2.70 | 2.66 | 2.70 | 1.67 |
| Gastroschisis | 1.63 | 1.89 | 2.59 | 2.56 | 2.26 | 2.83 |
| Unspecified Omphalocele/Gastroschisis | --- | --- | --- | --- | --- | --- |
| Prune belly sequence | 0.76 | 0.28 | 0.61 | 0.20 | 0.31 | 0.50 |
| Trisomy 13 | 1.23 | 1.12 | 1.65 | 1.33 | 1.90 | 1.74 |
| Trisomy 18 | 0.70 | 2.17 | 1.93 | 2.61 | 4.65 | 4.50 |
| Down syndrome, all ages (include age unknown) | 9.28 | 10.64 | 10.59 | 14.01 | 17.36 | 17.29 |
| <20 | 11.25 | 5.65 | 7.98 | 6.88 | 8.64 | 6.44 |
| 20-24 | 8.44 | 6.64 | 7.90 | 7.96 | 9.01 | 5.80 |
| 25-29 | 9.54 | 8.02 | 6.93 | 7.92 | 7.17 | 7.01 |
| 30-34 | 13.97 | 15.97 | 12.56 | 11.85 | 15.02 | 14.27 |
| 35-39 | 36.73 | 19.46 | 23.00 | 36.64 | 42.49 | 47.58 |
| 40-44 | 0.00 | 99.88 | 55.93 | 89.92 | 124.78 | 125.27 |
| 45+ | 0.00 | 0.00 | 0.00 | 425.53 | 301.72 | 55.56 |
| unknown | --- | --- | --- | --- | --- | --- |

nr = not reported

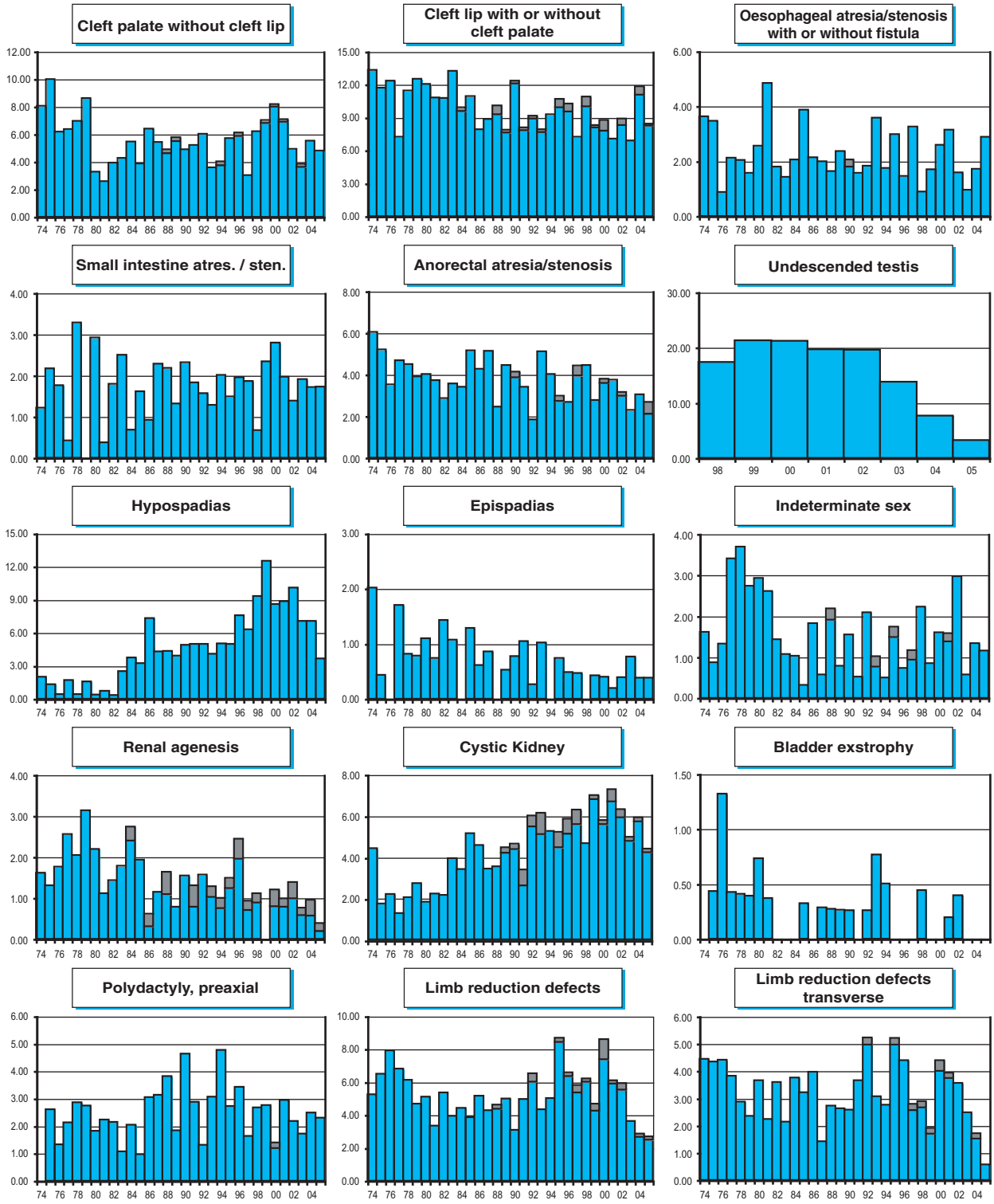
## USA-Atlanta: MACDP

'Time trends 1974-2005 (Birth prevalence rates per 10,000)



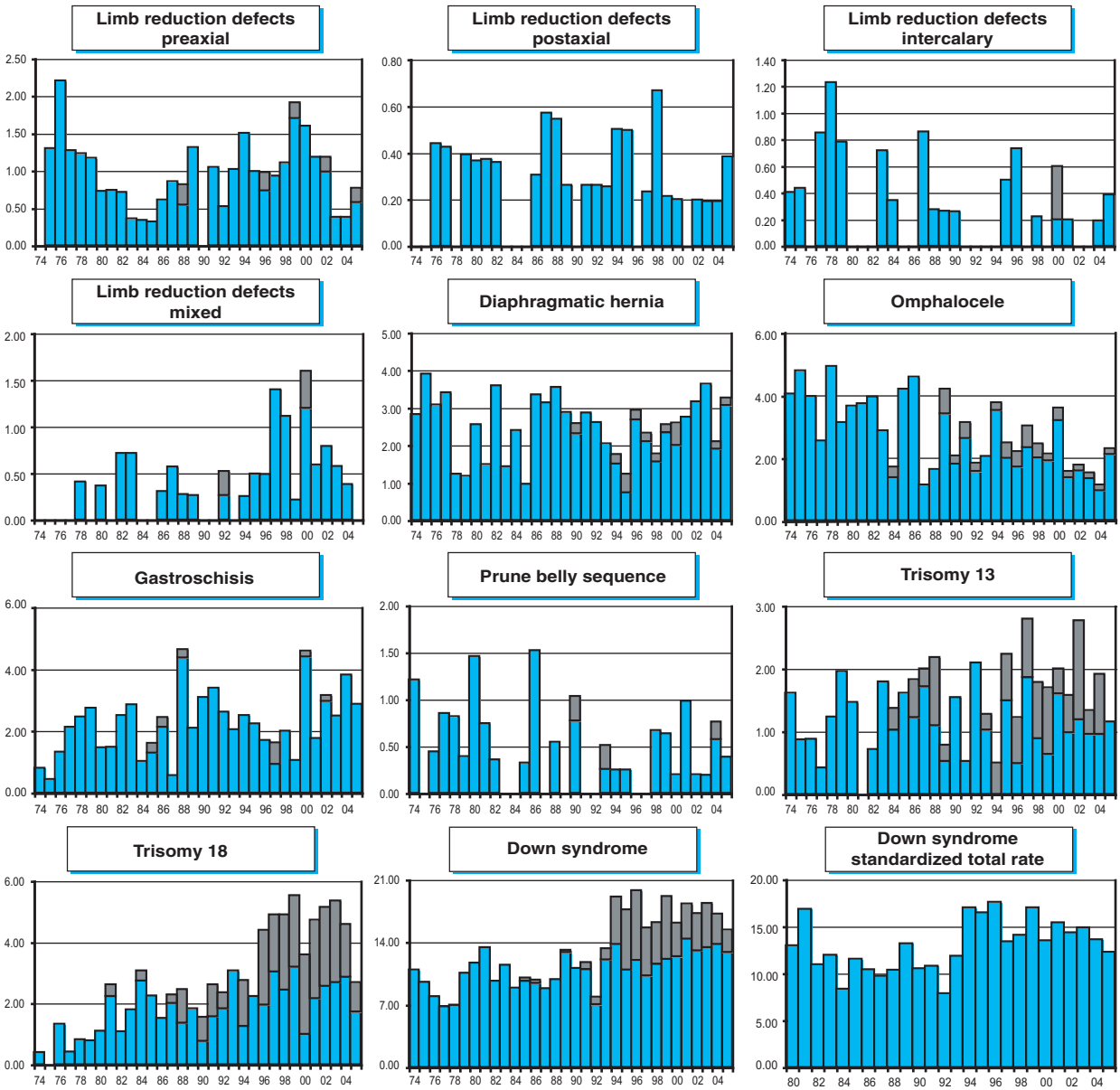

**Note:** L+S rates, ToP rates

## USA-Atlanta: MACDP




**Note:** L+S rates, ToP rates

## USA-Atlanta: MACDP




**Note:** L+S rates, ToP rates

## USA-Texas: BDES

Texas Birth Defects Epidemiology and Surveillance Branch

**History:**
BDES (Birth Defects Epidemiology and Surveillance) was established after an unusual cluster of anencephaly cases that occurred in Brownsville, Texas in 1991. Epidemiologic investigations revealed a higher than expected rate of neural tube defects among 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 1995, 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 age six 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.

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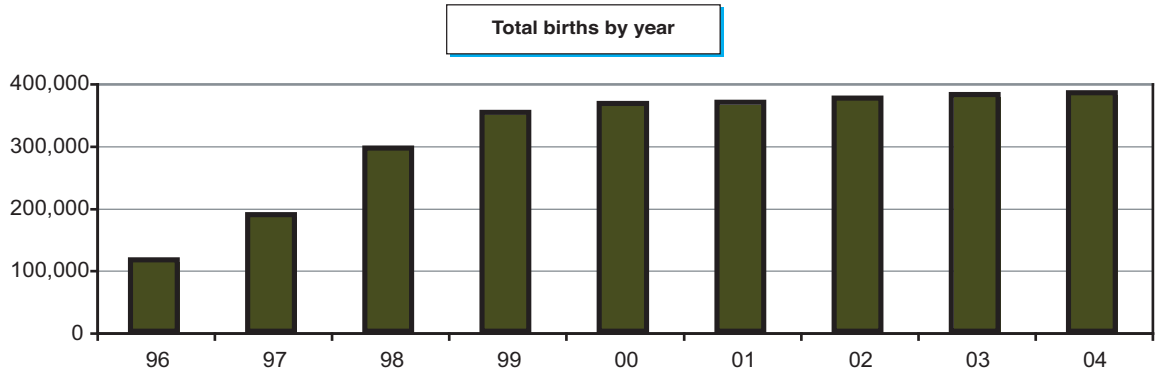
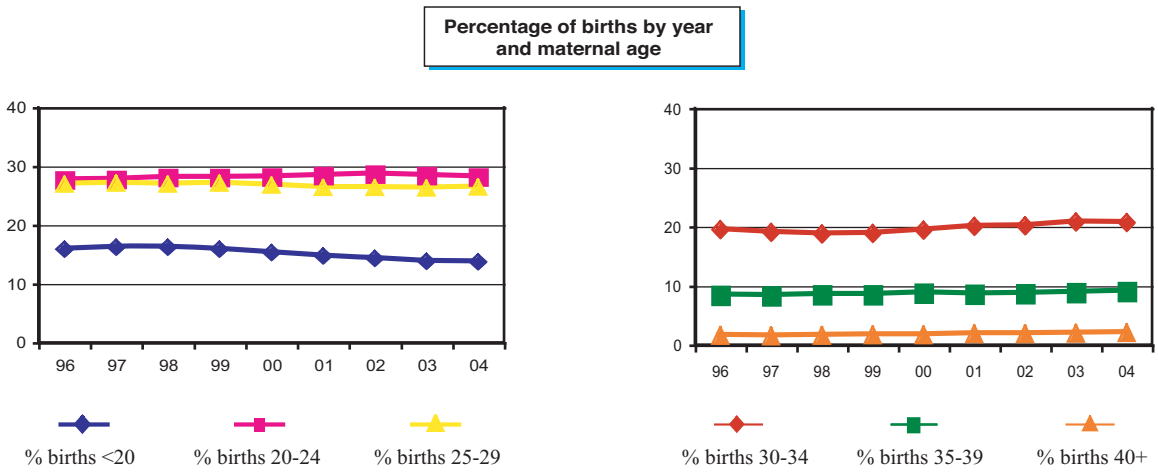
## USA-Texas: BDES




Terminations of Pregnancy (ToPs) in selected malformations (2002-2004)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 98 | 34.9 | Cystic kidney | 12 | 2.2 |
| Spina bifida | 20 | 4.9 | Limb reduction defects | 20 | 3.5 |
| Encephalocele | 10 | 12.2 | Diaphragmatic hernia | 4 | 1.3 |
| Holoprosencephaly | 15 | 11.0 | Omphalocele | 22 | 9.6 |
| Hydrocephaly | 21 | 3.0 | Gastroschisis | 12 | 2.5 |
| Hypoplastic left heart syndrome | 2 | 0.8 | Trisomy 13 | 18 | 13.8 |
| Cleft palate without cleft lip | 12 | 2.1 | Trisomy 18 | 61 | 23.8 |
| Cleft lip with or without cleft palate | 23 | 1.9 | Down syndrome | 63 | 4.5 |
| Renal agenesis | 19 | 8.7 | | | |

Total ToPs with births defects = 557 (Ratio ToPs/Births: 0.49 per 1,000)
*ToPs/ToPs+Births

# USA-Texas: BDES, 2004

| | |
|---|---|
| Live births (LB) | 380,905 |
| Stillbirths (SB) | 2,287 |
| Total births | 383,192 |
| Number of terminations of pregnancy (ToP) for birth defects | 184 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 28 | 31 | 27 | 2.24 |
| Spina bifida | 158 | 7 | 7 | 4.49 |
| Encephalocele | 19 | 0 | 2 | 0.55 |
| Microcephaly | 328 | 5 | 1 | 8.72 |
| Holoprosencephaly | 27 | 4 | 7 | 0.99 |
| Hydrocephaly | 218 | 9 | 4 | 6.03 |
| Anophthalmos | 8 | 2 | 1 | 0.29 |
| Microphthalmos | 86 | 0 | 2 | 2.30 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 12 | 0 | 0 | 0.31 |
| Microtia | 113 | 1 | 1 | 3.00 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 165 | 2 | 0 | 4.36 |
| Tetralogy of Fallot | 102 | 0 | 0 | 2.66 |
| Hypoplastic left heart syndrome | 76 | 1 | 0 | 2.01 |
| Coarctation of aorta | 166 | 1 | 1 | 4.38 |
| Choanal atresia, bilateral | 28 | 0 | 0 | 0.73 |
| Cleft palate without cleft lip | 192 | 1 | 2 | 5.09 |
| Cleft lip with or without cleft palate | 408 | 22 | 9 | 11.46 |
| Oesophageal atresia/stenosis with or without fistula | 60 | 0 | 0 | 1.57 |
| Small intestine atresia/stenosis | 65 | 0 | 1 | 1.72 |
| Anorectal atresia/stenosis | 159 | 5 | 2 | 4.33 |
| Undescended testis (36 weeks of gestation or later) | 399 | 3 | 1 | 10.52 |
| Hypospadias | 619 | 3 | 1 | 16.26 |
| Epispadias | 25 | 0 | 0 | 0.65 |
| Indeterminate sex | 16 | 11 | 4 | 0.81 |
| Renal agenesis | 61 | 8 | 4 | 1.91 |
| Cystic kidney | 197 | 1 | 5 | 5.30 |
| Bladder exstrophy | 10 | 0 | 1 | 0.29 |
| Polydactyly, preaxial | 111 | 2 | 2 | 3.00 |
| Total Limb reduction defects (include unspecified) | 164 | 9 | 7 | 4.70 |
| Transverse | 87 | 9 | 5 | 2.64 |
| Preaxial | 40 | 0 | 0 | 1.04 |
| Postaxial | 2 | 0 | 0 | 0.05 |
| Intercalary | 1 | 0 | 0 | 0.03 |
| Mixed | 29 | 0 | 1 | 0.78 |
| Unspecified | 5 | 0 | 1 | 0.16 |
| Diaphragmatic hernia | 92 | 5 | 1 | 2.56 |
| Omphalocele | 60 | 12 | 6 | 2.04 |
| Gastroschisis | 149 | 7 | 4 | 4.18 |
| Unspecified Omphalocele/Gastroschisis | 16 | 5 | 1 | 0.57 |
| Prune belly sequence | 12 | 0 | 0 | 0.31 |
| Trisomy 13 | 30 | 10 | 5 | 1.17 |
| Trisomy 18 | 51 | 21 | 22 | 2.45 |
| Down syndrome, all ages (include age unknown) | 457 | 18 | 16 | 12.81 |
| <20 | 38 | 1 | 1 | 7.63 |
| 20-24 | 64 | 1 | 0 | 6.02 |
| 25-29 | 63 | 3 | 1 | 6.62 |
| 30-34 | 98 | 2 | 6 | 13.39 |
| 35-39 | 112 | 7 | 3 | 35.40 |
| 40-44 | 77 | 4 | 5 | 117.50 |
| 45+ | 5 | 0 | 0 | 98.23 |
| unknown | 0 | 0 | 0 | 0.00 |

# USA-Texas: BDES, Previous years rates 1996 - 2004

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

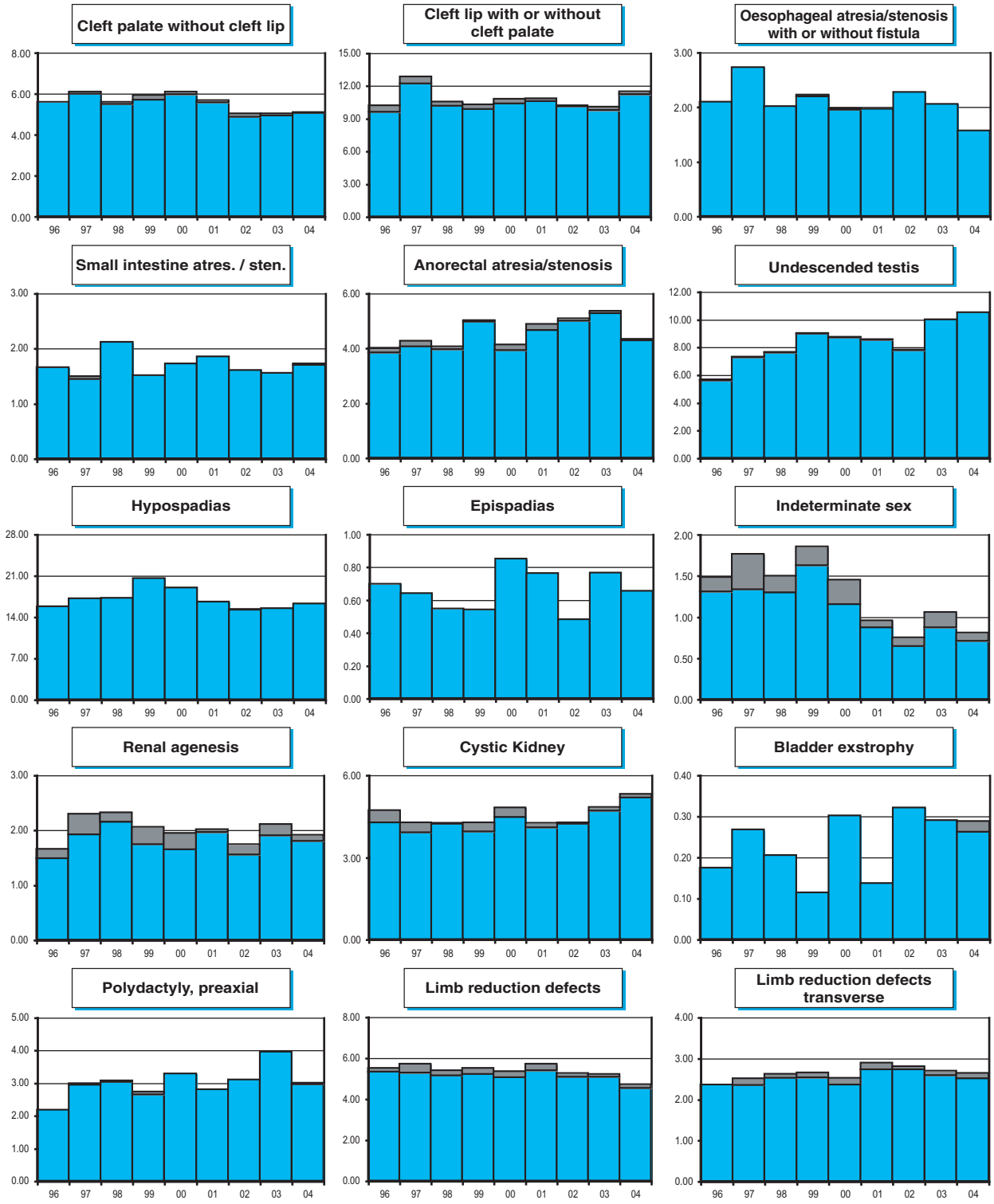
| | 1975-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000 | 2001-2004 |
|---|---|---|---|---|---|---|
| **Births** | | | | | **1,312,707** | **1,504,891** |
| Anencephaly | | | | | 3.23 | 2.51 |
| Spina bifida | | | | | 4.33 | 3.52 |
| Encephalocele | | | | | 1.06 | 0.77 |
| Microcephaly | | | | | 6.41 | 7.54 |
| Holoprosencephaly | | | | | 1.33 | 1.22 |
| Hydrocephaly | | | | | 7.24 | 6.03 |
| Anophthalmos | | | | | 0.36 | 0.30 |
| Microphthalmos | | | | | 2.49 | 2.53 |
| Unspecified Anophthalmos/Microphthalmos | | | | | --- | --- |
| Anotia | | | | | 0.22 | 0.29 |
| Microtia | | | | | 2.48 | 2.64 |
| Unspecified Anotia/Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | 4.84 | 4.78 |
| Tetralogy of Fallot | | | | | 2.99 | 3.35 |
| Hypoplastic left heart syndrome | | | | | 2.13 | 2.03 |
| Coarctation of aorta | | | | | 4.56 | 4.72 |
| Choanal atresia, bilateral | | | | | 1.26 | 1.01 |
| Cleft palate without cleft lip | | | | | 5.88 | 5.20 |
| Cleft lip with or without cleft palate | | | | | 10.82 | 10.63 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 2.16 | 1.96 |
| Small intestine atresia/stenosis | | | | | 1.71 | 1.68 |
| Anorectal atresia/stenosis | | | | | 4.36 | 4.90 |
| Undescended testis (36 weeks of gestation or later) | | | | | 8.08 | 9.22 |
| Hypospadias | | | | | 18.44 | 15.87 |
| Epispadias | | | | | 0.66 | 0.66 |
| Indeterminate sex | | | | | 1.61 | 0.89 |
| Renal agenesis | | | | | 2.08 | 1.94 |
| Cystic kidney | | | | | 4.46 | 4.66 |
| Bladder exstrophy | | | | | 0.21 | 0.26 |
| Polydactyly, preaxial | | | | | 2.95 | 3.22 |
| Total Limb reduction defects (include unspecified) | | | | | 5.45 | 5.22 |
| Transverse | | | | | 2.56 | 2.75 |
| Preaxial | | | | | 1.11 | 1.06 |
| Postaxial | | | | | 0.24 | 0.22 |
| Intercalary | | | | | 0.09 | 0.13 |
| Mixed | | | | | 1.28 | 0.83 |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 2.72 | 2.58 |
| Omphalocele | | | | | 2.29 | 2.07 |
| Gastroschisis | | | | | 3.85 | 4.24 |
| Unspecified Omphalocele/Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | 0.29 | 0.29 |
| Trisomy 13 | | | | | 1.24 | 1.14 |
| Trisomy 18 | | | | | 2.37 | 2.24 |
| Down syndrome, all ages (include age unknown) | | | | | 12.23 | 12.58 |
| <20 | | | | | 7.07 | 7.30 |
| 20-24 | | | | | 6.71 | 6.54 |
| 25-29 | | | | | 7.46 | 6.70 |
| 30-34 | | | | | 12.18 | 12.00 |
| 35-39 | | | | | 35.41 | 36.26 |
| 40-44 | | | | | 114.97 | 117.70 |
| 45+ | | | | | 156.82 | 168.61 |
| unknown | | | | | --- | --- |

## USA-Texas: BDES

'Time trends 1996-2004 (Birth prevalence rates per 10,000)

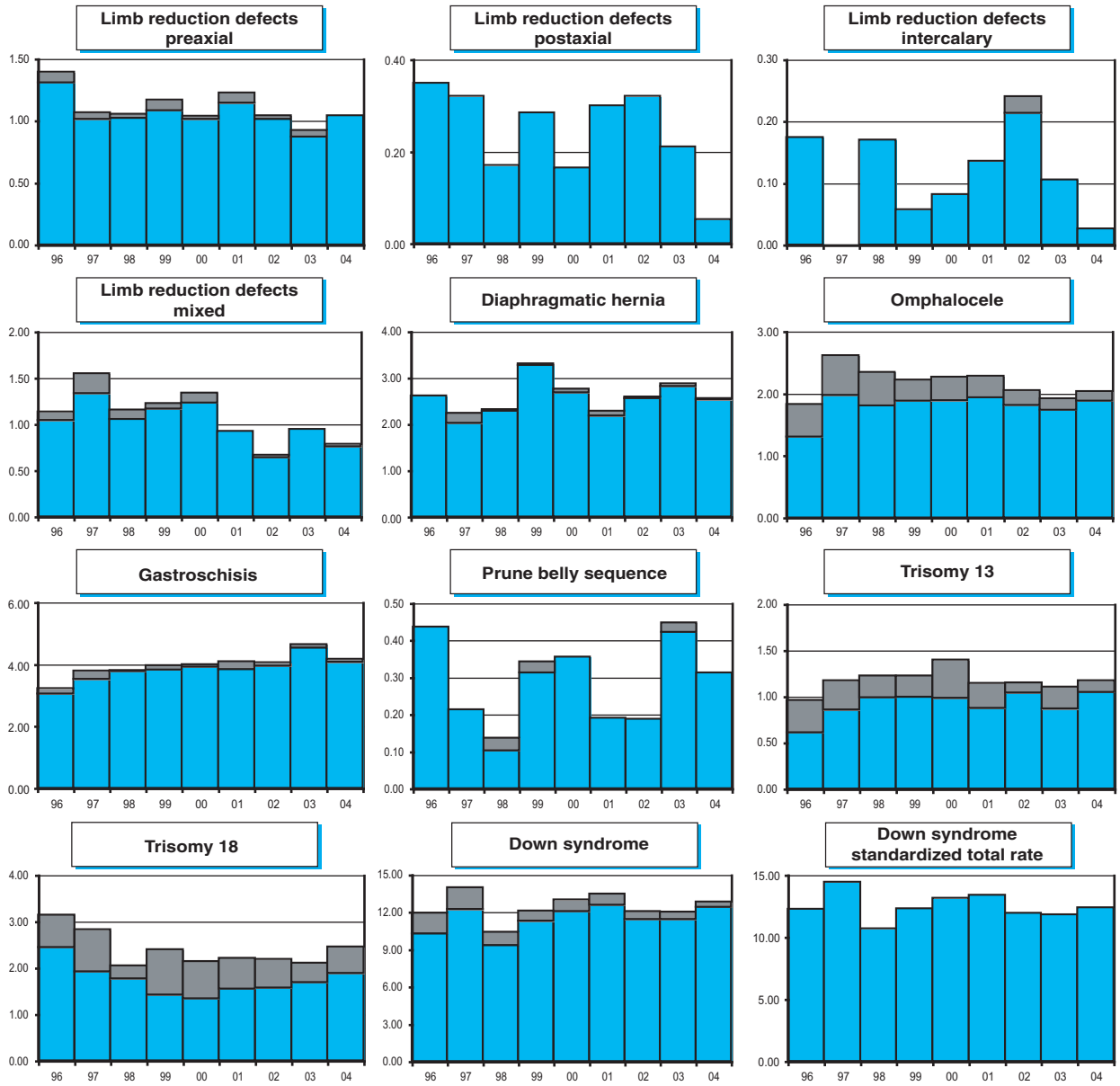

**Note:** L+S rates, ToP rates

## USA-Texas: BDES




**Note:** L+S rates, ToP rates

## USA-Texas: BDES




**Note:** L+S rates, ToP rates

## USA-Utah: UBDN

Utah Birth Defects Network (UBDN)

**History:**
The Utah Birth Defect Network (UBDN) evolved from a population-based pilot project on neural tube defects surveillance in 1994 to become a full, statewide, population-based surveillance program between 1994 and 1999. The UBDN currently monitors virtually all major structural birth defects occurring in Utah. Grants from the Centers for Disease Control and Prevention, the Utah Chapter of the March of Dimes, and Maternal Child Health Block Grants provided the funding for the UBDN to expand to its full surveillance system from the initial focus on neural tube defects (NTDs). The UBDN has ongoing legislative funding from the Utah Department of Health. In 2003 the UBDN became one of the ten Centers funded by the CDC to study causes of birth defects through the National Birth Defects Prevention Study. The UBDN is a full member of the Clearinghouse.

**Size and coverage:**
The UBDN is a state-wide population-based surveillance system that monitors approximately 50,000 births annually. All pregnancy outcomes are monitored, including stillbirths and pregnancy terminations of at least 20 weeks' gestation. Terminations less than 20 weeks with a diagnosis of a major birth defect are also included.

**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 health care providers and other agencies that voluntarily report a birth defect case to the UBDN. The UBDN has secured ongoing funding through the State Legislature for surveillance activities.

**Sources of ascertainment:**
The UBDN uses multiple sources of ascertainment, including delivery units, birth hospitals, pediatric departments, site champions, laboratories, prenatal diagnostic centers, hospital discharge data, and specialty clinics.

**Exposure information:**
Exposure information is obtained through the medical record abstraction process.

**Background information:**
General epidemiological data for all births are available through the Utah Department of Health's Office of Vital Records and Statistics. Background information for the Utah Birth Defect Network is available on the UBDN website at www.health.utah.gov/birthdefect.

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Salt Lake City, Utah 84114-4699 USA
**Phone:** 801 257 0566
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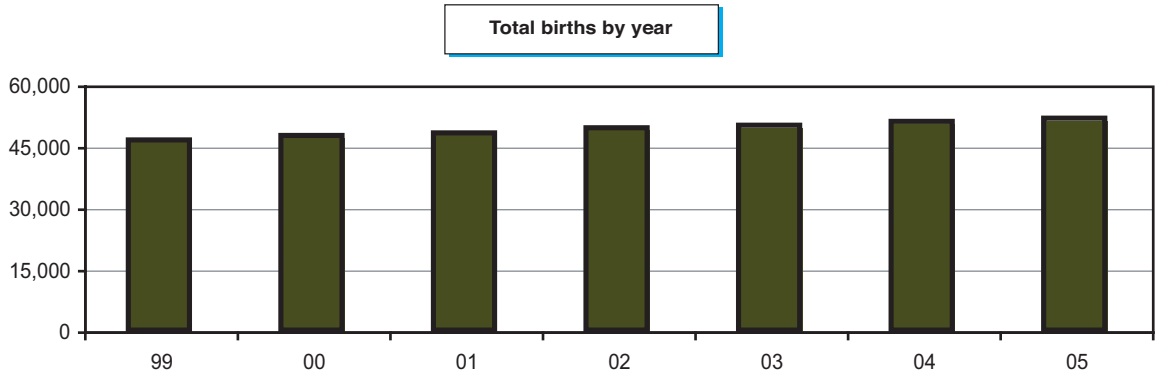
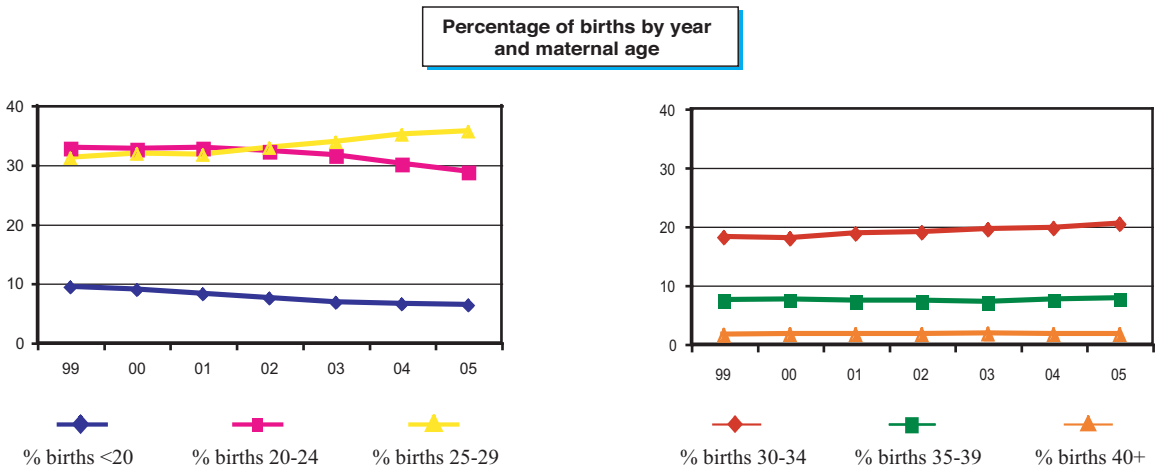
## USA-Utah: UBDN




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 21 | 56.8 | Cystic kidney | 11 | 13.1 |
| Spina bifida | 3 | 4.8 | Limb reduction defects | 11 | 10.1 |
| Encephalocele | 3 | 21.4 | Diaphragmatic hernia | 2 | 4.1 |
| Holoprosencephaly | 7 | 38.9 | Omphalocele | 8 | 23.5 |
| Hydrocephaly | 3 | 9.7 | Gastroschisis | 4 | 4.7 |
| Hypoplastic left heart syndrome | 1 | 1.9 | Trisomy 13 | 8 | 34.8 |
| Cleft palate without cleft lip | 2 | 2.0 | Trisomy 18 | 16 | 32.7 |
| Cleft lip with or without cleft palate | 13 | 6.8 | Down syndrome | 15 | 6.4 |
| Renal agenesis | 8 | 14.5 | | | |

Total ToPs with births defects§ = 94 (Ratio ToPs/Births: 0.91 per 1,000)
*ToPs/ToPs+Births

# USA-Utah: UBDN, 2005

| | |
|---|---|
| Live births (LB) | 51,517 |
| Stillbirths (SB) | 260 |
| Total births | 51,777 |
| Number of terminations of pregnancy (ToP) for birth defects | 38 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 4 | 4 | 8 | 3.09 |
| Spina bifida | 20 | 0 | 0 | 3.86 |
| Encephalocele | 3 | 2 | 1 | 1.16 |
| Microcephaly | 27 | 0 | 0 | 5.21 |
| Holoprosencephaly | 5 | 0 | 0 | 0.97 |
| Hydrocephaly | 9 | 1 | 0 | 1.93 |
| Anophthalmos | 1 | 0 | 3 | 0.77 |
| Microphthalmos | 3 | 0 | 0 | 0.58 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 2 | 0 | 0 | 0.39 |
| Microtia | 15 | 1 | 1 | 3.28 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 17 | 0 | 0 | 3.28 |
| Tetralogy of Fallot | 12 | 0 | 0 | 2.32 |
| Hypoplastic left heart syndrome | 19 | 1 | 0 | 3.86 |
| Coarctation of aorta | 34 | 1 | 0 | 6.76 |
| Choanal atresia, bilateral | 3 | 0 | 0 | 0.58 |
| Cleft palate without cleft lip | 31 | 1 | 1 | 6.37 |
| Cleft lip with or without cleft palate | 55 | 1 | 2 | 11.20 |
| Oesophageal atresia/stenosis with or without fistula | 15 | 0 | 1 | 3.09 |
| Small intestine atresia/stenosis | 12 | 0 | 0 | 2.32 |
| Anorectal atresia/stenosis | 19 | 0 | 1 | 3.86 |
| Undescended testis (36 weeks of gestation or later) | nr | nr | nr | nr |
| Hypospadias | 33 | 0 | 0 | 6.37 |
| Epispadias | 1 | 0 | 0 | 0.19 |
| Indeterminate sex | nr | nr | nr | nr |
| Renal agenesis | 13 | 3 | 3 | 3.67 |
| Cystic kidney | 25 | 0 | 4 | 5.60 |
| Bladder exstrophy | 1 | 0 | 0 | 0.19 |
| Polydactyly, preaxial | nr | nr | nr | nr |
| Total Limb reduction defects (include unspecified) | 38 | 4 | 5 | 9.08 |
| Transverse | 19 | 3 | 3 | 4.83 |
| Preaxial | 4 | 1 | 1 | 1.16 |
| Postaxial | 1 | 0 | 0 | 0.19 |
| Intercalary | 1 | 0 | 0 | 0.19 |
| Mixed | 3 | 0 | 2 | 0.97 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 16 | 0 | 1 | 3.28 |
| Omphalocele | 4 | 1 | 2 | 1.35 |
| Gastroschisis | 24 | 4 | 0 | 5.41 |
| Unspecified Omphalocele/Gastroschisis | 0 | 0 | 0 | 0.00 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 4 | 0 | 3 | 1.35 |
| Trisomy 18 | 7 | 6 | 3 | 3.09 |
| Down syndrome, all ages (include age unknown) | 64 | 4 | 3 | 13.71 |
| <20 | 3 | 0 | 0 | 9.24 |
| 20-24 | 10 | 0 | 0 | 6.72 |
| 25-29 | 17 | 2 | 0 | 10.32 |
| 30-34 | 11 | 0 | 0 | 10.47 |
| 35-39 | 14 | 1 | 3 | 45.78 |
| 40-44 | 8 | 1 | 0 | 119.21 |
| 45+ | 1 | 0 | 0 | 277.78 |
| unknown | 0 | 0 | 0 | 0.00 |

nr = not reported

# USA-Utah: UBDN, Previous years rates 1999 - 2005

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

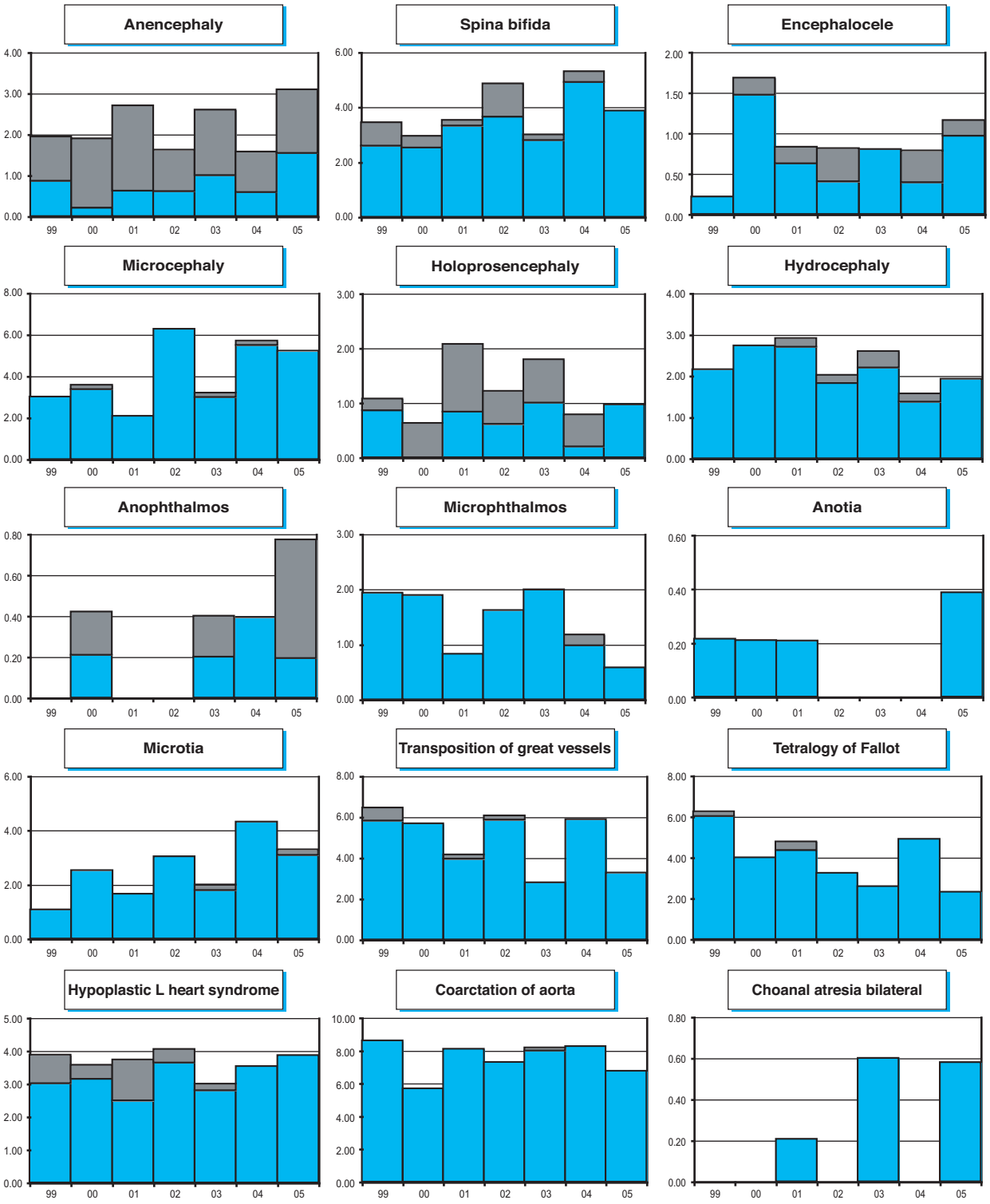
| | 1975-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000* | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | | **94,036** | **250,310** |
| Anencephaly | | | | | 1.91 | 2.32 |
| Spina bifida | | | | | 3.19 | 4.11 |
| Encephalocele | | | | | 0.96 | 0.88 |
| Microcephaly | | | | | 3.30 | 4.51 |
| Holoprosencephaly | | | | | 0.85 | 1.36 |
| Hydrocephaly | | | | | 2.45 | 2.20 |
| Anophthalmos | | | | | 0.21 | 0.32 |
| Microphthalmos | | | | | 1.91 | 1.24 |
| Unspecified Anophthalmos/Microphthalmos | | | | | --- | --- |
| Anotia | | | | | 0.21 | 0.12 |
| Microtia | | | | | 1.81 | 2.88 |
| Unspecified Anotia/Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | 6.06 | 4.43 |
| Tetralogy of Fallot | | | | | 5.10 | 3.56 |
| Hypoplastic left heart syndrome | | | | | 3.72 | 3.64 |
| Coarctation of aorta | | | | | 7.12 | 7.71 |
| Choanal atresia, bilateral | | | | | 0.00 | 0.28 |
| Cleft palate without cleft lip | | | | | 6.59 | 7.55 |
| Cleft lip with or without cleft palate | | | | | 14.99 | 13.18 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 2.76 | 2.56 |
| Small intestine atresia/stenosis | | | | | 1.28 | 1.32 |
| Anorectal atresia/stenosis | | | | | 3.19 | 3.56 |
| Undescended testis (36 weeks of gestation or later) | | | | | nr | nr |
| Hypospadias | | | | | 3.08 | 4.39 |
| Epispadias | | | | | 0.32 | 0.16 |
| Indeterminate sex | | | | | nr | nr |
| Renal agenesis | | | | | 3.30 | 3.68 |
| Cystic kidney | | | | | 4.68 | 5.71 |
| Bladder exstrophy | | | | | 0.32 | 0.20 |
| Polydactyly, preaxial | | | | | nr | nr |
| Total Limb reduction defects (include unspecified) | | | | | 5.74 | 6.55 |
| Transverse | | | | | 3.40 | 3.20 |
| Preaxial | | | | | 1.38 | 1.60 |
| Postaxial | | | | | 0.11 | 0.12 |
| Intercalary | | | | | 0.00 | 0.12 |
| Mixed | | | | | 0.53 | 1.04 |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 3.30 | 3.52 |
| Omphalocele | | | | | 2.76 | 2.44 |
| Gastroschisis | | | | | 3.72 | 5.27 |
| Unspecified Omphalocele/Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | 0.00 | 0.20 |
| Trisomy 13 | | | | | 1.17 | 1.64 |
| Trisomy 18 | | | | | 3.72 | 3.20 |
| Down syndrome, all ages (include age unknown) | | | | | 14.78 | 15.58 |
| <20 | | | | | 10.57 | 10.32 |
| 20-24 | | | | | 7.49 | 8.76 |
| 25-29 | | | | | 9.13 | 9.00 |
| 30-34 | | | | | 10.66 | 15.26 |
| 35-39 | | | | | 60.60 | 46.46 |
| 40-44 | | | | | 128.40 | 155.45 |
| 45+ | | | | | 217.39 | 531.40 |
| unspecified | | | | | --- | --- |

* data include less than 5 years

nr = not reported

## USA-Utah: UBDN

'Time trends 1999-2005 (Birth prevalence rates per 10,000)

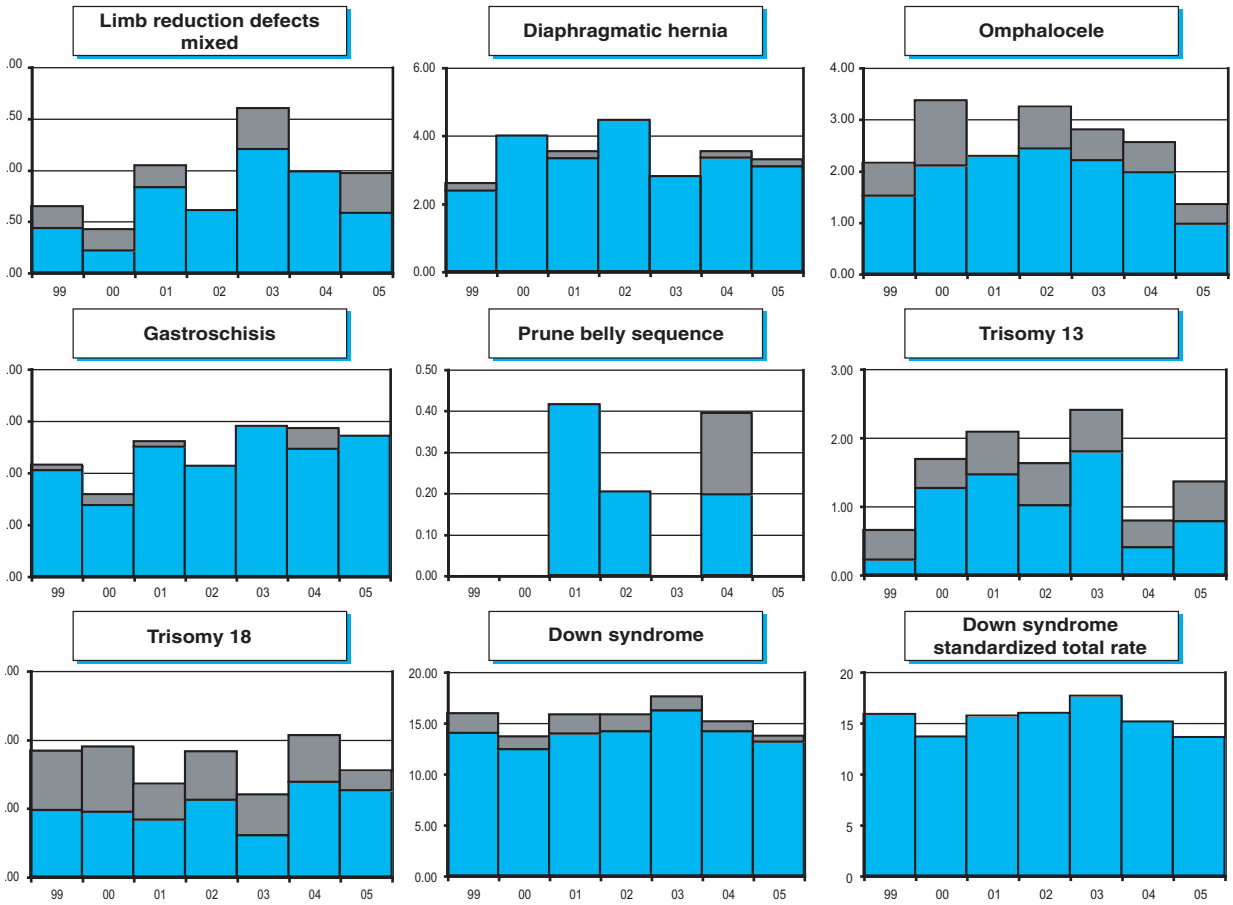



**Note:** L+S rates, ToP rates

## USA-Utah: UBDN


**Note:** L+S rates, ToP rates

## USA-Utah: UBDN




**Note:** L+S rates, ToP rates

## Wales: CARIS

Congenital Anomaly Register and Information Service (CARIS)

**History and Funding:**
Data collection commenced on 1st January 1998 and includes any baby where pregnancy ended after this date. CARIS joined EUROCAT in 1998 and ICBDSR in 2004. CARIS is based at Singleton Hospital, Swansea and is funded by the National Assembly for Wales. CARIS aims to collect data which can be used to describe the pattern of congenital anomalies across Wales. This should help:

- Build up and monitor the picture of congenital anomalies in Wales
- Assess interventions intended to help prevent or detect congenital anomalies
- Plan and co-ordinate provision of health services for affected babies and children
- Assess possible clusters of birth defects and their causes

**Population Coverage:**
The Registry covers the entire country of Wales (population-based = All mothers resident in defined geographic area) with an annual number of births of around 32,000.

**Sources of Ascertainment:**
Reporting is voluntary. The Register relies upon multi-source reporting including: antenatal clinics, delivery units, pediatric departments, ophthalmology, cytogenetics, pathology, orthopaedics, maxillo-facial and regional centres of pediatric surgery. Each delivery unit has a nominated co-ordinator to help ensure good reporting and chase for further details. CARIS staff also visit units to help with data collection. Registration covers all fetuses with prenatally diagnosed anomalies. There is no lower age of cut off, so the fetal losses and early terminations with anomalies are registered. All liveborn babies with structural anomalies are registered if diagnosed before their 1st birthday, but all chromosomal anomalies are registered, even if diagnosed later. Data exchange with the Mersey Register is also important as babies needing specialist services in North Wales are referred to Liverpool.

**Termination of Pregnancy:**
Termination of pregnancy is legal up to 24 weeks of gestation. Terminations of pregnancy are registered. If congenital anomaly is diagnosed, there is no upper gestational age limit for termination in cases of major anomaly.

**Stillbirth Definition and Early Fetal Deaths:**
Stillbirth definition: 24 weeks gestation (late fetal death after 23 completed weeks of gestation). Stillbirths of 24 weeks or more gestation are registered. Early fetal deaths/spontaneous abortions have no lower limit for inclusion on the register (earliest recorded go down to 8 weeks gestation. Autopsy rates were not given.

**Exposure Data Availability:**
Exposure information: information on maternal drug use, maternal and paternal diseases and occupations, outcomes of previous pregnancies is available. Folic acid supplementation before and during pregnancy is also collected.

**Denominators and Controls Information:**
Denominator date is obtained from the Office for National Statistics.

**Addresses and Staff:**
Dr Margery Morgan
Mr David Tucker
Congenital Anomaly Register and Information Service (CARIS)
Congenital Anomaly Register & Information Service,
Level 3 West Wing,
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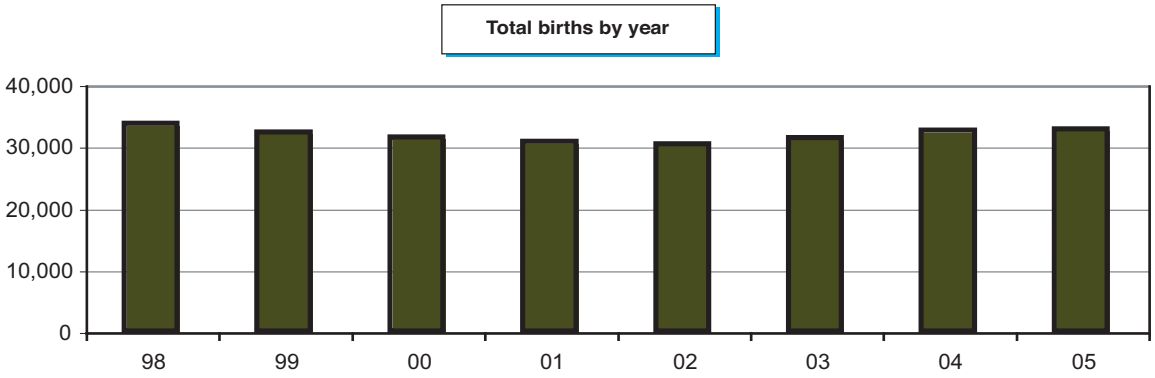
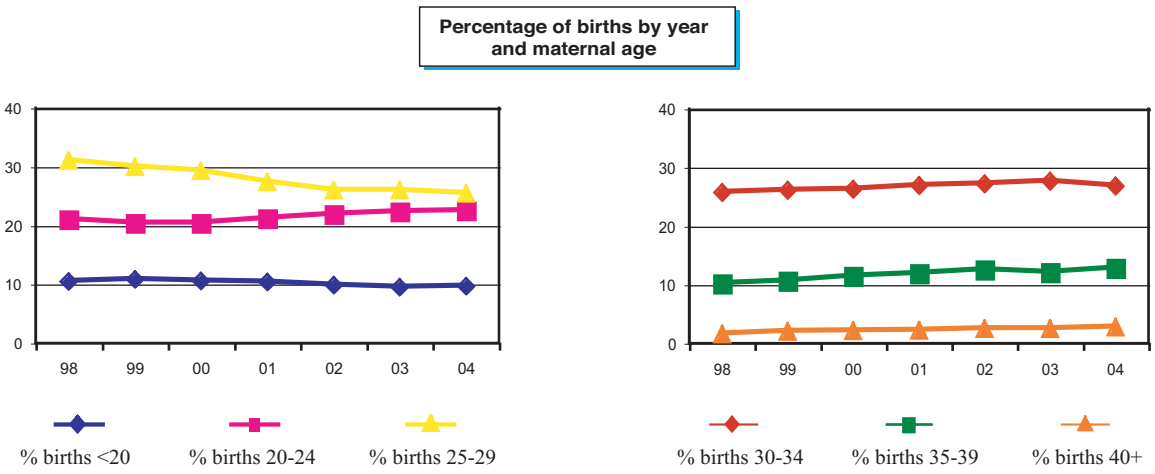
**E-mail:** dave.tucker@swansea-tr.wales.nhs.uk

## Wales: CARIS




Terminations of Pregnancy (ToPs) in selected malformations (2003-2005)
(Total cases: isolated + multiples + syndromes)

| Birth defects | ToPs | % of ToPs* | Birth defects | ToPs | % of ToPs* |
|---|---|---|---|---|---|
| Anencephaly | 53 | 94.6 | Cystic kidney | 21 | 26.6 |
| Spina bifida | 53 | 81.5 | Limb reduction defects | 18 | 25.0 |
| Encephalocele | 12 | 70.6 | Diaphragmatic hernia | 5 | 14.7 |
| Holoprosencephaly | 16 | 84.2 | Omphalocele | 24 | 55.8 |
| Hydrocephaly | 45 | 60.8 | Gastroschisis | 4 | 5.7 |
| Hypoplastic left heart syndrome | 13 | 40.6 | Trisomy 13 | 14 | 73.7 |
| Cleft palate without cleft lip | 12 | 13.2 | Trisomy 18 | 41 | 77.4 |
| Cleft lip with or without cleft palate | 13 | 12.0 | Down syndrome | 104 | 51.0 |
| Renal agenesis | 13 | 52.0 | | | |

Total ToPs with births defects = 501 (Ratio ToPs/Births: 5.19 per 1,000)
*ToPs/ToPs+Births

# Wales: CARIS, 2005

| | |
|---|---|
| Live births (LB) | 32,593 |
| Stillbirths (SB) | 175 |
| Total births | 32,768 |
| Number of terminations of pregnancy (ToP) for birth defects | 155 |

| Birth Defects | Number of cases | | | Rates*10,000 |
|---|---|---|---|---|
| | LB | SB | ToP | Total rate |
| Anencephaly | 1 | 0 | 17 | 5.49 |
| Spina bifida | 3 | 0 | 16 | 5.80 |
| Encephalocele | 2 | 0 | nr | 0.61 |
| Microcephaly | 5 | 0 | nr | 1.53 |
| Holoprosencephaly | 1 | 0 | nr | 0.31 |
| Hydrocephaly | 8 | 1 | 13 | 6.71 |
| Anophthalmos | 0 | 0 | nr | 0.00 |
| Microphthalmos | 2 | 0 | 0 | 0.61 |
| Unspecified Anophthalmos/Microphthalmos | 0 | 0 | 0 | 0.00 |
| Anotia | 1 | 0 | 0 | 0.31 |
| Microtia | 1 | 0 | 0 | 0.31 |
| Unspecified Anotia/Microtia | 0 | 0 | 0 | 0.00 |
| Transposition of great vessels | 16 | 0 | nr | 4.88 |
| Tetralogy of Fallot | 7 | 0 | nr | 2.14 |
| Hypoplastic left heart syndrome | 4 | 0 | nr | 1.22 |
| Coarctation of aorta | 16 | 0 | nr | 4.88 |
| Choanal atresia, bilateral | 0 | 0 | nr | 0.00 |
| Cleft palate without cleft lip | 35 | 0 | nr | 10.68 |
| Cleft lip with or without cleft palate | 30 | 0 | nr | 9.16 |
| Oesophageal atresia/stenosis with or without fistula | 10 | 1 | nr | 3.36 |
| Small intestine atresia/stenosis | 5 | 0 | 0 | 1.53 |
| Anorectal atresia/stenosis | 6 | 1 | nr | 2.14 |
| Undescended testis (36 weeks of gestation or later) | 5 | 0 | 0 | 1.53 |
| Hypospadias | 41 | 0 | 0 | 12.51 |
| Epispadias | 2 | 0 | 0 | 0.61 |
| Indeterminate sex | 4 | 0 | 0 | 1.22 |
| Renal agenesis | 0 | 1 | nr | 0.31 |
| Cystic kidney | 13 | 0 | nr | 3.97 |
| Bladder exstrophy | 0 | 0 | 0 | 0.00 |
| Polydactyly, preaxial | 3 | 0 | nr | 0.92 |
| Total Limb reduction defects (include unspecified) | 17 | 1 | nr | 5.49 |
| Transverse | 6 | 1 | 0 | 2.14 |
| Preaxial | 3 | 0 | nr | 0.92 |
| Postaxial | 0 | 0 | 0 | 0.00 |
| Intercalary | 2 | 0 | nr | 0.61 |
| Mixed | 6 | 0 | nr | 1.83 |
| Unspecified | 0 | 0 | 0 | 0.00 |
| Diaphragmatic hernia | 8 | 1 | nr | 2.75 |
| Omphalocele | 7 | 1 | nr | 2.44 |
| Gastroschisis | 16 | 0 | nr | 4.88 |
| Unspecified Omphalocele/Gastroschisis | 1 | 0 | nr | 0.31 |
| Prune belly sequence | 0 | 0 | 0 | 0.00 |
| Trisomy 13 | 1 | 0 | nr | 0.31 |
| Trisomy 18 | 2 | 4 | 12 | 5.49 |
| Down syndrome, all ages (include age unknown) | 42 | 1 | 33 | 23.19 |
| <20 | 0 | 1 | 0 | nr |
| 20-24 | 6 | 0 | nr | nr |
| 25-29 | 7 | 0 | nr | nr |
| 30-34 | 7 | 0 | nr | nr |
| 35-39 | 15 | 0 | 14 | nr |
| 40-44 | 5 | 0 | nr | nr |
| 45+ | 1 | 0 | nr | nr |
| unknown | 1 | 0 | 0 | 0.31 |

# Wales: CARIS, Previous years rates 1998 - 2005

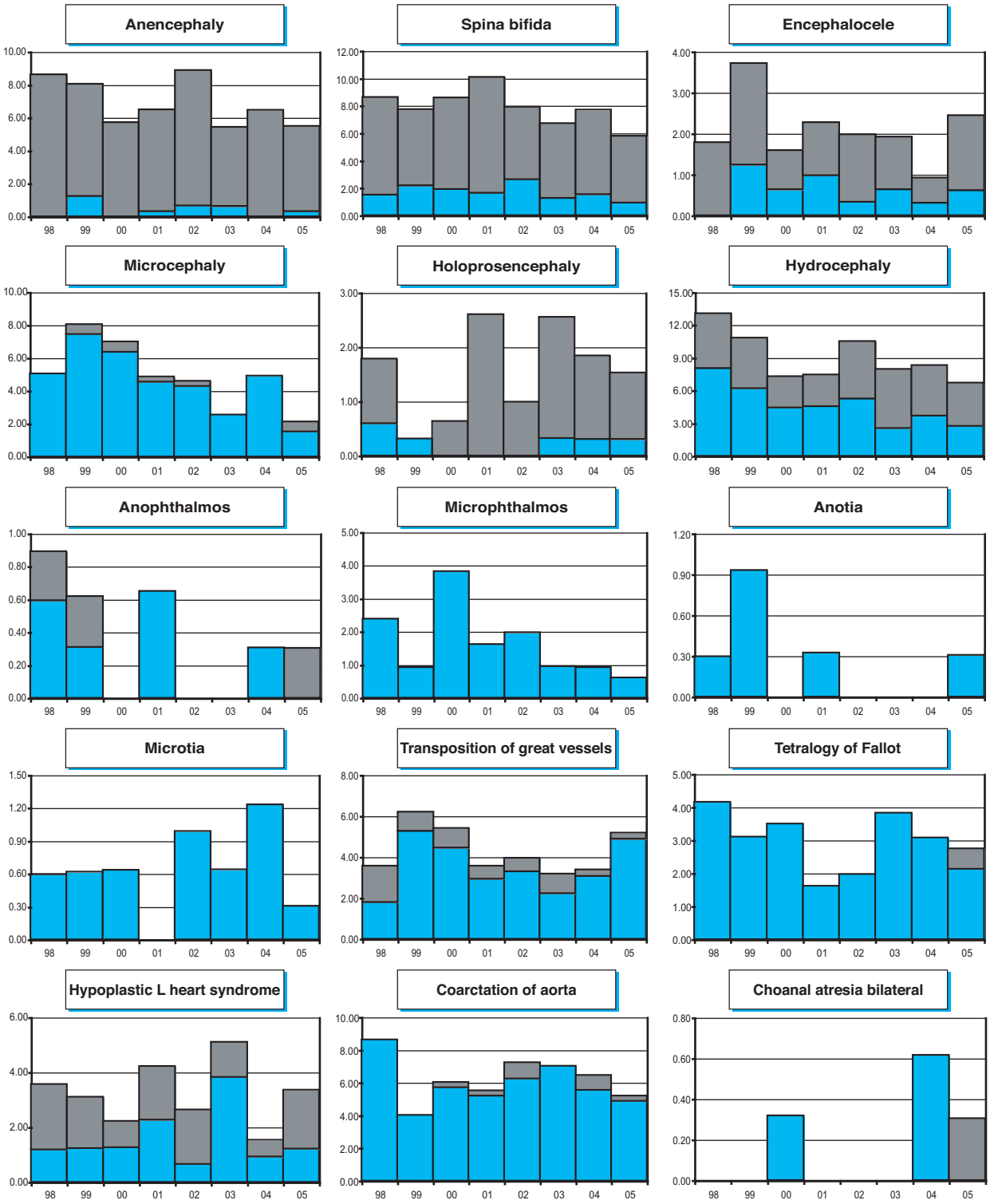
Birth prevalence rates: (LB+SB+TOP) * 10,000

| | 1975-1980 | 1981-1985 | 1986-1990 | 1991-1995 | 1996-2000* | 2001-2005 |
|---|---|---|---|---|---|---|
| **Births** | | | | | | |
| Anencephaly | | | | | 7.50 | 6.53 |
| Spina bifida | | | | | 8.32 | 7.61 |
| Encephalocele | | | | | 2.36 | 1.90 |
| Microcephaly | | | | | 6.68 | 3.80 |
| Holoprosencephaly | | | | | 0.92 | 1.90 |
| Hydrocephaly | | | | | 10.48 | 8.18 |
| Anophthalmos | | | | | 0.51 | 0.25 |
| Microphthalmos | | | | | 2.36 | 1.20 |
| Unspecified Anophthalmos/Microphthalmos | | | | | --- | --- |
| Anotia | | | | | 0.41 | 0.13 |
| Microtia | | | | | 0.62 | 0.63 |
| Unspecified Anotia/Microtia | | | | | --- | --- |
| Transposition of great vessels | | | | | 5.03 | 3.87 |
| Tetralogy of Fallot | | | | | 3.60 | 2.66 |
| Hypoplastic left heart syndrome | | | | | 2.98 | 3.36 |
| Coarctation of aorta | | | | | 6.27 | 6.28 |
| Choanal atresia, bilateral | | | | | 0.10 | 0.19 |
| Cleft palate without cleft lip | | | | | 8.84 | 9.26 |
| Cleft lip with or without cleft palate | | | | | 9.66 | 10.78 |
| Oesophageal atresia/stenosis with or without fistula | | | | | 1.03 | 1.78 |
| Small intestine atresia/stenosis | | | | | 4.01 | 1.84 |
| Anorectal atresia/stenosis | | | | | 5.03 | 3.87 |
| Undescended testis (36 weeks of gestation or later) | | | | | 3.60 | 2.09 |
| Hypospadias | | | | | 27.84 | 19.91 |
| Epispadias | | | | | 0.62 | 0.51 |
| Indeterminate sex | | | | | 0.31 | 0.76 |
| Renal agenesis | | | | | 7.19 | 3.74 |
| Cystic kidney | | | | | 10.58 | 9.45 |
| Bladder exstrophy | | | | | 0.41 | 0.19 |
| Polydactyly, preaxial | | | | | 0.51 | 0.89 |
| Total Limb reduction defects (include unspecified) | | | | | 10.17 | 8.49 |
| Transverse | | | | | 5.55 | 3.30 |
| Preaxial | | | | | 1.85 | 1.08 |
| Postaxial | | | | | 0.21 | 0.25 |
| Intercalary | | | | | 0.92 | 1.52 |
| Mixed | | | | | 0.51 | 1.65 |
| Unspecified | | | | | --- | --- |
| Diaphragmatic hernia | | | | | 3.70 | 3.68 |
| Omphalocele | | | | | 2.88 | 4.44 |
| Gastroschisis | | | | | 4.73 | 6.15 |
| Unspecified Omphalocele/Gastroschisis | | | | | --- | --- |
| Prune belly sequence | | | | | 0.10 | 0.06 |
| Trisomy 13 | | | | | 2.26 | 2.41 |
| Trisomy 18 | | | | | 4.52 | 4.94 |
| Down syndrome, all ages (include age unknown) | | | | | 18.90 | 20.60 |
| <20 | | | | | 10.64 | 7.34* |
| 20-24 | | | | | 6.48 | 6.54* |
| 25-29 | | | | | 10.57 | 11.60* |
| 30-34 | | | | | 18.15 | 15.33* |
| 35-39 | | | | | 55.52 | 47.94* |
| 40-44 | | | | | 130.83 | 187.27* |
| 45+ | | | | | 250.00 | 201.34* |
| unknown | | | | | --- | --- |

* data include less than 5 years

## Wales: CARIS

'Time trends 1998-2005 (Birth prevalence rates per 10,000)

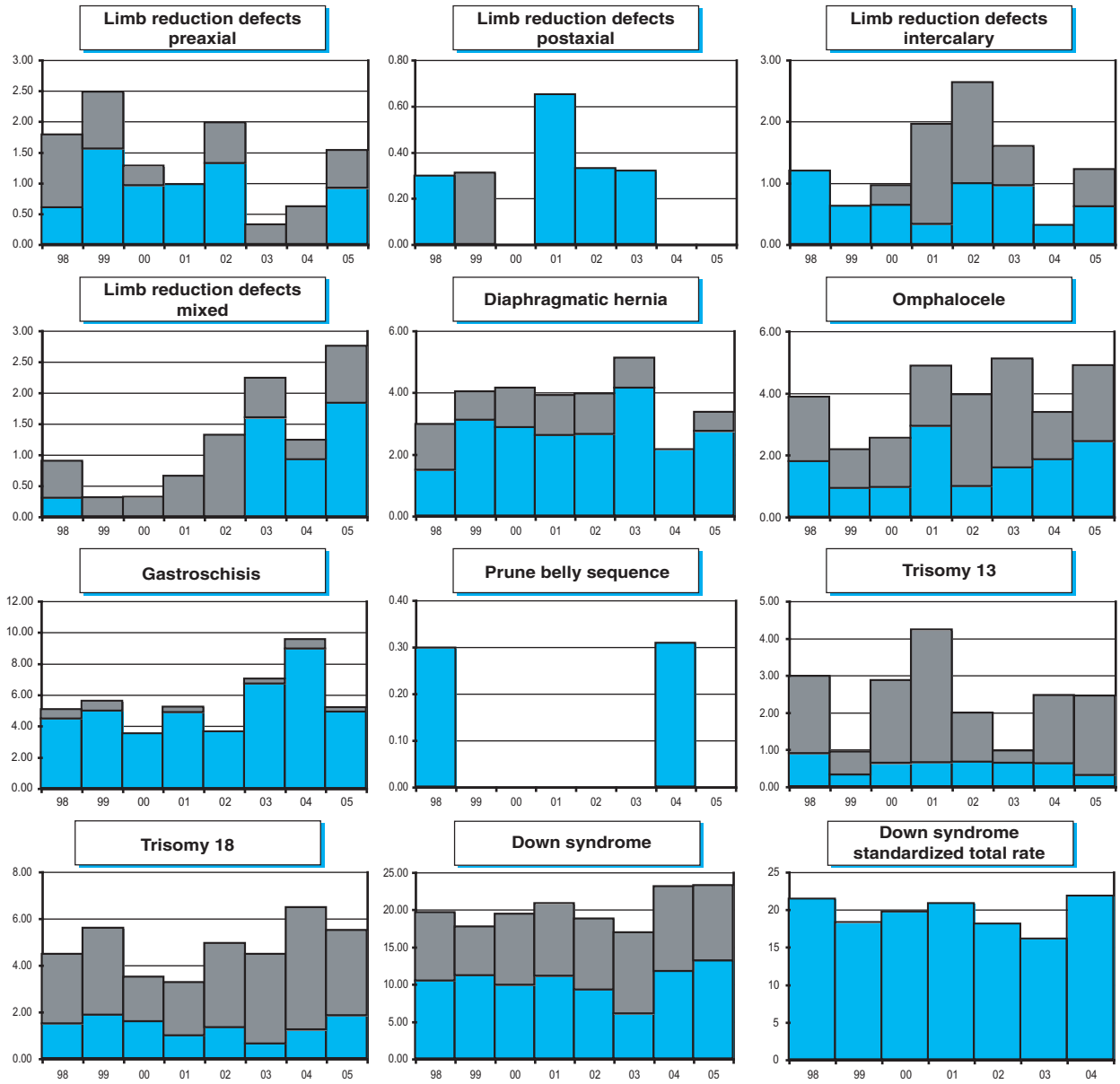



**Note:** L+S rates, ToP rates

## Wales: CARIS


**Note:** L+S rates, ToP rates

## Wales: CARIS




**Note:** L+S rates, ToP rates

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

## Australia:

Australian Congenital Malformation Monitoring System

**History:**
The mechanism for national monitoring of birth defects in Australia was established in 1981. The national programme became an associate member of the Clearinghouse in 1982 and full member in 1984. Australia has not contributed national data to the Clearinghouse for the last 2 years.
In Australia, the data have been provided to the national program by the state and territory health authorities primarily from their birth defects registers and perinatal data collections. However there are variations among state and territory data collections, in the definitions and classifications used, the duration of collection and the level of ascertainment. Therefore Australia has reviewed the existing system and has undertaken a project to develop a new system. They anticipate the resumption of data contribution to the ICBDSR in the foreseeable future.

**Size and coverage:**
All births of at least 20 weeks gestation or at least 400 grams birthweight in Australia are covered. In 2005, there were 272,419 births in Australia, an increase of 5.9% from the number reported in 2004.

**Legislation and funding:**
There is no national legislation requiring the reporting of birth defects at the national level. In some States, notification to their birth defect registry is required as part of their respective Public Health Acts. In some States and Territories, birth defect data is collected as part of another collection, and funding, if any is determined by the jurisdiction. The State and Territory Health Departments report to the AIHW National Perinatal Statistics Unit which is the national data custodian of the congenital anomalies data collection. The current funding for development of a national minimum dataset for the congenital anomalies is from the Australian Health Ministers Advisory Committee.

**Sources of ascertainment:**
The State and Territory birth defect data collections operate independently and there is enormous variation in the breadth of notification sources and level of ascertainment. Other sources of notification may include death certificates, autopsies, hospital morbidity databases, notification from health professionals, cytogenetic and prenatal screening. At the minimum, State and Territory birth defect registries and perinatal data collections send electronic notification to the central data custodian annually.

**Exposure information:**
Currently not available.

**Background information:**
In the absence of national legislation, there is variation in the scope, quality of data and ascertainment between the States and Territories. Under the current development project, Australia is working on development of an agreed national minimum data set for congenital anomalies. It's program of work also includes the development of clinical definitions for congenital anomalies a review of the scope of the collection and development of a nationally consistant classification system.

**Addresses and Staff:**
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2nd Floor, McNevin Dickson Building
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Randwick NSW 2031
Australia
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**Fax:** 61-2-93821025
**E-mail:** e.sullivan@unsw.edu.au
**Web:** www.npsu.unsw.edu.au

## South Africa: SABDSS

South Africa Birth Defects Surveillance Systems (SABDSS).

**History:**
The Programme started in 1988 and became a full member of the Clearinghouse in 1992.

**Size and coverage**:
The Programme is hospital based covering 15 sentinel sites over the country with approximately 50,000 annual birth or 5% of all births in South Africa.

**Legislation and funding:**
The Programme is funded by the Department of National Health. Participation in the Programme is voluntary.

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

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

**Background information:**
Total births for some participating hospitals are not accurately known.

**Addresses and Staff:**
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University of Cape Town-Medical School
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**Phone:** 27-21-4066482
**Fax:** 27-21-4066163
**E-mail:** db@phfm.vet.ac.za
**E-mail:** rauf@phfm.vet.ac.za

## United Arab Emirates

Program: Congenital abnormality study group

**History:**
The Program was initiated in 1992, but continuous monitoring did not begin until 1994. It is now an Associate Member of the ICBDMS.

**Size and coverage:**
The Programme covers about 8000 births a year and includes all births occurring in three major hospitals of the Al Ain Medical District, situated in the eastern part of the Abu Dhabi Emirate. It has a population of about 270,000. Still births with a weight of more than 500 gm are included.

**Legislation and funding:**
The Programme is funded by the Faculty of Medicine and Health Sciences of the UAE University.

**Sources of ascertainment:**
In each hospital, there is a neonatologist who examines, identifies abnormalities and records at birth in a template provided. The diagnosis is further assisted by a clinical geneticist/ dysmorphologist and pediatricians.

**Exposure information:**
Some basic information on exposure and maternal disease is collected in all cases.

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

**Activities**
Members of the Congenital Abnormality Study Group had regular meetings in Al Ain and discussed issues and concerns of congenital anomalies in the area. We also had meetings with speakers drawn from local scientists as well as overseas visitors. In conjunction with the Development and Genetics Priority Research Group of the Faculty of Medicine and Health Sciences, UAE University we had an international meeting this year (December 9-11, 2004). It was titled "International Genetics Congress:Global Challenge, Regional Focus on Advances in Community and Preventive Genetics. Participants came from 32 countries. There were 52 platform presentations and 114 poster presentations in all. Participants enjoyed good science and an excellent social program during the three days of the meeting. Procedings of the Congress are being published in Community Medicine as a supplementary issue with Lihadh Al Gazali (UAE) and Alan H Bittles (Edith Cowan University, Perth Australia) as Editors.

**Address for further information:**
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**Phone:** 971-3-7039 415
**Fax:** 971-3-7672022
**E-mail:** algazali@hotmail.com

Padmanabhan Krishna Rengasamy
**E-mail:** padamanabhanr@uaeu.ac.ae

*Selection of papers by Programme Directors and their collaborators are reported as following. The details are sent from the Programme Directors only for the listed Monitoring Systems. The collaborative publications, made by two or more ICBDSR members in any context, are first shown and not repeated in the specific registry section.*
*Papers can be obtained contacting authors.*

**Collaborative Publications**


Botto LD, Lisi A, Bower C, Canfield MA, Dattani N, De Vigan C, De Walle H, Erickson DJ, Halliday J, Irgens LM, Lowry RB, McDonnell R, Metneki J, Poetzsch S, Ritvanen A, Robert-Gnansia E, Siffel C, Stoll C, Mastroiacovo P. Trends of selected malformations in relation to folic acid recommendations and fortification: an international assessment. Birth Defects Res A Clin Mol Teratol. 2006; 76(10):693-705.

Botto LD, Robert-Gnansia E, Siffel C, Harris J, Borman B, Mastroiacovo P. Fostering international collaboration in birth defects research and prevention: a perspective from the International Clearinghouse for Birth Defects Surveillance and Research. Am J Public Health. 2006 May;96(5):774-80. Epub 2006 Mar 29.

Mastroiacovo P, Lisi A, Castilla EE, Martínez-Frías ML, Bermejo E, Marengo L, Kucik J, Siffel C, Halliday J, Gatt M, Annerèn G, Bianchi F, Canessa MA, Danderfer R, de Walle H, Harris J, Li Z, Lowry RB, McDonell R, Merlob P, Metneki J, Mutchinick O, Robert-Gnansia E, Scarano G, Sipek A, Pötzsch S, Szabova E, Yevtushok L. Gastroschisis and associated defects: an international study. Am J Med Genet A 2007;143:660-71.


**Australia VBDR**


Davidson N, King J, Riley M, Halliday J. Influence of pregnancy termination for birth defects on the perinatal mortality rate. Paediatric Perinatal Epidemiology, 19:50-55 (2005)

Jaques A, Sheffield L, Halliday J. Informed choice in women attending private clinics to undergo first trimester screening for Down syndrome. Prenatal Diagnosis, 25:656-664 (2005)

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