Birth Defects in Victoria 2005—2006

Victorian Perinatal Data Collection Unit
Statewide Quality Branch
Victoria 2008

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2008

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This report is also available on the Internet: http://www.health.vic.gov.au/perinatal

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Prof D. Penny	Cardiology
Assoc Prof D. Reddihough	Child Development and Rehab
Dr T. Tan	Orofacial clefts
Prof G. Werther	Endocrinolgy and Diabetes
Dr S. White	Dysmorphology

Provision of data for statistical and research purposes

Under the auspices of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM), the Victorian Perinatal Data Collection Unit (VPDCU) has, since 1982, collated information on all births in Victoria of at least 20 weeks gestation. The VPDCU also maintains the Birth Defects Register (VBDR) for Victorian children born since 1982. CCOPMM also maintains collections on perinatal, infant, child deaths (up to, but not including their 18th birthday), and maternal mortality. *CCOPMM readily provides data requested by health professionals; however, careful consideration is given to assure that the privacy of individuals is protected.*

The CCOPMM Chair reviews all requests for information from the VPDCU. If access to individual case records is requested, stringent conditions apply to safeguard security and confidentiality of any data released. Formal research proposals must conform to the National Health and Medical Research Council's *National Statement on Ethical Conduct in Research Involving Humans 1999.* Before any research project can access VPDCU data, a properly constituted Human Research Ethics Committee must have approved it.

All **requests** must be made **in writing** via e-mail, post or fax. A "Request for Access to Perinatal Data Form" may be accessed on the perinatal website at:

www.health.vic.gov.au/perinatal

All correspondence should be addressed to:

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CCOPMM encourages the use of information within this and other CCOPMM reports, providing appropriate acknowledgement of the source is made.

Abbreviations

a/s	atresia/stenosis
AIHW	Australian Institute of Health and Welfare
anom	anomalies
ASD	atrial septal defect
BPA	British Paediatric Association
CDH	congenital dislocation of hip
chrom	chromosomal
CI	confidence interval
CNS	central nervous system
COA	coarctation of aorta
DDH	developmental dysplasia of the hip
desc	described
dev	developmental
df	degrees of freedom
dysgen	dysgenesis
electr	electrolyte
end.	endocrine
GU	genitourinary
HLHS	hypoplastic left heart syndrome
inc	includes
ICD	International Classification of Diseases
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
LGA	Local Government Area
LL	lower limit
MCH Nurse	Maternal & Child Health Nurse
metab	metabolic
MHW	Mercy Hospital for Women
MMC	Monash Medical Centre
MS	musculoskeletal
nec	not elsewhere classified
NND	neonatal death
NOS	not otherwise specified
NPSU	National Perinatal Statistics Unit
nutri	nutritional
p	probability
PDA	patent ductus arteriosus
PR(s)	prevalence rate(s)
pns	peripheral nervous system
RCH	Royal Children's Hospital
red	reduction
RR	relative risk
RWH	Royal Women's Hospital
s/insuff	stenosis/insufficiency
SB	stillbirth
spec	specified
sternocleido	sternocleidomastoid
TGV	transposition of great vessels
TOP	termination of pregnancy
trans	translocation
udt	undescended testis
UL	upper limit
unspe	unspecified
VBDR	Victorian Birth Defects Register
VPDCU	Victorian Perinatal Data Collection Unit
VSD	ventricular septal defect

Definitions

A birth defect is any abnormality of prenatal origin, either present following conception or occurring before the end of pregnancy. This includes structural, functional, genetic, chromosomal and biochemical abnormalities.

Birth refers to both livebirths and stillbirths (see definitions below).

Birth defect cases refers to the number of liveborn or stillborn infants, or terminations at any gestation affected by at least one birth defect.

Birth prevalence has as the denominator the number of still and livebirths at and after 20 weeks gestation. **Overall prevalence** includes terminations of birth defects before 20 weeks gestation.

Confinements are the number of pregnancies resulting in at least one birth.
(NB: Number of confinements does not equal number of births. One confinement may result in two births ie. twins).

Isolated anomalies are defined as cases with a single condition or a single condition associated with one or more minor anomalies listed on the exclusion list (Appendix B), but which have been included periodically throughout the history of the VBDR.

Livebirth is the complete expulsion or extraction from its mother of a baby of at least 20 weeks gestation or, if gestation is unknown, weighing at least 400g who, after being born, breathes or shows any evidence of life such as a heartbeat.

Multiple different system defects are those involving two or more body systems (eg. orofacial cleft condition plus one or more defects from systems other than digestive.)

Multiple same system defects are those involving one body system only (eg. orofacial cleft condition plus another digestive system birth defect.)

Neonatal death refers to a death occurring within 28 days of livebirth in an infant whose gestation was at least 20 weeks or, if gestation is unknown, weighing at least 400g.

Perinatal death is a stillbirth or neonatal death.

Stillbirth is the complete expulsion or extraction from its mother of a baby of at least 20 weeks gestation or, if the gestation is unknown, weighing at least 400g who did not, at any time after delivery, breathe or show any evidence of life such as a heartbeat.

Highlights 2005—2006

• Combined data for 2005-2006 show that there was a birth defect in 4.2% of all births. This now represents approximately 2,700 babies each year in Victoria.	*page 15, Table 3.1*
• For 12% of the birth defects, the pregnancy was terminated before 20 weeks. Another 8% of babies with a birth defect die in the perinatal period. This includes 5% terminations of pregnancy at 20 weeks or more and 3% true perinatal deaths.	*page 29 Fig 4.3*
Prevalence data: • The **three most common** birth defects are often seen as isolated abnormalities and are not incompatible with life, even when associated with another birth defect: hypospadias (89% isolated, 99% survival), obstructive defects of the renal pelvis (77% isolated, 96% survival), ventricular septal defect (45% isolated, 84% survival). The majority of these cases are notified from hospital inpatient listings.	*page 17 Table 3.4* *pages 68,74 & 50*
• **Trisomy 21, or Down syndrome**, is the next most common birth defect, and continues to increase in overall prevalence primarily due to advancing maternal age of the population and use of first trimester screening. This early screening identifies some fetuses that previously would have spontaneously miscarried and not entered the statistics, but instead are counted as terminations of pregnancy. Prevalence of both **Trisomy 13** and **Trisomy 18** has also increased, again most likely due to ascertainment of non-viable fetuses through early prenatal screening and pregnancy termination.	*page 86* *pages 88 & 90*
• There has been no decline in the number of babies liveborn with Trisomy 21.	*page 86*
• Prevalence of **neural tube defects** (NTDs), particularly that of spina bifida, has decreased since 1995. However, since 1998 there has been no further decline. Overall prevalence of NTDs was 12.6/10,000 in 2005-2006.	*page 40*
• **Cardiac defects** combined comprise 24% of all birth defects. The significant increase in prevalence of **hypoplastic left heart syndrome** since 1993 is due to interstate residents giving birth in Victoria in preparation for neonatal surgery.	*Page 18 Fig 3.5 Page 52*
• Prevalence of **oespohageal atresia** has increased in recent years, and this has also been reported elsewhere, but the reason is unknown.	*Page 62*
• **Obstructive defects of the renal pelvis** also continues to increase, most likely related to increasing prenatal ultrasound detection.	*Page 74*
Maternal age: • 4.4% of women aged 35-39 years and 6.1% of women 40 years or more, had a baby with a birth defect. Specific significant associations with advanced maternal age were: ventricular septal defect (VSD), cleft palate, cystic kidney disease, developmental dysplasia of the hip (DDH), Trisomy 21, Trisomy 18, and Trisomy 13.	*Page 31 Table 5.1* *Pages 50, 56, 72, 76, 86,88 & 90*
• Younger maternal age was significantly associated with all neural tube defects, gastroschisis, and obstructive defects of the renal pelvis (ODRP).	*Pages 40, 84 & 74*
Maternal country of birth: • 5.4% of women born in the Middle East had a baby with a birth defect (increased prevalence, compared with Australian born women, of microcephalus, hydrocephalus, VSD, hypospadias, ODRP). Asian-born women had a lower prevalence of spina bifida, hypospadias, and DDH, and a higher prevalence of anorectal atresia and/or stenosis and Trisomy 18. African-born women had a lower prevalence of DDH and a higher prevalence of Trisomy 18.	*Page 31* *Pages 42, 44, 50, 68, 76* *Pages 36, 68, 76, 66, & 90* *Page 76 and 90*

Other maternal characteristics: • Unpartnered women, women living in the North West metropolitan region and women having their first baby are all more likely to have a baby with a birth defect than their counterparts. *	*Pages 32-33*
Sex of baby: • 4.7% of males compared with 3.7% of female babies had a birth defect. Higher prevalence in males was seen for transposition of great vessels, hypoplastic left heart syndrome, cleft lip and palate, anorectal atresia and/or stenosis, renal agenesis and dysgenesis, ODRP, cystic kidney disease, diaphragmatic hernia and Trisomy 21. DDH is three times more prevalent in females compared to males.	*Page 28* *Page 46, 52, 60, 66, 70, 72, 74, 80, 86* *Page 76*
Plurality: • 4.6% of multiple births had a birth defect: The only significant increased prevalence rates were for hydrocephalus and VSD.	*Page 29* *Page 44 & 50*

**Regional differences in prevalence of individual birth defects have not been included in this report as it is not possible to interpret these data without detailed statistical analysis.*

1. Introduction

This latest report on birth defects in 2005 and 2006 updates all the previous reports[1-5] . See our revised website for electronic versions of all reports.

www.health.vic.gov.au/perinatal

1.1 Background

The Victorian Perinatal Data Collection Unit (VPDCU), established under the Health Act6[6], 1958, operates under the aegis of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM). One of the fundamental purposes of the VPDCU was the establishment and maintenance of a Victorian Birth Defects Register (VBDR). The ongoing maintenance of the VBDR is enshrined in the legislation pertaining to CCOPMM and is an ongoing function of CCOPMM.

1.2 Purposes of the VBDR

The purposes of this state-wide population based surveillance system are to:

1. determine how often birth defects are occurring in Victoria and identify changing health service needs (prevalence and survival data),

2. give statistical information to organisations responsible for planning and providing health care facilities for those with birth defects, or who provide information to those concerned about having a baby with a birth defect,

3. provide information for epidemiological research to increase knowledge of aetiology and preventability of birth defects,

4. assess effectiveness of primary prevention and screening programs for birth defects,

5. respond to community and health service provider concerns about perceived clusters or changes in frequency of birth defects.

6. provide data to the National Perinatal Statistics Unit (NPSU) and to the International Clearinghouse for Birth Defects Surveillance and Research.

1.3 Definition of a birth defect

A birth defect is any abnormality of prenatal origin, either present following conception or occurring before the end of pregnancy. This includes structural, functional, genetic, chromosomal and biochemical abnormalities.

The VBDR collects data on all birth defects for livebirths, stillbirths and terminations of pregnancy occurring since January 1, 1982, irrespective of the age at diagnosis, up to 18 years of age.

Some other registers use the terms congenital abnormality, congenital malformation or congenital anomaly instead of birth defect. We continue to use the term birth defects in line with the International Clearinghouse for Birth Defects Surveillance and Research to whom we send our data, along with many registers around the world.

1.4 Sources of notification

Data are obtained from multiple sources (Table 1.1). For the period 2005—2006 there were 8,604 notifications for a total of 5,766 cases (excluding notifications for isolated minor conditions excluded from this report). This approximates to 1.5 notifications per case.

Table 1.1 Sources of notification, 2005—2006

Notification source	Number	Percent(%)
VPDCU – birth forms	3,459	40.2
Hospital sources	3,255	37.8
Perinatal death certificates	465	5.4
Autopsy reports	207	2.4
Cytogenetics reports	793	9.2
Maternal and child health nurses	346	4.0
Other professionals	75	0.9
Other (e.g. parent)	3	0.0
Unknown	1	0.0
Total*	**8,604**	**100.0**

**This excludes 716 notifications for isolated minor anomalies not included in this report.*

Since 1992 the VBDR has been updated from hospital inpatient listings from the Royal Children's Hospital (RCH) detailing all children born since 1982 who have subsequently been admitted to the RCH with a birth defect. We have also obtained listings of all children born since 1982 who have visited the RCH Cardiology Unit and Genetic Health Services Victoria, either as an inpatient or outpatient. This procedure has also been adopted for Monash Medical Centre where the inpatient listings date back to 1992.

In 2005—2006 we have added cases obtained from several of the Metropolitan Level 2 hospitals. This resulted in additional cases that would otherwise have been missed.

1.5 Data items

All notifications of birth defects (excluding terminations of pregnancy before 20 weeks gestation and interstate births) are linked to the Perinatal birth form to obtain an obstetric history for each case. Midwives complete this form as part of the mandatory notification system to the VPDCU for every birth in Victoria (see Definitions). The data items routinely maintained on the VBDR are listed in Appendix C. Further data items are available for each case of 20 weeks gestation or more as required.

1.6 Data quality

Over the years the data quality of the VBDR has been assessed by validation studies[7-10] . The most recent study[9] completed in 2003 noted further improvement in overall notification to 88%. Ascertainment of all terminations remains difficult[10]. Another validation study is underway in 2008.

1.7 Confidentiality

There is a strict policy in place to guide use of the data held in the VBDR for statistical and research purposes. The data are held in a secure place, with any hard copy information under lock and key and password protected on stand alone computers (non-networked). Foremost consideration is that the release of data by the Council will not endanger the confidentiality of the information. Human Research Ethics Committee approval must be obtained for projects that use birth defect data. Identifiable data are not provided in any report.

2. Explanatory notes relating to this report

2.1 Defects versus cases

Section 3.7 describes *each individual birth defect* (defect rate). Sections 4, 5 & 6 describe *birth defect cases* (case rate) - the number of liveborn or stillborn infants, or termination of pregnancy affected by at least one birth defect. Thus, the number of birth defects exceeds the number of cases.

2.2 Births versus confinements

There is an important difference between the number of births and the number of confinements. Confinement is defined as the final phase of pregnancy during which labour and birth occur. Any one confinement can result in more than one birth, such as is the case with twins. Tables related to infant characteristics use births as the denominator. Tables related to maternal factors use confinements as the denominator. **See Appendix D for the denominator figures used for infant and maternal statistics.**

2.3 BPA Classification of diseases

Conditions have been classified using the British Paediatric Association (BPA) Classification of Diseases - Perinatal Supplement, compatible with International Classification of Diseases - 9th revision.

With syndromes it is the practice of the VBDR to code the syndrome **and** all of its manifestations.

2.4 Twenty-eight selected defects

As in our previous reports[1-5], this report includes detailed information on twenty-eight selected defects that are either "lethal, have significant consequences for surviving children and their families, or are relatively common"[11-12].

2.5 Exclusions

In this report, information is included on cases notified to the VBDR by **December 31, 2007**. Certain isolated minor defects such as inguinal hernia, hydrocele, undescended testes (>=37 weeks gestation), vesico-ureteric reflux, and all conditions listed in the Exclusions List (Appendix B) have been excluded from this report. This may account for some of the differences between reports from some other States, where higher birth defect prevalence arises from the inclusion of such congenital anomalies.

2.6 Terminations of pregnancy (TOPs)

'Termination of pregnancy (TOP)' usually refers to an induced abortion before 20 weeks gestation in the presence of a birth defect in the fetus. However, since 1996, the report has also included '**termination of pregnancy at 20 weeks or more** (TOP ≥ 20 weeks)', to accurately reflect birth outcomes for pregnancies induced at these later gestations in the presence of a fetal birth defect.

2.7 Statistics – see Appendix D for denominator data

For each prevalence rate (PR), a 95% Poisson confidence interval (if number of cases < 400) or a 95% Binomial confidence interval (if number of cases >= 400) has been calculated. Due to space constraints, these confidence intervals are not generally shown (in Section 6), but statistically significant differences are pointed out in the text. This allows for comparisons between rates to be made and significant differences to be recognised; if the confidence intervals do not overlap, then the rates will be significantly different at the 5% level ($p < 0.05$).

Pooled prevalence rates for selected infant and maternal characteristics of each of the twenty-eight selected defects have been calculated on cases notified between 2001—2006. This increases sample size to account for annual fluctuations in numbers. Each prevalence rate is presented as N/10,000 pregnancies.

$$\text{Prevalence rate} = \frac{\text{Number of birth defects in 2001—2006}}{\text{Total births (live \& still) plus TOPs < 20 weeks in 2001—2006}}$$

Some prevalence rates that appear considerably different from others (eg. in Cleft lip and palate, the 2001-2006 PR for South American-born women is 47.7/10,000 pregnancies compared to the PR for Australian-born women of 6.1) do not reach levels of significance due to small numbers involved.

Chi square for linear trend has been calculated using Epi Info 2002[13]. Relative risks in sections 4 and 5 have also been calculated using Epi Info 2002.

Trends

For each of the 28 selected defects linear trends for the period 1995-2006 were plotted. Additionally, to look for more recent trends, the data for the period 2001-2006 were also plotted. Wherever the regression co-efficient (R) of the trend line was > 0.4, chi-square values and p-values were calculated.

3. Summary tables

3.1 Birth defects by year, 1983—2006

The increase in overall prevalence until 1990 probably reflects improving ascertainment, but the fluctuations since 1990 are more likely to reflect true prevalence, as the methods of ascertainment since then have been the same. There may be a small increase in ascertainment in 2006 due to the inclusion of defects notified from some Level 2 Hospitals (which had not been previously notified). For 2005—2006 the overall prevalence was 420/10,000 (4.2%).

Table 3.1 Birth defects by year, 1983—2006

Year	Total births, 20 weeks and later	Defects, 20 weeks and later	Defects before 20 weeks (terminations)	N/10,000 Pregnancies (including terminations)	%
1983	60,628	1,675	2	276.6	2.8
1984	60,737	1,714	9	283.6	2.8
1985	61,189	1,599	18	264.2	2.6
1986	61,253	1,625	80	278.0	2.8
1987	61,566	1,640	55	275.1	2.8
1988	63,666	1,889	103	312.4	3.1
1989	64,255	1,990	123	328.2	3.3
1990	66,878	2,208	133	349.3	3.5
1991	65,248	2,295	140	372.4	3.7
1992	66,305	2,345	152	375.7	3.8
1993	64,737	2,281	203	382.5	3.8
1994	64,932	2,331	250	396.0	4.0
1995	63,717	2,497	257	430.5	4.3
1996	62,951	2,234	272	396.4	4.0
1997	62,308	2,343	298	421.8	4.2
1998	62,091	2,372	275	424.4	4.2
1999	62,690	2,609	295	461.1	4.6
2000	62,564	2,653	292	468.5	4.7
2001	62,149	2,393	307	432.3	4.3
2002	63,133	2,548	327	453.0	4.5
2003	63,552	2,646	356	469.7	4.7
2004	63,700	2,710	342	476.6	4.8
2005	66,654	2,413	338	410.6	4.1
2006	69,856	2,654	361	429.4	4.3
Total	1,526,570*	53,664	4988	383.0	3.8

**These figures may differ from the number of births presented in Births in Victoria reports[15] due to the inclusion of terminations of pregnancy greater than 20 weeks for psychosocial reasons and birth defects, that have been excluded from the latest Births in Victoria report.[15]*

3.2 Comparison of overall prevalence of birth defects, 1987—2006

Table 3.2 Comparison of overall prevalence of birth defects, 1989—2006

Defect	1987—1991 (N/10,000)	1992—1996 (N/10,000)	1997—2001 (N/10,000)	2002—2006 (N/10,000)
Total births + TOPs (n)	*322,167*	*323,776*	*313,269*	*328,619*
Anencephaly	5.0	7.1	5.6	5.8
Spina bifida	8.2	8.3	7.1	6.2
Encephalocele	1.4	2.1	1.6	1.4
Microcephalus	2.9	2.9	2.7	2.6
Hydrocephalus	6.5	8.4	10.0	9.4
Transposition of great vessels	4.9	5.9	5.4	5.8
Tetralogy of Fallot	3.3	4.2	4.1	3.9
Ventricular septal defect	27.5	31.0	30.5	34.7
Hypoplastic left heart syndrome	2.6	2.5	3.3	4.1
Coarctation of aorta	5.6	4.8	3.6	4.5
Cleft palate	6.6	8.0	7.8	8.9
Cleft lip	4.0	3.6	4.1	3.9
Cleft lip and palate	6.6	6.2	7.1	6.1
Oesophageal atresia and/or stenosis	2.9	4.0	3.3	3.1
Anorectal atresia and/or stenosis	2.9	2.3	3.3	3.2
Small intestinal atresia and/or stenosis	3.5	4.9	4.3	4.0
Hypospadias*	61.0	51.8	64.8	70.1
Renal agenesis and dysgenesis	5.1	5.7	6.3	6.8
Cystic kidney disease	4.0	5.7	6.9	6.6
Obstructive defects of renal pelvis	10.3	21.5	37.8	39.7
Developmental dysplasia of hip	31.1	28.3	26.9	30.2
Limb reduction defects	6.0	7.2	5.8	5.6
Diaphragmatic hernia	3.2	4.0	3.3	3.1
Exomphalos	3.4	3.1	3.3	3.3
Gastroschisis	1.3	2.1	2.8	2.0
Trisomy 21 (Down syndrome)	15.9	18.5	25.1	34.7
Trisomy 13 (Patau syndrome)	1.3	2.2	2.8	3.4
Trisomy 18 (Edward syndrome)	3.7	5.2	6.3	8.6

**These figures have used only male births plus male TOPs as the denominator.*

3.3 Order of prevalence of individual birth defects, 2005—2006

The following table shows the order of prevalence of twenty-eight selected birth defects, not cases, presented in section six of this report for 2005—2006. These are many of the defects reported by the International Clearinghouse for Birth Surveillance and Research[12].

Table 3.3 Order of prevalence of twenty eight selected birth defects, 2005—2006

Defect	n/10,000	1 in x number of births + TOPs
Hypospadias*	74	135
Obstructive defects of renal pelvis	40	250
Ventricular septal defect	32.2	311
Trisomy 21	29.5	339
Developmental dysplasia of hip	27.5	364
Trisomy 18	8.4	1190
Hydrocephalus	8.1	1235
Cleft Palate	8.0	1250
Cystic kidney	6.8	1471
Renal agenesis/dysgenesis	6.6	1515
Transposition of great vessels	6.3	1587
Spina bifida	6.0	1667
Cleft lip and palate	5.5	1818
Anencephaly	5.5	1818
Coarctation of aorta	4.9	2041
Limb reduction defects	4.8	2083
Hypoplastic left heart syndrome	4.5	2222
Anorectal atresia and/or stenosis	4.4	2273
Trisomy 13	3.9	2564
Cleft lip	3.9	2564
Oesophageal atresia and/or stenosis	3.8	2632
Tetralogy of Fallot	3.7	2703
Exomphalos	3.1	3226
Intestinal atresia and/or stenosis	3.2	3125
Diaphragmatic hernia	2.8	3571
Gastroschisis	2.3	4348
Microcephalus	1.8	5556
Encephalocele	1.2	8333

**This figure used male babies only as the denominator, as hypospadias is a condition that affects only males. In previous reports[1-5] , all births were used as the denominator.*

3.4 Relative frequency of birth defects by body system, 2005—2006[14]

Figure 3.1 Relative frequency of birth defects by body system, 2005—2006

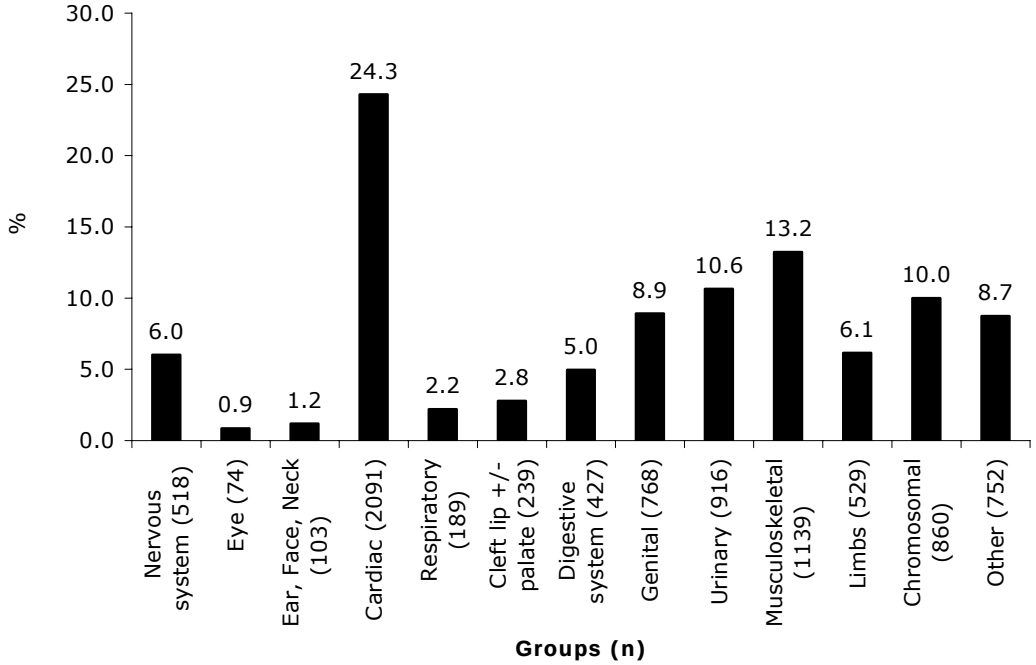

Figure 3.1 presents the distribution of individual birth defects**,** not cases, affecting fetuses/babies in 2005—2006, **grouped according to body system**. Cardiac conditions comprise the greatest proportion of all birth defects (24.3%), followed by musculoskeletal (13.2%) and urinary system defects (10.6%).

3.5 Summary of associated defects for twenty-five selected defects, 2005—2006

The table below reflects the associations with other birth defects for twenty-five selected defects: that is, whether they are isolated conditions, or associated with chromosomal anomalies or occur as multiple same system (eg. two or more cardiac defects) or multiple different system defects (eg. cleft lip and ventricular septal defect).

Table 3.4 Summary of patterns of birth defects for twenty-five selected defects, 2005—2006

Defect	Total	% Isolated	% Chromosomal	% Other (multiple same system & multiple different systems)
Anencephaly	75	82.7	4	13.4
Spina bifida	82	73.2	7.3	19.5
Encephalocele	17	70.6	5.9	23.5
Microcephalus	25	36	8	56
Hydrocephalus	111	42.3	12.6	45
Transposition of great vessels	86	4.7	7	88.4
Tetralogy of Fallot	46	21.7	10.9	67.4
Ventricular septal defect	441	44.7	12.5	42.8
Hypoplastic left heart syndrome	61	45.9	9.8	44.3
Coarctation of aorta	67	16.4	10.4	73.1
Cleft palate	110	56.4	8.2	35.5
Cleft lip	54	77.8	5.6	16.7
Cleft lip and palate	75	66.7	20	13.3
Oesophageal atresia and/or stenosis	52	30.8	3.8	65.4
Small intestinal atresia and/or stenosis	44	59.1	6.8	34.1
Anorectal atresia and/or stenosis	60	35	8.3	56.7
Hypospadias	522	89.1	0.6	10.3
Renal agenesis and dysgenesis	90	47.8	11.1	41.1
Cystic kidney disease	92	54.3	7.6	38
Obstructive defects of renal pelvis	550	76.5	2.5	20.9
Congenital dislocation of hip	377	94.4	0.3	5.3
Limb reduction defects	66	31.8	10.6	57.6
Diaphragmatic hernia	38	26.3	7.9	65.8
Exomphalos	41	14.6	31.7	53.7
Gastroschisis	31	71	9.8	19.3

3.6 Termination of pregnancy for a birth defect before 20 weeks gestation, 1995—2006

Between 1995—2006 there were 3,720 terminations of pregnancy for birth defects before 20 weeks gestation reported to the VBDR, 60.1% for a chromosomal anomaly and 17.2% for central nervous system (CNS) defects (mainly anencephaly or spina bifida).

For the period 2005—2006, 68% of terminations were for a chromosomal anomaly and 13% for CNS defects.

Table 3.5 Terminations of pregnancy for a birth defect before 20 weeks gestation by body system, 1995—2006

Defect / body system	1995-1996 N=529 %	1997-1998 N=573 %	1999-2000 N=587 %	2001-2002 N=634 %	2003-2004 N=698 %	2005-2006 N=699 %
Chromosomal	45.7	52.6	60.5	65.5	65.3	70.6
Central nervous	22.9	21.5	16.7	15.5	13.9	12.6
Cardiac	1.9	2.1	2.7	1.4	1.7	0.9
Respiratory	0.2	0.3	0.0	0.0	0.1	0.0
Digestive	0.4	0.5	0.5	0.5	0.1	0.1
Genitourinary	2.3	1.6	3.1	1.6	1.3	0.4
Musculoskeletal	4.2	3.7	4.8	2.7	2.0	2.7
Multiple systems	13.6	11.0	6.5	5.5	7.4	4.4
Other	7.2	4.7	3.6	4.9	5.6	5.6
Not stated	1.7	1.9	1.7	2.5	2.4	2.6
Total	100.0	100.0	100.0	100.0	100.0	100.0

3.7 Birth defects by four digit code, 2005—2006

The following figures refer to individual birth defects, ***not cases.*** The codes are compatible with the British Paediatric Association Classification of Diseases Supplement to ICD – 9th Revision.

It is possible for one case to have two or more conditions within a particular code range. (These codes are marked with an "*".) Therefore the number of individual birth defects may exceed the number of cases.

NB: Conditions which are notified to the International Clearinghouse for Birth Defect Surveillance and Research are indicated by shading in the following table.

Table 3.6 Birth defects by four digit code, 2005—2006

Codes	Defects	Number	N/10,000
740*	**Anencephalus & similar anomalies**	**76**	**5.5**
7400#	Anencephalus	75	5.5
7401	Craniorachischisis	1	0.1
7402	Iniencephaly	0	0.0
741*	**Spina bifida**	**82**	**6.0**
742	**Other nervous system (NS)**	**360**	**26.3**
7420	Encephalocele	17	1.2
7421	Microcephalus	25	1.8
7422	Brain reduction	118	8.6
7423*	Hydrocephalus	111	8.1
7424-9	Other	63	4.6
330-7	Hereditary & degenerative diseases	13	0.9
343-9	Cerebral palsy	4	0.3
340-2/3499	Other disorders of CNS	0	0.0
350-9	Disorders of peripheral NS	9	0.7
743	**Eye**	**74**	**5.4**
7430	Anophthalmos	4	0.3
7431	Microphthalmos	6	0.4
7432	Buphthalmos	4	0.3
74330-1/3-9	Other lens	1	0.1
74332	Cataract	14	1.0
7434-9	Other	45	3.3
744	**Ear, face & neck**	**103**	**7.5**
7440	Ear-affecting hearing	17	1.2
74400	*Auditory canal*	*8*	*0.6*
74401	*Absent auricle*	*4*	*0.3*
74402-9	*Other*	*5*	*0.4*
7441-3	Other ear	43	3.1
74410-9	*Accessory auricle*	*19*	*1.4*
74421	*Microtia*	*11*	*0.8*
74420/2-9	*Other ear*	*13*	*0.9*
7444-9	Face & neck	44	3.2
389	Deafness	8	0.6

 # Anencephalus includes absence of brain, acrania, anencephaly and hemianencephaly.

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Table 3.6 Birth defects by four digit code, 2005—2006 (cont.)

Codes	Defects	Number	N/10,000
745	**Bulbus cordis & cardiac septal closure**	**952**	**69.5**
7450	Common truncus	19	1.4
7451*	Transposition of great vessels	93	6.8
7452	Tetralogy/Pentalogy of Fallot	46	3.4
7453	Common ventricle	8	0.6
7454*	Ventricular septal defect	442	32.3
7455	Atrial septal defect	290	21.2
7456	Endocardial cushion	54	3.9
7457-9	Other	0	0.0
746	**Other heart**	**527**	**38.5**
425	Cardiomyopathy	4	0.3
426	Conduction disorder	4	0.3
7460	Pulmonary valve	137	10.0
74600	*Atresia*	*30*	*2.2*
74601	*Stenosis*	*102*	*7.4*
74602-9	*Other*	*5*	*0.4*
7461	Tricuspid atresia/stenosis	70	5.1
7462	Ebstein anomaly	3	0.2
7463-4	Aortic valve stenosis/insufficiency	35	2.6
7465-6	Mitral stenosis/insufficiency	73	5.3
7467	Hypoplastic left heart syndrome	61	4.5
7468	Other specified	113	8.2
7469	Unspecified	27	2.0
747	**Circulatory**	**612**	**44.7**
7470	Patent ductus arteriosus	336	24.5
7471	Coarctation of aorta	67	4.9
7472	Other aorta	55	4.0
7473	Pulmonary artery	82	6.0
7474	Great veins	46	3.4
7475	Single umbilical artery	0	0.0
7476	Peripheral vascular	8	0.6
7478	Other specified	18	1.3
7479	Unspecified	0	0.0
748	**Respiratory**	**189**	**13.8**
7480	Choanal atresia	20	1.5
7481	Other nose	15	1.1
7482-3	Larynx/trachea/bronchus	104	7.6
7484-6	Lung	50	3.6
7488-9	Other respiratory	0	0.0
749*	**Cleft palate/lip**	**239**	**17.4**
7490	Cleft palate	110	8.0
7491	Cleft lip	54	3.9
7492	Cleft lip & palate	75	5.5
750	**Upper alimentary tract**	**162**	**11.8**
7503	Oesophageal atresia/stenosis	52	3.8

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Table 3.6 Birth defects by four digit code, 2005—2006 (cont.)

Codes	Defects	Number	N/10,000
7501-2/4-9	Other	110	8.0
751	**Other Digestive**	**265**	**19.3**
7510	Meckels diverticulum	2	0.1
7511	Small intestine atresia/stenosis	46	3.4
75110	*Duodenal atresia/stenosis*	*19*	*1.4*
75111	*Jejunal atresia/stenosis*	*12*	*0.9*
75112	*Ileal atresia/stenosis*	*7*	*0.5*
75119	*Unspecified atresia/stenosis*	*8*	*0.6*
7512*	Large intestine rectal anal atresia/stenosis	64	4.7
75120	*Large intestine atresia/stenosis*	*4*	*0.3*
75121-2	*Rectal atresia/stenosis*	*5*	*0.4*
75123-4	*Anal atresia/stenosis*	*55*	*4.0*
7513	Hirschsprung's disease	23	1.7
7514	Intestinal fixation	26	1.9
7515-9	Other digestive	64	4.7
524-579	Other	40	2.9
752	**Genital organs**	**768**	**56.1**
7520-1	Ovaries/fallopian etc	8	0.6
7522-3	Uterus	9	0.7
7524	Cervix/vagina/external genitalia	22	1.6
7525@	Undescended testes	72	5.3
75260/3-5	Hypospadias	522	38.1
75261	Epispadias	9	0.7
75262	Chordee	55	4.0
7527	Indeterminate sex	22	1.6
7528	Other specified	49	3.6
7529	Unspecified	0	0.0
753	**Urinary**	**916**	**66.9**
7530	Renal agenesis/dysgenesis	100	7.3
75300	*Bilateral*	*24*	*1.8*
75301	*Unilateral*	*66*	*4.8*
7531#	Cystic kidney disease	92	6.7
75311-3	*Polycystic*	*30*	*2.2*
75316	*Multicystic*	*48*	*3.5*
75310/4/8	*Other*	*14*	*1.0*
7532#	Obstructive defects renal pelvis/ureter	570	41.6
75320	*Hydronephrosis*	*531*	*38.8*
75321-9	*Other*	*39*	*2.8*
7533	Other specified kidney disorders	68	5.0
7534	Other specified disorders of ureter	19	1.4
7535	Exstrophy of bladder	3	0.2
7536	Urethra bladder neck atresia/stenosis	30	2.2
7537	Urachus	3	0.1

@ Undescended testes in term babies is included in VBDR, but isolated cases have been excluded for the purposes of this report as a minor malformation. This figure refers to cases of undescended testes which are not isolated but occur with other defects.

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Table 3.6 Birth defects by four digit code, 2005—2006 (cont.)

Codes	Defects	Number	N/10,000
7538	Other bladder/urethra	15	1.1
7539	Unspecified	5	0.4
592-608	Other	12	0.9
754	**Certain musculoskeletal**	**775**	**56.6**
7540	Of skull, face & jaw	33	2.4
7541	Of sternocleidomastoid	0	0.0
7542	Of spine	5	0.4
75430	Congenital dislocation of hip	377	27.5
75431-2	Other hip	10	0.7
7544	Genu recurvatum/bowing	5	0.4
7545-7	Of feet	339	24.7
7548	Other	6	0.4
755*	**Limbs**	**529**	**38.6**
7550	Polydactyly	178	13.0
7551	Syndactyly	141	10.3
7552	Reduction deformities upper limb	52	3.8
7553	Reduction deformities lower limb	34	2.5
7554	Reduction deformities unspecified limb	2	0.1
7555	Other upper limb	65	4.7
7556	Other lower limb	37	2.7
75580	Arthrogryposis multiplex congenita	8	0.6
75581-8	Other specified	10	0.7
7559	Unspecified	2	0.1
756	**Other musculoskeletal**	**364**	**26.6**
7560	Skull, face & bones	116	8.5
75600	*Craniosynostosis*	*69*	*5.0*
75601-2/4-9	*Other*	*25*	*1.8*
75603	*Pierre Robin syndrome*	*22*	*1.6*
7561	Spine	49	3.6
7562-3	Ribs & sternum	22	1.6
7564	Chondrodystrophy	25	1.8
75643	*Achondroplasia*	*6*	*0.4*
75644	*Other dwarfing*	*7*	*0.5*
75640-9/5-9	*Other*	*12*	*0.9*
7565	Osteodystrophies	25	1.8
75650	*Osteogenesis*	*13*	*0.9*
75651-9	*Other*	*12*	*0.9*
7566	Diaphragm	43	3.1
75660/2-9	*Other*	*5*	*0.4*
75661	*Diaphragmatic hernia*	*38*	*2.8*
7567	Abdominal wall	75	5.5
75670	*Exomphalos*	*41*	*3.0*
75671	*Gastroschisis*	*31*	*2.3*
75672-9	*Other*	*3*	*0.2*
7568	Other specified of muscle/ tendon/fascia/	9	0.7
7569	Unspecified	0	0.0

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Table 3.6 Birth defects by four digit code, 2005—2006 (cont.)

Codes	Defects	Number	N/10,000
757	**Integument**	**86**	**6.3**
7570	Hereditary oedema	1	0.1
7571	Icthyosis congenita	6	0.4
7572	Dermatoglyphic anomalies	0	0.0
7573	Other specified anomalies skin	71	5.2
7574	Specified anomalies of hair	2	0.1
7575	Specified anomalies of nails	3	0.2
7576	Specified anomalies of breast	0	0.0
7578	Other specified anomalies of integument	1	0.1
7579	Unspecified	2	0.1
758	**Chromosomal**	**860**	**62.8**
7580	Trisomy 21	405	29.6
7581	Trisomy 13	54	3.9
7582	Trisomy 18	114	8.3
7583	Autosomal deletion	46	3.4
7585	Other autosomal	92	6.7
7586	Turner's syndrome	63	4.6
7587	Klinefelter's syndrome	17	1.2
7588	Other sex chromosomes	31	2.3
7589	Unspecified	38	2.8
759	**Other**	**180**	**13.1**
7590	Spleen	14	1.0
7591	Adrenal gland	8	0.6
7592	Other endocrine gland	9	0.7
7593	Situs inversus	9	0.7
7594	Conjoined twins	3	0.2
7595	Tuberous sclerosus	4	0.3
7596	Harmartoses NEC	3	0.2
7597	Multiple so described	5	0.4
7598	Other specified syndromes	97	7.1
7599	Unspecified	28	2.0
	Other	**79**	**5.8**
76076	Fetal alcohol syndrome	1	0.1
76079	Maternal conditions	0	0.0
7710-2	Congenital infection	17	1.2
77121	*Toxoplasmosis*	*0*	*0.0*
77119	*Cytomegalovirus*	*15*	*1.1*
7710/2-9	*Other*	*2*	*0.1*
7780	Hydrops fetalis (non-immune)	54	3.9
7834	Developmental delay	7	0.5
140-239	**Neoplasms**	**134**	**9.8**
140-208	Malignant	20	1.5
210-229	Benign	84	6.1
22809	*Haemangioma*	*22*	*1.6*

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Table 3.6 Birth defects by four digit code, 2003—2004 (cont.)

Codes	Defects	Number	N/10,000
22819	*Cystic hygroma*	*55*	*4.0*
2100-2279	*Other*	*7*	*0.5*
235-9	Uncertain behaviour	30	2.2
240-279	**Endocrine/nutritional/metabolic**	**222**	**16.2**
2439	Congenital hypothyroidism	52	3.8
250-9	Other endocrine glands	14	1.0
260-9	Nutritional deficiencies	6	0.4
2701	Phenylketonuria	18	1.3
2700/2-9	Other disorders of amino-acid	24	1.8
271	Of carbohydrate metabolism	4	0.3
272	Of lipid metabolism	1	0.1
273	Of plasma protein metabolism	5	0.4
275	Of mineral metabolism	5	0.4
2770	Cystic fibrosis	46	3.4
2771-9	Other metabolism	39	2.8
279	Of immunity	8	0.6
280-9	**Diseases of blood**	**48**	**3.5**
282	Hereditary haemolytic anaemia	42	3.1
281/3-5	Other anaemias	0	0.0
286	Coagulation defects	5	0.4
287-9	Other	1	0.1
315-318	**Developmental delay**	**3**	**0.2**

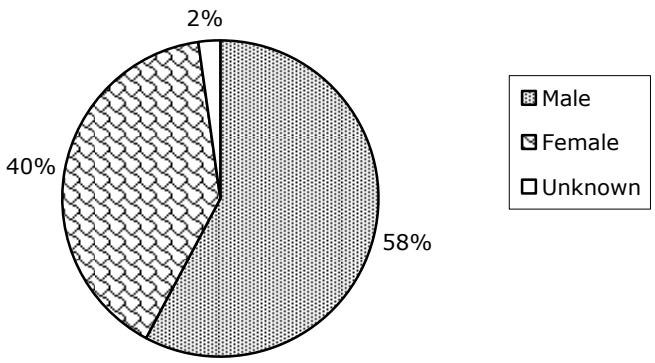
4. Birth defect cases by selected infant characteristics

Table 4.1 Birth defect cases by number of defects per case 2005—2006

Number of birth defects	No. cases	%
1	4,351	*75.5*
2	787	*13.6*
3	296	*5.1*
4+	332	*5.8*
Total	5,766	*100.0*

- 5,766 birth defect cases were notified to the VBDR, comprising 8,604 individual birth defects.

Figure 4.1 Proportion of birth defect cases by sex, 2005—2006



- Birth defects were more commonly reported in males than in females (ie 4.7% of all males versus 3.5% of all females). This gives a relative risk of having a male baby with a birth defect of 1.16 (95% CI 1.13-1.18, p<0.0001).

- There were 4 cases of indeterminate sex, as distinct from unknown. All births of indeterminate sex are considered to be birth defects.

Table 4.2 Birth defect cases by birthweight (excluding TOPs < 20wks) , 2005—2006

Weight (grams)	No. birth defects	%	Total no. births	%
<1000	527	10.4	1,546	1.1
1000-2499	525	10.4	8,142	6.0
2500 +	4,011	79.2	126,787	92.9
Unknown	4	0.1	35	0.0
Total	5,067	100.0	136,510	100.0

- Birth defects were recorded among 10.9% (1,052/9,688) of low birthweight infants (<2,500 grams) compared with 3.2% (4,011/126,787) of infants with birthweight of 2,500 grams or more.

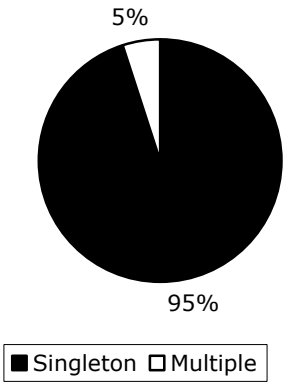
- Relative risk of a having a low birthweight baby with a birth defect is 3.16 (95% CI 2.99,3.75, p<0.0001) compared with babies weighing 2,500 grams or more.

Table 4.3 Birth defect cases by gestational age (excluding TOPs < 20wks), 2005—2006

Gestation (wks)	No. birth defects	%	Total No. births	%
20-27	378	7.5	1,501	1.1
28-31	163	3.2	1,053	0.8
32-36	579	11.4	8,526	6.2
37-41	3,893	76.8	123,577	90.5
> 41	54	1.1	1,837	1.3
Unknown	0	0.0	16	0.0
Total	5,067	100.0	136,510	100.0

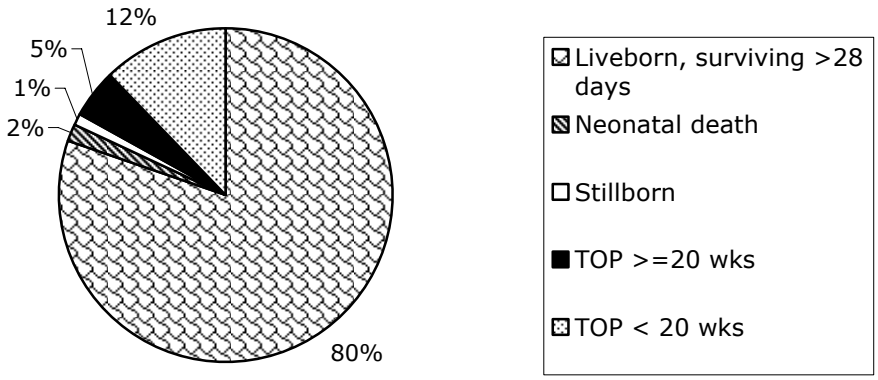
- 10.1% (1,120/11,080) of premature infants (< 37 weeks) had a birth defect compared to 3.1% (3,947/125,414) among term and post-term infants (>= 37 weeks).
- Relative risk of a birth defect for a pre-term birth compared with a term birth is 2.92 (95% CI 2.76, 3.08, p<0.0001).

Figure 4.2 Proportion of birth defect cases by plurality, 2005—2006



- 4.1% (5,484/132,714) of singleton births had a birth defect.
- 4.6% (282/5,034) of multiple births (twins, triplets, quads) had a birth defect.
- Relative risk of a birth defect for multiple versus singleton births is 1.35 (95% CI 1.20-1.52, p <0.0001).

Figure 4.3 Perinatal mortality associated with birth defect cases, 2005—2006



- Perinatal mortality is high among infants with birth defects.
- 12.1% of all pregnancies with a birth defect were terminated before 20 weeks gestation and another 4.9% were terminated at 20 weeks or greater.
- Of babies born at 20 weeks or more with a birth defect, 62 (1.1%) were stillborn and 95 (1.6%) were neonatal deaths. This corresponds to a perinatal mortality rate of **32.8/1,000 births** with a birth defect (excluding TOPs for a birth defect both before and after 20 weeks gestation).
- Relative risk of a perinatal death for a baby with a birth defect compared to a baby without a birth defect is 4.16 (3.59,4.82, $p < 0.0001$). (This figure excludes terminations of pregnancy for a birth defect both before and after 20 weeks gestation, and terminations of pregnancy at 20 weeks or more for psychosocial reasons.)

NB: These mortality figures comprise all birth defect cases, including those where the cause of death may not be directly related to the birth defect.

5. Birth defect cases by selected maternal characteristics

Table 5.1 Birth defect cases by maternal age group, 2005—2006

Age group (yrs)	No. birth defects	%	Total no. confinements + TOPs	%	% of birth defects within each group	95% Confidence Interval	
						LL	UL
<20	175	3.1	3,806	2.8	4.6	3.9	5.3
20-24	629	11.2	15,280	11.3	4.2	3.8	4.4
25-29	1,326	23.5	33,826	25.1	3.9	3.7	4.1
30-34	1,912	33.9	49,216	36.5	3.9	3.7	4.1
35-39	1,196	21.2	27,221	20.2	4.4	4.2	4.6
40+	317	5.6	5,227	3.9	6.1	5.4	6.7
Unknown	77	1.4	91	0.1	n/a	n/a	n/a
Total	5,632	100.0	134,667	100.0	4.2	4.1	4.3

- The prevalence of birth defects was statistically significantly higher among mothers aged 35 to 39 years (4.4%) and women aged 40 years or more (6.1%) compared to women less than 35 years of age.

- Women aged 35 years and over comprised 26.8% of all mothers who had a pregnancy affected by a birth defect and had a relative risk of 1.14 compared to younger women (95%CI 1.09-1.19,p<0.0001).

- If all mothers with pregnancies affected by chromosomal abnormalities are excluded, then the proportion of women aged 35 or more who had a pregnancy affected by a birth defect was 22.7%, giving a relative risk of having a child with a birth defect of 0.95 compared to younger women (95% CI 0.90-1.00, p=0.06). Therefore, there is no overall increased risk for older women having a pregnancy affected by a birth defect other than for chromosomal anomalies.

Table 5.2 Birth defect cases by maternal country of birth, 2005—2006

Country of birth groups	No. birth defects	%	Total no. confinements + TOPs	%	% of birth defects within each group	95% Confidence Interval	
						LL	UL
Australia	4,127	73.3	101,354	75.3	4.1	3.9	4.2
Oceania/NZ	141	2.5	3,632	2.7	3.8	3.3	4.5
UK inc Eire	158	2.8	3,494	2.6	4.5	3.8	5.2
Europe	154	2.7	3,953	2.9	3.9	3.3	4.5
Middle East	162	2.9	3,007	2.2	5.4	4.6	6.2
Asia	556	9.9	13,674	10.2	4.1	3.4	4.4
Nth America	30	0.5	822	0.6	3.7	2.4	4.9
Sth America	27	0.5	842	0.6	3.2	2.0	4.4
Africa	137	2.4	3,219	2.4	4.3	3.6	4.9
Unknown	140	2.5	670	0.5	n/a	n/a	n/a
Total	5,632	100.0	134,667	100.0	4.2	4.1	4.3

- Women from the Middle East are more likely to have babies with a birth defect than women from any of the other country of birth groups.

Table 5.3 Birth defect cases by Department of Human Services Regions, 2005—2006

Region#	No. birth defects	%	Total no. confinements + TOPS	%	% of birth defects within each group	95% Confidence Interval LL	UL
Barwon	351	6.2	8,393	6.2	4.2	3.8	4.6
Grampians	178	3.2	5,072	3.8	3.5	3.0	4.0
Loddon Mallee	286	5.1	7,351	5.5	3.9	3.5	4.3
Hume	278	4.9	6,299	4.7	4.4	3.9	4.9
Gippsland	228	4.0	5,565	4.1	4.1	3.6	4.6
North West Metro	1,921	34.1	43,492	32.3	4.4	4.2	4.6
E Metro	892	15.8	22,957	17.0	3.9	3.6	4.1
S Metro	1,317	23.4	32,397	24.1	4.1	3.9	4.3
Other*	177	3.1	3,138	2.3	5.6	4.8	6.5
Unknown	3	0.1	3	0.0	n/a	n/a	n/a
Total	5,631	100.0	134,667	100.0	4.2	4.1	4.3

 # Region is based on mothers' postcode of residence at the time of birth.
** Refers to women who reside outside Victoria but who give birth at a Victorian hospital.*

- Women from the North West Metropolitan Region are more likely to have a baby born with a birth defect when compared to all other Regions (RR 1.05, 95% CI 1.01-1.09, p<0.018).

NB: The following three tables have lower overall proportions of birth defects because terminations of pregnancy before 20 weeks have been excluded.

Table 4.4 Birth defect cases by Aboriginality* (excluding TOPs < 20wks), 2005—2006

Aboriginality	No. cases	%	Total no. confinements	%	% of birth defects within each group	95% Confidence Interval LL	UL
Yes	48	1.0	1,094	0.8	4.4	3.2	5.6
No	4,889	99.0	132,839	99.2	3.7	3.6	3.8
Unknown	1	0.0	41	0.0	n/a	n/a	n/a
Total	4938	100.0	133,974	100.0	3.7	3.6	3.8

**Recent reports[15] have indicated poor reporting of Aboriginality to the VPDCU, leading to possible under ascertainment of birth defects*

- There is no difference in prevalence of birth defects in pregnancies of women reported as Koori compared to non-Koori women.

Table 5.5 Birth defect cases by marital status (excluding TOPs < 20wks), 2005—2006

Marital status	No. cases	%	Total no. confinements	%	% of birth defects within each group	95% Confidence Interval	
						LL	UL
Single	680	13.8	16,732	12.5	4.1	3.8	4.4
Divorced	28	0.6	469	0.4	6.0	3.8	8.1
Widowed	3	0.1	45	0.0	6.7	0	1.4
Separated	31	0.6	713	0.5	4.3	2.9	5.8
Married	3,427	69.4	95,404	71.2	3.6	3.5	3.7
De facto	745	15.1	20,296	15.1	3.7	3.41	3.
Unknown	24	0.5	315	0.2	n/a	n/a	n/a
Total	4,938	100.	133,974	100.0	3.7	3.6	3.8

- If marital status is divided into unpartnered (single, separated, divorced, widowed) versus partnered (defacto, married), there is a statistically significant increased relative risk of 1.13 (95% CI 1.06-1.21, p<0.001) for unpartnered women having a baby with a birth defect.

Table 5.6 Birth defect cases by parity (excluding TOPs < 20wks), 2005—2006

Parity	No. cases	%	Total no. confinements	%	% of birth defects within each group	95% Confidence Interval	
						LL	UL
One*	2,257	45.7	57,809	43.1	3.9	3.7	4.1
Two	1,530	31.0	45,772	34.2	3.3	3.2	3.5
Three	690	14.0	19,862	14.8	3.5	3.2	3.7
Four	275	5.6	6,591	4.9	4.2	3.7	4.7
Five	98	2.0	2,238	1.7	4.4	3.5	5.2
Six or more	88	1.8	1,700	1.3	5.2	4.1	6.2
Unknown	0	0.0	2	0.0	n/a	n/a	n/a
Total	4,938	100.0	133,974	100.0	3.7	3.6	3.8

**First birth at 20 weeks or more.*

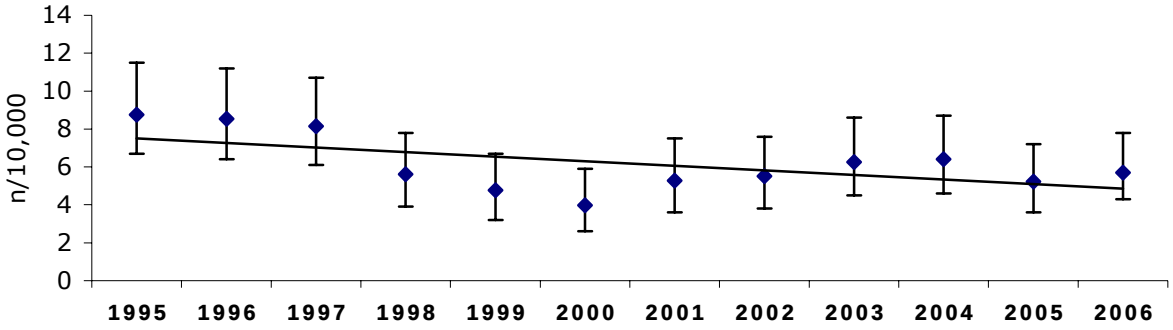
- Primiparous women are more likely to have babies with birth defects than multiparous women (RR 1.07, (95% CI 1.04-1.10, p<0.0001).

6.1 Anencephaly

British Paediatric Association code 740.00 – 740.29

Definition: Total or partial absence of the cranial vault, the covering skin and the brain tissue.

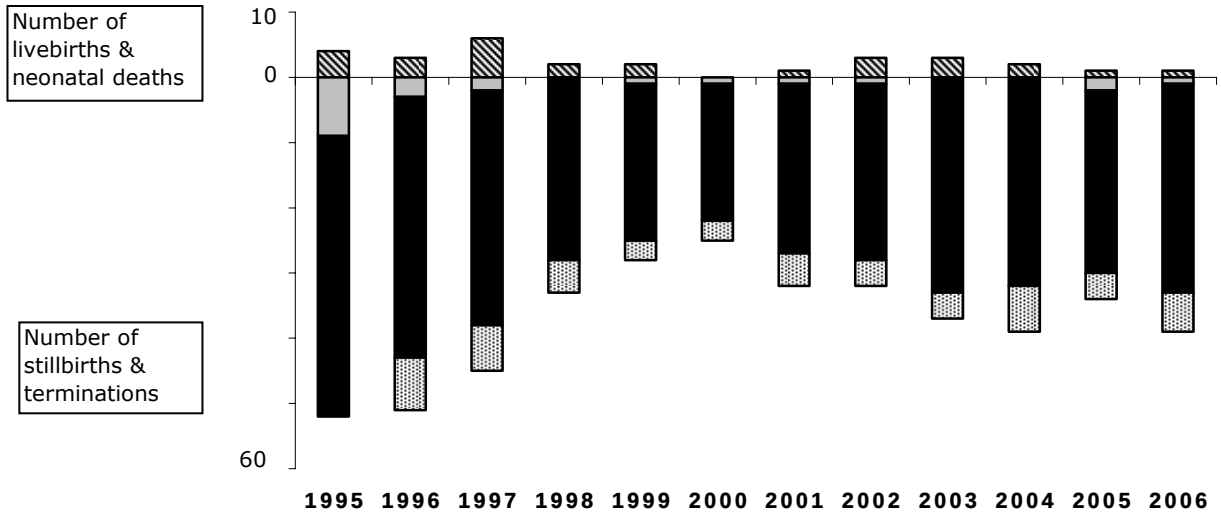
Trend in prevalence rates, 1995-2006



N / 10,000	8.8	8.5	8.1	5.6	4.8	4.0	5.3	5.5	6.3	6.4	5.2	5.7

- From 1995 to 2006 there has been no statistically significant change in the prevalence of anencephaly. In particular, there has been no decrease since 1998.

Number of cases and pregnancy outcomes, 1995-2006



Total number	56	54	51	35	30	25	33	35	40	41	35	40
Outcome	**Percent (%)**											
Neonatal death	7	6	12	6	7	0	3	9	8	5	3	3
Survived > 28 days	0	0	0	0	0	0	0	0	0	0	0	0
Stillbirth	16	6	4	0	3	4	3	3	0	0	6	3
TOP < 20 weeks	77	74	71	80	80	84	79	77	83	78	80	80
TOP >= 20 weeks*	0	15	14	14	10	12	15	11	10	17	11	15

**These cases are not identified in the dataset prior to 1996*

- Most pregnancies affected by this condition were terminated before 20 weeks gestation, with the number fluctuating around 80% per year.

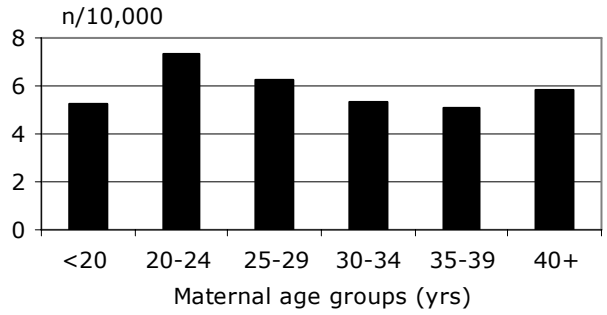
Anencephaly and associated birth defects, 2005-2006

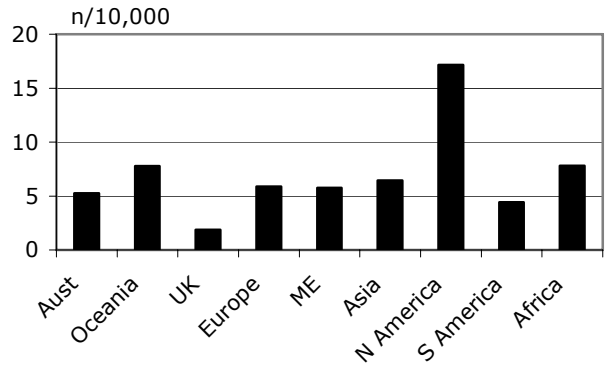
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	59	2	1	0	62	82.7
Chromosomal	3	0	0	0	3	4.0
Other same system	2	0	0	0	2	2.7
Other different systems	6	1	1	0	8	10.7
Total	70	3	2	0	75	100.0

Five year summary of the prevalence of anencephaly and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=224)	**No.**	**%**	**PR**
Sex			
Male	52	*23.2*	2.6
Female	54	*24.1*	2.8
Indeterminate/ Unknown*	118	*52.7*	
Plurality#			
Singleton	217	*96.9*	5.8
Multiple	7	*3.1*	5.0
Maternal (n=224)			
Maternal Age (yrs)			
<20	6	*2.7*	5.3
20-24	33	*14.7*	7.3
25-29	63	*28.1*	6.2
30-34	76	*33.9*	5.4
35-39	36	*16.1*	5.1
40+	8	*3.6*	5.8
Unknown	2	*0.9*	
Country of birth			
Australia	153	*68.3*	5.3
Oceania inc NZ	8	*3.6*	7.8
UK inc Eire	2	*0.9*	1.9
Europe	7	*3.1*	5.9
Middle East	5	*2.2*	5.8
Asia	24	*10.7*	6.4
Nth America	4	*1.8*	17.2
Sth America	1	*0.4*	4.4
Africa	6	*2.7*	7.8
Unknown	14	*6.3*	

**Large number of unknowns due to many TOPs*
 # excludes unknown





- Although some PRs were higher than others, due to small sample size there were no statistically significant associations for sex of infant, plurality, maternal age or country of birth for 2001-2006.

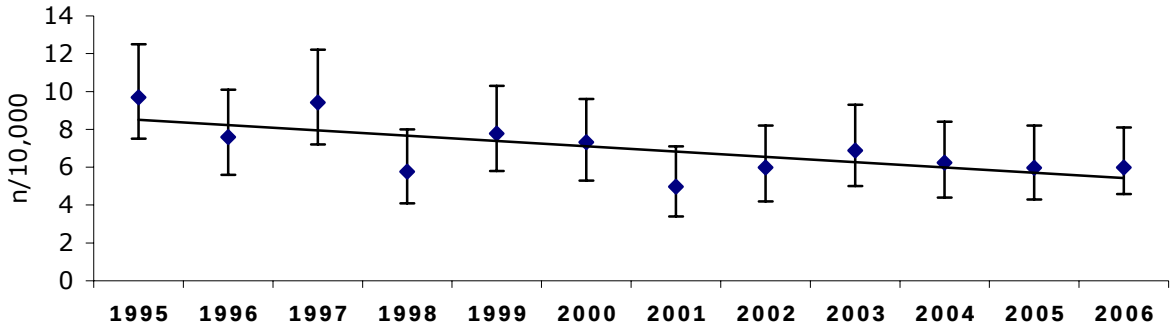
6.2 Spina bifida

British Paediatric Association code 741.00 – 741.99

Definition: Herniation or exposure of the spinal cord and/or meninges through an incompletely closed spine. Hydrocephalus may or may not be present.

NB: Unlike previous reports, spina bifida is not counted when present with anencephaly. (For 2005-2006 this involves only one case).

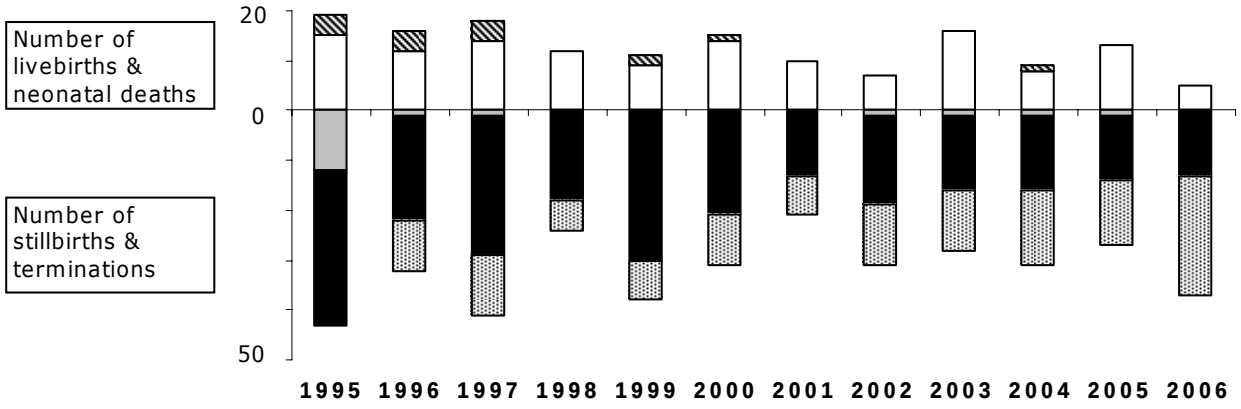
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N / 10,000	9.7	7.6	9.4	5.8	7.8	7.3	5.0	6.0	6.9	6.2	6.0	6.0

- From 1995 to 2006 there has been a statistically significant decrease in spina bifida, however, there has been no statistically significant decrease since 1998.

Number of cases and pregnancy outcomes, 1995-2006



Total number	62	48	59	36	49	46	31	38	44	40	40	42
Outcome	**Percent (%)**											
Neonatal death	6	8	7	0	4	2	0	0	0	3	0	0
Survived > 28 days	24	25	24	33	18	30	32	18	36	20	33	12
Stillbirth	19	2	2	0	0	0	0	3	2	3	3	0
TOP < 20 weeks	50	44	47	50	61	46	42	47	34	38	33	31
TOP >= 20 weeks*	0	21	20	17	16	22	26	32	27	38	33	57

**These cases are not identified in the dataset prior to 1996*

- Most pregnancies affected by this condition were terminated, with over half being terminated after 20 weeks gestation in 2006.

Spina bifida and associated birth defects, 2005-2006

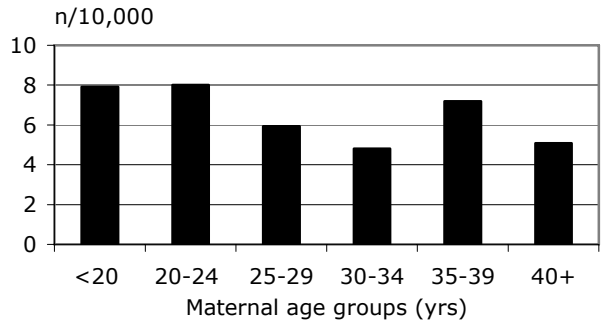
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	47	0	0	13	60	73.1
Chromosomal	4	1	0	1	6	7.3
Other same system	5	0	0	1	6	7.3
Other different systems	7	0	0	3	10	12.2
Total	63	1	0	18	82	100.0

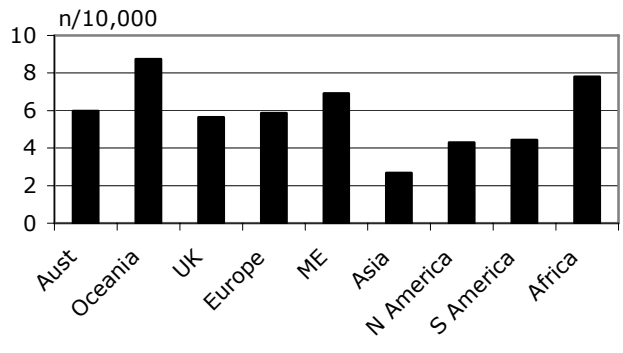
Five year summary of the prevalence of spina bifida and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=235)	**No.**	**%**	**PR**
Sex			
Male	115	*48.9*	5.7
Female	102	*43.4*	5.4
Indeterminate/ Unknown	18	*7.7*	
Plurality#			
Singleton	232	*98.7*	6.2
Multiple	3	*1.3*	2.2
Maternal (n=235)			
Maternal Age (yrs)			
<20	9	*3.8*	7.9
20-24	36	*15.3*	8.0
25-29	60	*25.5*	5.9
30-34	68	*28.9*	4.8
35-39	51	*21.7*	7.2
40+	7	*3.0*	5.1
Unknown	4	*1.7*	
Country of birth			
Australia	173	*73.6*	6.0
Oceania inc NZ	9	*3.8*	8.7
UK inc Eire	6	*2.6*	5.6
Europe	7	*3.0*	5.9
Middle East	6	*2.6*	6.9
Asia	10	*4.3*	2.7*
Nth America	1	*0.4*	4.3
Sth America	1	*0.4*	4.4
Africa	6	*2.6*	7.8
Unknown	16	*6.8*	

#excludes unknown

**statistically significant, p=0.01*





- The PR for Asian-born women was significantly lower than for Australian-born women. There were no statistically significant associations for sex of infant, plurality or maternal age for 2001-2006.

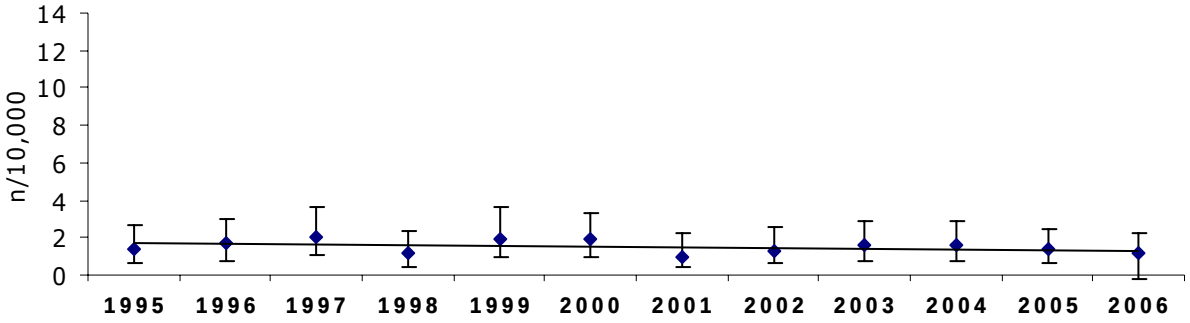
6.3 Encephalocele

British Paediatric Association code 742.00 – 742.09

Definition: Herniation of the brain and/or meninges through a defect in the skull.

NB: Unlike previous reports, encephalocele is not counted when present with anencephaly or spina bifida (For 2005-2006 there were no cases in this category).

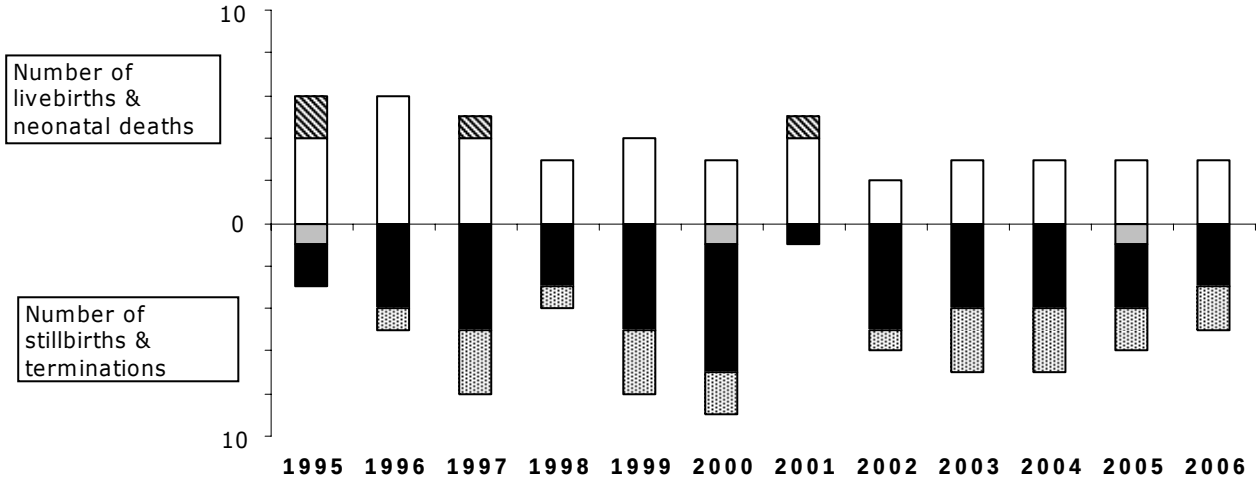
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	**1.4**	**1.7**	**2.1**	**1.1**	**1.9**	**1.9**	**1.0**	**1.3**	**1.6**	**1.6**	**1.3**	**1.1**

- The prevalence of this neural tube defect is much lower than anencephaly or spina bifida. From 1995 to 2006 there has been no statistically significant change in the prevalence of encephalocele.

Number of cases and pregnancy outcomes, 1995-2006



Total number	**9**	**11**	**13**	**7**	**12**	**12**	**6**	**8**	**10**	**10**	**9**	**8**
Outcome	**Percent (%)**											
Neonatal death	22	0	8	0	0	0	17	0	0	0	0	0
Survived > 28 days	44	55	31	43	33	25	67	25	30	30	33	38
Stillbirth	11	0	0	0	0	8	0	0	0	0	11	0
TOP < 20 weeks	22	36	38	43	42	50	17	63	40	40	33	38
TOP >= 20 weeks*	0	9	23	14	25	17	0	13	30	30	22	25

**These cases are not identified in the dataset prior to 1996*

- More than half of pregnancies affected by this condition were terminated since 1997.

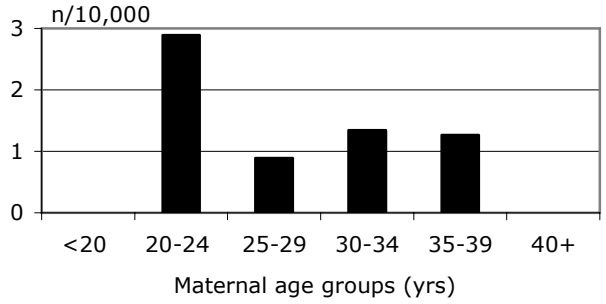
Encephalocele and associated birth defects, 2005-2006

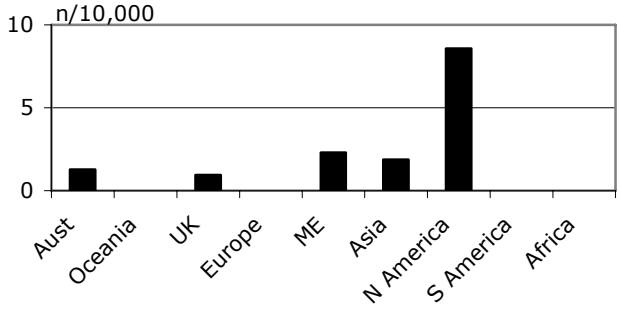
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	7	0	0	5	12	70.6
Chromosomal	1	0	0	0	1	5.9
Other same system	1	0	0	0	1	5.9
Other different systems	1	1	0	1	3	17.6
Total	10	1	0	6	17	100.0

Five year summary of the prevalence of encephalocele and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=51)	**No.**	**%**	**PR**
Sex			
Male	21	*41.2*	1.0
Female	22	*43.1*	1.2
Indeterminate/ Unknown	8	*15.7*	
Plurality#			
Singleton	49	*96.1*	1.3
Multiple	2	*3.9*	1.4
Maternal (n=51)			
Maternal Age (yrs)			
<20	0	*0.0*	0.0
20-24	13	*25.5*	2.9
25-29	9	*17.6*	0.9
30-34	19	*37.3*	1.3
35-39	9	*17.6*	1.3
40+	0	*0.0*	0.0
Unknown	1	*2.0*	
Country of birth			
Australia	37	*72.5*	1.3
Oceania inc NZ	0	*0.0*	0.0
UK inc Eire	1	*2.0*	0.9
Europe	0	*0.0*	0.0
Middle East	2	*3.9*	2.3
Asia	7	*13.7*	1.9
Nth America	2	*3.9*	8.6
Sth America	0	*0.0*	0.0
Africa	0	*0.0*	0.0
Unknown	2	*3.9*	

#excludes unknown





- Although some PRs were higher than others, due to small sample size there were no statistically significant associations for sex of infant, plurality, maternal age or country of birth for 2001-2006.

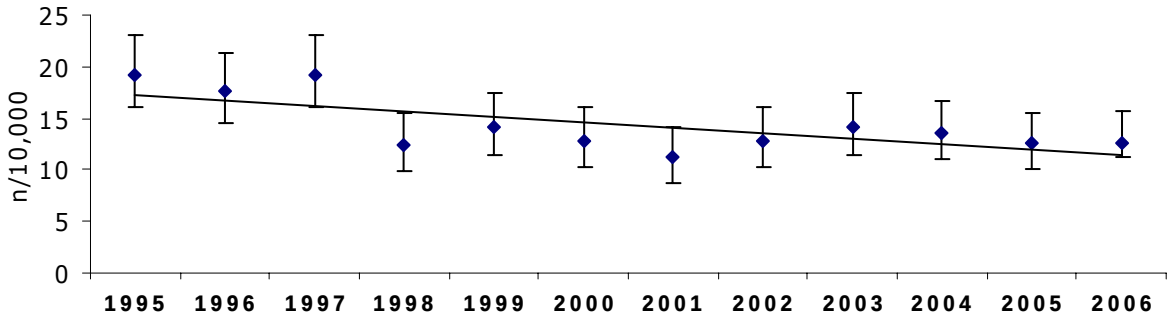
6.4 All neural tube defects combined

British Paediatric Association code 740.00 – 742.09

Definition: Includes all cases of anencephalus, spina bifida and encephalocele.

NB: These figures include all cases with neural tube defects (NTDs), not all NTD conditions ie. If a case has both anencephaly and spina bifida it will only be counted once.

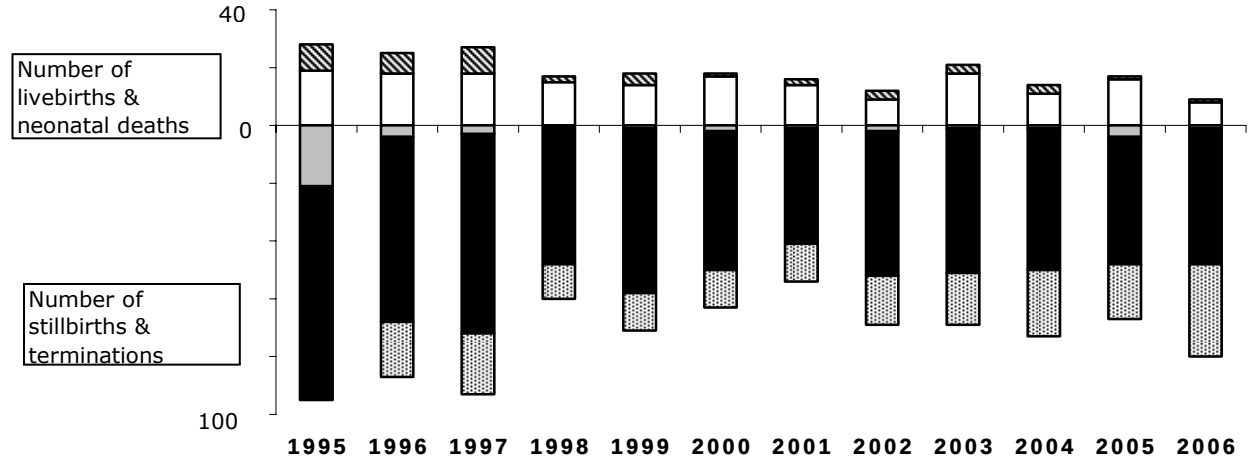
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	19.2	17.7	19.2	12.3	14.1	12.9	11.2	12.8	14.1	13.6	12.5	12.8

- From 1995 to 2006 there has been a statistically significant decline in the prevalence of all neural tube defects combined, however, there has been no statistically significant decrease since 1998.

Number of cases and pregnancy outcomes, 1995-2006



Total number	**123**	**112**	**120**	**77**	**89**	**81**	**70**	**81**	**90**	**87**	**84**	**90**
Outcome	**Percent (%)**											
Neonatal death	7	6	8	3	4	1	3	4	3	3	1	1
Survived > 28 days	15	16	15	19	16	21	20	11	20	13	19	9
Stillbirth	17	4	3	0	1	2	1	2	1	1	5	1
TOP < 20 weeks	60	57	58	62	64	59	57	62	56	56	52	53
TOP >= 20 weeks*	0	17	18	16	15	16	19	21	20	26	23	36

**These cases are not identified in the dataset prior to 1996*

- On average, 84% of all pregnancies affected by this condition were terminated either before or after 20 weeks gestation over the past five years.

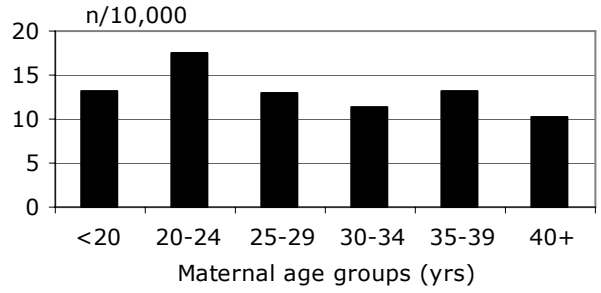
All neural tube defects (combined) and associated birth defects, 2005-2006

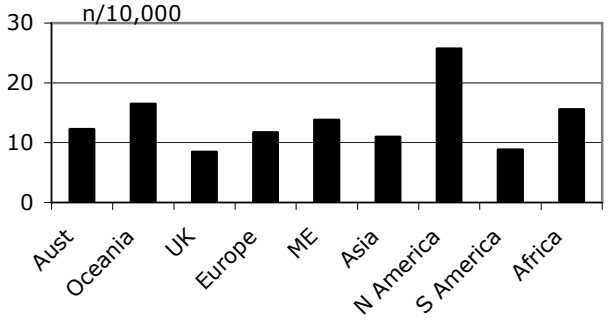
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	114	2	1	18	135	77.6
Chromosomal	8	1	0	1	10	5.7
Multiple NTD	1	0	0	0	1	0.6
Other same system	6	0	0	1	7	4.0
Other different systems	14	2	1	4	21	12.1
Total	143	5	2	24	174	100.0

Five year summary of the prevalence of all neural defects (combined) and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=502)	**No.**	**%**	**PR**
Sex			
Male	188	*37.5*	9.4
Female	171	*34.1*	9.0
Indeterminate/ Unknown	143	*28.5*	
Plurality#			
Singleton	491	*97.8*	13.0
Multiple	11	*2.2*	7.9
Maternal (n=502)			
Maternal Age (yrs)			
<20	15	*3.0*	13.2
20-24	79	*15.8*	17.6*
25-29	132	*26.3*	13.0
30-34	161	*32.1*	11.4
35-39	94	*18.7*	13.1
40+	14	*2.8*	10.2
Unknown	7	*1.4*	
Country of birth			
Australia	357	*71.1*	12.3
Oceania inc NZ	17	*3.4*	16.5
UK inc Eire	9	*1.8*	8.5
Europe	14	*2.8*	11.7
Middle East	12	*2.4*	13.8
Asia	41	*8.2*	11.0
Nth America	6	*1.2*	25.7
Sth America	2	*0.4*	8.9
Africa	12	*2.4*	15.6
Unknown	32	*6.4*	

#excludes unknown
**statistically significant, p=0.001*





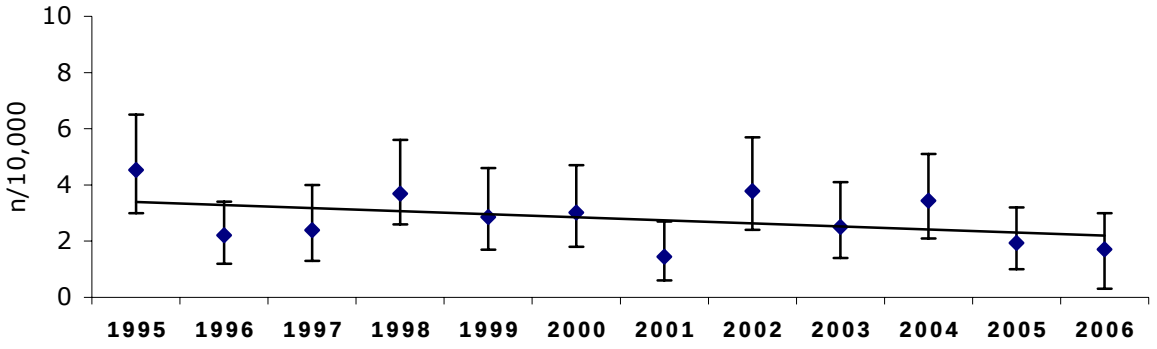
- The PR for women aged 20-24 years was significantly higher than for women aged 30-34 years. There were no statistically significant associations for sex of infant, plurality or country of birth for 2001-2006.

6.5 Microcephalus

British Paediatric Association code 742.19

Definition: A small cranium defined by an occipito-frontal circumference three standard deviations below the age-sex appropriate distribution curves.

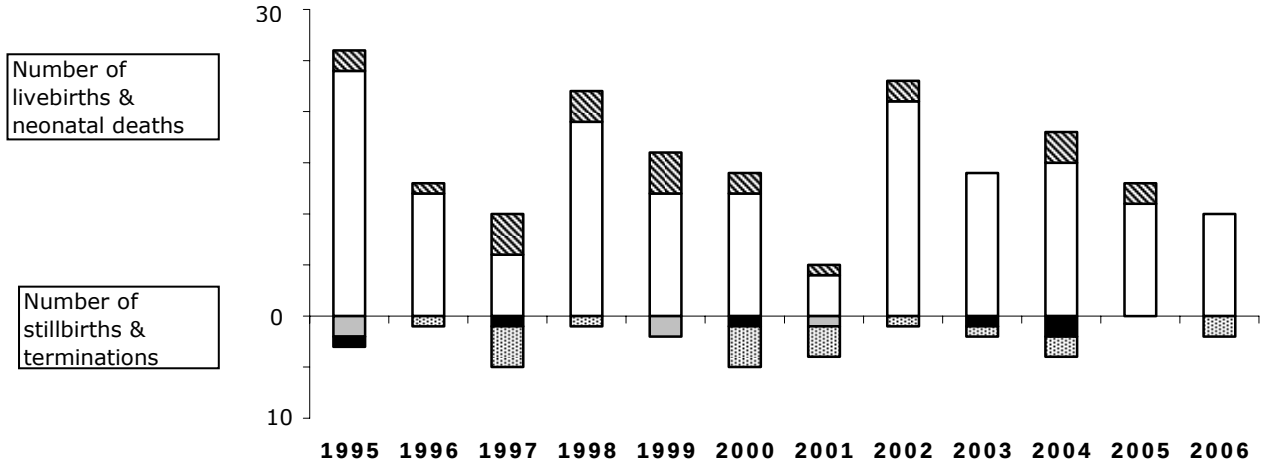
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	4.5	2.2	2.4	3.7	2.9	3.0	1.4	3.8	2.5	3.4	1.9	1.7

- From 1995 to 2006 there has been no statistically significant change in the prevalence of microcephalus

Number of cases and pregnancy outcomes, 1995-2006



Total number	29	14	15	23	18	19	9	24	16	22	13	12
Outcome	**Percent (%)**											
Neonatal death	7	7	27	13	22	11	11	8	0	14	15	0
Survived > 28 days	83	86	40	83	67	63	44	88	88	68	85	83
Stillbirth	7	0	0	0	11	0	11	0	0	0	0	0
TOP < 20 weeks	3	0	7	0	0	5	0	0	6	9	0	0
TOP >= 20 weeks*	0	7	27	4	0	21	33	4	6	9	0	17

**These cases are not identified in the dataset prior to 1996*

- On average, 76% of babies with this condition survived beyond 28 days in 2001 - 2006.

Microcephalus and associated birth defects, 2005-2006

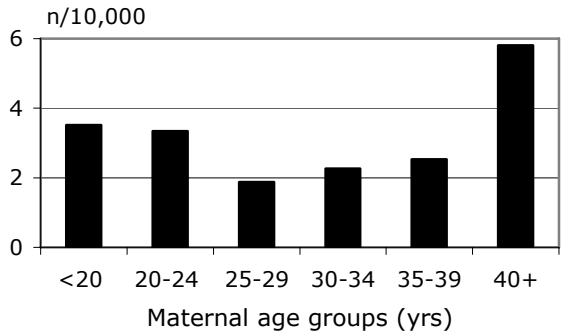
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	9	9	36.0
Chromosomal	0	0	0	2	2	8.0
Other same system	1	0	0	2	3	12.0
Other different systems	1	0	2	8	11	44.0
Total	2	0	2	21	25	100.0

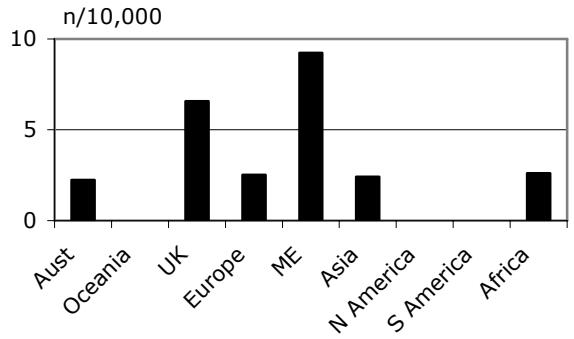
Five year summary of the prevalence of microcephalus and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=96)	**No.**	**%**	**PR**
Sex			
Male	44	*45.8*	2.2
Female	51	*53.1*	2.7
Indeterminate/ Unknown	1	*1.0*	
Plurality#			
Singleton	92	*95.8*	2.4
Multiple	4	*4.2*	2.9
Maternal (n=96)			
Maternal Age (yrs)			
<20	4	*4.2*	3.5
20-24	15	*15.6*	3.3
25-29	19	*19.8*	1.9
30-34	32	*33.3*	2.3
35-39	18	*18.8*	2.5
40+	8	*8.3*	5.8
Unknown	0	*0.0*	
Country of birth			
Australia	65	*67.7*	2.2
Oceania inc NZ	0	*0.0*	0.0
UK inc Eire	7	*7.3*	6.6
Europe	3	*3.1*	2.5
Middle East	8	*8.3*	9.2*
Asia	9	*9.4*	2.4
Nth America	0	*0.0*	0.0
Sth America	0	*0.0*	0.0
Africa	2	*2.1*	2.6
Unknown	2	*2.1*	

#excludes unknown

**statistically significant, p<0.0001*





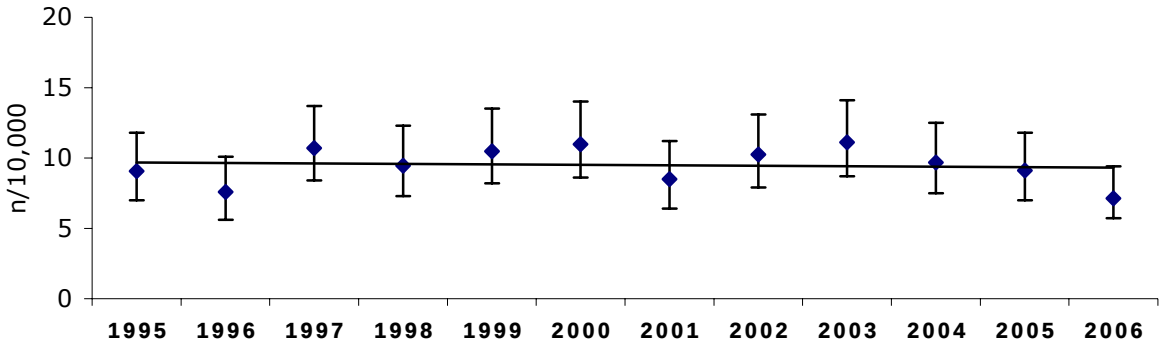
- The PR for Middle-Eastern born women was significantly higher than for Australian-born women. Although some PRs were higher than others, due to small sample size there were no statistically significant associations for sex of infant, plurality or maternal age for 2001-2006.

6.6 Hydrocephalus

British Paediatric Association code 742.30 – 742.39

Definition: Dilatation of the cerebral vessels not associated with primary brain atrophy of brain with or without enlargement of the head and diagnosed at birth. These cases exclude hydrocephalus associated with spina bifida or encephalocele.

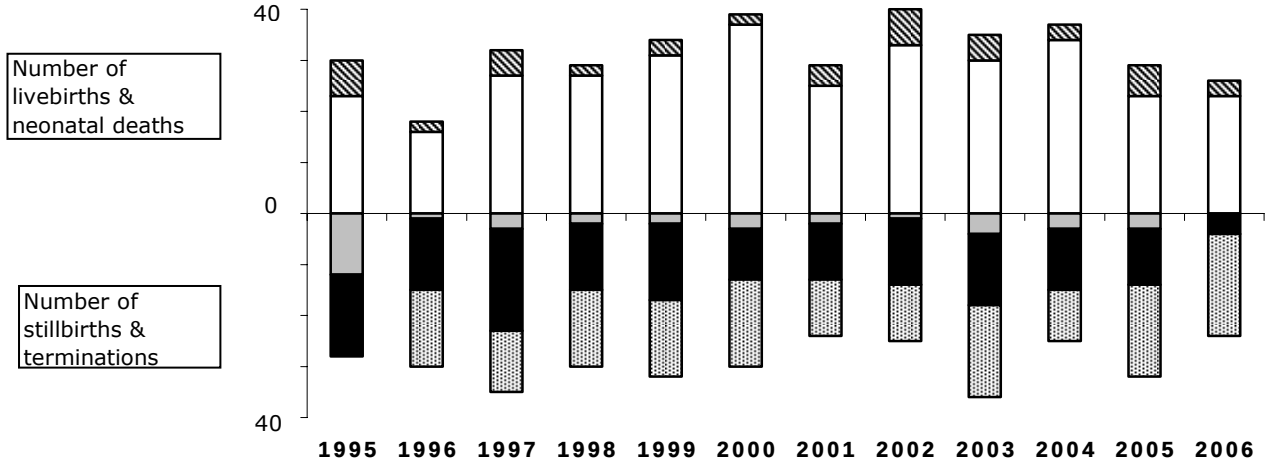
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	9.1	7.6	10.7	9.5	10.5	11.0	8.5	10.2	11.1	9.7	9.1	7.1

- From 1995 to 2006 there has been no statistically significant change in the prevalence of hydrocephalus.

Number of cases and pregnancy outcomes, 1995-2006



Total number	58	48	67	59	66	69	53	65	71	62	61	50
Outcome	**Percent (%)**											
Neonatal death	12	4	7	3	5	3	8	11	7	5	10	6
Survived > 28 days	40	33	40	46	47	54	47	51	42	55	38	46
Stillbirth	21	2	4	3	3	4	4	2	6	5	5	0
TOP < 20 weeks	28	29	30	22	23	14	21	20	20	19	18	8
TOP >= 20 weeks*	0	31	25	25	23	25	21	17	25	16	30	40

**These cases are not identified in the dataset prior to 1996*

- On average, 47% of babies with this condition survived more than 28 days in 2001 – 2006.

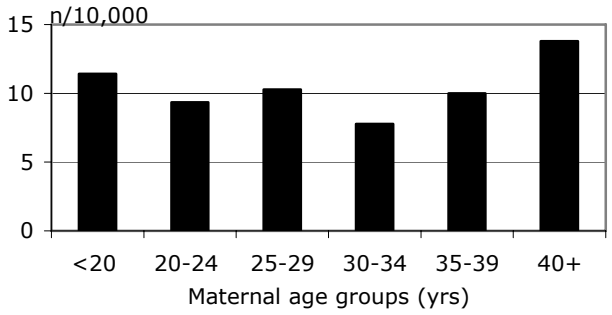
Hydrocephalus and associated birth defects, 2005-2006

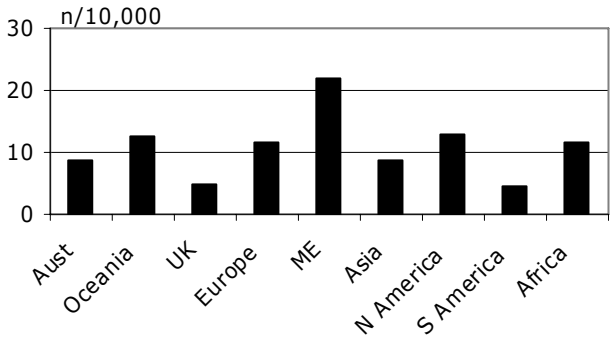
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	15	1	3	28	47	42.3
Chromosomal	10	1	2	1	14	12.6
Other same system	9	0	0	3	12	10.8
Other different systems	19	1	4	14	38	34.2
Total	53	3	9	46	111	100.0

Five year summary of the prevalence of hydrocephalus and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=362)	**No.**	**%**	**PR**
Sex			
Male	203	*56.1*	10.1
Female	153	*42.3*	8.0
Indeterminate/ Unknown	6	*1.7*	
Plurality#			
Singleton	324	*89.5*	8.6
Multiple	38	*10.5*	27.4*
Maternal (n=360)			
Maternal Age (yrs)			
<20	13	*3.6*	11.4
20-24	42	*11.6*	9.3
25-29	104	*28.9*	10.3
30-34	110	*30.6*	7.8
35-39	71	*19.7*	10.0
40+	19	*5.3*	13.8
Unknown	1	*0.3*	
Country of birth			
Australia	251	*69.3*	8.6
Oceania inc NZ	13	*3.6*	12.6
UK inc Eire	5	*1.4*	4.7
Europe	14	*3.9*	11.7
Middle East	19	*5.2*	21.9**
Asia	33	*9.1*	8.8
Nth America	3	*0.8*	12.9
Sth America	1	*0.3*	4.4
Africa	9	*2.5*	11.7
Unknown	12	*3.3*	

#excludes unknown
** statistically significant, p<0.0001*
***statistically significant, P<0.001*





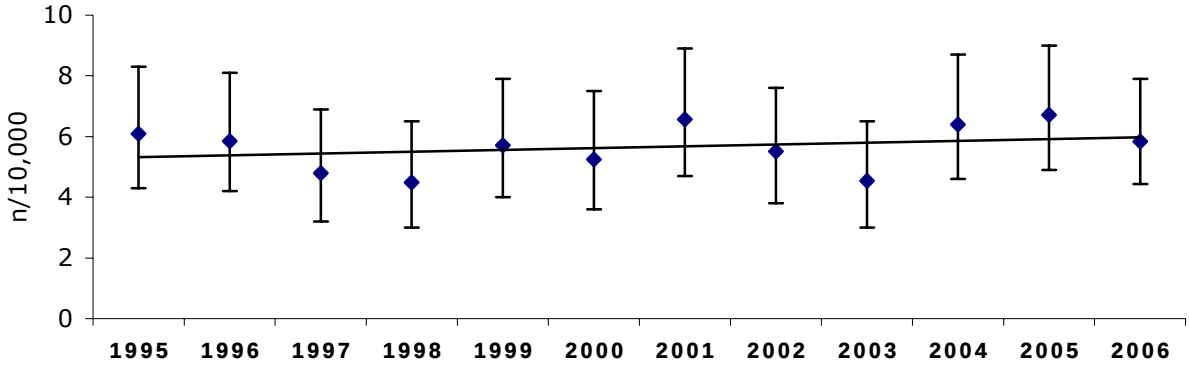
- Multiple births had a significantly higher PR than singleton births. The PR for women born in the Middle East was significantly higher than for women born in Australia, UK including Eire and Asia. There were no statistically significant associations for sex of infant or maternal age for 2001-2006.

6.7 Transposition of great vessels

British Paediatric Association code 745.10 – 745.19

Definition: The aorta exits from the right ventricle and the pulmonary artery from the left ventricle, with or without other cardiac defects. Includes double outlet right ventricle, so-called corrected transposition.

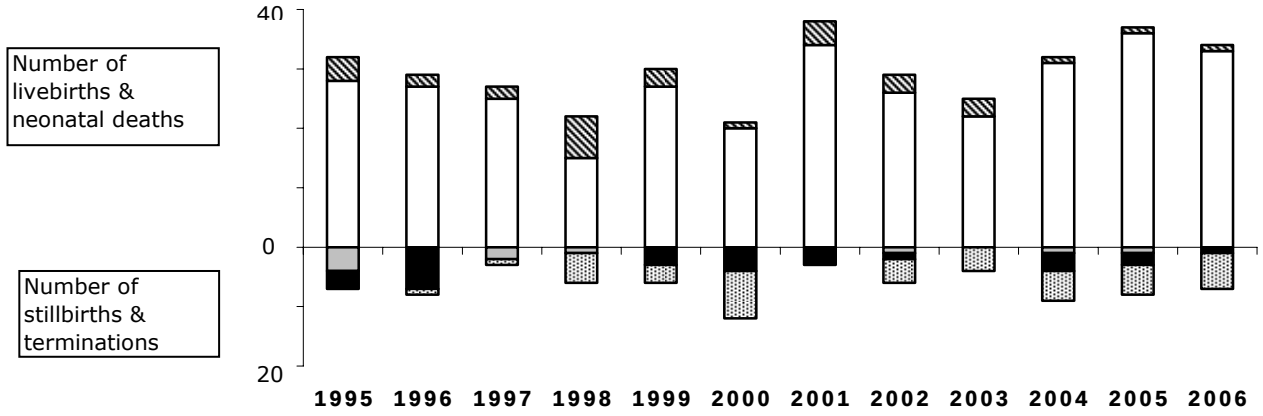
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	6.1	5.9	4.8	4.5	5.7	5.3	6.6	5.5	4.5	6.4	6.7	5.8

- From 1995 to 2006 there has been no significant change in the prevalence of transposition of the great vessels.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	39	37	30	28	36	33	41	35	29	41	45	41
Outcome	**Percent (%)**											
Neonatal death	10	5	7	25	8	3	10	9	10	2	2	2
Survived > 28 days	72	73	83	54	75	61	83	74	76	76	80	80
Stillbirth	10	0	7	4	0	0	0	3	0	2	2	0
TOP < 20 weeks	8	19	0	0	8	12	7	3	0	7	4	2
TOP >= 20 weeks*	0	3	3	18	8	24	0	11	14	12	11	15

**These cases are not identified in the dataset prior to 1996*

- On average, 80% of babies with this condition survived beyond 28 days in 2001 - 2006.

Transposition of great vessels and associated birth defects, 2005-2006

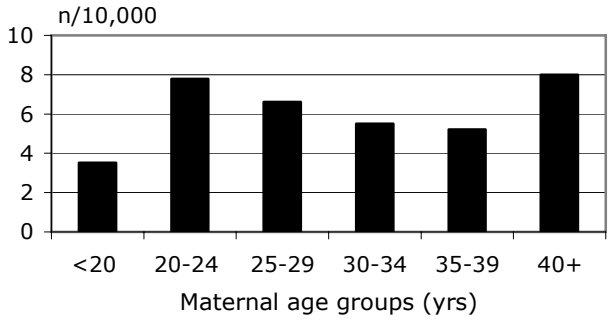
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	4	4	4.7
Chromosomal	3	0	1	2	6	7.0
Other same system	4	0	1	52	57	66.3
Other different systems	7	1	0	11	19	22.1
Total	14	1	2	69	86	100.0

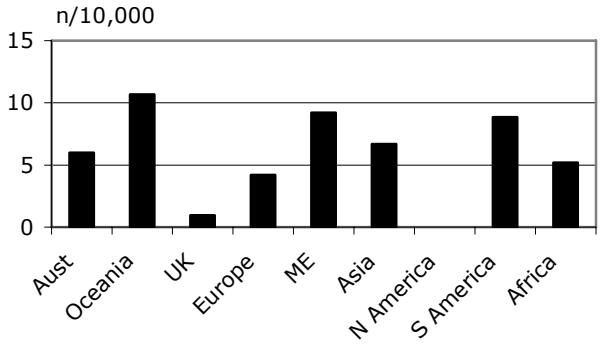
Five year summary of the prevalence of transposition of great vessels and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=232)	**No.**	**%**	**PR**
Sex			
Male	142	*61.2*	7.1*
Female	88	*37.9*	4.6
Indeterminate/ Unknown	2	*0.9*	
Plurality#			
Singleton	227	*97.8*	6.0
Multiple	5	*2.2*	3.6
Maternal (n=232)			
Maternal Age (yrs)			
<20	4	*1.7*	3.5
20-24	35	*15.1*	7.8
25-29	67	*28.9*	6.6
30-34	78	*33.6*	5.5
35-39	37	*15.9*	5.2
40+	11	*4.7*	8.0
Unknown	0	*0.0*	
Country of birth			
Australia	175	*75.4*	6.0
Oceania inc NZ	11	*4.7*	10.7
UK inc Eire	1	*0.4*	0.9
Europe	5	*2.2*	4.2
Middle East	8	*3.4*	9.2
Asia	25	*10.8*	6.7
Nth America	0	*0.0*	0.0
Sth America	2	*0.9*	8.9
Africa	4	*1.7*	5.2
Unknown	1	*0.4*	

#excludes unknown

** statistically significant, p<0.01*





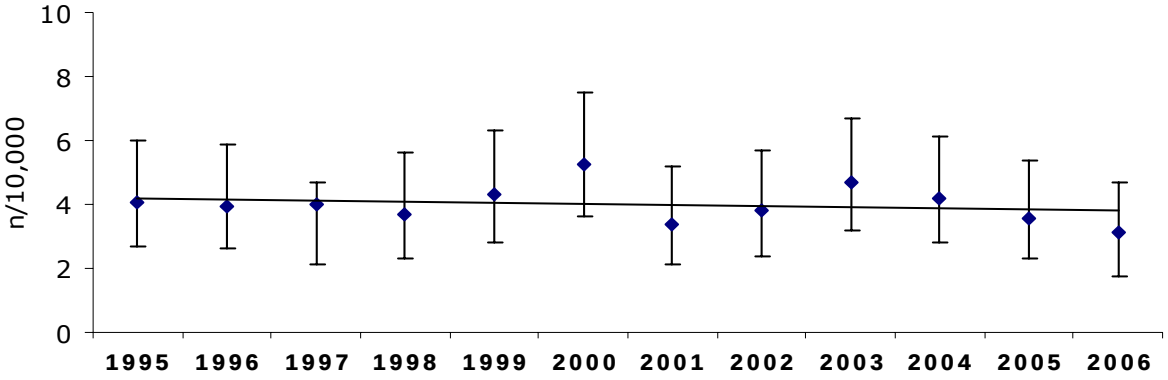
- PR for males was significantly higher than for females. Although some PRs were higher than others, due to small numbers there were no statistically significant associations for plurality, country of birth or maternal age for 2001-2006.

6.8 Tetralogy of Fallot

British Paediatric Association code 745.20

Definition: A condition characterised by ventricular septal defect, overriding aorta, infundibular pulmonary stenosis and often right ventricular hypertrophy.

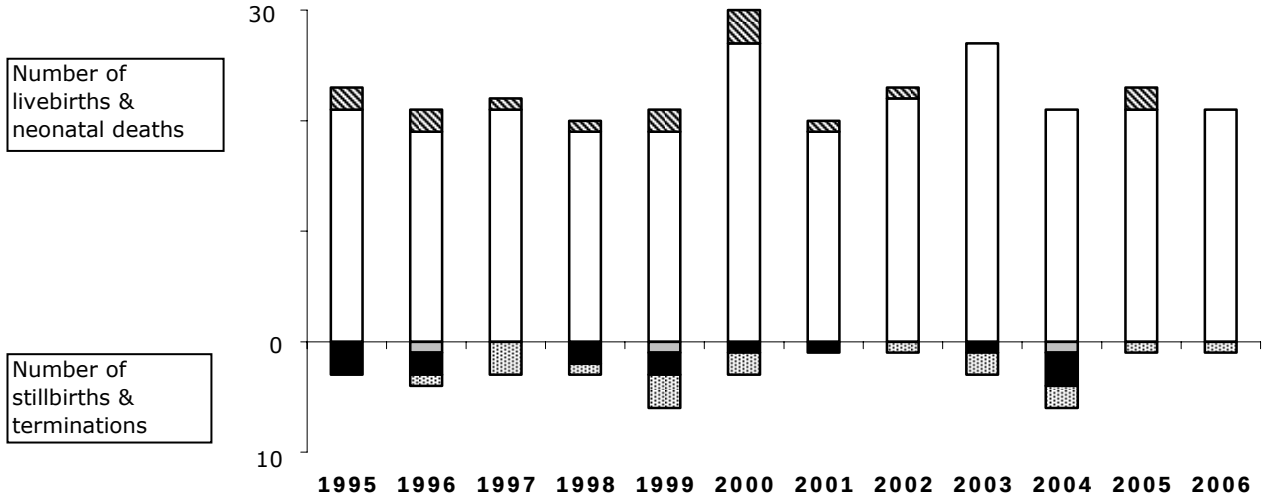
Trend in prevalence rates, 1995-2006



N/10,000	4.1	4.0	4.0	3.7	4.3	5.3	3.4	3.8	4.7	4.2	3.6	3.1

- From 1995 to 2006 there has been no significant change in the prevalence of Tetralogy of Fallot.

Number of cases and pregnancy outcomes, 1995-2006



Total number	26	25	25	23	27	33	21	24	30	27	24	22
Outcome	**Percent (%)**											
Neonatal death	8	8	4	4	7	9	5	4	0	0	8	0
Survived > 28 days	81	76	84	83	70	82	90	92	90	78	88	95
Stillbirth	0	4	0	0	4	0	0	0	0	4	0	0
TOP < 20 weeks	12	8	0	9	7	3	5	0	3	11	0	0
TOP >= 20 weeks*	0	4	12	4	11	6	0	4	7	7	4	5

**These cases are not identified in the dataset prior to 1996*

- On average, 89% of babies with this condition have survived beyond 28 days in 2001 - 2006.

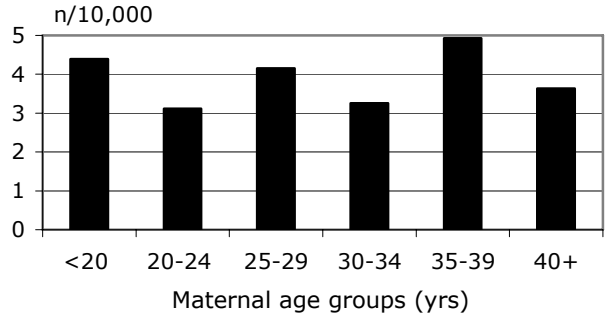
Tetralogy of Fallot and associated birth defects, 2005-2006

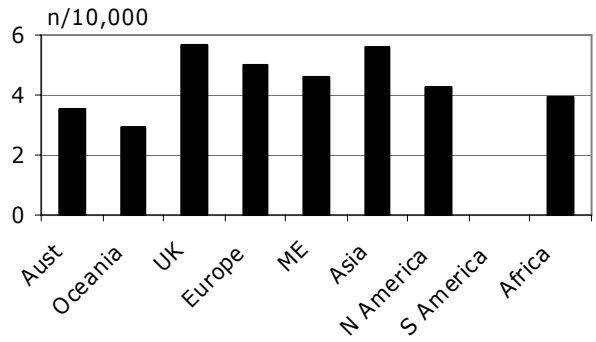
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	10	10	21.7
Chromosomal	2	0	1	2	5	10.9
Other same system	0	0	0	15	15	32.6
Other different systems	0	0	1	15	16	34.8
Total	2	0	2	42	46	100.0

Five year summary of the prevalence of Tetralogy of Fallot and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=148)	**No.**	**%**	**PR**
Sex			
Male	88	*59.5*	4.4
Female	59	*39.9*	3.1
Indeterminate/ Unknown	1	*0.7*	
Plurality#			
Singleton	139	*93.9*	3.7
Multiple	9	*6.1*	6.5
Maternal (n=148)			
Maternal Age (yrs)			
<20	5	*3.4*	4.4
20-24	14	*9.5*	3.1
25-29	42	*28.4*	4.1
30-34	46	*31.1*	3.3
35-39	35	*23.6*	4.9
40+	5	*3.4*	3.6
Unknown	1	*0.7*	
Country of birth			
Australia	103	*69.6*	3.5
Oceania inc NZ	3	*2.0*	2.9
UK inc Eire	6	*4.1*	5.6
Europe	6	*4.1*	5.0
Middle East	4	*2.7*	4.6
Asia	21	*14.2*	5.6
Nth America	1	*0.7*	4.3
Sth America	0	*0.0*	0.0
Africa	3	*2.0*	3.9
Unknown	1	*0.7*	

*excludes unknown





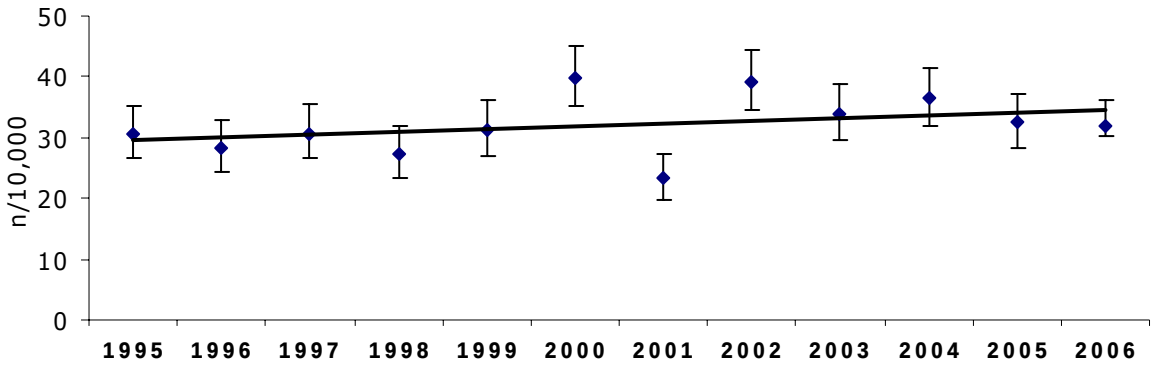
- There were no statistically significant associations for sex of infant, plurality, maternal age or country of birth for 2001-2006.

6.9 Ventricular septal defect

British Paediatric Association code 745.40 – 745.49

Definition: A defect in the septum between the left and right ventricles of the heart, which permits blood to be shunted between them. Excludes VSD as part of Tetralogy of Fallot.

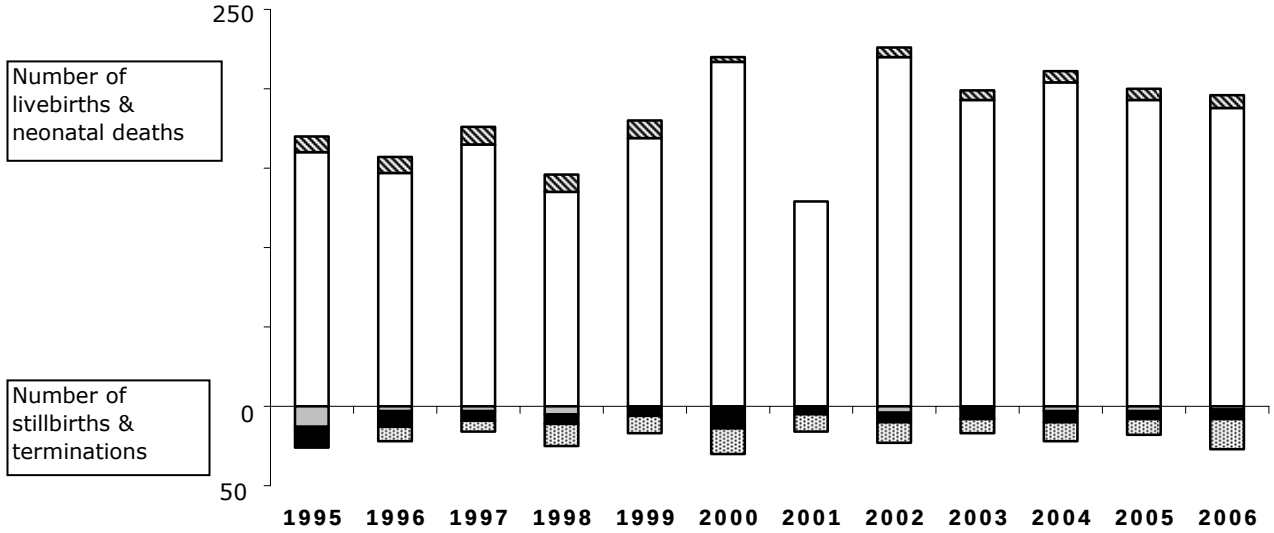
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	**30.6**	**28.3**	**30.7**	**27.4**	**31.3**	**39.8**	**23.2**	**39.2**	**33.8**	**36.4**	**32.5**	**31.8**

- From 1995 to 2006 there has been no statistically significant change in the prevalence of ventricular septal defect.

Number of cases and pregnancy outcomes, 1995-2006



Total number	**196**	**179**	**192**	**171**	**197**	**250**	**145**	**249**	**216**	**233**	**218**	**223**
Outcome	**Percent (%)**											
Neonatal death	5	6	6	6	6	1	0	2	3	3	3	4
Survived > 28 days	82	82	86	79	86	87	89	88	89	88	89	84
Stillbirth	7	2	2	3	1	0	0	2	0	1	1	1
TOP < 20 weeks	7	6	3	4	3	6	3	2	3	3	2	3
TOP >= 20 weeks*	0	5	4	8	6	6	8	5	4	5	5	9

**These cases are not identified in the dataset prior to 1996*

- On average, 88% of babies with this condition survived beyond 28 days in 2001-2006.

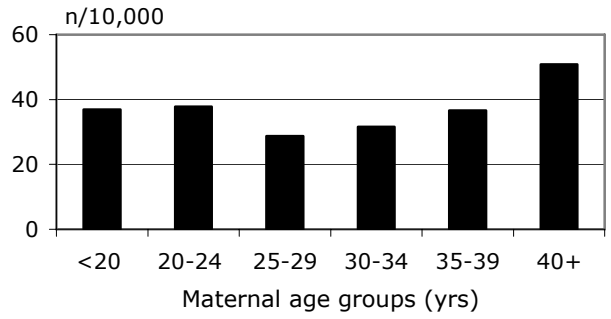
Ventricular septal defect and associated birth defects, 2005-2006

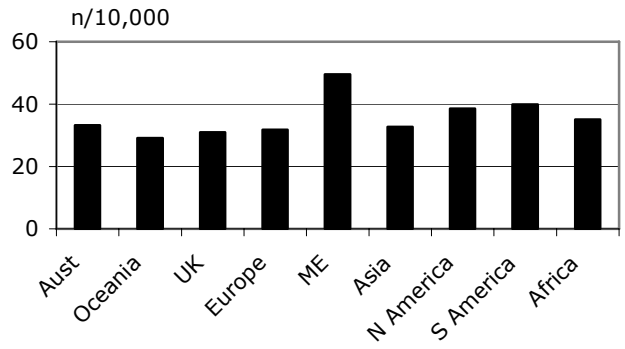
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	197	197	44.7
Chromosomal	20	2	8	25	55	12.5
Other same system	10	2	1	108	121	27.4
Other different systems	10	1	6	51	68	15.4
Total	40	5	15	381	441	100.0

Five year summary of the prevalence of ventricular septal defect and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=1,284)	**No.**	**%**	**PR**
Sex			
Male	642	*50.0*	32.0
Female	635	*49.5*	33.4
Indeterminate/ Unknown	7	*0.5*	
Plurality#			
Singleton	1,217	*94.8*	32.3
Multiple	67	*5.2*	48.3*
Maternal (n=1,282)			
Maternal Age (yrs)			
<20	42	*3.3*	36.9
20-24	170	*13.2*	37.8
25-29	291	*22.7*	28.7
30-34	447	*34.9*	31.6
35-39	260	*20.3*	36.6**
40+	70	*5.1*	50.8**
Unknown	2	*0.2*	
Country of birth			
Australia	967	*75.4*	33.2
Oceania inc NZ	30	*2.3*	29.1
UK inc Eire	33	*2.6*	31.0
Europe	38	*3.0*	31.8
Middle East	43	*3.4*	49.6***
Asia	122	*9.5*	32.7
Nth America	9	*0.7*	38.6
Sth America	9	*0.7*	39.9
Africa	27	*2.1*	35.1
Unknown	4	*0.3*	

#excludes unknown
**statistically significant, p=0.001*
***statistically significant, p=0.001*
****statistically significant, p<0.01*





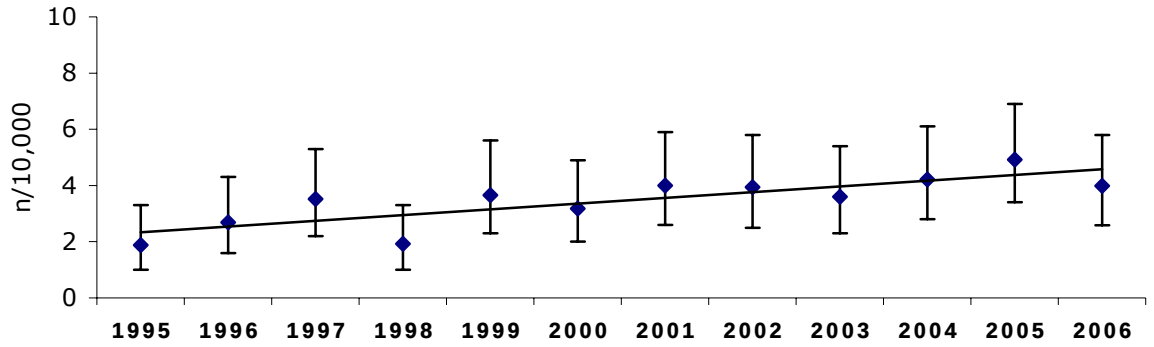
- PR for multiple births was significantly higher than for singletons. The PR was significantly higher for women aged 35 years or more compared to women aged 25-29 years. The PR for women born in the Middle East was significantly higher than for Australian born women. There were no significant associations for sex of infant.

6.10 Hypoplastic left heart syndrome

British Paediatric Association code 746.7

Definition: Hypoplastic left ventricle associated with aorta and/or mitral valve atresia.

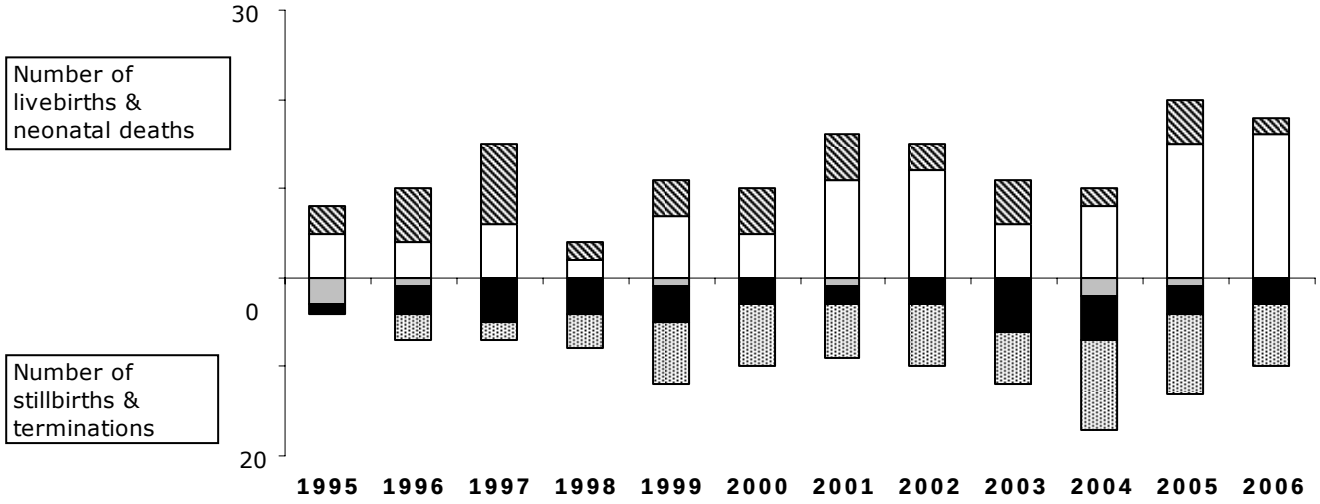
Trend in prevalence rates, 1995-2006



N/10,000	1.9	2.7	3.5	1.9	3.7	3.2	4.0	3.9	3.6	4.2	4.9	4.0

- From 1995 to 2006 there has been a statistically significant increase in the prevalence of hypoplastic left heart syndrome. This increase is thought to be related to non-Victorian residents coming to Victoria (RWH/RCH) for birth and subsequent treatment, following prenatal diagnosis of HLHS. However from 2001-2006 there has been no significant increase of this condition.

Number of cases and pregnancy outcomes, 1995-2006



Total number	12	17	22	12	23	20	25	25	23	27	33	28
Outcome	**Percent (%)**											
Neonatal death	25	35	41	17	17	25	20	12	22	7	15	7
Survived > 28 days	42	24	27	17	30	25	44	48	26	30	45	57
Stillbirth	25	6	0	0	4	0	4	0	0	7	3	0
TOP < 20 weeks	8	18	23	33	17	15	8	12	26	19	9	11
TOP >= 20 weeks*	0	18	9	33	30	35	24	28	26	37	27	25

**These cases are not identified in the dataset prior to 1996*

- Approximately 50% of pregnancies with this condition were terminated since 1998 and over the last five years the survival rate beyond 28 days has ranged from 26 to 57% of all cases.

Hypoplastic left heart syndrome and associated birth defects, 2005-2006

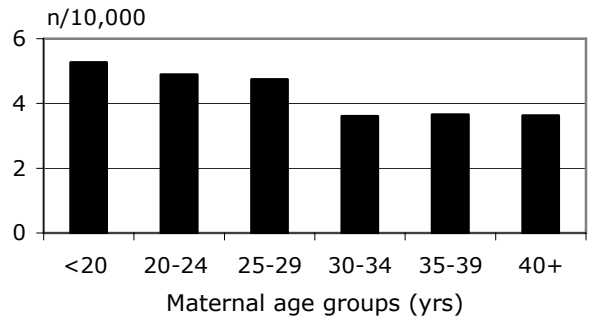
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	12	0	3	13	28	45.9
Chromosomal	6	0	0	0	6	9.8
Other same system	2	0	1	12	15	24.6
Other different systems	2	1	3	6	12	19.7
Total	22	1	7	31	61	100.0

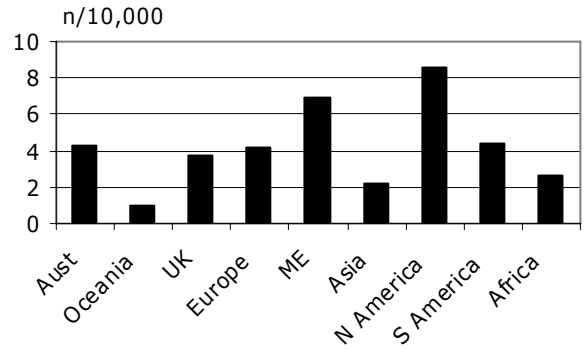
Five year summary of the prevalence of hypoplastic left heart syndrome and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=161)	**No.**	**%**	**PR**
Sex			
Male	103	*64.0*	5.1*
Female	53	*32.9*	2.8
Indeterminate/ Unknown	5	*3.1*	
Plurality#			
Singleton	157	*97.5*	4.2
Multiple	4	*2.5*	2.9
Maternal (n=161)			
Maternal Age (yrs)			
<20	6	*3.7*	5.3
20-24	22	*13.7*	4.9
25-29	48	*29.8*	4.7
30-34	51	*31.7*	3.6
35-39	26	*16.1*	3.7
40+	5	*3.1*	3.6
Unknown	3	*1.9*	
Country of birth			
Australia	124	*77.0*	4.3
Oceania inc NZ	1	*0.6*	1.0
UK inc Eire	4	*2.5*	3.8
Europe	5	*3.1*	4.2
Middle East	6	*3.7*	6.9
Asia	8	*5.0*	2.1
Nth America	2	*1.2*	8.6
Sth America	1	*0.6*	4.4
Africa	2	*1.2*	2.6
Unknown	8	*5.0*	

#excludes unknown

**statistically significant, p<0.001*





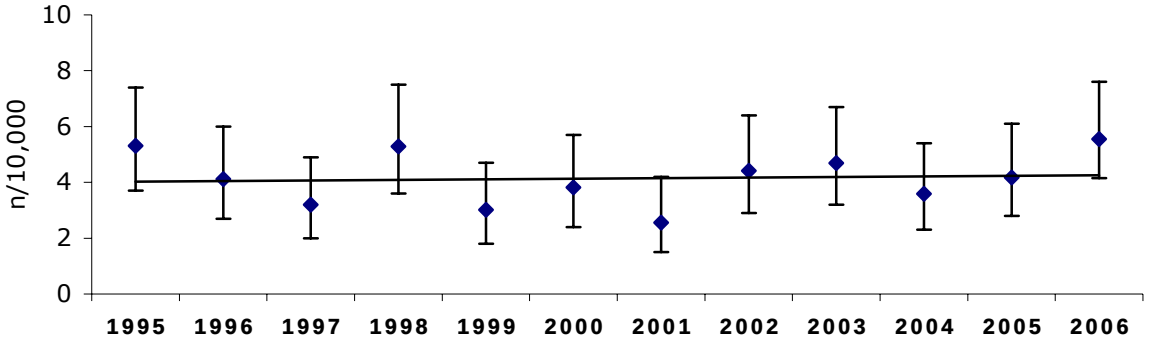
- PR for males was significantly higher than for females. There were no statistically significant associations for plurality, maternal age or country of birth for 2001-2006.

6.11 Coarctation of aorta

British Paediatric Association code 746.10 – 747.19

Definition: An obstruction in the descending aorta, almost invariably (98%) at the insertion of the ductus arteriosus.

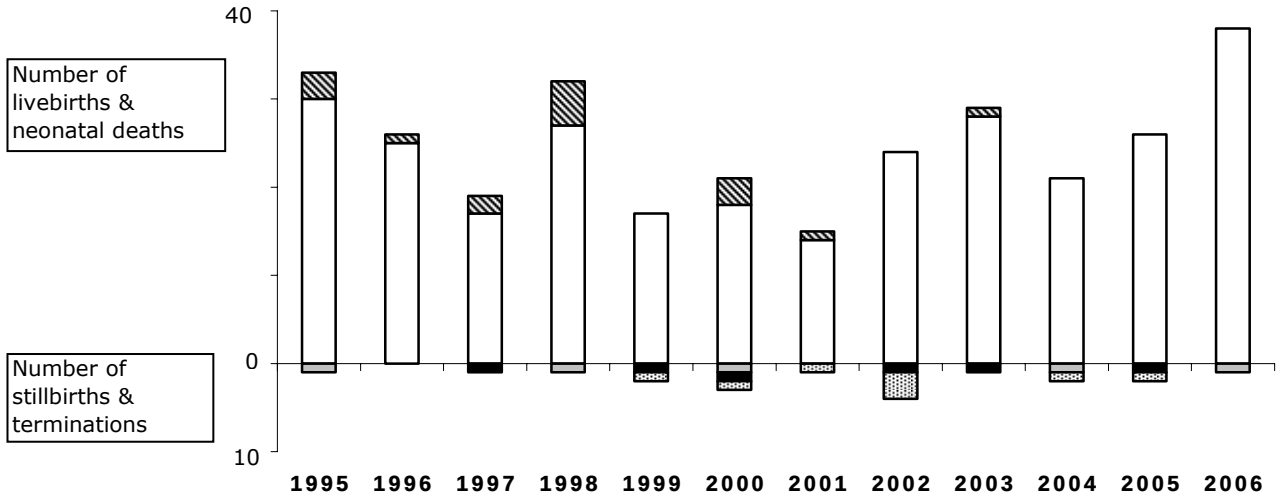
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	5.3	4.1	3.2	5.3	3.0	3.8	2.6	4.4	4.7	3.6	4.2	5.6

- From 1995 to 2006 there has been no statistically significant change in the prevalence of coarctation of aorta.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	34	26	20	33	19	24	16	28	30	23	28	39
Outcome	**Percent (%)**											
Neonatal death	9	4	10	15	0	13	6	0	3	0	0	0
Survived > 28 days	88	96	85	82	89	75	88	86	93	91	93	97
Stillbirth	3	0	0	3	0	4	0	0	0	4	0	3
TOP < 20 weeks	0	0	5	0	5	4	0	4	3	0	4	0
TOP >= 20 weeks*	0	0	0	0	5	4	6	11	0	4	4	0

**These cases are not identified in the dataset prior to 1996*

- On average, 91% of babies with this condition survived beyond 28 days from 2001-2006.

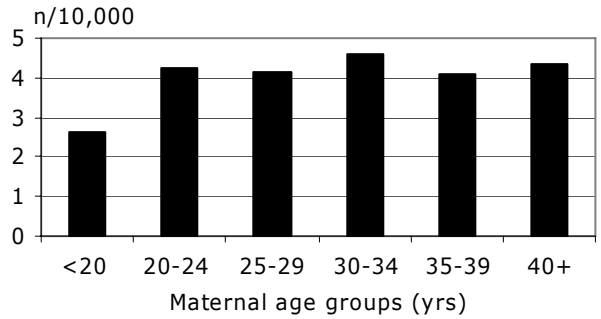
Coarctation of aorta and associated birth defects, 2005-2006

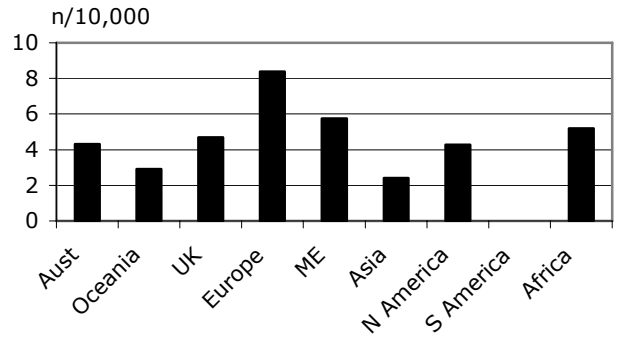
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	1	0	10	11	16.4
Chromosomal	1	0	0	6	7	10.4
Other same system	1	0	0	41	42	62.7
Other different systems	0	0	0	7	7	10.4
Total	2	1	0	64	67	100.0

Five year summary of the prevalence of coarctation of aorta and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=164)	**No.**	**%**	**PR**
Sex			
Male	93	*56.7*	4.6
Female	71	*43.3*	3.7
Indeterminate/ Unknown	0	*0.0*	
Plurality#			
Singleton	159	*97.0*	4.2
Multiple	5	*3.0*	3.6
Maternal (n=164)			
Maternal Age (yrs)			
<20	3	*1.8*	2.6
20-24	19	*11.6*	4.2
25-29	42	*25.6*	4.1
30-34	65	*39.6*	4.6
35-39	29	*17.7*	4.1
40+	6	*3.7*	4.4
Unknown	0	*0.0*	
Country of birth			
Australia	126	*76.8*	4.3
Oceania inc NZ	3	*1.8*	2.9
UK inc Eire	5	*3.0*	4.7
Europe	10	*6.1*	8.4
Middle East	5	*3.0*	5.8
Asia	9	*5.5*	2.4
Nth America	1	*0.6*	4.3
Sth America	0	*0.0*	0.0
Africa	4	*2.4*	5.2
Unknown	1	*0.6*	

#excludes unknown





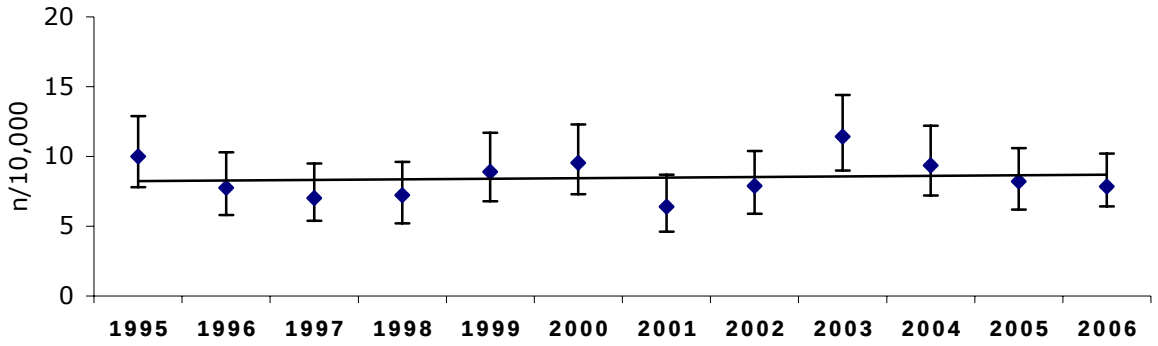
- There were no statistically significant associations for sex of infant, plurality, maternal age or country of birth for 2001-2006.

6.12 Cleft palate

British Paediatric Association code 749.00 – 749.09

Definition: A closure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip. Excludes cleft palate with cleft lip, cleft uvula, functional short palate and high narrow palate.

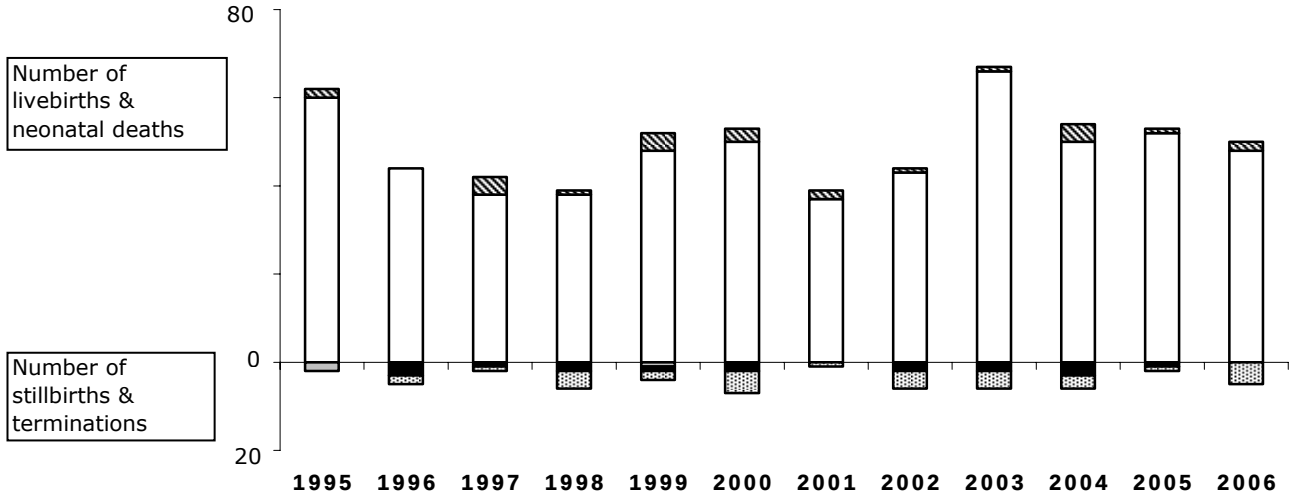
Trend in prevalence rates, 1995-2006



N/10,000	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	10.0	**7.8**	**7.0**	**7.2**	**8.9**	**9.5**	**6.4**	**7.9**	**11.4**	**9.4**	**8.2**	**7.8**

- From 1995 to 2006 there has been no statistically significant change in the prevalence of cleft palate.

Number of cases and pregnancy outcomes, 1995-2006



Total number	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**2003**	**2004**	**2005**	**2006**
Outcome	**Percent (%)**											
Neonatal death	3	0	9	2	7	5	5	2	1	7	2	4
Survived > 28 days	94	90	86	84	86	83	93	86	90	83	95	87
Stillbirth	3	0	0	0	2	0	0	0	0	0	0	0
TOP < 20 weeks	0	6	2	4	2	3	0	4	3	5	2	0
TOP >= 20 weeks*	0	4	2	9	4	8	3	8	5	5	2	9

**These cases are not identified in the dataset prior to 1996*

- On average, 89% of babies with this condition survived beyond 28 days in 2001 - 2006.

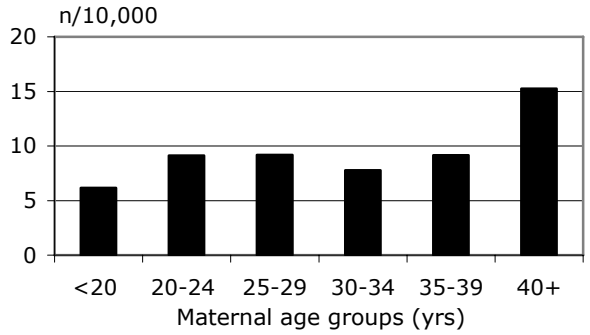
Cleft palate and associated birth defects, 2005-2006

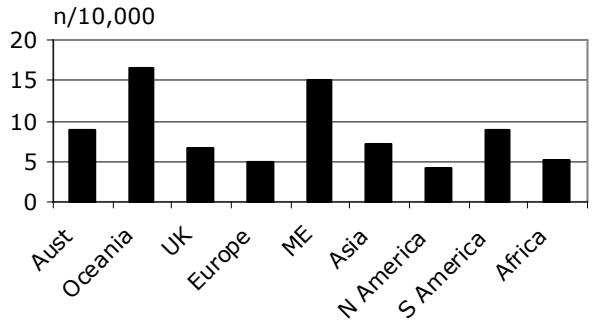
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	62	62	56.4
Chromosomal	4	0	1	4	9	8.2
Pierre Robin syndrome	0	0	0	20	20	18.2
Other same system	0	0	0	1	1	0.9
Other different systems	3	0	2	13	18	16.4
Total	7	0	3	100	110	100.0

Five year summary of the prevalence of cleft palate and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=338)	**No.**	**%**	**PR**
Sex			
Male	151	*44.7*	7.5
Female	187	*55.3*	9.8
Indeterminate/ Unknown	0	*0.0*	
Plurality#			
Singleton	326	*96.4*	8.6
Multiple	12	*3.6*	8.6
Maternal (n=337)			
Maternal Age (yrs)			
<20	7	*2.1*	6.2
20-24	41	*12.1*	9.1
25-29	93	*27.5*	9.2
30-34	110	*32.5*	7.8
35-39	65	*19.2*	9.2
40+	21	*6.2*	15.3*
Unknown	0	*0.0*	0.0
Country of birth			
Australia	259	*76.6*	8.9
Oceania inc NZ	17	*5.0*	16.5
UK inc Eire	7	*2.1*	6.6
Europe	6	*1.8*	5.0
Middle East	13	*3.8*	15.0
Asia	27	*8.0*	7.2
Nth America	1	*0.3*	4.3
Sth America	2	*0.6*	8.9
Africa	4	*1.2*	5.2
Unknown	1	*0.3*	

#excludes unknown
**statistically significant, p<0.01*





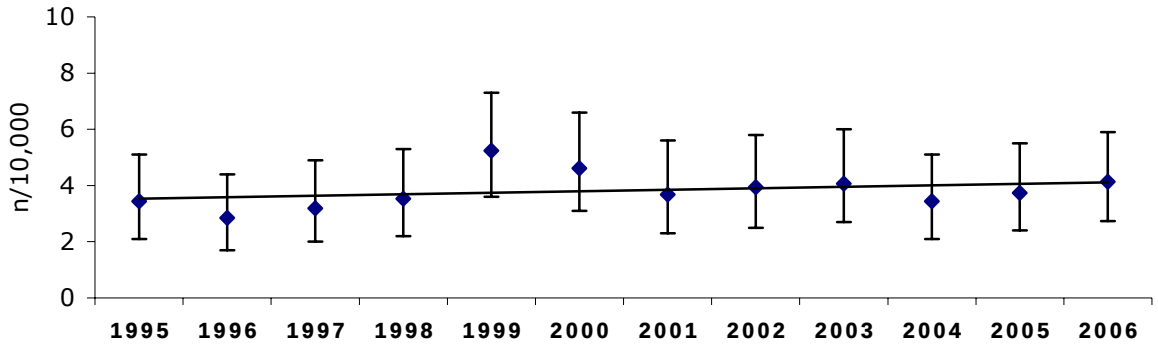
- PR for women aged 40 years or more was significantly higher than for women aged 30-34. There were no statistically significant associations for sex of infant, plurality or country of birth for 2001-2006.

6.13 Cleft lip

British Paediatric Association code 749.10 – 749.19

Definition: Partial or complete clefting of the upper lip, without clefting of the alveolar ridge and palate.

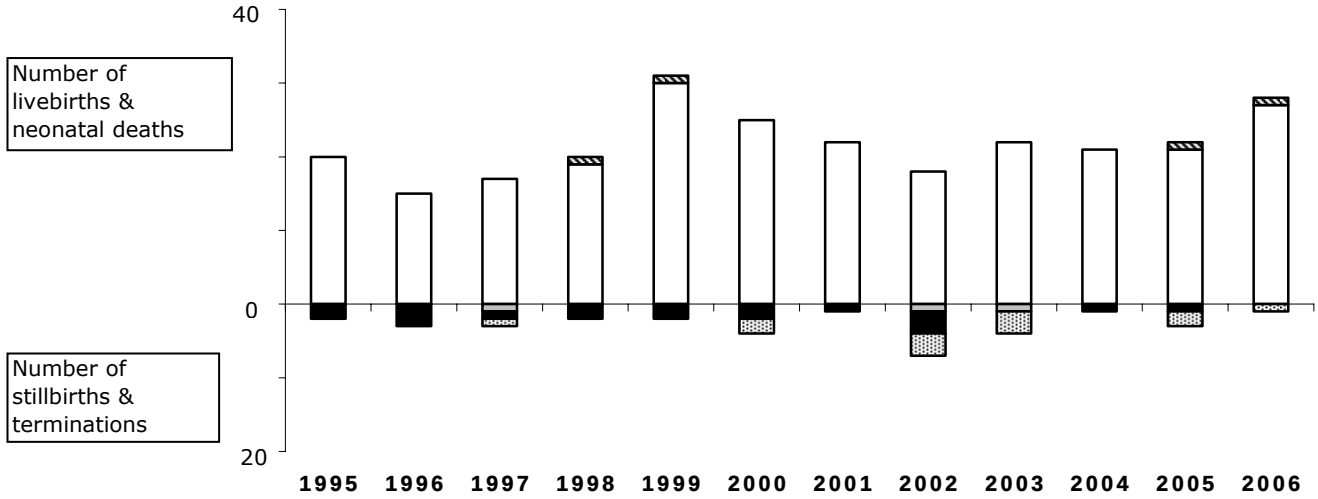
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	3.4	2.8	3.2	3.5	5.2	4.6	3.7	3.9	4.1	3.4	3.7	4.1

- From 1995 to 2006 there has been no statistically significant change in the prevalence of cleft lip.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	22	18	20	22	33	29	23	25	26	22	25	29
Outcome	**Percent (%)**											
Neonatal death	0	0	0	5	3	0	0	0	0	0	4	3
Survived > 28 days	91	83	85	86	91	86	96	72	85	95	84	93
Stillbirth	0	0	5	0	0	0	0	4	4	0	0	0
TOP < 20 weeks	9	17	5	9	6	7	4	12	0	5	4	0
TOP >= 20 weeks*	0	0	5	0	0	7	0	12	12	0	8	3

**These cases are not identified in the dataset prior to 1996*

- On average 87% babies with this condition survived beyond 28 days in 2001 - 2006.

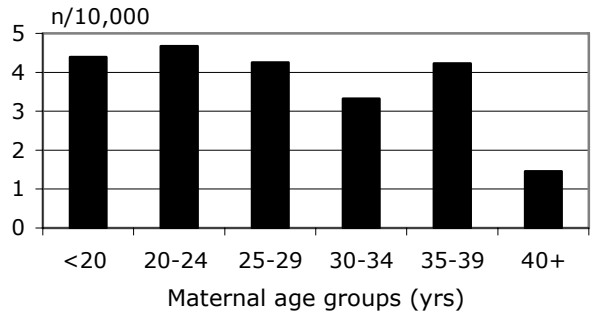
Cleft lip and associated birth defects, 2005-2006

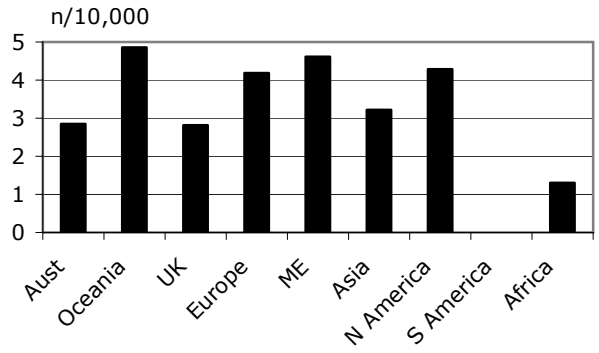
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	42	42	77.8
Chromosomal	0	0	2	1	3	5.6
Other same system	0	0	0	0	0	0.0
Other different systems	4	0	0	5	9	16.7
Total	4	0	2	48	54	100.0

Five year summary of the prevalence of cleft lip and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=150)	**No.**	**%**	**PR**
Sex			
Male	93	*62.0*	4.6
Female	57	*38.0*	3.0
Indeterminate/ Unknown	0	*0.0*	
Plurality#			
Singleton	143	*95.3*	3.8
Multiple	7	*4.7*	5.0
Maternal (n=149)			
Maternal Age (yrs)			
<20	5	*3.3*	4.4
20-24	21	*14.0*	4.7
25-29	43	*28.7*	4.2
30-34	47	*31.3*	3.3
35-39	30	*20.0*	4.2
40+	2	*1.3*	1.5
Unknown	1	*0.7*	
Country of birth			
Australia	113	*75.3*	3.9
Oceania inc NZ	1	*0.7*	1.0
UK inc Eire	4	*2.7*	3.8
Europe	5	*3.3*	4.2
Middle East	4	*2.7*	4.6
Asia	15	*10.0*	4.0
Nth America	0	*0.0*	0.0
Sth America	0	*0.0*	0.0
Africa	4	*2.7*	5.2
Unknown	3	*2.0*	

#excludes unknown





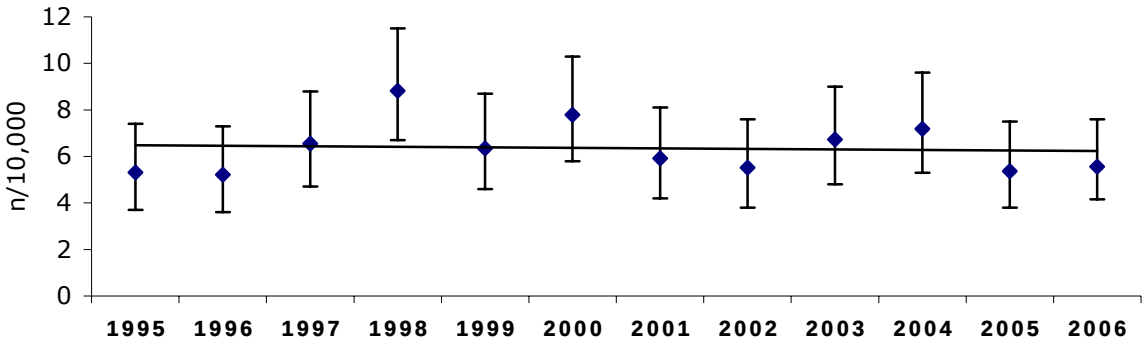
- There were no statistically significant associations for sex of infant plurality, maternal age or country of birth for 2001-2006.

6.14 Cleft lip and palate

British Paediatric Association code 749.20 – 749.29

Definition: Partial or complete clefting of the upper lip, the alveolar ridge and the cleft palate.

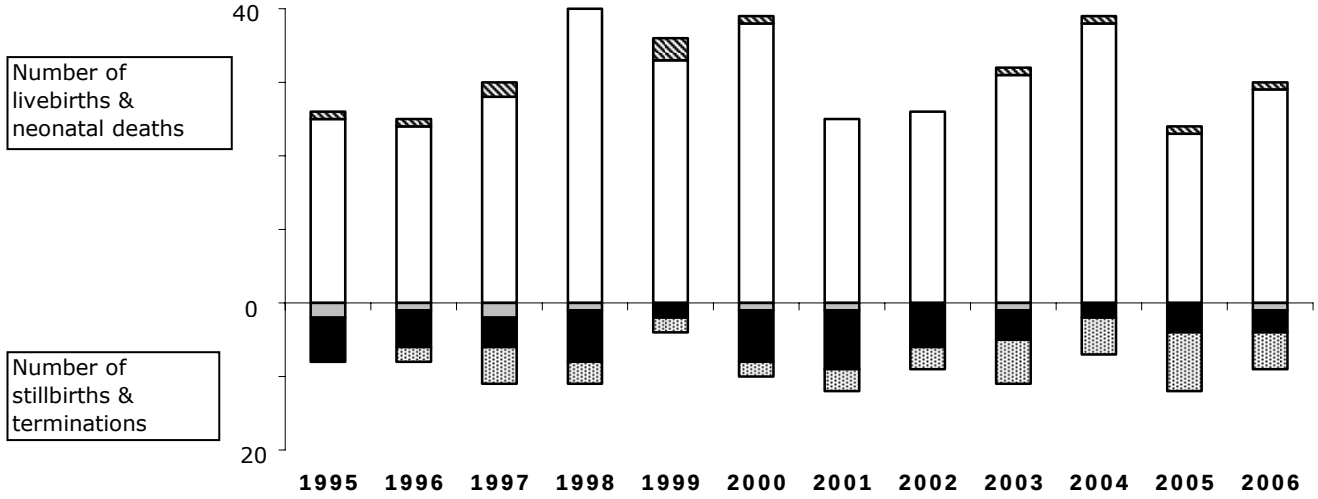
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	5.3	5.2	6.5	8.8	6.4	7.8	5.9	5.5	6.7	7.2	5.4	5.6

- From 1995 to 2006 there has been no statistically significant change in the prevalence of cleft lip and palate.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Outcome	**Percent (%)**											
Neonatal death	3	3	5	7	8	2	0	0	2	2	3	3
Survived > 28 days	74	73	68	73	83	78	68	74	72	83	64	74
Stillbirth	6	3	5	2	0	2	3	0	2	0	0	3
TOP < 20 weeks	18	15	10	13	5	14	22	17	9	4	11	8
TOP >= 20 weeks*	0	6	12	5	5	4	8	9	14	11	22	13

**These cases are not identified in the dataset prior to 1996*

- On average 72.5 % of babies with this condition survived beyond 28 days in 2001-2006.

Cleft lip and palate and associated birth defects, 2005-2006

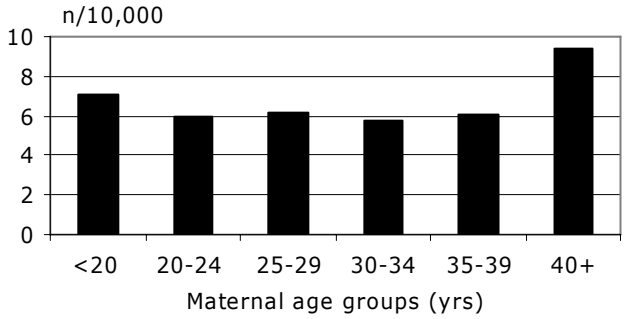
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	4	1	0	45	50	66.7
Chromosomal	12	0	2	1	15	20.0
Other same system	0	0	0	0	0	0.0
Other different systems	4	0	0	6	10	13.3
Total	20	1	2	52	75	100.0

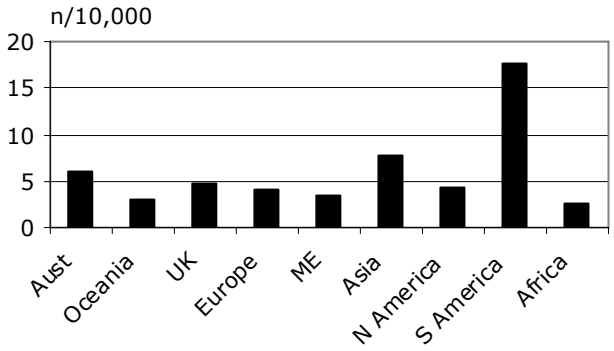
Five year summary of the prevalence of cleft lip and palate and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=236)	**No.**	**%**	**PR**
Sex			
Male	149	*63.1*	7.4*
Female	83	*35.2*	4.4
Indeterminate/ Unknown	4	*1.7*	
Plurality#			
Singleton	226	*95.8*	6.0
Multiple	10	*4.2*	7.2
Maternal (n=235)			
Maternal Age (yrs)			
<20	8	*3.4*	7.0
20-24	27	*11.4*	6.0
25-29	62	*26.3*	6.1
30-34	81	*34.3*	5.7
35-39	43	*18.2*	6.1
40+	13	*5.5*	9.4
Unknown	1	*0.4*	
Country of birth			
Australia	177	*75.0*	6.1
Oceania inc NZ	3	*1.3*	2.9
UK inc Eire	5	*2.1*	4.7
Europe	5	*2.1*	4.2
Middle East	3	*1.3*	3.5
Asia	29	*12.3*	7.8
Nth America	1	*0.4*	4.3
Sth America	4	*1.7*	17.7
Africa	2	*0.8*	2.6
Unknown	6	*2.5*	

#excludes unknown

**statistically significant, p<0.0001*





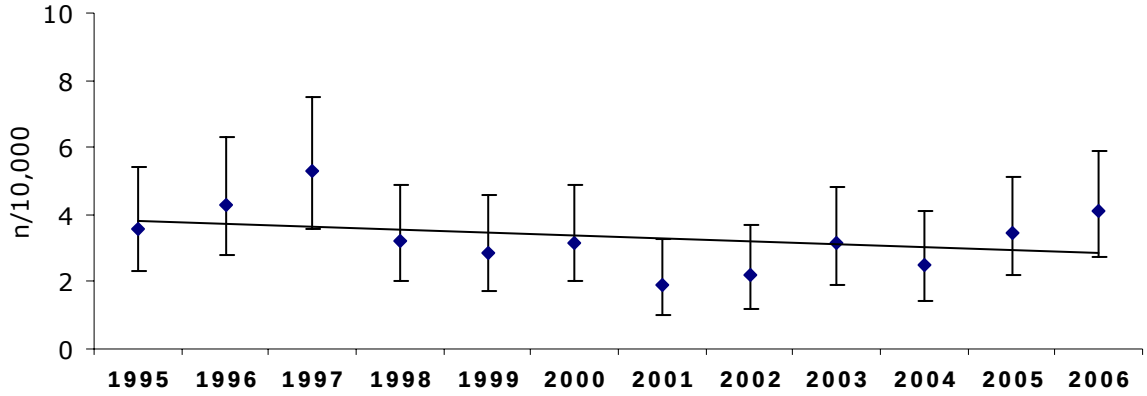
- PR for males was significantly higher than for females. Although some PRs were higher than others, due to small sample size there were no statistically significant associations for plurality, maternal age or country of birth for 2001-2006.

6.15 Oesophageal atresia and/or stenosis

British Paediatric Association code 750.30 – 750.38

Definition: Absence of continuity or narrowing of the oesophagus, with or without tracheal fistula.

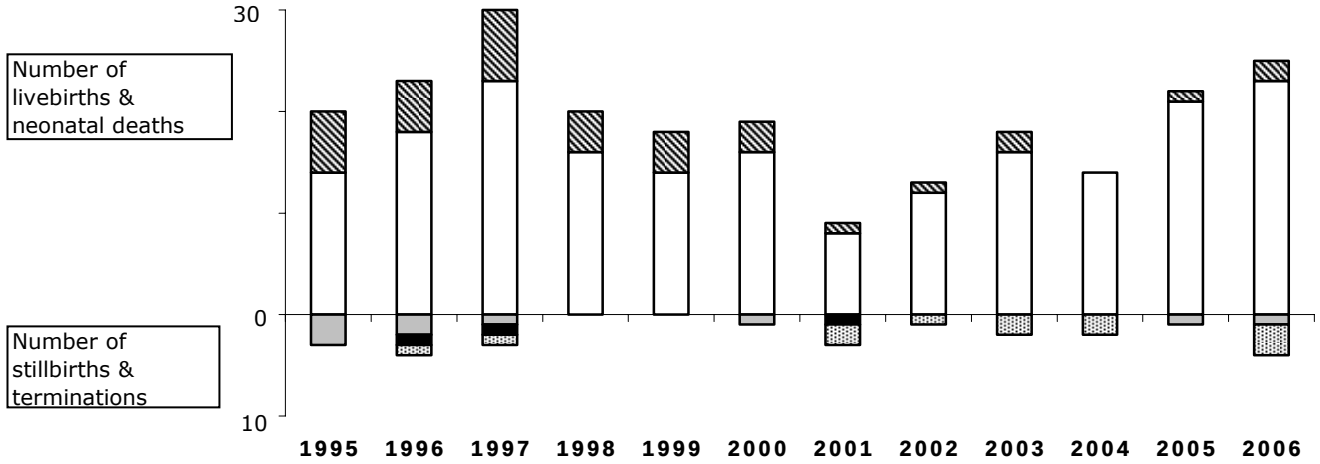
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	3.6	4.3	5.3	3.2	2.9	3.2	1.9	2.2	3.1	2.5	3.4	4.1

- From 1995 to 2006 there has been no statistically significant change in the prevalence of oesophageal atresia and/or stenosis. However from 2001-2006 there has been a significant increase of this condition.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	23	27	33	20	18	20	12	14	20	16	23	29
Outcome	**Percent (%)**											
Neonatal death	26	19	21	20	22	15	8	7	10	0	4	7
Survived > 28 days	61	67	70	80	78	80	67	86	80	88	91	79
Stillbirth	13	7	3	0	0	5	0	0	0	0	4	3
TOP < 20 weeks	0	4	3	0	0	0	8	0	0	0	0	0
TOP >= 20 weeks*	0	4	3	0	0	0	17	7	10	13	0	10

**These cases are not identified in the dataset prior to 1996*

- On average 82% of babies with this condition survived beyond 28 days in 2001 – 2006.

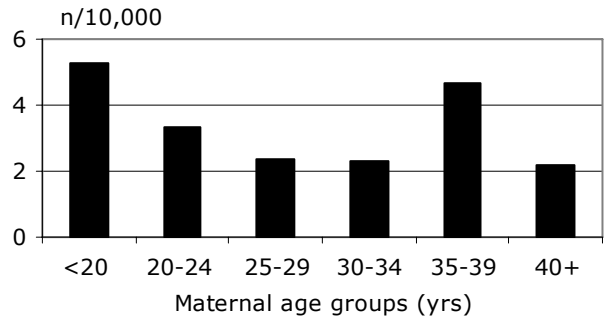
Oesophageal atresia and/or stenosis and associated birth defects, 2005-2006

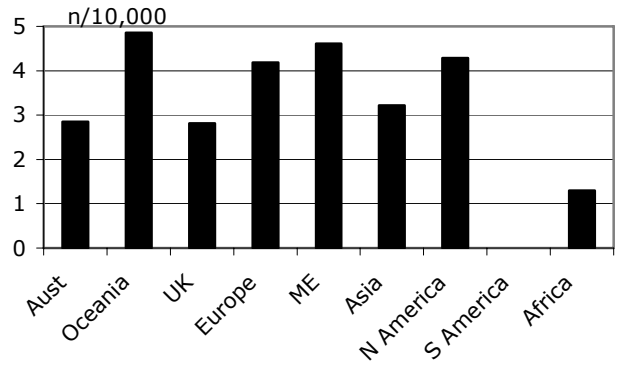
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	2	1	13	16	30.8
Chromosomal	1	0	0	1	2	3.8
Other same system	0	0	0	2	2	3.8
Other different systems	2	0	2	28	32	61.5
Total	3	2	3	44	52	100.0

Five year summary of the prevalence of oesophageal atresia and/or stenosis and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=114)	**No.**	**%**	**PR**
Sex			
Male	72	*63.2*	3.6
Female	42	*36.8*	2.2
Indeterminate/ Unknown	0	*0.0*	
Plurality#			
Singleton	110	*96.5*	2.9
Multiple	4	*3.5*	2.9
Maternal (n=114)			
Maternal Age (yrs)			
<20	6	*5.3*	5.3
20-24	15	*13.2*	3.3
25-29	24	*21.1*	2.4
30-34	33	*28.9*	2.3
35-39	33	*28.9*	4.6
40+	3	*2.6*	2.2
Unknown	0	*0.0*	
Country of birth			
Australia	83	*72.8*	2.8
Oceania inc NZ	5	*4.4*	4.9
UK inc Eire	3	*2.6*	2.8
Europe	5	*4.4*	4.2
Middle East	4	*3.5*	4.6
Asia	12	*10.5*	3.2
Nth America	1	*0.9*	4.3
Sth America	0	*0.0*	0.0
Africa	1	*0.9*	1.3
Unknown	0	*0.0*	

#excludes unknown





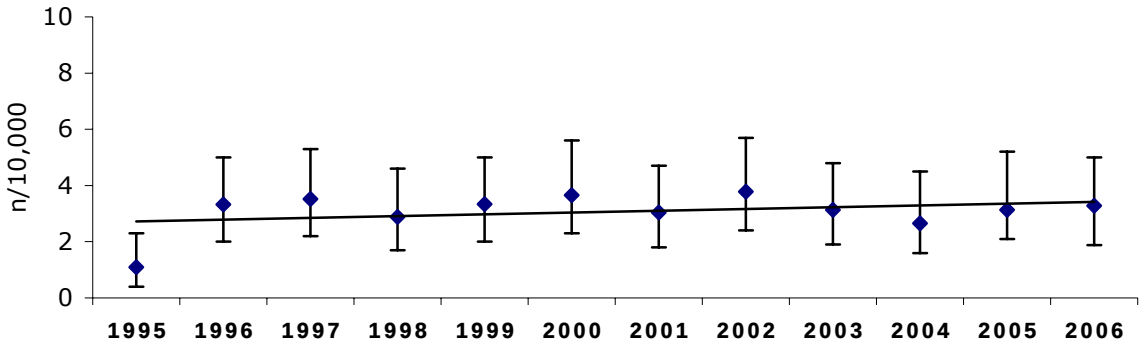
- There were no statistically significant associations for sex or infant, plurality, maternal age or country of birth for 2001-2006.

6.16 Small intestinal atresia and/or stenosis

British Paediatric Association code 751.10 – 751.19

Definition: Completion or partial occlusion of the lumen of a segment of the small intestine. It can involve a single area or multiple areas of the duodenum, jejunum or ileum.

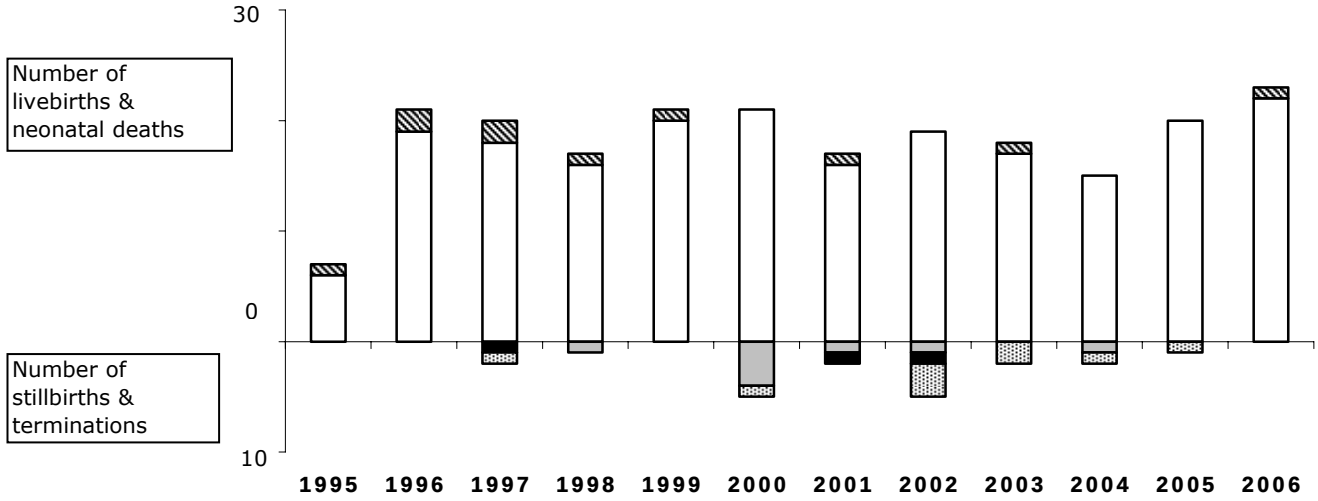
Trend in prevalence rates, 1995-2006



N/10,000	1.1	3.3	3.5	2.9	3.3	3.7	3.0	3.8	3.1	2.7	3.1	3.3

- From 1995 to 2006 there has been no statistically significant change in the prevalence of small intestinal atresia and/or stenosis.

Number of cases and pregnancy outcomes, 1995-2006



Total number	7	21	22	18	21	23	19	24	20	17	21	23
Outcome	**Percent (%)**											
Neonatal death	14	10	9	6	5	0	5	0	5	0	0	4
Survived > 28 days	86	90	82	89	95	91	84	79	85	88	95	96
Stillbirth	0	0	0	6	0	4	5	4	0	6	0	0
TOP < 20 weeks	0	0	5	0	0	0	5	4	0	0	0	0
TOP >= 20 weeks*	0	0	5	0	0	4	0	13	10	6	5	0

**These cases are not identified in the dataset prior to 1996*

- On average, 88% of babies with this condition survived beyond 28 days in 2001-2006.

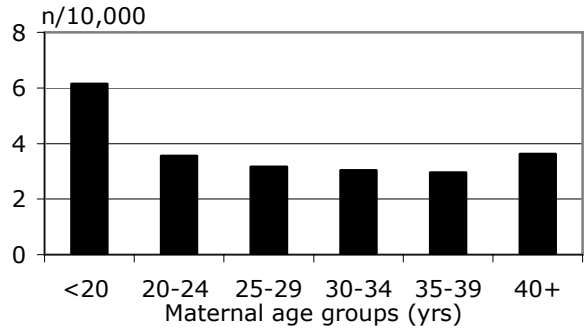
Small intestinal atresia and/or stenosis and associated birth defects, 2005-2006

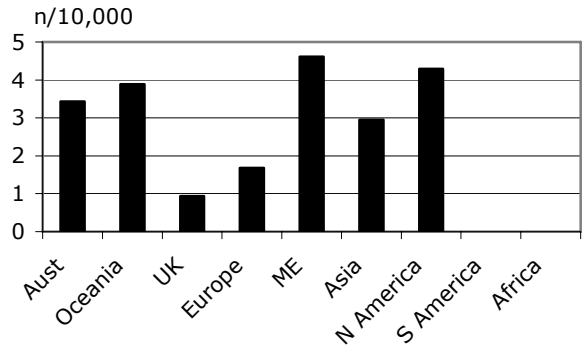
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	1	0	0	25	26	59.1
Chromosomal	0	0	0	3	3	6.8
Other same system	0	0	0	6	6	13.6
Other different systems	0	0	1	8	9	20.5
Total	1	0	1	42	44	100.0

Five year summary of the prevalence of small intestinal atresia and/or stenosis and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=124)	**No.**	**%**	**PR**
Sex			
Male	61	*49.2*	3.0
Female	63	*50.8*	3.3
Indeterminate/ Unknown	0	*0.0*	
Plurality#			
Singleton	121	*97.6*	3.2
Multiple	3	*2.4*	2.2
Maternal (n=124)			
Maternal Age (yrs)			
<20	7	*5.6*	6.2
20-24	16	*12.9*	3.6
25-29	32	*25.8*	3.2
30-34	43	*34.7*	3.0
35-39	21	*16.9*	3.0
40+	5	*4.0*	3.6
Unknown	0	*0.0*	
Country of birth			
Australia	100	*80.6*	3.4
Oceania inc NZ	4	*3.2*	3.9
UK inc Eire	1	*0.8*	0.9
Europe	2	*1.6*	1.7
Middle East	4	*3.2*	4.6
Asia	11	*8.9*	2.9
Nth America	1	*0.8*	4.3
Sth America	0	*0.0*	0.0
Africa	0	*0.0*	0.0
Unknown	1	*0.8*	

#excludes unknown





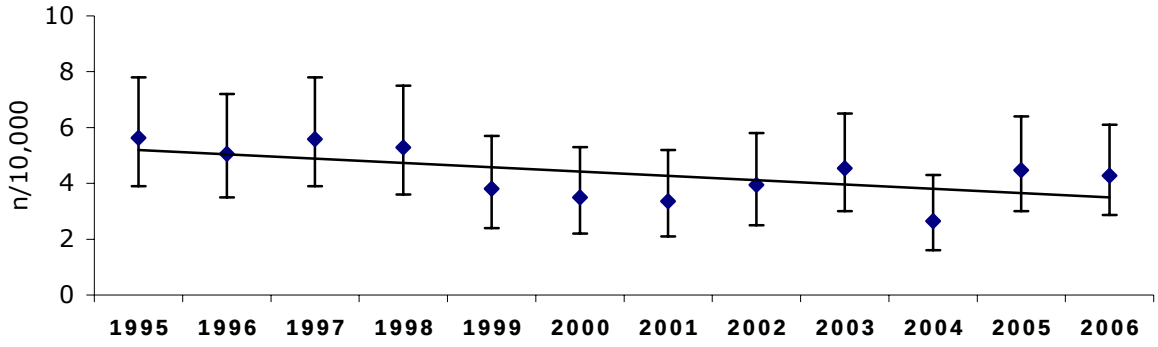
- Although some PRs were higher than others, due to small sample size there were no statistically significant associations for infant sex, plurality, maternal age or country of birth for 2001-2006.

6.17 Anorectal atresia and/or stenosis

British Paediatric Association code 751.21 – 751.24

Definition: Absence of continuity of the anorectal canal or of a communication between rectum and anus, or narrowing of the anal canal, with or without fistula to neighbouring organs. Excludes ectopic anus.

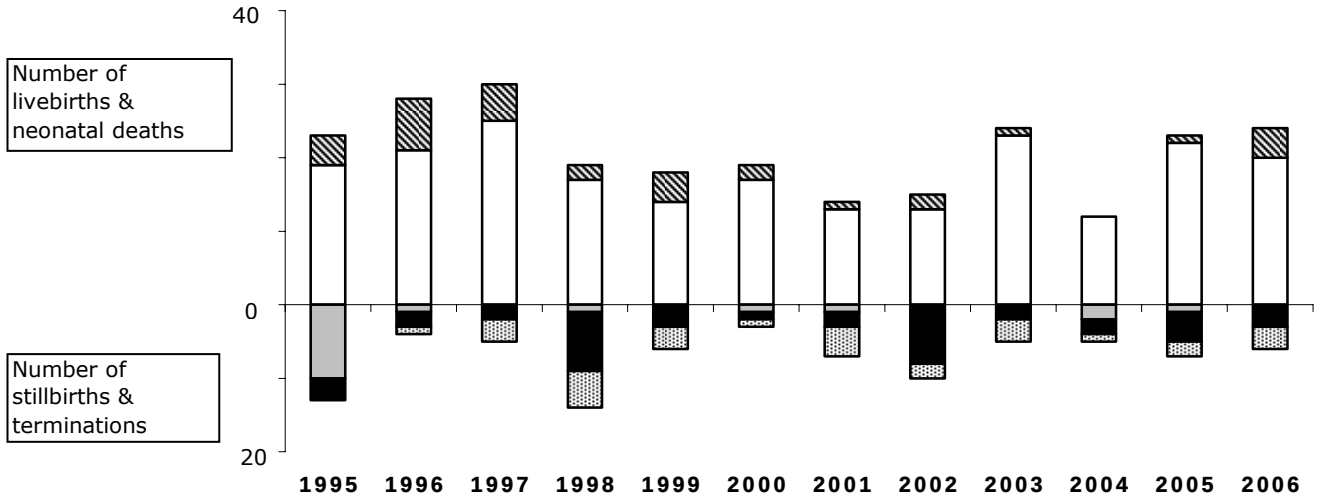
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	5.6	5.1	5.6	5.3	3.8	3.5	3.4	3.9	4.5	2.7	4.5	4.3

- From 1995 to 2006 there has been no statistically significant change in the prevalence of anorectal atresia and/or stenosis.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
Outcome	**Percent (%)**											
Neonatal death	11	22	14	6	17	9	5	8	3	0	3	13
Survived > 28 days	53	66	71	52	58	77	62	52	79	71	73	67
Stillbirth	28	3	0	3	0	5	5	0	0	12	3	0
TOP < 20 weeks	8	6	6	24	13	5	10	32	7	12	13	10
TOP >= 20 weeks*	0	3	9	15	13	5	19	8	10	6	7	10

**These cases are not identified in the dataset prior to 1996*

- On average, 67% of babies with this condition survived beyond 28 days in 2001 – 2006.

Anorectal atresia and/or stenosis and associated birth defects, 2005-2006

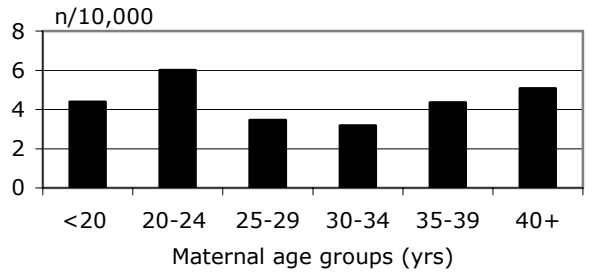
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	21	21	35.0
Chromosomal	4	0	0	1	5	8.3
Other same system	0	0	0	0	0	0.0
Other different systems	8	1	5	20	34	56.7
Total	12	1	5	42	60	100.0

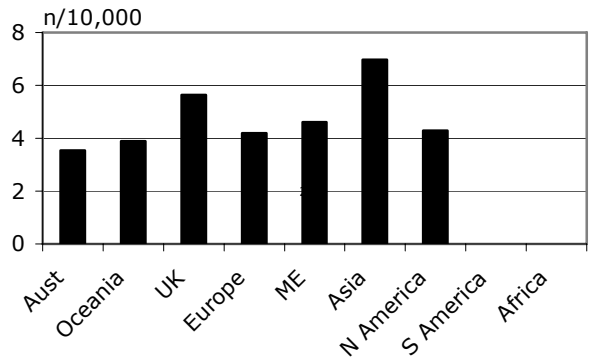
Five year summary of the prevalence of anorectal atresia and/or stenosis and selected infant and maternal characteristics, 2001 - 2006

	2001—2006		
Infant (n=152)	**No.**	**%**	**PR**
Sex			
Male	94	*61.8*	4.7
Female	56	*36.8*	2.9
Indeterminate/ Unknown	2	*1.3*	
Plurality#			
Singleton	142	*93.4*	3.8
Multiple	10	*6.6*	7.2
Maternal (n=150)			
Maternal Age (yrs)			
<20	5	*3.3*	4.4
20-24	27	*18.0*	6.0
25-29	35	*23.3*	3.5
30-34	45	*30.0*	3.2
35-39	31	*20.7*	4.4
40+	7	*4.7*	5.1
Unknown	0	*0.0*	
Country of birth			
Australia	103	*68.7*	3.5
Oceania inc NZ	4	*2.7*	3.9
UK inc Eire	6	*4.0*	5.6
Europe	5	*3.3*	4.2
Middle East	4	*2.7*	4.6
Asia	26	*17.3*	7.0*
Nth America	1	*0.7*	4.3
Sth America	0	*0.0*	0.0
Africa	0	*0.0*	0.0
Unknown	1	*0.7*	

#excludes unknown

** statistically significant, p<0.01*





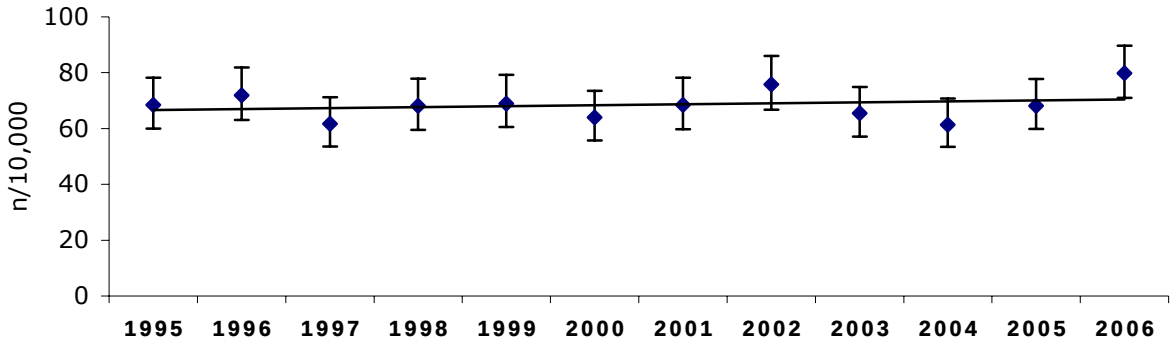
- The PR for women born in Asia was significantly higher than for women born in Australia. There were no statistically significant associations for sex or infant, plurality or maternal age in 2001-2006.

6.18 Hypospadias

British Paediatric Association code 752.60

Definition: Opening of the urethra on the ventral side of the penis, distally to the sulcus.

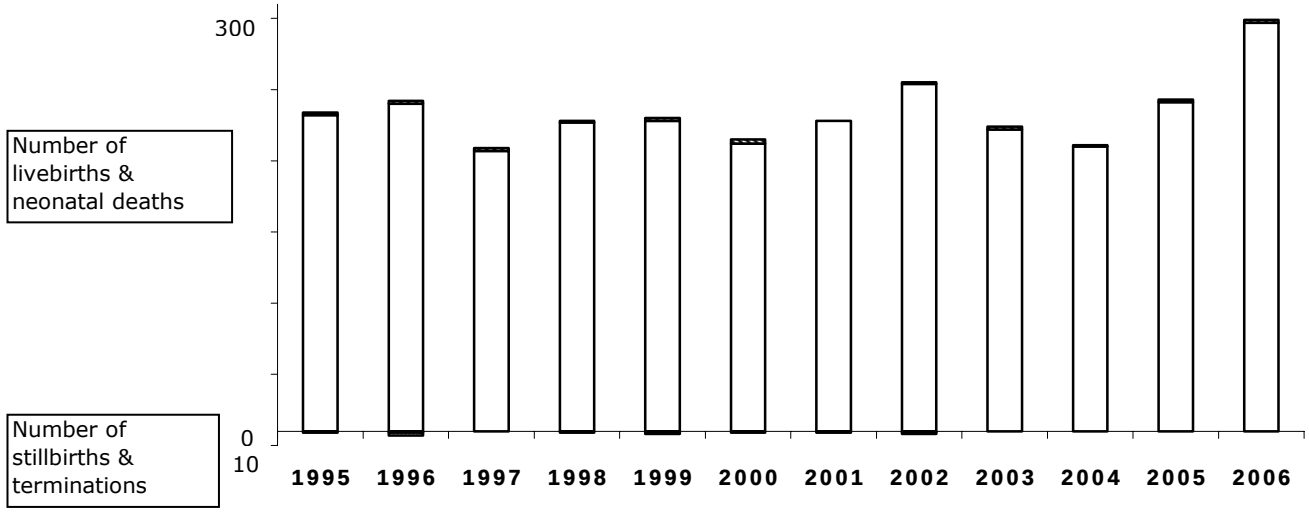
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	68.5	71.9	61.8	68.1	69.0	64.0	68.4	75.8	65.4	61.4	68.2	79.8

- From 1995 to 2006 there has been no statistically significant change in the prevalence of hypospadias.

Number of cases and pregnancy outcomes, 1995-2006



Total number	225	235	199	219	222	206	219	247	214	201	233	289
Outcome	**Percent (%)**											
Neonatal death	1	1	1	0	1	1	0	0	1	0	1	1
Survived > 28 days	99	98	99	99	98	98	100	99	99	100	99	99
Stillbirth	0	0	0	0	0	0	0	0	0	0	0	0
TOP < 20 weeks	0	0	0	0	0	0	0	0	0	0	0	0
TOP >= 20 weeks*	0	1	0	0	0	0	0	0	0	0	0	0

**These cases are not identified in the dataset prior to 1996*

- On average, 99% of babies with this condition survived beyond 28 days in 2001 – 2006.

Hypospadias and associated birth defects, 2005-2006

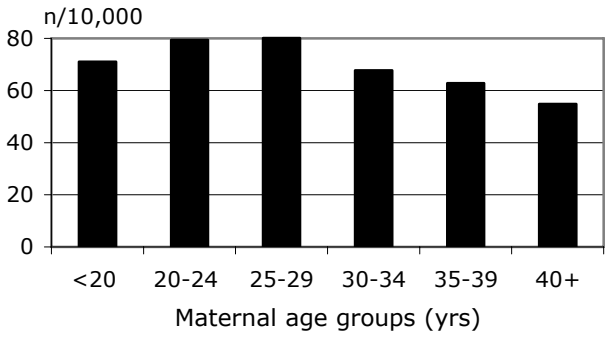
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	1	464	465	89.1
Chromosomal	0	0	0	3	3	0.6
Other same system	0	0	0	10	10	1.9
Other different systems	0	0	3	41	44	8.4
Total	0	0	4	518	522	100.0

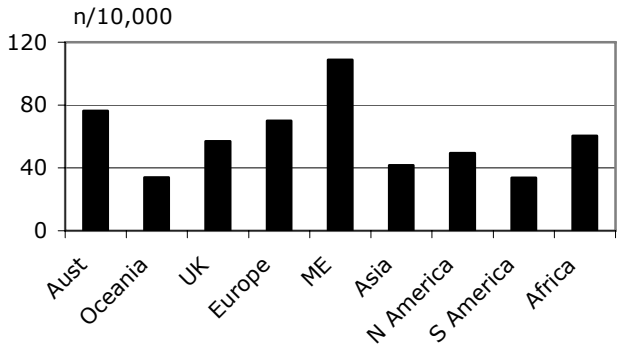
Five year summary of the prevalence of hypospadias and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=1,403)	**No.**	**%**	**PR**
Sex			
Male	1400	*99.8*	69.9
Female	n/a		
Indeterminate/ Unknown	3	*0.2*	
Plurality#			
Singleton	1339	*95.4*	69.2
Multiple	64	*4.6*	91.5
Maternal (n=1,399)			
Maternal Age (yrs)			
<20	42	*3.0*	71.1
20-24	183	*13.1*	79.4
25-29	416	*29.7*	80.1
30-34	491	*35.1*	67.7
35-39	229	*16.4*	62.9
40+	38	*2.6*	54.8
Unknown	0	*0.0*	
Country of birth			
Australia	1138	*81.3*	76.3
Oceania inc NZ	18	*1.3*	33.8*
UK inc Eire	31	*2.2*	56.9
Europe	43	*3.1*	70.1
Middle East	48	*3.4*	108.8
Asia	80	*5.7*	41.7*
Nth America	6	*0.4*	49.4
Sth America	4	*0.3*	33.6
Africa	24	*1.7*	60.3
Unknown	7	*0.5*	

#excludes unknown

** statistically significant, p<0.0001*





- The PR for women born in Asia and Oceania including New Zealand was significantly lower than for women born in Australia and the Middle East. There were no statistically significant associations for plurality or maternal age in 2001-2006.

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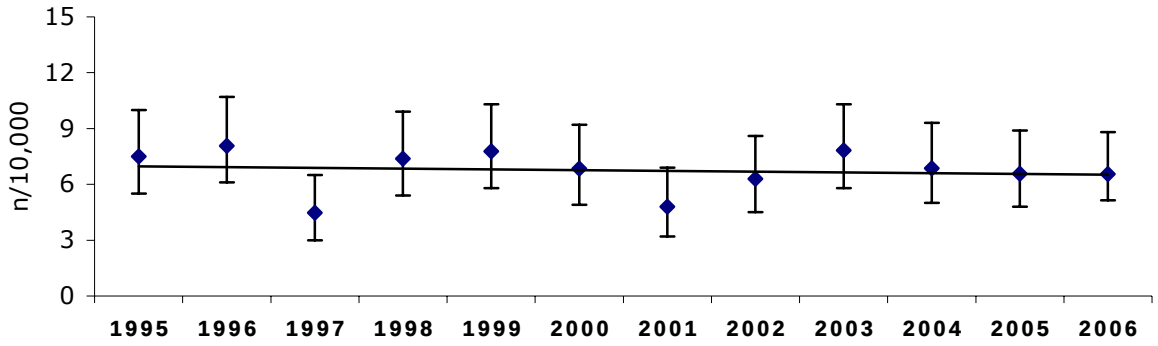
6.19 Renal agenesis and dysgenesis

British Paediatric Association code 753.00 – 753.09

Definition: This heterogenous group includes bilateral or unilateral absence of the kidneys or severe dysplasia.

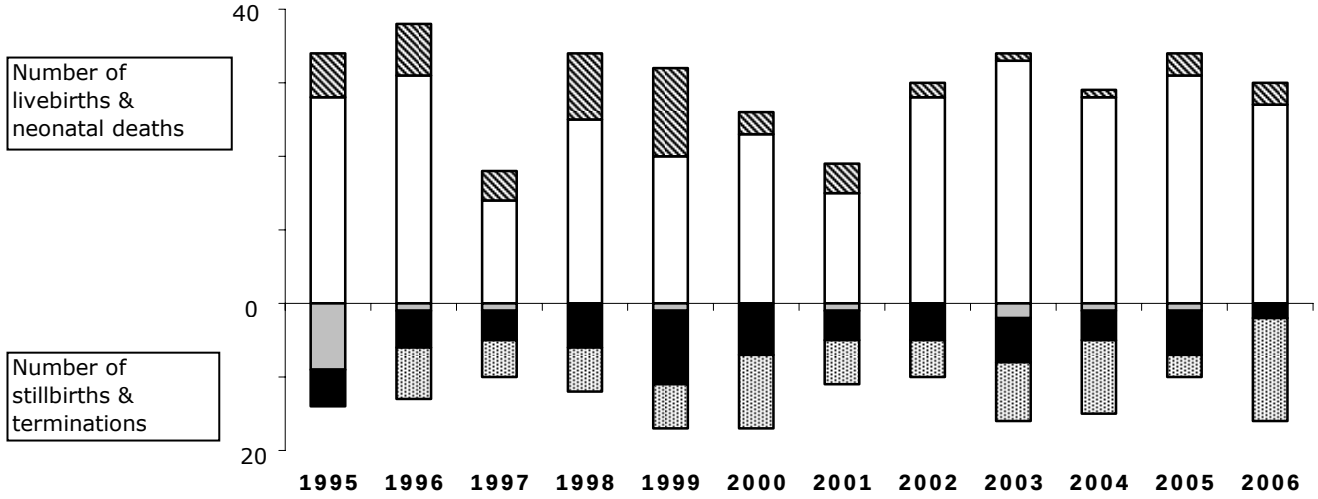
Trend in prevalence rates, 1995-2006



N / 10,000	7.5	8.1	4.5	7.4	7.8	6.8	4.8	6.3	7.8	6.9	6.6	6.6

- From 1995 to 2006 there has been no statistically significant change in the prevalence of renal agenesis and dysgenesis.

Number of cases and pregnancy outcomes, 1995-2006



Total number	48	51	28	46	49	43	30	40	50	44	44	46
Outcome	**Percent (%)**											
Neonatal death	13	14	14	20	24	7	13	5	2	2	7	7
Survived > 28 days	58	61	50	54	41	53	50	70	66	64	70	59
Stillbirth	19	2	4	0	2	0	3	0	4	2	2	0
TOP < 20 weeks	10	10	14	13	20	16	13	13	12	9	14	4
TOP >= 20 weeks*	0	14	18	13	12	23	20	13	16	23	7	30

**These cases are not identified in the dataset prior to 1996*

- On average, 63% of babies with this condition survived beyond 28 days in 2001-2006.

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Renal agenesis and dysgensis and associated birth defects, 2005-2006

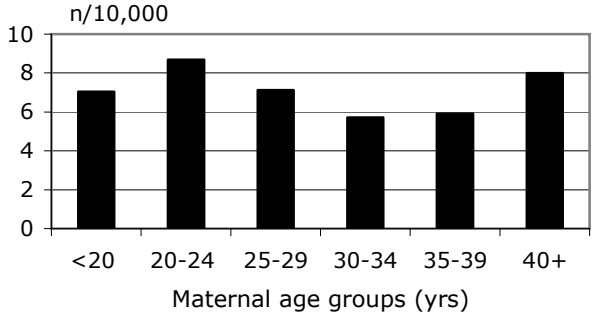
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	6	0	4	33	43	47.8
Chromosomal	5	1	1	3	10	11.1
Other same system	5	0	0	13	18	20.0
Other different systems	9	0	1	9	19	21.1
Total	25	1	6	58	90	100.0

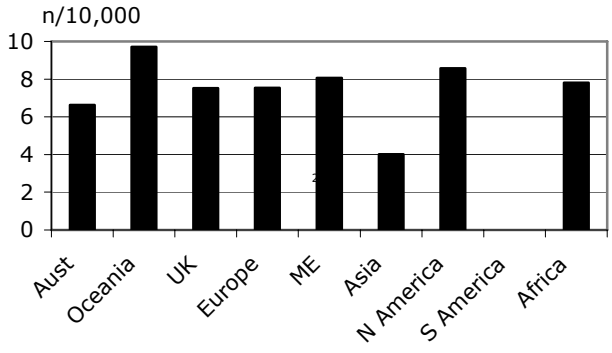
Five year summary of the prevalence of renal agenesis and dysgenesis and selected infant and maternal characteristics, 2001 - 2006

	2001—2006		
Infant (n=254)	**No.**	**%**	**PR**
Sex			
Male	166	*65.4*	8.3*
Female	81	*31.9*	4.3
Indeterminate/ Unknown	7	*2.8*	
Plurality#			
Singleton	245	*96.5*	6.5
Multiple	9	*3.5*	6.5
Maternal (n=253)			
Maternal Age (yrs)			
<20	8	*3.2*	7.0
20-24	39	*15.4*	8.7
25-29	72	*28.5*	7.1
30-34	81	*32.0*	5.7
35-39	42	*16.6*	5.9
40+	11	*4.3*	8.0
Unknown	0	*0.0*	
Country of birth			
Australia	193	*76.3*	6.6
Oceania inc NZ	10	*4.0*	9.7
UK inc Eire	8	*3.2*	7.5
Europe	9	*3.6*	7.5
Middle East	7	*2.8*	8.1
Asia	15	*5.9*	4.0
Nth America	2	*0.8*	8.6
Sth America	0	*0.0*	0.0
Africa	6	*2.4*	7.8
Unknown	3	*1.2*	

#excludes unknown

** statistically significant, p<0.0001*





- The PR for males was significantly higher than for females. There were no statistically significant associations for plurality, maternal age or country of birth in 2001-2006.

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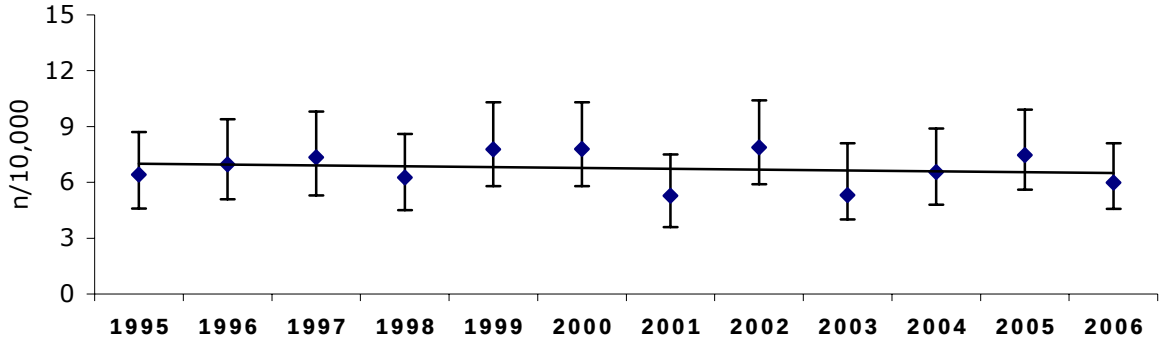
6.20 Cystic kidney disease

British Paediatric Association code 753.10 – 753.18

Definition: This covers a wide range of birth defects with renal cysts of varying size and extent, occurring bilaterally or unilaterally. Polycystic and multicystic kidney disease are both included.

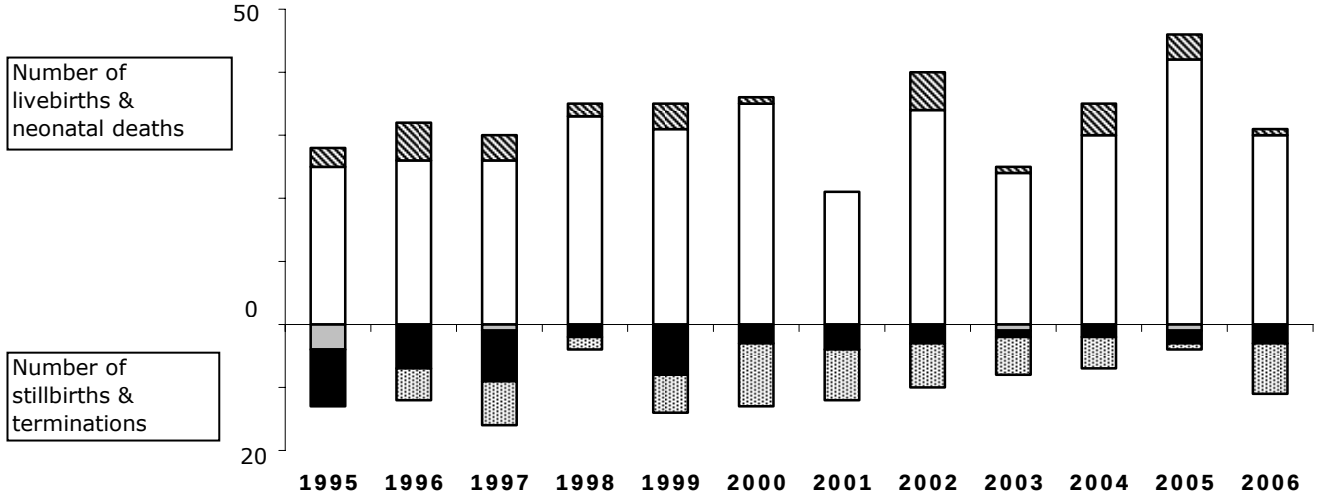
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	6.4	7.0	7.3	6.3	7.8	7.8	5.3	7.9	5.3	6.6	7.5	6.0

- From 1995 to 2006 there has been no statistically significant change in the prevalence of cystic kidney disease.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	41	44	46	39	49	49	33	50	34	42	50	42
Outcome	**Percent (%)**											
Neonatal death	7	14	9	5	8	2	0	12	3	12	8	2
Survived > 28 days	61	59	57	85	63	71	64	68	71	71	84	71
Stillbirth	10	0	2	0	0	0	0	0	6	0	2	0
TOP < 20 weeks	22	16	17	5	16	6	12	6	3	5	4	7
TOP >= 20 weeks*	0	11	15	5	12	20	24	14	18	12	2	19

**These cases are not identified in the dataset prior to 1996*

- On average, 72% of babies with this condition survived beyond 28 days in 2001-2006.

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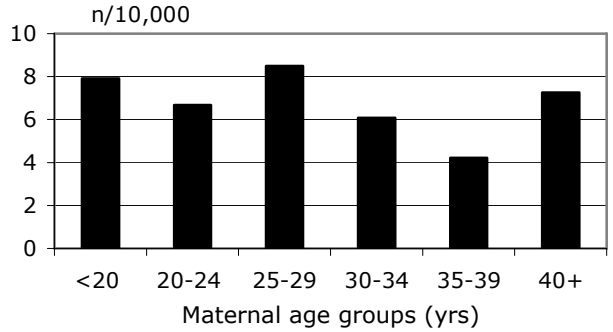
Cystic kidney disease and associated birth defects, 2005-2006

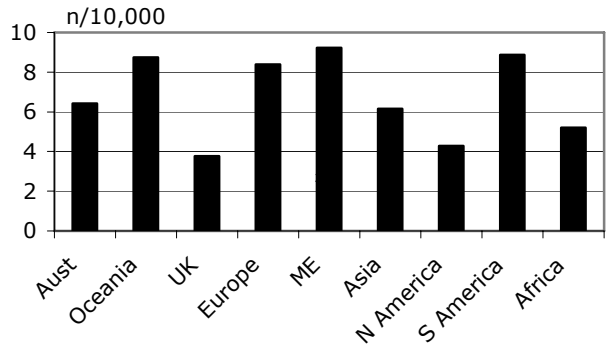
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	2	1	2	45	50	54.3
Chromosomal	5	0	1	1	7	7.6
Other same system	5	0	2	13	20	21.7
Other different systems	2	0	0	13	15	16.3
Total	14	1	5	72	92	100.0

Five year summary of the prevalence of cystic kidney disease and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=251)	**No.**	**%**	**PR**
Sex			
Male	154	*61.4*	7.7*
Female	96	*38.2*	5.0
Indeterminate/ Unknown	1	*0.4*	
Plurality#			
Singleton	240	*95.6*	6.4
Multiple	11	*4.4*	7.9
Maternal (n=251)			
Maternal Age (yrs)			
<20	9	*3.6*	7.9
20-24	30	*12.0*	6.7
25-29	86	*34.3*	8.5
30-34	86	*34.3*	6.1
35-39	30	*12.0*	4.2**
40+	10	*4.0*	7.3
Unknown	0	*0.0*	
Country of birth			
Australia	187	*74.5*	6.4
Oceania inc NZ	9	*3.6*	8.7
UK inc Eire	4	*1.6*	3.8
Europe	10	*4.0*	8.4
Middle East	8	*3.2*	9.2
Asia	23	*9.2*	6.2
Nth America	1	*0.4*	4.3
Sth America	2	*0.8*	8.9
Africa	4	*1.6*	5.2
Unknown	3	*1.2*	

#excludes unknown
** statistically significant, p<0.001*
***statistically significant, p<0.001*





- The PR for males was significantly higher than for females. The PR for women aged 35-39 years was significantly lower than for women aged 25-29 years. There were no statistically significant associations for plurality or country of birth in 2001-2006.

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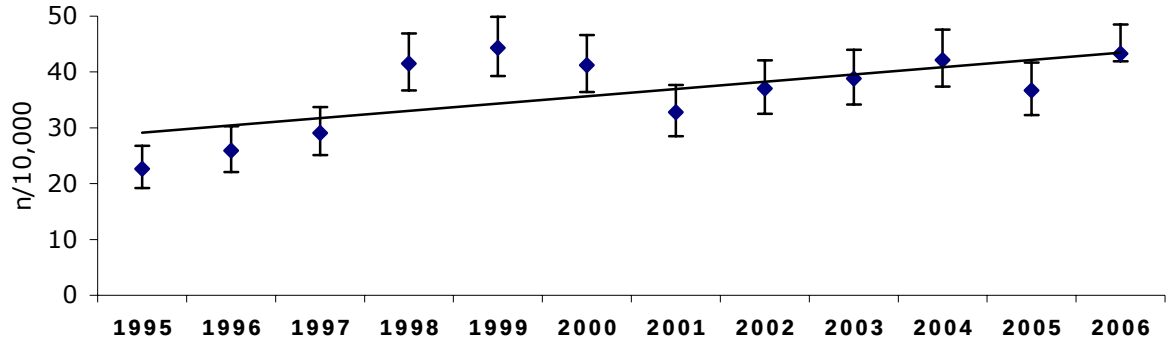
6.21 Obstructive defects of the renal pelvis

British Paediatric Association code 753.20 – 753.29

Definition: This heterogenous group includes hydronephrosis and any other defect that results in dilatation of the renal collecting system, bilaterally or unilaterally.

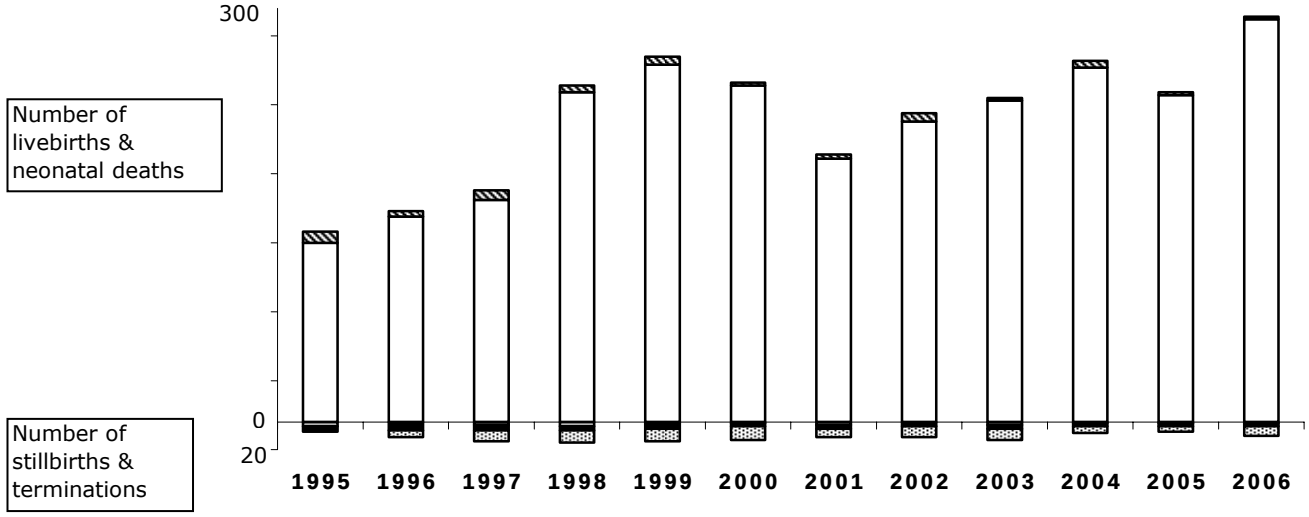
Trend in prevalence rates, 1995-2006



N/10,000	22.7	25.9	29.1	41.5	44.3	41.2	32.8	37.0	38.8	42.2	36.7	43.3

- From 1995 to 2006 there has been a statistically significant increase in the prevalence of obstructive defects of the renal pelvis, which has persisted over the last five years.

Number of cases and pregnancy outcomes, 1995-2006



Total number	145	164	182	259	279	259	205	235	248	270	246	304
Outcome	**Percent (%)**											
Neonatal death	6	2	4	2	2	1	1	3	1	2	1	1
Survived > 28 days	90	91	88	92	93	94	93	93	94	95	96	96
Stillbirth	2	0	1	1	0	0	1	0	1	0	0	0
TOP < 20 weeks	3	4	2	1	1	1	1	1	1	1	1	1
TOP >= 20 weeks*	0	3	4	3	3	4	3	3	3	2	2	2

**These cases are not identified in the dataset prior to 1996*

- On average 95% of babies with this condition have survived beyond 28 days since 1998.

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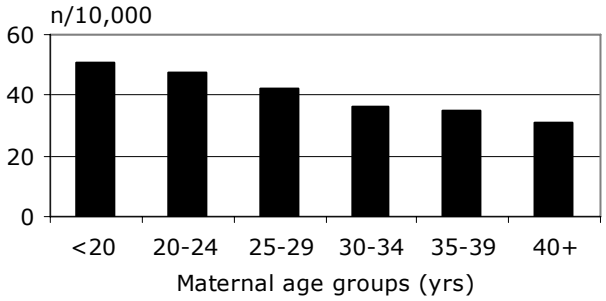
Obstructive defects of the renal pelvis and associated birth defects, 2005-2006

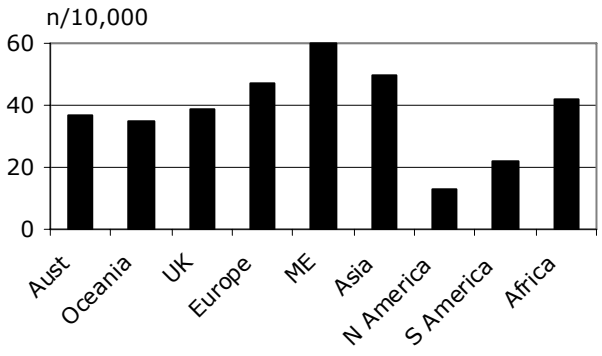
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	1	0	420	421	76.5
Chromosomal	4	0	1	9	14	2.5
Other same system	2	0	0	48	50	9.1
Other different systems	9	1	3	52	65	11.8
Total	15	2	4	529	550	100.0

Five year summary of the prevalence of obstructive defects of the renal pelvis and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=1,508)	**No.**	**%**	**PR**
Sex			
Male	1121	*74.3*	55.9*
Female	381	*25.3*	20.0
Indeterminate/ Unknown	6	*0.4*	
Plurality#			
Singleton	1449	*96.1*	38.4
Multiple	59	*3.9*	42.5
Maternal (n=1,498)			
Maternal Age (yrs)			
<20	58	*3.9*	51.0
20-24	213	*14.2*	47.4**
25-29	426	*28.4*	42.1
30-34	510	*34.0*	36.0
35-39	248	*16.6*	34.9
40+	43	*2.6*	31.2
Unknown	0	*0.0*	
Country of birth			
Australia	1077	*71.9*	37.0
Oceania inc NZ	36	*2.4*	35.0
UK inc Eire	41	*2.7*	38.5
Europe	56	*3.7*	46.9
Middle East	52	*3.5*	59.9***
Asia	186	*12.4*	49.8***
Nth America	3	*0.2*	12.9
Sth America	5	*0.3*	22.2
Africa	32	*2.1*	41.6
Unknown	10	*0.7*	

#excludes unknown
** statistically significant, p<0.0001*
*** statistically significant, p<0.0001*
****statistically significant, p<0.001*





- The PR for males was significantly higher than for females. The PR for women aged 20-24 was significantly higher than for women aged 30 years or more. The PR for women born in the Middle East and Asia was significantly higher than for women born in Australia and North America. There were no statistically significant associations for plurality in 2001-2006.

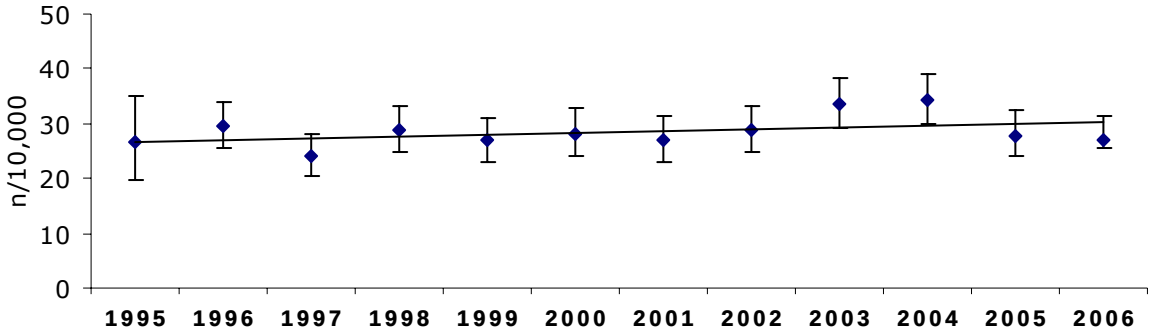
6.22 Developmental dysplasia of the hip

(previously reported as congenital dislocated hips)

British Paediatric Association code 754.30

Definition: The femoral head is displaced (or displaceable) from the acetabulum of the pelvis. This is not to be confused with 'clicky hips' which are not included in the VBDR.

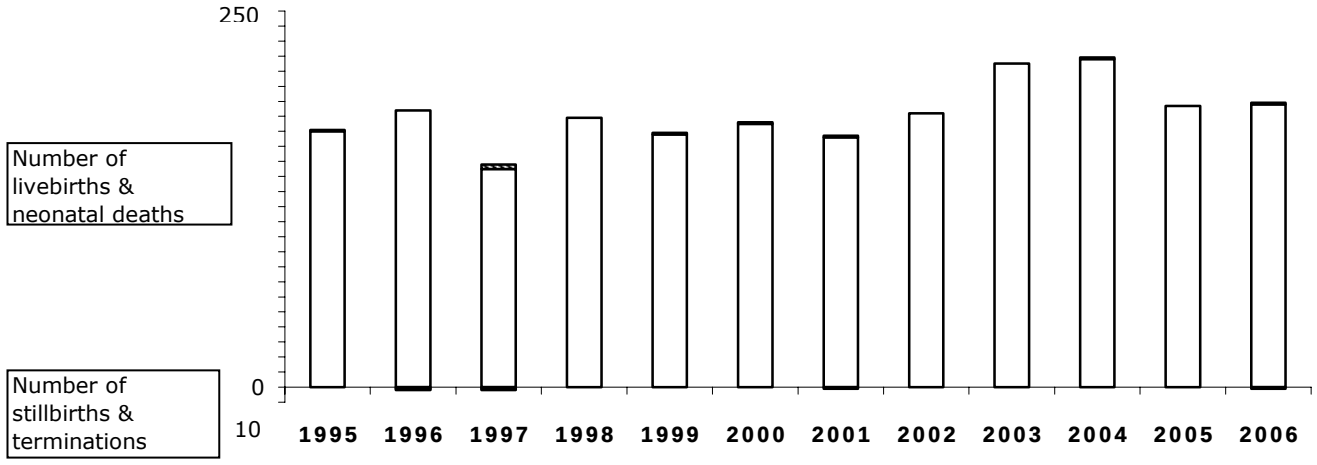
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	26.7	29.4	24.0	28.7	26.8	28.0	26.9	28.7	33.6	34.2	27.9	27.1

- From 1995 to 2006 there has been no statistically significant change in the prevalence of developmental dysplasia of the hip.

Number of cases and pregnancy outcomes, 1995-2006



Total number	171	186	150	179	169	176	168	182	215	219	187	190
Outcome	**Percent (%)**											
Neonatal death	1	0	2	0	1	1	1	0	0	0	0	1
Survived > 28 days	99	99	97	100	99	99	99	100	100	100	100	99
Stillbirth	0	1	1	0	0	0	0	0	0	0	0	0
TOP < 20 weeks	0	1	0	0	0	0	0	0	0	0	0	0
TOP >= 20 weeks*	0	0	1	0	0	0	1	0	0	0	0	1

**These cases are not identified in the dataset prior to 1996*

- Almost all babies with this condition survived beyond 28 days.

Developmental dysplasia of the hip and associated birth defects, 2005-2006

Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	0	0	0	356	356	94.4
Chromosomal	0	0	0	1	1	0.3
Other same system	0	0	0	4	4	1.1
Other different systems	1	0	1	14	16	4.2
Total	1	0	1	375	377	100.0

Five year summary of the prevalence of developmental dysplasia of the hip and selected infant and maternal characteristics, 2001 - 2006

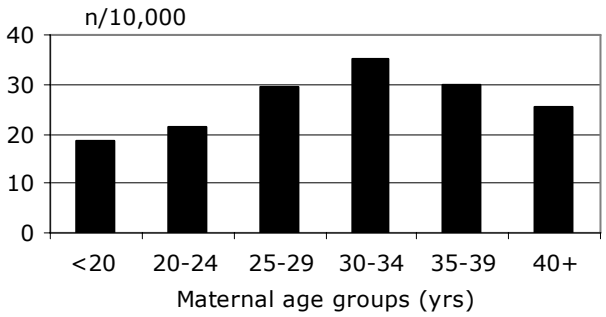
2001—2006			
Infant (n=1,161)	**No.**	**%**	**PR**
Sex			
Male	268	*23.1*	13.4
Female	892	*76.8*	46.9*
Indeterminate/ Unknown	1	*0.1*	
Plurality#			
Singleton	1138	*98.0*	30.2
Multiple	23	*2.0*	16.6
Maternal (n=1,158)			
Maternal Age (yrs)			
<20	21	*1.8*	18.5
20-24	96	*8.3*	21.4
25-29	297	*25.6*	29.3
30-34	496	*42.8*	35.1**
35-39	213	*18.4*	30.0
40+	35	*3.0*	25.4
Unknown	0	*0.0*	
Country of birth			
Australia	943	*81.4*	32.4
Oceania inc NZ	25	*2.2*	24.3
UK inc Eire	43	*3.7*	40.4
Europe	27	*2.3*	22.6
Middle East	18	*1.6*	20.7
Asia	75	*6.5*	20.1***
Nth America	6	*0.5*	25.7
Sth America	7	*0.6*	31.0
Africa	12	*1.0*	15.6***
Unknown	2	*0.2*	

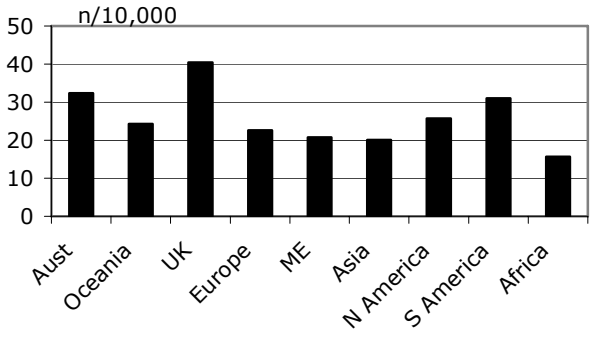
#excludes unknown

** statistically significant, p<0.0001*

*** statistically significant, p<0.0001*

**** statistically significant, p<0.0001*





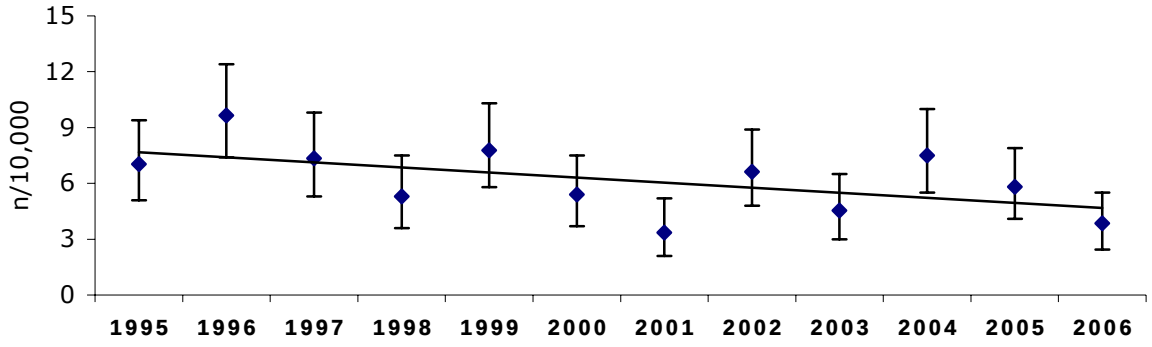
- The PR for females was significantly higher than for males. The PR for women aged 30-34 years was significantly higher than for women aged 24 years or less. The PR for Asian-born and African-born women was significantly lower than for women born in Australia or the UK including Eire.

6.23 Limb reduction defects

British Paediatric Association code 755.20 – 755.49

Definition: There is a wide range of severity from partial absence of a phalanx to complete absence of a major skeletal structure such as a whole limb. These may, for other purposes, be analysed in groups: transverse, longitudinal, intercalary, multiple or unspecified.

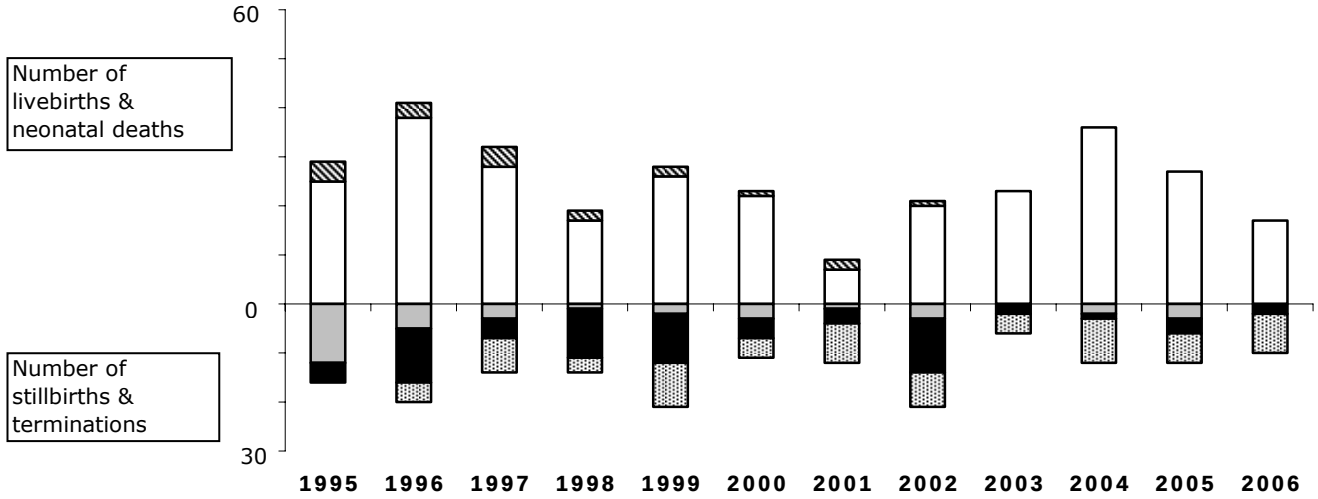
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	7.0	9.6	7.3	5.3	7.8	5.4	3.4	6.6	4.5	7.5	5.8	3.8

- From 1995 to 2006 there has been no statistically significant change in the prevalence of limb reduction defects.

Number of cases and pregnancy outcomes, 1995-2006



Total number	45	61	46	33	49	34	21	42	29	48	39	27
Outcome	**Percent (%)**											
Neonatal death	9	5	9	6	4	3	10	2	0	0	0	0
Survived > 28 days	56	62	61	52	53	65	33	48	79	75	69	63
Stillbirth	27	8	7	3	4	9	5	7	0	4	8	0
TOP < 20 weeks	9	18	9	30	20	12	14	26	7	2	8	7
TOP >= 20 weeks*	0	7	15	9	18	12	38	17	14	19	15	30

**These cases are not identified in the dataset prior to 1996*

- On average 62% of babies with this condition survived beyond 28 days in 2001-2006.

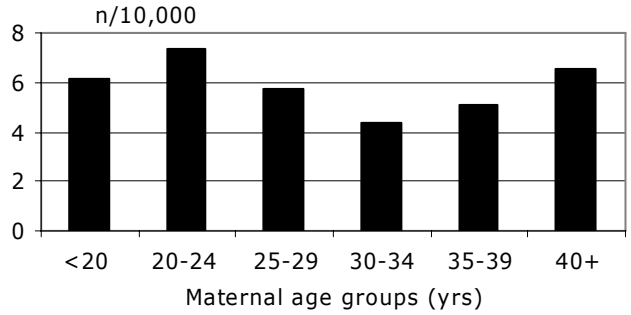
Limb reduction defects and associated birth defects, 2005-2006

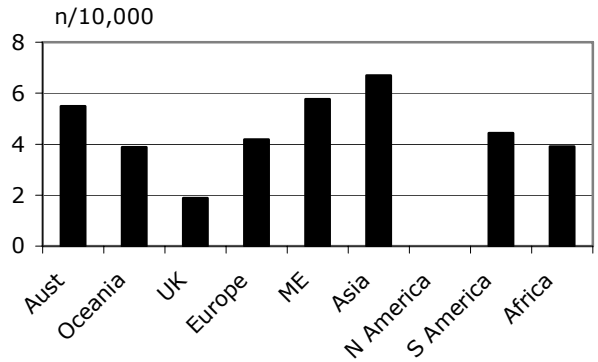
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	4	0	0	17	21	31.8
Chromosomal	6	0	0	1	7	10.6
Other same system	1	0	0	13	14	21.2
Other different systems	8	3	0	13	24	36.4
Total	19	3	0	44	66	100.0

Five year summary of the prevalence of limb reduction defects and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=206)	**No.**	**%**	**PR**
Sex			
Male	102	*49.5*	5.1
Female	96	*46.6*	5.0
Indeterminate/ Unknown	8	*3.9*	
Plurality#			
Singleton	197	*95.6*	5.2
Multiple	9	*4.4*	6.5
Maternal (n=206)			
Maternal Age (yrs)			
<20	7	*3.4*	6.2
20-24	33	*16.0*	7.3
25-29	58	*28.2*	5.7
30-34	62	*30.1*	4.4
35-39	36	*17.5*	5.1
40+	9	*4.4*	6.5
Unknown	1	*0.5*	
Country of birth			
Australia	160	*77.7*	5.5
Oceania inc NZ	4	*1.9*	3.9
UK inc Eire	2	*1.0*	1.9
Europe	5	*2.4*	4.2
Middle East	5	*2.4*	5.8
Asia	25	*12.1*	6.7
Nth America	0	*0.0*	0.0
Sth America	1	*0.5*	4.4
Africa	3	*1.5*	3.9
Unknown	1	*0.5*	

#excludes unknown





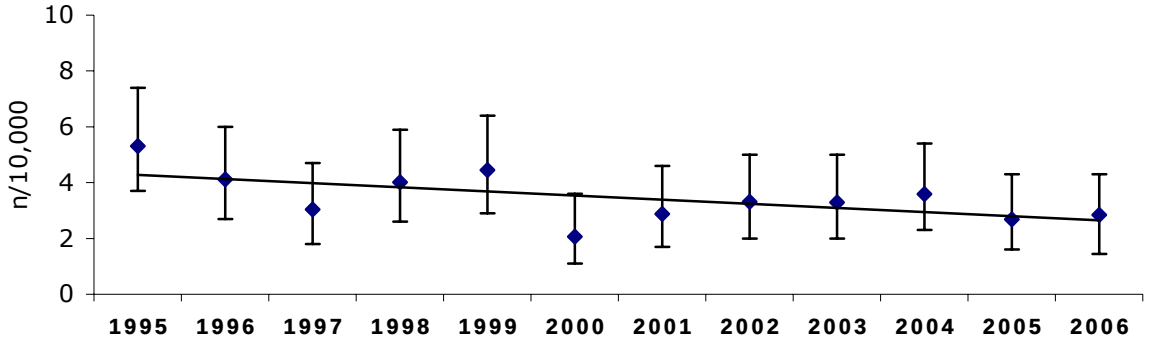
- There were no statistically significant associations for sex of infant, plurality, maternal age or maternal country of birth in 2001-2006.

6.24 Diaphragmatic hernia

British Paediatric Association code 756.61

Definition: Herniation into the thorax of abdominal organs through a defect of the diaphragm.

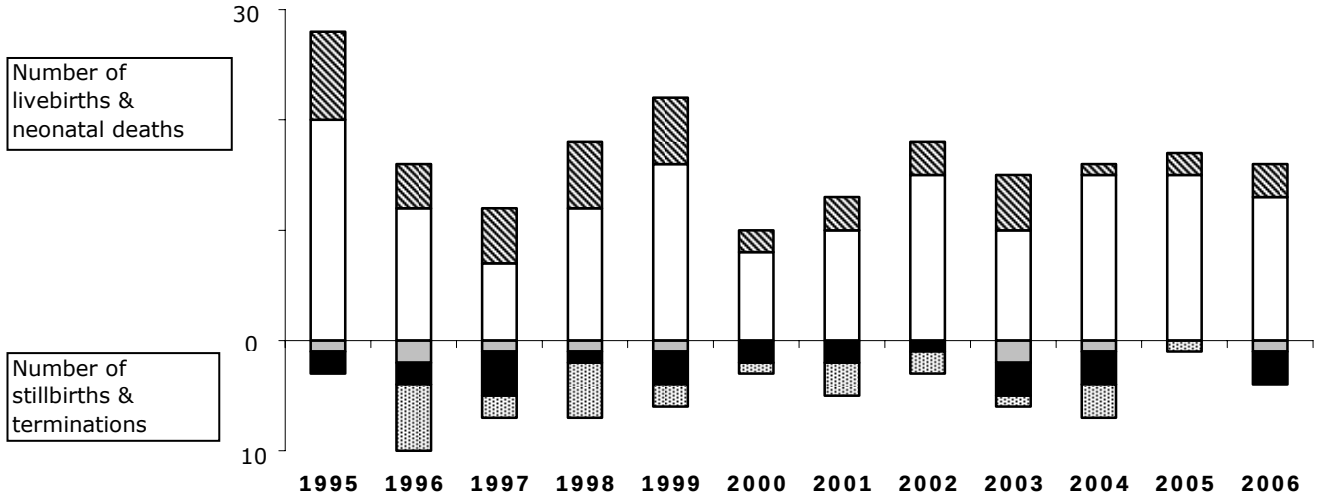
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	5.3	4.1	3.0	4.0	4.4	2.1	2.9	3.3	3.3	3.6	2.7	2.8

- From 1995 to 2006 there has been no statistically significant change in the prevalence of diaphragmatic hernia.

Number of cases and pregnancy outcomes, 1995-2006



Total number	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
	34	26	19	25	28	13	18	21	21	23	18	20
Outcome	**Percent (%)**											
Neonatal death	24	15	26	24	21	15	17	14	24	4	11	15
Survived > 28 days	59	46	37	48	57	62	56	71	48	65	83	65
Stillbirth	12	8	5	4	4	0	0	0	10	4	0	5
TOP < 20 weeks	6	8	21	4	11	15	11	5	14	13	0	15
TOP >= 20 weeks*	0	23	11	20	7	8	17	10	5	13	6	0

**These cases are not identified in the dataset prior to 1996*

- On average, 65% of babies with this condition survived beyond 28 days in 2001-2006.

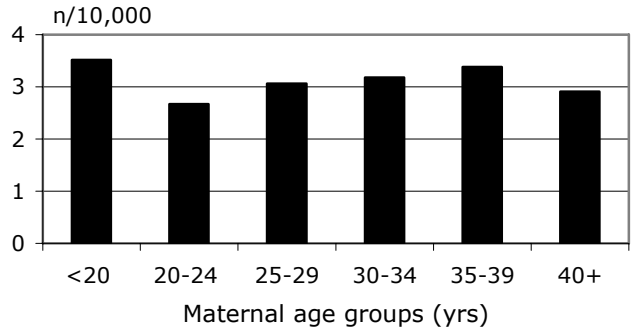
Diaphragmatic hernia and associated birth defects, 2005-2006

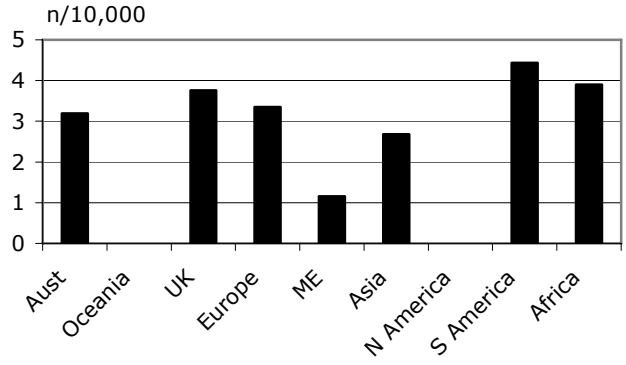
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	2	1	2	5	10	26.3
Chromosomal	1	0	2	0	3	7.9
Other same system	0	0	0	0	0	0.0
Other different systems	1	0	1	23	25	65.8
Total	4	1	5	28	38	100.0

Five year summary of the prevalence of diaphragmatic hernia and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=121)	**No.**	**%**	**PR**
Sex			
Male	77	*63.6*	3.8*
Female	41	*33.9*	2.2
Indeterminate/ Unknown	3	*2.5*	
Plurality#			
Singleton	117	*96.7*	3.1
Multiple	4	*3.3*	2.9
Maternal (n=121)			
Maternal Age (yrs)			
<20	4	*3.3*	3.5
20-24	12	*9.9*	2.7
25-29	31	*25.6*	3.1
30-34	45	*37.2*	3.2
35-39	24	*19.8*	3.4
40+	4	*3.3*	2.9
Unknown	1	*0.8*	
Country of birth			
Australia	93	*76.9*	3.2
Oceania inc NZ	0	*0.0*	0.0
UK inc Eire	4	*3.3*	3.8
Europe	4	*3.3*	3.4
Middle East	1	*0.8*	1.2
Asia	10	*8.3*	2.7
Nth America	0	*0.0*	0.0
Sth America	1	*0.8*	4.4
Africa	3	*2.5*	3.9
Unknown	5	*4.1*	

#excludes unknown
** statistically significant, p<0.01*





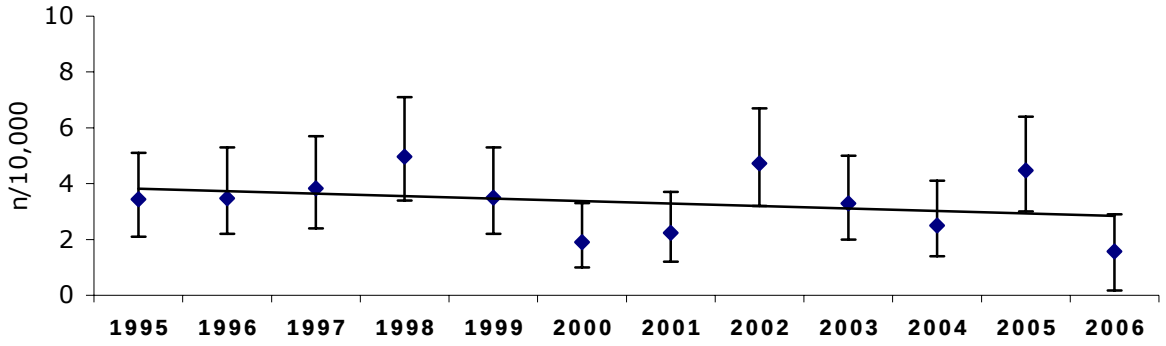
- PR for males was significantly higher than for females. There were no statistically significant associations for plurality, maternal age or maternal country of birth in 2001-2006.

6.25 Exomphalos

British Paediatric Association code 756.70

Definition: Herniation of abdominal contents through the umbilical insertion and covered by membrane which may or may not be intact.

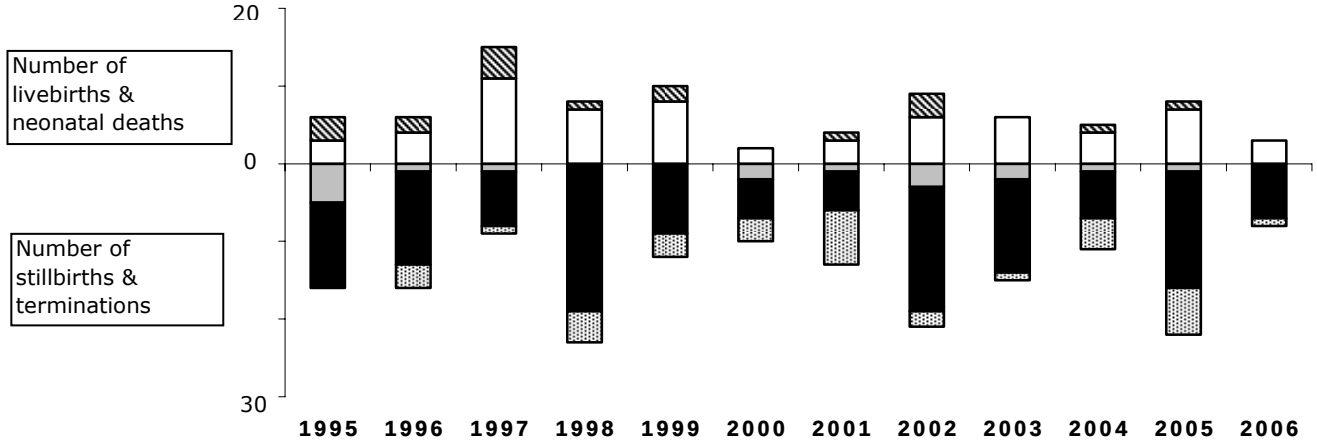
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	3.4	3.5	3.8	5.0	3.5	1.9	2.2	4.7	3.3	2.5	4.5	1.6

- From 1995 to 2006 there has been no statistically significant change in the prevalence of exomphalos.

Number of cases and pregnancy outcomes, 1995-2006



Total number	22	22	24	31	22	12	14	30	21	16	30	11
Outcome	**Percent (%)**											
Neonatal death	14	9	17	3	9	0	7	10	0	6	3	0
Survived > 28 days	14	18	46	23	36	17	21	20	29	25	23	27
Stillbirth	23	5	4	0	0	17	7	10	10	6	3	0
TOP < 20 weeks	50	55	29	61	41	42	36	53	57	38	50	64
TOP >= 20 weeks*	0	14	4	13	14	25	29	7	5	25	20	9

**These cases are not identified in the dataset prior to 1996*

- On average 24% of babies with this condition survived beyond 28 days in 2001-2006. Increasing rate of TOP was probably due to prenatal detection of cases with a chromosomal abnormality (32%).

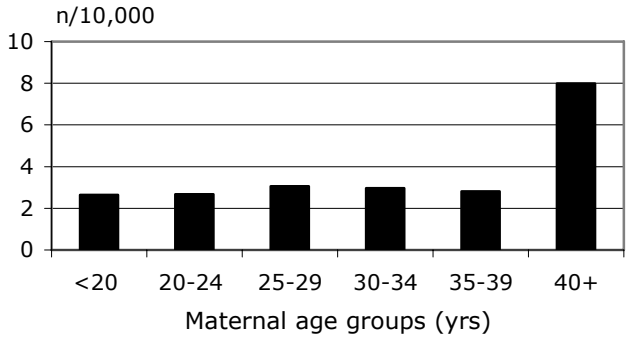
Exomphalos and associated birth defects, 2005-2006

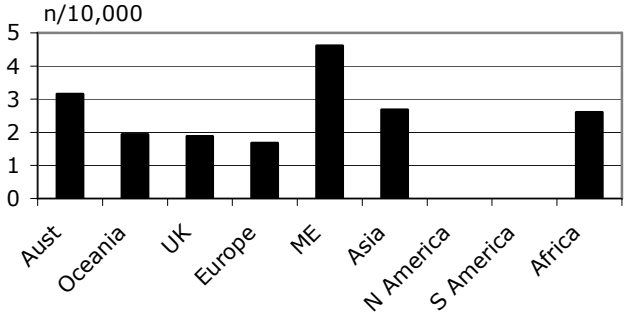
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	4	0	0	2	6	14.6
Chromosomal	12	0	1	0	13	31.7
Other same system	0	0	0	0	0	0.0
Other different systems	13	1	0	8	22	53.7
Total	29	1	1	10	41	100.0

Five year summary of the prevalence of exomphalos and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=122)	**No.**	**%**	**PR**
Sex			
Male	60	*49.2*	3.0
Female	49	*40.2*	2.6
Indeterminate/ Unknown	13	*10.6*	
Plurality#			
Singleton	115	*94.3*	3.0
Multiple	7	*5.7*	5.0
Maternal (n=121)			
Maternal Age (yrs)			
<20	3	*2.5*	2.6
20-24	12	*9.9*	2.7
25-29	31	*25.6*	3.1
30-34	42	*34.7*	3.0
35-39	20	*16.5*	2.8
40+	11	*9.1*	8.0
Unknown	2	*1.7*	
Country of birth			
Australia	92	*76.0*	3.2
Oceania inc NZ	2	*1.7*	1.9
UK inc Eire	2	*1.7*	1.9
Europe	2	*1.7*	1.7
Middle East	4	*3.3*	4.6
Asia	10	*8.3*	2.7
Nth America	0	*0.0*	0.0
Sth America	0	*0.0*	0.0
Africa	2	*1.7*	2.6
Unknown	7	*5.8*	

#excludes unknown





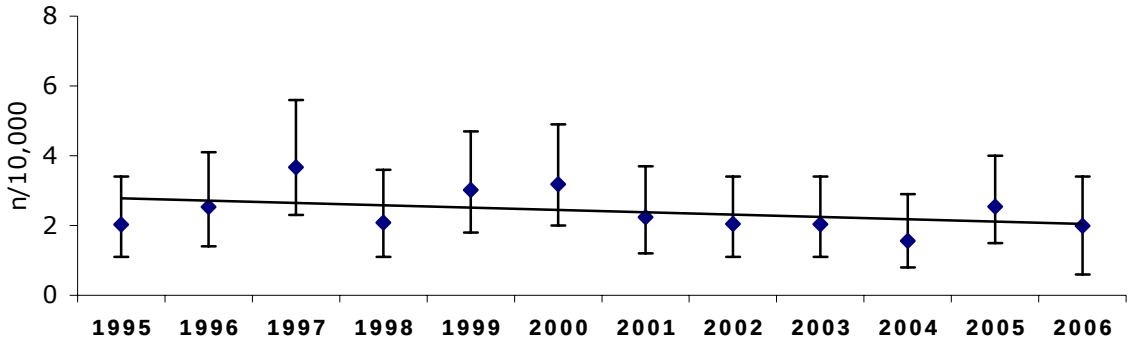
- Although some PRs were higher than others, due to small sample size there were no statistically significant associations for sex of infant, plurality, maternal age or maternal country of birth in 2001-2006.

6.26 Gastroschisis

British Paediatric Association code 756.71

Definition: Visceral herniation through a right sided abdominal wall defect to an intact umbilical cord, and not covered by a membrane.

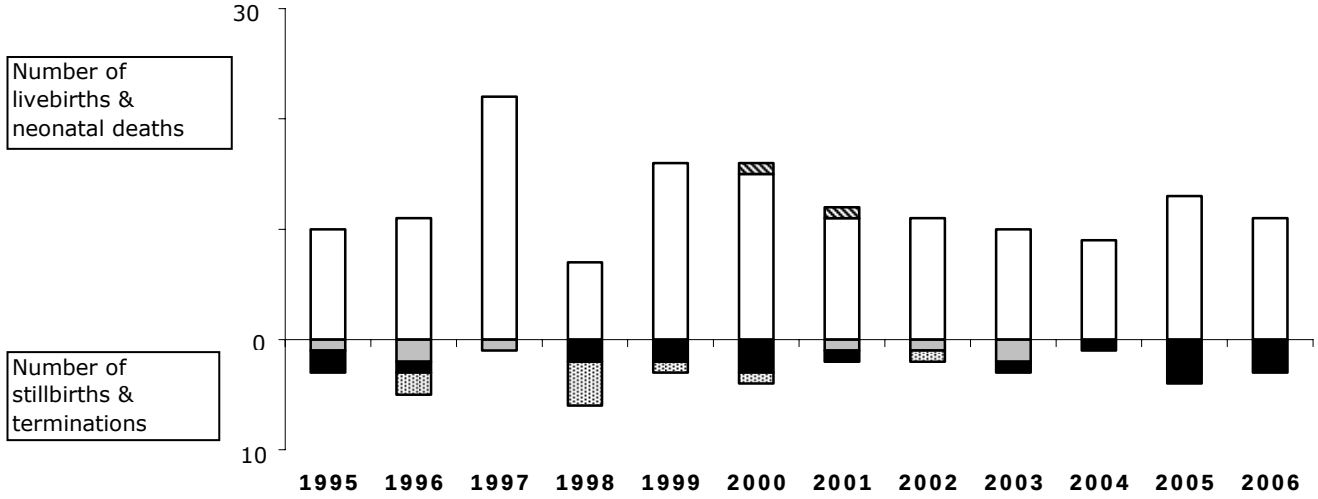
Trend in prevalence rates, 1995-2006



N / 10,000	2.0	2.5	3.7	2.1	3.0	3.2	2.2	2.0	2.0	1.6	2.5	2.0

- From 1995 to 2006 there has been no statistically significant change in the prevalence of gastroschisis.

Number of cases and pregnancy outcomes, 1995-2006



Total number	13	16	23	13	19	20	14	13	13	10	17	14
Outcome	**Percent (%)**											
Neonatal death	0	0	0	0	0	5	7	0	0	0	0	0
Survived > 28 days	77	69	96	54	84	75	79	85	77	90	76	79
Stillbirth	8	13	4	0	0	0	7	8	15	0	0	0
TOP < 20 weeks	15	6	0	15	11	15	7	0	8	10	24	21
TOP >= 20 weeks*	0	13	0	31	5	5	0	8	0	0	0	0

**These cases are not identified in the dataset prior to 1996*

- On average, 81% of babies with this condition survived beyond 28 days in 2001-2006.

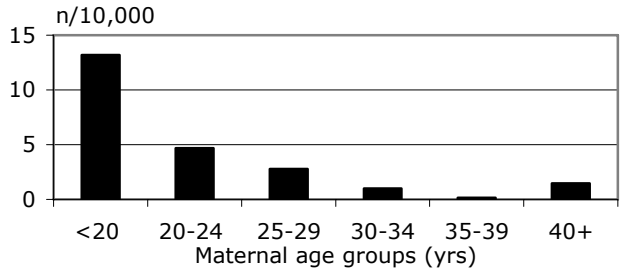
Gastroschisis and associated birth defects, 2005-2006

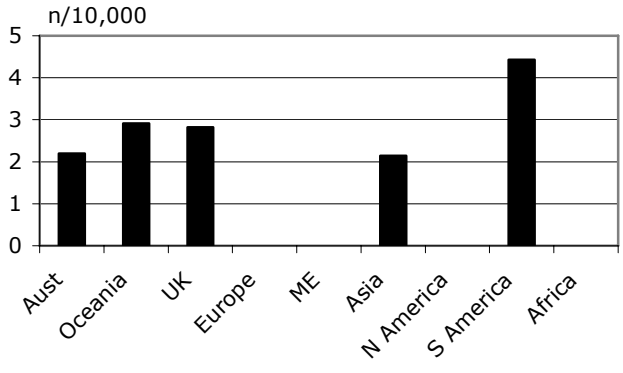
Type	TOP (pre & post 20 weeks)	Stillbirth	Neonatal Death	Surviving > 28 Days	Total	% of Total
Isolated anomaly	1	0	0	21	22	71.0
Chromosomal	3	0	0	0	3	9.8
Other same system	1	0	0	0	1	3.2
Other different systems	2	0	0	3	5	16.1
Total	7	0	0	24	31	100.0

Five year summary of the prevalence of gastroschisis and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=81)	**No.**	**%**	**PR**
Sex			
Male	45	*55.6*	2.2
Female	35	*43.2*	1.8
Indeterminate/ Unknown	1	*1.2*	
Plurality#			
Singleton	80	*98.8*	2.1
Multiple	1	*1.2*	0.7
Maternal (n=81)			
Maternal Age (yrs)			
<20	15	*18.5*	13.2*
20-24	21	*25.9*	4.7*
25-29	28	*34.6*	2.8
30-34	14	*17.3*	1.0
35-39	1	*1.2*	0.1
40+	2	*2.5*	1.5
Unknown	0	*0.0*	
Country of birth			
Australia	64	*79.0*	2.2
Oceania inc NZ	3	*3.7*	2.9
UK inc Eire	3	*3.7*	2.8
Europe	0	*0.0*	0.0
Middle East	0	*0.0*	0.0
Asia	8	*9.9*	2.1
Nth America	0	*0.0*	0.0
Sth America	1	*1.2*	4.4
Africa	0	*0.0*	0.0
Unknown	2	*2.5*	

#excludes unknown
**statistically significant, p<0.0001*





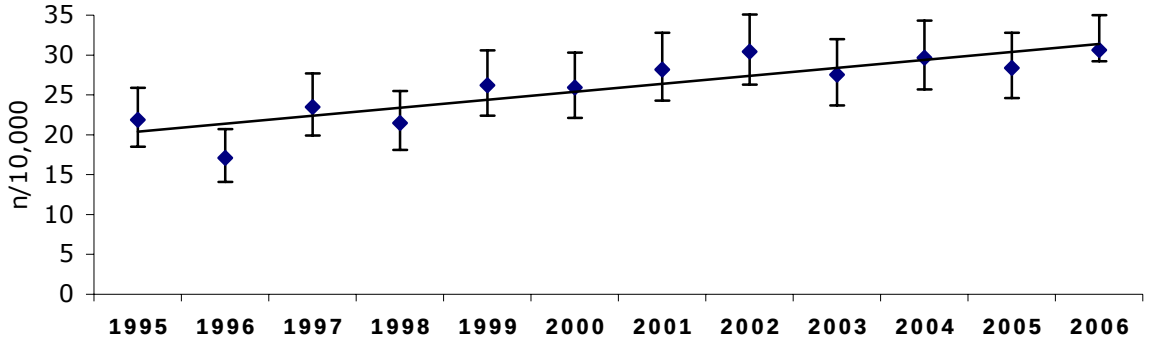
- The PR for women aged 24 years and less was significantly higher than for women aged 25 years and over. Although some PRs were higher than others, due to small sample size there were no statistically significant associations for sex of infant, plurality or maternal country of birth in 2001-2006.

6.27 Trisomy 21

British Paediatric Association code 758.00 – 758.09

Definition: Down Syndrome – additional chromosome 21.

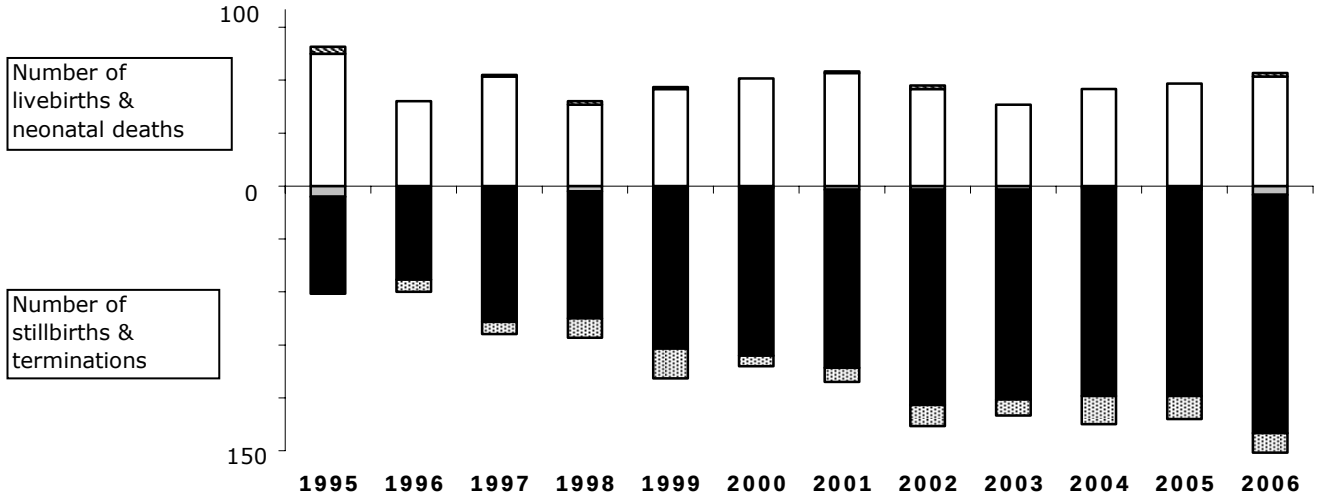
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	21.9	17.1	23.5	21.5	26.2	25.9	28.2	30.4	27.5	29.7	28.4	30.6

- From 1995 to 2006 there has been a statistically significant increase in the prevalence of Trisomy 21. However, this significant increase was not seen from 2001-2006.

Number of cases and pregnancy outcomes, 1995-2006



Total number	140	108	147	134	165	163	176	193	176	190	190	215
Outcome	**Percent (%)**											
Neonatal death	3	0	1	1	1	0	1	1	0	0	1	1
Survived > 28 days	54	44	42	34	33	37	36	28	26	29	31	29
Stillbirth	4	0	0	2	1	1	1	1	1	1	1	2
TOP < 20 weeks	39	49	52	54	55	58	57	63	68	62	62	63
TOP >= 20 weeks*	0	6	5	8	10	4	5	6	5	8	7	5

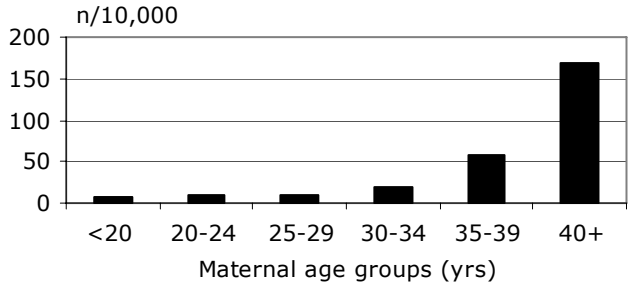
**These cases are not identified in the dataset prior to 1996*

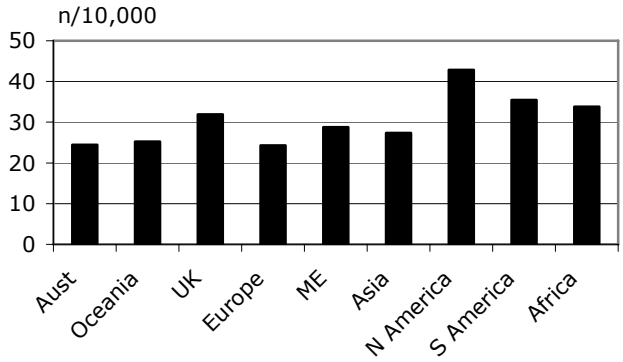
- On average 30% of babies with this condition survived beyond 28 days in 2001-2006. The proportion of TOPs has remained stable over the last five years.

Five year summary of the prevalence of Trisomy 21 and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=1,140)	**No.**	**%**	**PR**
Sex			
Male	634	*55.6*	31.6*
Female	486	*42.6*	25.6
Indeterminate/ Unknown	20	*1.8*	
Plurality#			
Singleton	1109	*97.3*	29.4
Multiple	31	*2.7*	22.3
Maternal (n=1,139)			
Maternal Age (yrs)			
<20	7	*0.6*	6.2
20-24	46	*4.0*	10.2
25-29	92	*8.1*	9.1
30-34	257	*22.5*	18.2**
35-39	419	*36.8*	59.0**
40+	231	*20.3*	167.8**
Unknown	87	*7.6*	
Country of birth			
Australia	712	*62.5*	24.4
Oceania inc NZ	26	*2.3*	25.3
UK inc Eire	34	*3.0*	31.9
Europe	29	*2.5*	24.3
Middle East	25	*2.2*	28.8
Asia	102	*9.0*	27.3
Nth America	10	*0.9*	42.9
Sth America	8	*0.7*	35.4
Africa	26	*2.3*	33.8
Unknown	167	*14.6*	

#excludes unknown
**statistically significant, p<0.001*
***statistically significant, P<0.0001*





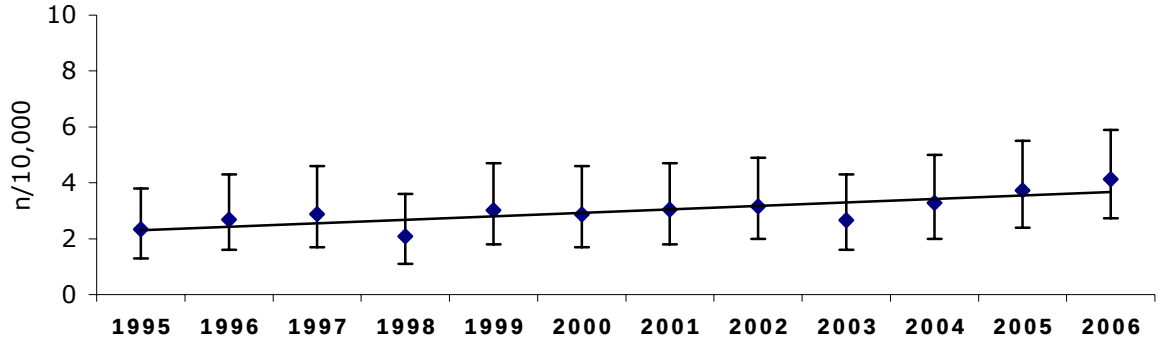
- PR for males was significantly higher than for females. The PR for women aged 30 years and more was significantly higher than for women aged less than 30. There were no statistically significant associations for plurality or maternal country of birth in 2001-2006.
- There were an additional 16 cases of Trisomy 21 identified through prenatal diagnosis in 2005 – 2006 that have not been reported to the VBDR. They are presumed to be miscarriages or un-notified pregnancy terminations before 20 weeks gestation

6.28 Trisomy 13

British Paediatric Association code 758.10 – 758.19

Definition: Patau Syndrome – additional chromosome 13.

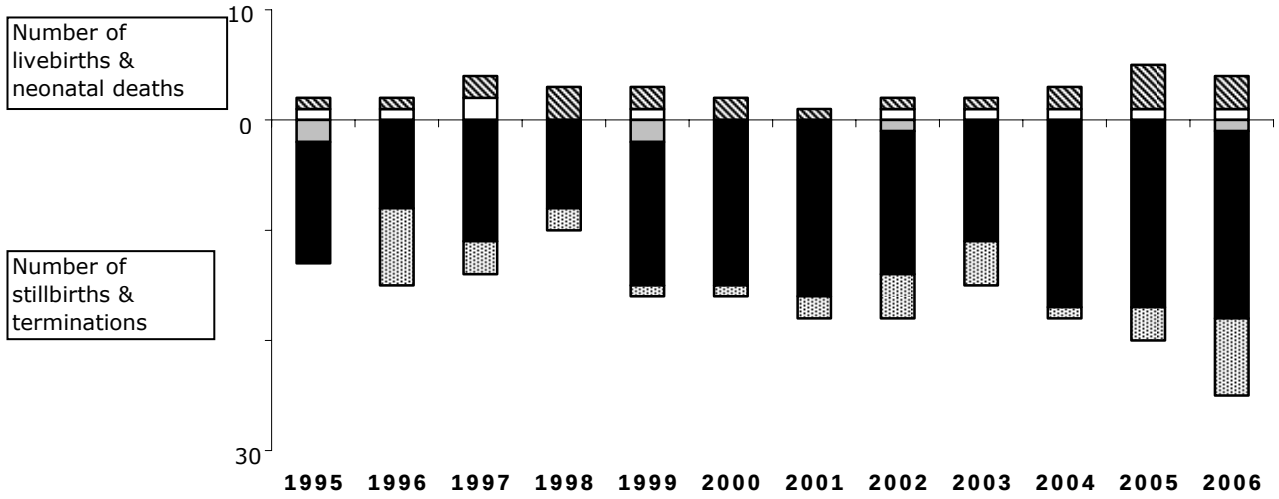
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	2.3	2.7	2.9	2.1	3.0	2.9	3.0	3.2	2.7	3.3	3.7	4.1

- From 1995 to 2006 there has been a statistically significant increase in the prevalence of Trisomy 13. This significant increase was also seen from 2001-2006.

Number of cases and pregnancy outcomes, 1995-2006



Total number	15	17	18	13	19	18	19	20	17	21	25	29
Outcome	**Percent (%)**											
Neonatal death	7	6	11	23	11	11	5	5	6	10	16	10
Survived > 28 days	7	6	11	0	5	0	0	5	6	5	4	3
Stillbirth	13	0	0	0	11	0	0	5	0	0	0	3
TOP < 20 weeks	73	47	61	62	68	83	84	65	65	81	68	59
TOP >= 20 weeks*	0	41	17	15	5	6	11	20	24	5	12	24

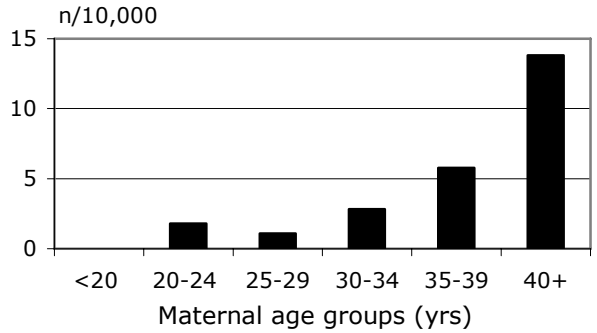
**These cases are not identified in the dataset prior to 1996*

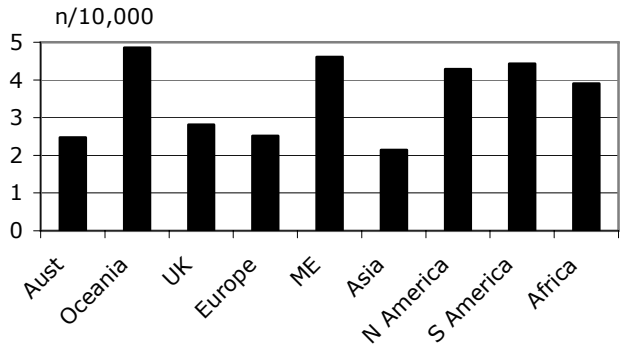
- Less than 5% of babies with this condition survived beyond 28 days in 2001-2006.

Five year summary of the prevalence of Trisomy 13 and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=131)	**No.**	**%**	**PR**
Sex			
Male	70	*53.4*	3.5
Female	55	*42.0*	2.9
Indeterminate/ Unknown	6	*4.6*	
Plurality#			
Singleton	131	*100.0*	3.5
Multiple	0	*0.0*	0.0
Maternal (n=131)			
Maternal Age (yrs)			
<20	0	*0.0*	0.0
20-24	8	*6.1*	1.8
25-29	11	*8.4*	1.1
30-34	40	*30.5*	2.8*
35-39	41	*31.3*	5.8*
40+	19	*14.5*	13.8*
Unknown	12	*9.2*	
Country of birth			
Australia	72	*55.0*	2.5
Oceania inc NZ	5	*3.8*	4.9
UK inc Eire	3	*2.3*	2.8
Europe	3	*2.3*	2.5
Middle East	4	*3.1*	4.6
Asia	8	*6.1*	2.1
Nth America	1	*0.8*	4.3
Sth America	1	*0.8*	4.4
Africa	3	*2.3*	3.9
Unknown	31	*23.7*	

#excludes unknown
**statistically significant, p<0.0001*





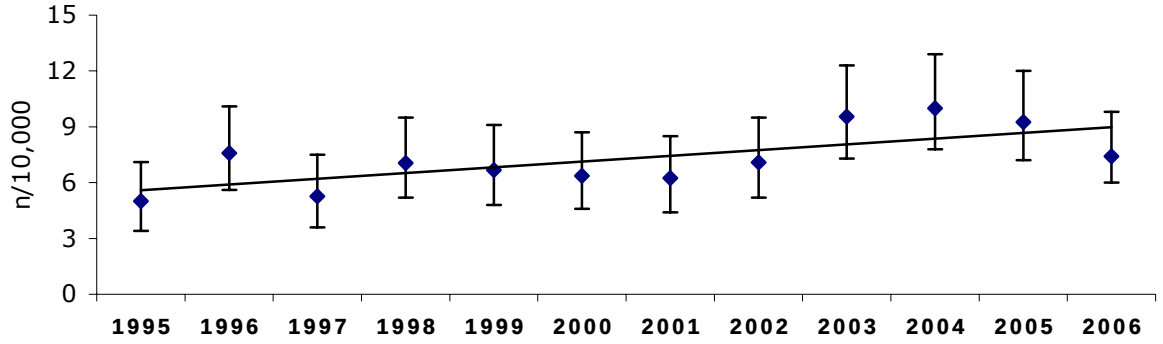
- The PR for women aged 30 years and more was significantly higher than for women aged less than 30. There were no statistically significant associations for sex of infant, plurality or maternal country of birth in 2001-2006.
- There were an additional 3 cases of Trisomy 13 identified through prenatal diagnosis in 2005 – 2006 that have not been reported to the VBDR. They are presumed to be miscarriages or un-notified pregnancy terminations before 20 weeks gestation

6.29 Trisomy 18

British Paediatric Association code 758.20 – 758.29

Definition: Edward Syndrome – additional chromosome 18.

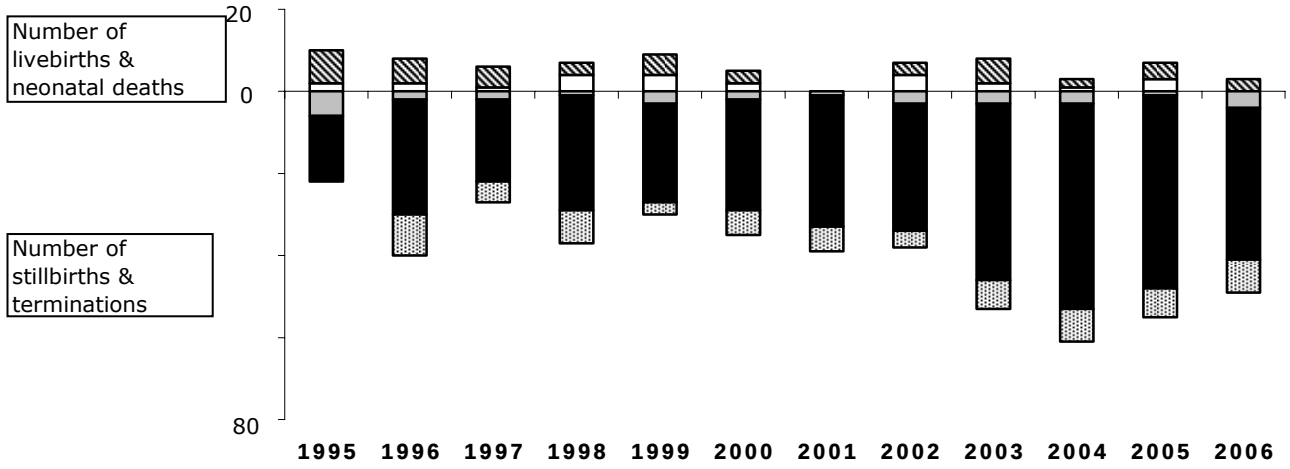
Trend in prevalence rates, 1995-2006



	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006
N/10,000	5.0	7.6	5.3	7.1	6.7	6.4	6.2	7.1	9.5	10.0	9.3	7.4

- From 1995 to 2006 there has been a statistically significant increase in the prevalence of Trisomy 18. However, this significant increase was not seen from 2001-2006.

Number of cases and pregnancy outcomes, 1995-2006



Total number	32	48	33	44	42	40	39	45	61	64	62	52
Outcome	**Percent (%)**											
Neonatal death	25	13	15	7	12	8	0	7	10	3	6	6
Survived > 28 days	6	4	3	9	10	5	0	9	3	2	5	0
Stillbirth	19	4	6	2	7	5	3	7	5	5	2	8
TOP < 20 weeks	50	58	61	64	57	68	82	69	70	78	76	71
TOP >= 20 weeks*	0	21	15	18	7	15	15	9	11	13	11	15

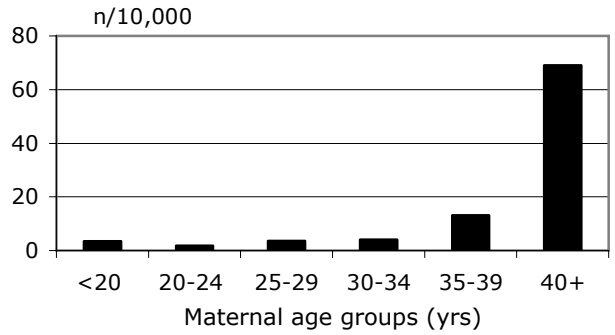
**These cases are not identified in the dataset prior to 1996*

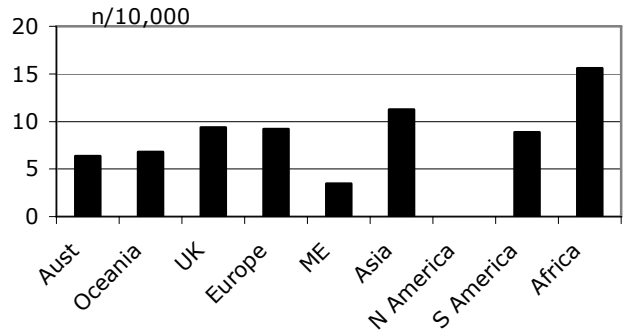
- Less than 5% of babies with this condition survived beyond 28 days in 2001-2006.

Five year summary of the prevalence of Trisomy 18 and selected infant and maternal characteristics, 2001 - 2006

2001—2006			
Infant (n=323)	**No.**	**%**	**PR**
Sex			
Male	145	*44.9*	7.2
Female	169	*52.3*	8.9
Indeterminate/ Unknown	9	*2.8*	
Plurality#			
Singleton	315	*97.5*	8.4
Multiple	8	*2.5*	5.8
Maternal (n=323)			
Maternal Age (yrs)			
<20	4	*1.2*	3.5
20-24	8	*2.5*	1.8
25-29	36	*11.1*	3.6
30-34	57	*17.6*	4.0
35-39	93	*28.8*	13.1*
40+	95	*29.4*	69.0*
Unknown	30	*9.3*	
Country of birth			
Australia	185	*57.3*	6.4
Oceania inc NZ	7	*2.2*	6.8
UK inc Eire	10	*3.1*	9.4
Europe	11	*3.4*	9.2
Middle East	3	*0.9*	3.5
Asia	42	*13.0*	11.3**
Nth America	0	*0.0*	0.0
Sth America	2	*0.6*	8.9
Africa	12	*3.7*	15.6**
Unknown	51	*15.8*	

#excludes unknown
**statistically significant, p<0.0001*
***statistically significant, p<0.001*





- The PR for women aged 35 years and more was significantly higher than for women aged less than 34 years. The PR for Asian-born and African born women was significantly higher than for Australian-born women. There were no statistically significant associations for sex of infant or plurality in 2001-2006.
- There were an additional 2 cases of Trisomy 18 identified through prenatal diagnosis in 2005 – 2006 that have not been reported to the VBDR. They are presumed to be miscarriages or un-notified pregnancy terminations before 20 weeks gestation

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Appendix A Activities of the Victorian Birth Defects Register since July 2006 (until June 2008)

The following table summarises projects that have used (or are currently underway and using) VBDR data since July 2006.

No.	Project Title	Collaborators
1	Demographics of Victorian women who have prenatal diagnosis in 1992, 1998 , 2002 and mapping of prenatal diagnosis across Victoria	Funded by MCRI
2	First information needs of families with a baby with Down Syndrome	Perpetual Foundation
3	Prevalence of Fetal Alcohol Syndrome in Victoria, Australia	Public Health Fellow project
4	Antenatal diagnosis of cardiac malformations	Advanced Medical Science project
5	How to measure alcohol in pregnancy	Funded by MCRI
6	Follow-up of 1st and 2nd trimester maternal serum screening in Victoria	Funded by DHS
7	Screening for Klinefelter's Syndrome	PhD project
8	Trends in birth outcome and survival of Down syndrome Victoria, Australia	HIM student
10	Updating predictors of having a baby with neural tube defects	Paediatric Trainee project
11	A registry and clinical study of birth defects and syndromes associated with cleft lip/palate	Genetic Fellow project
12	Health & development in children with chromosome rearrangements detected before birth	Masters of Health Science project
13	Non-users of genetic services - why don't they use services	Funded by DHS
14	Perinatal outcomes of singleton IVF	Funded by the BUPA Foundation
15	Exposure to siblings in early life and risk of Crohn's Disease in childhood	Funded by MCRI
16	Validation of the VBDR	HIM student/In-house project
17	An audit of developmental dysplasia of the hip in Victoria, Australia, 2007	In house project
18	Scoping of disability data collection in Victoria	Funded by DHS, DEECD and MCRI
19	Follow-up of prenatal detection and management of fetal abnormality- Phase I & Phase 2	Advanced Medical Science project
20	Investigation of an increased rate of birth defects in a regional area of Victoria and a possible birth defects cluster within this region	Public Health Fellow

GHSV: Genetic Health Services Victoria

MCRI: Murdoch Childrens Research Institute

RCH: Royal Children's Hospital

RWH: Royal Women's Hospital

Provision of data to the International Clearinghouse of Birth Defects Surveillance and Research (ICBDSR)

Victorian BDR data are again included in their annual report, which is available on the website: www.icbd.org/. This report summarises data from more than 40 member programs across five continents.

Data from the VBDR have also been provided for a number of collaborative studies with the ICBDM:

- International collaborative research on Craniofacial Anomalies (supported by WHO Human Genetics Program) - ongoing
- Very rare birth defects project –

Leoncini E et al, Frequency of holoprosencephaly in the International Clearinghouse for Birth Defects Surveillance Systems: searching for population variations, (paper submitted)

Requests for unpublished data since July 2006—June 2008

Examples of ad-hoc requests for unpublished data from July 2006 to July 2008.

- Obstetrics & Gynaecology Fellow - requested information on x-linked hydrocephalus for conference presentation.
- Department of Primary Industry - requested information on limb reduction defects in children in specific rural areas, as they had seen an outbreak of hemimelia in claves -> lead to a 'cluster investigation'.
- Researcher, MCRI - requested advice on classification of specific birth defects for the development of an NH & MRC research grant.
- Interstate BDR - requested information on fetal alcohol syndrome (FAS) numbers for an APSU publication on FAS.
- Cleft Registry, RCH - requested birth data to link to their cleft cases, to provide more complete data on their Registry.
- Epidemiologist, DHS – requested data on prevalence of Down Syndrome in women aged 35 years or more for inclusion in a report on preventable disease.
- Department of paediatrics and adolescent gynaecology – requested trends in the prevalence of specific conditions to analyse changes in workload over time.
- Masters student – requested trends in prevalence in conjoined twins for project.
- PhD student, University of Melbourne – requested information on undescended testes (UDT) and associated anomalies for a PhD on testicular descent and limb bud development.
- Medical Defence Association of Victoria – requested data on the number of birth defects diagnosed antenatally to assist in development of guidelines.
- Down Syndrome Association – requested information on the number of pregnancies and livebirths affected by Down Syndrome in Metropolitan Melbourne over time, for inclusion in a funding request.
- Urology Department, RCH – requested data on the month of birth of hypospadias cases from 1983 to current to investigate association between month of birth and the development of this condition.
- ICU, RCH – requested information on the prevelance of TOPs for HLHS from 1995-2005 to analyse decisions parents make with regard to HLHS outcomes.
- Aussie Hands Foundation – requested data on the prevalence of specific hand anomalies for inclusion on their website.
- Obstetrics Department – requested data on TOPs at their hospital to undertake an audit of cases.
- Barwon Health, DHS – requested data on number of liveborn spina bifida cases over a number of years for planning of service provision for "Transition of Young Adults with Complex Medical Conditions".
- Neonatologist, NETS – requested updated data on diaphragmatic hernia to continue study on evaluating changes in its treatment.
- Researcher, MCRI – requested birth defect data to link with their Crohn's cases and controls to determine if any associated anomalies.
- ICHBDSR – requested information on maternal age and outcome of Down Syndrome cases from 1993-2005 for a journal article.
- Professor of Paediatrics, USA University – requested information on trends in prevalence of Hirschsprung's Disease for international comparisons.

Publications, 2006—2008 (using BDR data or specific to VBDR interests)

1. Vallino-Napoli LD, Riley MM, Halliday **J**L. An epidemiologic study of orofacial clefts with other birth defects in Victoria, Australia. *Cleft Palate-Craniofacial Journal*, 43:571-576 (2006).

2. Muggli EE, McCloskey D and Halliday**, JL**. Health behaviour modelling for prenatal diagnosis in Australia: A geodemographic framework for health service utilisation and policy development. *BMC Health Services Research*, 6:109 (2006)

3. Muggli EE, Collins VR and Halliday **JL**. Mapping uptake of prenatal diagnosis for Down syndrome across Victoria, Australia. *ANZJOG*, 46:492-500 (2006)

4. 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, Poetzch S, Ritvanen A, Robert-Gnansia E, Siffel C, Stoll C, and Mastroiacovo P. Trends of Selected Malformations in Relation to Folic Acid Recommendations and Fortification: an International Assessment. *Birth Defects Research (Part A)*, 76:693-705 (2006)

5. Muggli EE and Halliday, JL. Folic Acid and Risk of Twinning: A systematic review of the recent literature, July 1994-July 2006. Med J Aus, 186:243-248 (2007)

6. Allen K, Riley M, Goldfeld S and Halliday J. Estimating the Prevalence of Fetal Alcohol Syndrome in Victoria using routinely collected administrative data. Aus NZ J Public Health, 31:62-66 (2007)

7. Halliday, J. Outcomes of IVF conceptions - are they different? Best Practice & Research Clinical Obstetrics and Gynaecology: 21,1:67-81 (2007)

8. Jaques AM, Halliday J, Francis I, Bonacquisto L, Forbes R, Cronin A and Sheffield L. Follow-up and evaluation of the Victorian first trimester combined screening program for Down syndrome and trisomy 18 BJOG. 2007 Jul;114(7):812-8 (2007)

9. Colleen Chew, JL Halliday, MM Riley, DJ Penny. A Population-based study of Antenatal Detection of Congenital Heart Disease by Ultrasound *Ultrasound Obstet Gynecol* 29,619-624 (2007)

10. Morley R, Halliday J, Donath S. Low to moderate alcohol consumption in pregnancy: how can we get better evidence? *MJA* 187 (5) 315 *(*Sept 2007)

11. Collins VR, Muggli EE, Riley M, Palma S, Halliday JL. Is Down Syndrome a disappearing birth defect? *J Pediatrics* 152_20-4 (2008)

12. Nagle C**,** Gunn J, Bell R, Lewis S, Meiser B, Metcalfe S, Ukoumunne O and Halliday J. Use of a decision aid for prenatal testing of fetal abnormalities to improve women's informed decision making: a cluster randomised controlled trial [ISRCTN22532458] BJOG 115:339-347 (2008)

13. Herlihy, A. S.Halliday, J. Is paternal age playing a role in the changing prevalence of Klinefelter syndrome? Eur J Hum Genet May 21, 2008 [Epub ahead of print]

14. Nagle C, Lewis S, Meiser B, Gunn J, Halliday J and Bell, R. Exploring general practitioners' experience of informing women about prenatal screening tests for foetal abnormalities: a qualitative focus group studyBMC Health Serv Res 8 (1): 114 (2008)

Papers accepted by peer-reviewed journals

15. Jaques A, Collins V, Pitt J, Halliday J. Coverage of the Victorian newborn screening program in 2003: a retrospective population study (accepted J Paed & Child Health, Mar 2008)

16. Halliday J, Collins V, Riley M, Youssef D and Muggli E. Has prenatal screening influenced the prevalence of co-morbidities with Down Syndrome and subsequent survival rates? (accepted Paediatrics April 2008)

17. du Plessis L, Halliday J, Fletcher A, Riley M and Hunt R. What has happed with neural tube defects and women' understanding of folate in Victoria since 1998? (accepted MJA May 2008)

Reports (2006-2008)

In 2007, there were two (four page) **VBDR Bulletins** produced highlighting 2005 data of interest as an interim process for disseminating data. These are available on the Victorian Perinatal Data Collection Unit website: **www.health.vic.gov.au/perinatal**

In 2006, the VPDCU was involved with the Environmental Health Unit, Public Health, DHS, and the Cancer Council of Victoria in a cluster study regarding a landfill in Tullamarine. The result of this study was a report:

Lynch, Riley M, Davey M-A and Thursfield V. *Investigating health concerns in populations living near the Cleanaway landfill in Victoria*, Environmental Health Unit, Public Health Branch, Victorian Government Department of Human Services, Melbourne, 2006.

There has been continued monitoring of prenatal diagnosis by the Murdoch Childrens Research Institute (MCRI) in collaboration with Victorian Perinatal Data Collection Unit. An annual report is produced by: Evelyne Muggli and Jane Halliday. Prenatal Diagnosis in Victoria – 2006
Prenatal Diagnosis in Victoria – 2007

These are also available on the website and on request from:

Public Health Genetics
MCRI
Royal Children's Hospital
Parkville, 3052
tel: 83416260

Appendix B: List of exclusions

NB: There has been some variation in this list of exclusions between 1983—2002. Some excluded conditions may be included in this report if they were previously not excluded AND occur with other birth defect.

Abnormal palmar creases
Accessory nipples
Anal fissure
Balanced autosomal translocation (unless occurring with structural defects)
Birth injuries
Birth marks (smaller than 4cm, not including giant naevus)
Bowing of legs (unless severe)
Blocked tear ducts (dacrostenosis)
Brushfield spots
Cephalhaematoma
Cleft gum
Clicky hips
Clinodactyly
Craniotabes (unless severe)
Dermatogplyphic abnormalities
Ear abnormalities (minor)
Epicanthic folds
Gastro-oesophageal reflux
Haemangioma (<4 cm wide)
Hernia - inguinal, umbilical
High-arched palate
Hydrocele
Hypertelorism
Imperforate hymen
Laryngeal stridor
Larygomalacia
Low slung/set ears
Macroglossia (large tongue)
Meckel's diverticulum
Meconium ileus
Mental retardations (unless occurring with a syndrome/structural defect)
Metatarsus varus
Micrognathia (unless severe)
Mongolian spots
Occiput, flat/prominent
Patent ductus arteriosus (< 37 weeks)
Philtrum, long/short
Plagiocephaly
Pre-auricular sinus
Prominent forehead
Protruding tongue
Ptosis
Retrognathia (unless severe)
Rocker-bottom feet (prominent heels)
Sacral pits, dimples, sinuses
Short sternum
Simian creases
Single umbilical artery/2 vessels in cord
Skin folds/tags
Slanting eyes
Small mouth

Spina bifida occulta
Sternomastoid tumour
Subluxating knee joint
Talipes (positional)
Toe anomalies - minor
Tongue tie
Torticollis
Ureteric reflux (ultrasound diagnosed)
Webbing of 2nd & 3rd toes/fingers
Wide suture lines

Appendix C: Routine data items contained in the Victorian Birth Defects Register

Maternal data: postcode, date of birth, method of prenatal diagnosis, name

Infant /fetus data: hospital of birth, date of birth (or termination), sex, birthweight, plurality, rank, discharge status, date of death (if applicable), BPA Codes for congenital defects, position code, source of notification

Other data items available from linkage to the Perinatal Morbidity Statistics Form:

(1) **Maternal items:** UR number, local government area, region, country of birth, aboriginality, discharge date and status, marital status, number of previous pregnancies, date of completion of last pregnancy, outcome of last pregnancy, maternal medical conditions, obstetric complications, indication(s) for operative delivery, complications of labour birth and postnatal, procedures and operations, type of labour, presentation, method of delivery

(2) **infant data items**: apgar, time to establish respiration, resuscitation methods, neonatal morbidity

Appendix D: Denominator figures for infant and maternal Characteristics for twenty eight selected defects

Appendix D.1 Denominator statistics for maternal age, by confinements, for twenty eight selected defects, 2001—2006, 2005—2006

Maternal age group	2001—2006	2005—2006
< 20	11,214	3,806
20-24	44,944	15,280
25-29	101,220	33,826
30-34	141,489	49,216
35-39	71,026	27,221
40+	13,768	5,227
Unknown	223	91
Total	384,048	134,667

Appendix D.2 Denominator statistics for maternal country of birth, by confinements, for twenty eight selected defects, 2001—2006, 2005—2006

Country of birth	2001—2006	2005—2006
Australia	291,2199	101,354
Oceania inc NZ	10,297	3,632
UK inc Eire	10,648	3,494
Europe	11,938	3,953
Middle East	8,678	3,007
Asia	37,321	13,674
Nth America	2,332	822
Sth America	2,257	842
Africa	7,687	3,219
Unknown	1,561	670
Total	384,048	134,667

Appendix D.3 Denominator statistics for sex, all pregnancies, for twenty eight selected defects, 2001—2006, 2005—2006

Sex	2001—2006	2005—2006
Male	200,420	70,385
Female	190,169	66,666
Indeterminate	27	4
Unknown	458	153
Total	391,074	137,208

Appendix D.4 Denominator statistics for plurality, all pregnancies, for twenty eight selected Defects, 2001—2006, 2005—2006

Plurality	2001—2006	2005—2006
Singleton	377,163	132,174
Twin	13,505	4,896
Triplet	361	130
Quad	20	8
Unknown	25	0
Total	391,074	137,208