Birth Defects in Victoria 1983–1998

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Abbreviations

a/s	atresia/stenosis
AIHW	Australian Institute of Health and Welfare
anom	anomalies
ASD	atrial septal defect
BDR	Birth Defects/Congenital Malformations Register
CDH	congenital dislocation of hip
chrom	chromosomal
CI	confidence interval
CMR	Birth Defects/Congenital Malformations Register
cns	central nervous system
COA	coarctation of aorta
desc	described
dev	developmental
df	degrees of freedom
dysgen	dysgenesis
electr	electrolyte
end	endocrine
g	grams
HLHS	hypoplastic left heart syndrome
inc	includes
ICD	International Classification of Diseases
LGA	Local Government Area
MCH Nurse	Maternal & Child Health Nurse
metab	metabolic
MHW	Mercy Hospital for Women
MMC	Monash Medical Centre
nec	not elsewhere classified
NND	neonatal death
NPSU	National Perinatal Statistics Unit
nutri	nutritional
p	probability
PDA	patent ductus arteriosus
PDCU	Perinatal Data Collection Unit
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
tris	trisomy
udt	undescended testicle
unspe	unspecified
VCGS	Victorian Clinical Genetics Service
VSD	ventricular septal defect

Definitions

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 400 grams who, after being born, breathes or shows any evidence of life such as a heartbeat.

Birth defects are structural or anatomical abnormalities that are present at birth, usually resulting from abnormal development in the first trimester of pregnancy.

Birth defect cases refers to the number of liveborn or stillborn infants affected by at least one 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 400 grams.

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 400 grams. who did not, at any time after delivery, breathe or show any evidence of life such as a heartbeat.

Termination of pregnancy in this report, refers to an induced abortion by medical or mechanical means before 20 weeks gestation.

Birth prevalence has as the numerator, the number of still and livebirths at and after 20 weeks gestation. **Overall prevalence** includes terminations of malformations before 20 weeks gestation.

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Highlights

- In the 16 year study period, 1983 to 1998, there have been 32,445 babies born with a birth defect at or after 20 weeks gestation. Another 2,402 were identified as terminations of pregnancy before 20 weeks gestation. This gives an overall prevalence of 344 / 10,000 or 3.4%. From 1995–1998 the overall prevalence was 388 / 10,000 or 3.9%
- The four most common defects for the period 1995–1998 are hypospadias, ventricular septal defect, congenital dislocation of the hip and obstructive defects of the renal pelvis.
- For the first time in 1998 there was a substantial decline in both anencephaly and spina bifida. This may be a chance fluctuation or it may indicate effective nutritional intake of periconceptional folate. The overall prevalence of these birth defects in pregnancy is 1 / 1,724 for anencephaly, 1 / 1,190 for spina bifida or 1 / 704 for either neural tube defect. There are now less than 15 babies per year liveborn with spina bifida.
- Advanced maternal age is significantly associated with hydrocephalus, exomphalos, Down syndrome and other chromosome abnormalities.
- Teenage mothers have a significantly increased risk of having babies with spina bifida, hydrocephalus and gastroschisis.
- There are some significant gender differences in birth prevalence with males at higher risk compared with females. Birth defects significantly associated with males include cardiac (transposition of great vessels, Tetralogy of Fallot, coarctation of aorta) or renal (anorectal atresia, renal agenesis / dysgenesis, obstructive defects of renal pelvis). Males are also at increased risk of cleft lip and palate and Down syndrome, while females have an increased risk for anencephaly, cleft palate and congenital dislocation of the hip.
- Congenital dislocation of the hip is a very common birth defect, reported in 28.6 / 10,000 births or 1 / 350 births (1983–1998), with female babies having a fourfold increased risk for this condition over male babies.
- The number of cases reported with obstructive defects of the renal pelvis continues to increase annually. This is due to increased reporting secondary to ultrasound detection in pregnancy.
- The number of babies living beyond 28 days with hypoplastic left heart and diaphragmatic hernia has not increased. There has been an improved survival rate for babies with renal agenesis / dysgenesis with 55% between 1995–1998 surviving beyond 28 days compared with only 33% in 1983–1994.
- The overall prevalence of Down syndrome has increased, due to there being a larger number of older women becoming pregnant. At the same time, the proportion of fetuses with Down syndrome that are detected in pregnancy has markedly increased. There are now approximately 65 liveborn babies with Down syndrome each year in Victoria.
- The proportion of all babies with Down syndrome born to women less than 35 years has decreased so that in 1995–1998 it was 44%, not over 60% as in earlier years. This shift in proportions is probably due to affected fetuses, that would once have spontaneously aborted and never been identified as affected, now being identified by prenatal testing in older women in the first and early second trimester.
- A number of birth defects are associated with a high frequency of chromosome abnormalities: e.g. Tetralogy of Fallot (14%) and exomphalos (26%). Overall , 6.7% of birth defects have a chromosomal abnormality which represents 0.4% of all pregnancies.
- The prevalence of having a baby affected by a birth defect increases with increasing age. Women aged 35 years or more, 11.7% of all mothers for the period 1983–1998, comprise 15% of all mothers who have a pregnancy affected by a birth defect for this same period. They have a relative risk of having a baby with a birth defect of 1.34 (1.3–1.38) compared with younger women. If the chromosomal abnormalities—Down Sydrome, Patau Syndrome and Edward Syndrome—are excluded, the relative risk for women aged 35 years or more having a baby with a birth defect decreases to 1.13 (1.10–1.17).
- Women from the Middle East are significantly more likely to have a baby with a birth defect than women from any of the other country of birth groups. This is not related to advanced maternal age and may reflect genetic factors.

- Women from Rural Regions (grouped) have a statistically significant decreased likelihood of having a baby with a birth defect when compared to Metropolitan Regions (grouped) (RR 0.9, 96% CI 0.88–0.92, $p<0.0001$). This is a small decrease and may relate to ascertainment issues as much as a biological risk, such as the fact that they are generally younger than Metropolitan mothers.
- The prevalence of birth defects in pregnancies of women identified as Koori in the Perinatal Morbidity Statistics System is significantly less than for those identified as non-Koori (RR 0.77, 95% CI 0.66–0.91, $p=0.001$).
- Babies from multiple births are more likely to have a birth defect than babies from singleton births (RR 1.55, 98% CI 1.48–1.64, $p<0.0001$).

1. Introduction

1.1 Background

With the decline of mortality and morbidity in children from other causes (eg infection), birth defects are now more prominent as a serious health hazard of childhood. Almost 4% of Victorian babies are born each year with a malformation and they contribute substantially to the amount of perinatal mortality and morbidity.

In 1979 the Commonwealth Government agreed in principle to collect more information about births and malformations. It was decided that the States would be responsible for setting up their own systems and the Commonwealth would establish a National Perinatal Statistics Unit, to collate information from all the states and provide an overall picture. The Victorian Perinatal Data Collection Unit (PDCU) was established under the Health Act of 1982, and operates under the aegis of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (the Council). Under this same Health Act, 1982, one of the fundamental purposes for the establishment of the PDCU was the formation of a Birth Defects / Congenital Malformations Register (BDR).

This report presents a 16 year overview of key maternal and infant factors associated with birth defects. In many instances the data for the past 4 years are presented separately to highlight changes that have occurred over time. This report provides supplementary information to our previous report, *Congenital Malformations in Victoria 1983–1994 (1)*. Differences in prevalence data for some birth defects are often influenced by the inclusion of pregnancy termination data in this most recent report.

1.2 Purposes

The Victorian Birth Defects / Congenital Malformations Register is a state-wide population based surveillance system established 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 for birth defects,
5. respond to community concerns about perceived clusters or changes in frequency of birth defects.

The BDR was established in 1984 and collects information on all infants born in Victoria since 1982.

1.3 Inclusion Criteria

Notifiable malformations are defined, for Victorian data purposes, as structural defects or chromosomal abnormalities present at birth. We also collect information on inborn errors in metabolism, haematological disorders, congenital infections, neoplasms, and developmental delay. There are certain isolated minor malformations (such as inguinal hernia, and hydrocele) that are not notifiable and these are listed in Appendix. A.

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

For the purposes of this report, information is included on all cases notified to the BDR **by December 31, 1999**. However, certain isolated minor defects, such as undescended testes (>=37 weeks gestation) and vesico-ureteric reflux, which are reported in the BDR, have been excluded

from this report. This is consistent with the reporting of birth defects in Congenital Malformations in Australia (2), but differs from reports from other state birth defect registries, where higher birth defect prevalence rates include such minor malformations.

Differences with the Annual Report (s) of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (3) and Congenital Malformations in Victoria, 1983–1994 (1)

Many differences in prevalence data exists between this current report and the Annual Report(s) of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity and *Congenital Malformations in Victoria, 1983–1994 (1)*. This can primarily be attributed to the time period being reported in each report. The BDR continues to include data on children who are reported with a birth defect up until the age of 15 years. Therefore new cases are being added (or sometimes deleted) each year. Each report is current at the time of publication, but is never "complete".

There number of birth defects included in this report is approximately 300 less than that reported in the 1998 Council Annual Report (3) due to the exclusion of all cases with isolated undescended testes greater than 36 weeks gestation and vesicoureteric reflux.

Differences with AIHW NPSU Report , Congenital Malformations Australia (2)

There are considerable differences between the Victorian data contained in the Australian Institute of Health & Welfare (AIHW) National Perinatal Statistics Unit (NPSU) Report and the data presented here. These differences reflect both different inclusion criteria and variations in coding practice. We believe this report more accurately reflects the birth prevalence of birth defects in the State of Victoria.

1.4 Sources of Notification

The BDR is a voluntary notification system. Data are obtained from multiple sources. Table 1.1 details the number of notifications of birth defect cases for 1989–1998 births. The number of notifications exceeds the number of cases due to multiple reporting of cases from different sources.

Data on sources of notification have been maintained only since 1989. For the period 1989–1998 there were 40,524 notifications for a total of 24,316 cases. This approximates to 1.7 notifications per case.

Table 1.1 Sources of Notification, 1989–1998

Notification Source	Number	Percent(%)
PDCU—perinatal forms	16,391	40.4
Perinatal Death Certificate	1,968	4.9
Autopsy Report	1,266	3.1
Cytogenetics Report	2,369	5.8
Maternal and Child Health Nurse	3,584	8.8
Hospital listings	14,593	36.0
Other Professionals	290	0.7
Other (e.g. parent)	23	0.1
Unknown	40	0.1
Total	**40,524**	**100.0**

The Annual Report of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity tabulates these figures and has recorded the changing proportion of notifications from these sources over the years. The latest Report (3) demonstrates that 58% of notifications were from the perinatal form, completed by midwives for every baby born in Victoria and sent to the PDCU.

1.5 Data Items

The BDR collects maternal, infant and fetal demographic data on all cases. All notifications (excluding terminations of pregnancy pre 20 weeks and interstate births) are linked to the Perinatal Morbidity Statistics Form to obtain a complete obstetric history for each case. The data items routinely maintained on the BDR are listed in Appendix B. Further data items are available for each case of 20 weeks or more from the PDCU where all the information from the Perinatal Morbidity Statistics Form is recorded.

1.6 Data Quality

Because of the voluntary nature of the BDR, it has been necessary to assess data quality by validation studies (4,5). The first, conducted in 1986, discovered that the proportion of birth defects notified to the BDR improved from 35% to 48% during the period 1982–1985. The second study, conducted in 1993, also reported a marked improvement in notification rates from 50% to 86% during the period 1989–1992.

Since 1992 considerable time and effort has been expended in improving the quality of the data in the BDR. The Register 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 RCH with a birth defect. We have also obtained listings of all children born since 1982 who have visited the RCH Cardiology Unit and the Victorian Clinical Genetics Service (VCGS) Metabolic Clinic, either as an inpatient or outpatient. This procedure has also been adopted for Monash Medical Centre where the inpatient listings date back to 1992.

1.7 Explanatory Notes

Cases verses Defects

Section 2 describes each *individual malformation*. Sections 4 & 5 describe birth defect cases—the number of liveborn or stillborn infants affected by at least one birth defect. Thus, the number of birth defects exceeds the number of cases.

Classification of Diseases

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

With *Syndromes* it is the policy of the PDCU to code the syndrome **and** all of its manifestations.

Births and Confinements

There is an important difference between the number of births and the number of confinements (sometimes referred to as 'maternities'). 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. This differs from the previous report (1) where only births were used as the denominator for both infant and maternal characteristics.

Maternal Country of Birth

For the purposes of this report we have separated women born in Asia from those born in the Middle East. In the previous report (1) these women were combined in one category called "Asia including Middle East".

1.8 Terminations of Pregnancy

In this report termination of pregnancy refers to an induced abortion by medical or surgical means before 20 weeks gestation. If there is an induced abortion at 20 weeks or more, or if the gestation is unknown and the birthweight is 400 grams or more, it is required to be registered as a birth and is classified as a stillbirth by the PDCU and the BDR.

Notification of terminations for malformations only began in 1986. While the capture of these data in the early years was very incomplete, each subsequent year has shown a marked improvement in ascertainment of cases. There has also been an increase in the number of terminations because of the increased use of prenatal diagnosis.

Since 1986 a PDCU staff member has been responsible for reviewing the medical records of all patients with a termination for malformations at both Monash Medical Centre and the Royal Women's Hospital. A letter is sent to all other hospitals with maternity services to obtain information on patients who have had a termination for a malformation This procedure has an ICD code which enables extraction of these specific records.

This report differs from the previous report (1), because data on terminations for malformations have been included for all variables except birthweight. This was felt to be appropriate due to the improved data quality, and the importance of providing a complete picture of the prevalence of certain congenital defects where a high proportion of affected pregnancies are terminated.

1.9 Statistics

For each rate figure , a 95% Poisson confidence interval (if number of cases < 400) or a 95% Binomial confidence interval (if number of cases >= 400) has been calculated. 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 ($p < 0.05$). P values have not been estimated. Relative risks in sections 4 and 5 have been calculated using Epi Info Version 6.1.

1.10 Regional Variations

Victoria is divided into a number of Health Regions for planning and policy purposes. Between 1983–1998 changes have occurred in regional boundaries three times. Therefore, to provide regional trend data over this time necessitates allocation of certain postcodes to specific regions where regional and / or local government area (LGA) boundaries have changed.

For the purposes of this report, all postcodes from 1983 to 1998 have been assigned to current (1998) regional boundaries. This changes the proportion of births in each Region from the 1992–1995 boundaries in the following way: Barwon nil, Grampians +0.5%, Loddon Mallee +0.5%, Hume -0.2%, Gippsland nil, Western Metro -2.3%, Northern Metro + 1.5%, Eastern Metro +1.1%, Southern Metro -1.1%.

An example of how the re-distribution of postcodes can affect the proportion of cases in each Region between 1992–1995 and 1998, is tabulated below using spina bifida as an example.

Table 1.2: An Example: a Proportion of Spina Bifida Cases in Health Regions, 1992–1995 and 1998

Region	1992–1995 (%)	1998(%)
Barwon S W	6.0	5.7
Grampians	3.6	3.5
Loddon Mallee	6.6	7.0
Hume	5.6	5.5
Gippsland	6.6	6.8
Western Metro	17.1	15.2
Northern Metro	15.6	17.0
Eastern Metro	14.3	14.8
Southern Metro	23.0	22.7
Other	1.6	1.6

It should also be noted that the reassignment of postcodes from 1983–1996 to the current regional boundaries is only an approximation. In reality some postcodes are divided between two or more Regions in proportions ranging from 10% to 99%. In this report, postcodes between 1983–1996 have been allocated exclusively to one Region (see Appendix D.) Postcodes for 1997 and 1998 are correctly proportionately assigned to Regions as this is achieved during the creation of the Perinatal Morbidity Statistics file. *Any conclusions drawn from differences in the prevalence of malformations between Regions should be made with due consideration of the Regional boundary changes and allocation of postcodes.*

2. Summary Tables

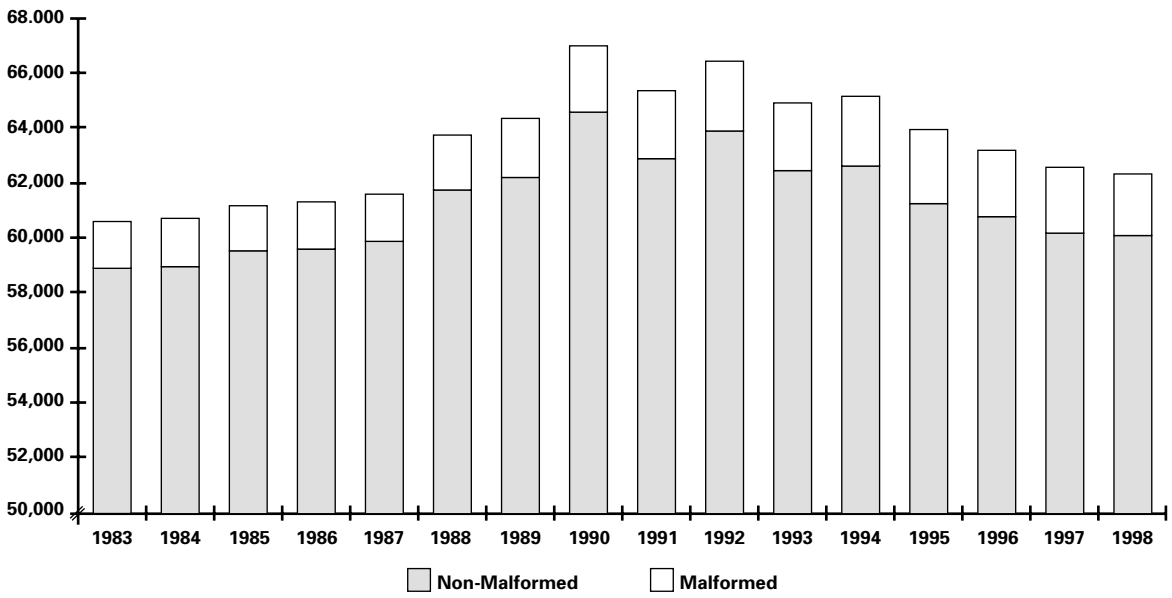
2.1 Birth Defects by Year, 1983–1998

The combined birth prevalence of birth defects over 16 years is 344.2 / 10,000 or 3.4%. The increase 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. From 1995–1998 the overall prevalence was 388 / 10,000 or 3.8%

Table 2.1 Birth Defects by Year, 1983–1998

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	1,700	2	280.7	2.8
1984	60,737	1,746	11	289.3	2.9
1985	61,189	1,622	18	268.0	2.7
1986	61,253	1,634	83	280.3	2.8
1987	61,566	1,657	60	279.7	2.8
1988	63,666	1,889	109	313.8	3.1
1989	64,255	2,023	126	334.4	3.3
1990	66,878	2,269	143	360.7	3.6
1991	65,248	2,332	142	379.2	3.8
1992	66,305	2,382	157	382.9	3.8
1993	64,737	2,255	207	380.3	3.8
1994	64,932	2,284	253	390.7	3.9
1995	63,717	2,436	259	423.0	4.2
1996	62,951	2,145	269	383.5	3.8
1997	62,308	2,100	294	384.2	3.8
1998	62,091	1,971	269	360.8	3.6
Total	**1,012,461**	**32,445**	**2,402**	**344.2**	**3.4**

Figure 2.1 Number of Birth Defects as a Proportion of Total Births, 1983–1998



2.2 Comparison of Prevalence of Birth Defects, 1983–1994 and 1995–1998

The following table shows changes in the prevalence of birth defects since the production of the previous report (1).

Table 2.2 Comparison of Prevalence of Birth Defects, 1983–1994 and 1995–1998

Defect	Number (1983–1994)	Prevalence/ 10,000	Number (1995–1998)	Prevalence/ 10,000
Total pregnancies (births + TOPs)	*762,705*		*252,158*	
Anencepahly	401	5.3	185	7.3
Spina Bifida	649	8.5	205	8.1
Encephalocele	149	2.0	38	1.5
Microcephalus	241	3.2	69	2.7
Hydrocephalus	497	6.5	224	8.9
Transposition of Great Vessels	372	4.9	134	5.3
Tetralogy of Fallot	256	3.4	97	3.8
Ventricular Septal Defect	2,049	26.9	734	29.1
Hypoplastic Left Heart Syndrome	205	2.7	63	2.5
Coarctation of Aorta	447	5.9	101	4.0
Cleft Palate	556	7.3	196	7.8
Cleft Lip	298	3.9	83	3.3
Cleft Lip and Palate	504	6.6	157	6.2
Oesophageal Atresia and/or Stenosis	274	3.6	103	4.1
Anorectal Atresia and/or Stenosis	292	3.8	135	5.4
Hypospadias	1,848	24.2	809	32.1
Renal Agenesis and Dysgenesis	368	4.8	165	6.5
Cystic Kidney Disease	302	4.0	167	6.6
Obstructive Defects of Renal Pelvis	804	10.5	705	28.0
Congenital Dislocation of Hip	2,245	29.4	657	26.1
Limb Reduction Defects	461	6.0	181	7.2
Diaphragmatic Hernia	253	3.3	104	4.1
Exomphalos	218	2.9	97	3.8
Gastroschisis	100	1.3	65	2.6
Trisomy 21	1,195	15.7	525	20.8
Trisomy 13	108	1.4	62	2.5
Trisomy 18	266	3.5	156	6.2

2.3 Order of Prevalence of Twenty Seven Selected Birth Defects, 1995–1998

The following table shows the order of prevalence of twenty-seven selected defects presented in section three of this report for 1995–1998. These are the defects adopted by the International Clearinghouse for Birth Defects Monitoring System and are those reported by the National Perinatal Statistics Unit (2). The three most common defects for this period were hypospadias, ventricular septal defect and obstructive defects of the renal pelvis.

Table 2.3 Order of Prevalence of Twenty Seven Selected Birth Defects, 1995–1998

Anomaly	N/10,000	1 in x Number of Births
Hypospadias	32.1	312
Ventricular Septal Defect	29.1	344
Obstructive Defects of the Renal Pelvis	28.0	357
Congenital Dislocation of Hip	26.1	383
Trisomy 21	20.8	481
Hydrocephalus	8.9	1,124
Spina Bifida	8.1	1,235
Cleft Palate	7.8	1,282
Anencephaly	7.3	1,370
Limb Reduction Defects	7.2	1,389
Cystic Kidney	6.6	1,515
Renal Agenesis and Dysgenesis	6.5	1,538
Cleft Lip and Palate	6.2	1,613
Trisomy 18	6.2	1,613
Anorectal Atresia and/or Stenosis	5.4	1,852
Transposition of Great Vessels	5.3	1,887
Oesophageal Atresia and/or Stenosis	4.1	2,439
Diaphragmatic Hernia	4.1	2,439
Coarctation of Aorta	4.0	2,500
Tetralogy of Fallot	3.8	2,632
Exomphalos	3.8	2,632
Cleft Lip	3.3	3,030
Microcephalus	2.7	3,704
Gastroschisis	2.6	3,846
Hypoplastic Left Heart Syndrome	2.5	4,000
Trisomy 13	2.5	4,000
Encephalocele	1.5	6,667

2.4 Summary of Patterns of Birth Defects, 1983–1998

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

Table 2.4 Summary of Patterns of Birth Defects, 1983–1998

Anomaly	Total	% Isolated	% Chromosomal	% Other (Multiple Same System & Multiple Different Systems, Multiple NTD)
Anencephaly	586	72.7	1.2	26.1
Spina Bifida	854	63.2	3.5	33.3
Encephalocele	187	45.5	2.7	51.0
Microcephalus	310	29.4	14.2	56.4
Hydrocephalus	721	42.7	12.2	45.1
Transposition of Great Vessels	506	9.1	5.5	85.4
Tetralogy of Fallot	353	21.3	13.9	65.0
Ventricular Septal Defect	2,783	37.4	14.7	47.9
Hypoplastic Left Heart Syndrome	268	36.6	10.8	52.6
Coarctation of Aorta	548	17.9	8.2	74.0
Cleft Palate	748	59.0	9.0	32.0
Cleft Lip	380	85.0	3.4	11.6
Cleft Lip & Palate	662	68.1	11.3	20.5
Oesophageal Atresia and/or Stenosis	377	35.8	11.9	52.3
Anorectal Atresia and/or Stenosis	427	31.6	7.3	61.2
Hypospadias	2,657	86.4	1.4	12.3
Renal Agenesis/Dysgenesis	533	31.1	6.0	16.9
Cystic Kidney	469	38.2	7.5	54.3
Obstructive Defects Renal Pelvis	1,545	61.5	5.1	33.4
Congenital Dislocation of Hip	2,902	92.5	0.8	6.7
Limb Reduction Defects	642	40.8	8.7	50.4
Diaphragmatic Hernia	357	48.5	9.0	42.6
Exomphalos	315	27.0	25.7	47.3
Gastroschisis	165	67.3	1.8	30.4

2.5 Birth Defects by Major Anatomical System, Victoria, 1983–1998

The following figures refer to individual birth defects, not cases. The order of the birth defects reflects that determined by the coding system.

Table 2.5 Birth Defects by Major Anatomical Site, 1983–1998

Codes	Defects	Number	% of All Birth Defects	N/10,000 Pregnancies
Total		**55,743**		
740-2	Nervous System	4,519	8.1	44.5
743	Eye	814	1.5	8.0
744	Ear, Face & Neck	1,081	1.9	10.7
745-6	Heart	9,595	17.2	94.5
747	Circulatory System	4,348	7.8	42.8
748	Respiratory System	1,584	2.8	15.6
749	Cleft Palate/Lip	1,810	3.2	17.8
750-1	Digestive System	3,475	6.2	34.2
752	Genital Organs	3,992	7.2	39.3
753	Urinary System	3,928	7.0	38.7
755	Limbs	3,711	6.7	36.6
754/6	Other Musculoskeletal	8,563	15.4	84.4
757	Integument	467	0.8	4.6
758	Chromosomal	3,746	6.7	36.9
759	Other & Unspecified	1,289	2.3	12.7
760	Maternal Conditions	9	0.0	0.1
7710-2	Congenital Infections	87	0.2	0.9
140-239	Neoplasms	448	0.8	4.4
240-279	Endocrine/Nutritional/Metabolic	1,375	2.5	13.5
280-289	Disease of Blood	284	0.5	2.8
315/8	Devolopmental Delay	222	0.4	2.2
7780	Hydrops	326	0.6	3.2
7786*	Hydrocele	70	0.1	0.7

* Hydrocele is a minor condition that was collected in the BDR until approximately 1994–1995. All isolated cases have been excluded from this report. This figure refers to cases of hydrocoele that are associated with other malformations

2.6 Birth Defects by Four Digit Code, 1983–1998

The following figures refer to individual birth defects, not cases.

Table 2.6 Birth Defects by Four Digit Code, 1983–1998

Codes	Defects	Number	N/10,000 Pregnancies
740*	**Anencephalus & Similar Anomalies**	**647**	**6.4**
7400#	Anencephalus	607	6.0
7401	Craniorachischisis	29	0.3
7402	Iniencephaly	11	0.1
741*	**Spina Bifida**	**947**	**9.3**
742	**Other Nervous System (NS)**	**2,925**	**28.8**
7420	Encephalocele	187	1.8
7421	Microcephalus	310	3.1
7422	Brain Reduction	458	4.5
7423*	Hydrocephalus	728	7.2
7424-9	Other	450	4.4
330-7	Hereditary & Degenerative Diseases of Central NS	105	1.0
343-9	Cerebral Palsy	625	6.2
340-2/3499	Other Disorders of CNS	11	0.1
350-9	Disorders of Peripheral N S	51	0.5
743	**Eye**	**814**	**8.0**
7430	Anophthalmos	20	0.2
7431	Microphthalmos	94	0.9
7432	Buphthalmos	49	0.5
74330-1/3-9	Other Lens	16	0.2
74332	Cataract	169	1.7
7434-9	Other	466	4.6
744	**Ear, Face & Neck**	**1,201**	**11.8**
7440	Ear-Affecting Hearing	120	1.2
74400	*Auditory Canal*	*66*	*0.7*
74401	*Absent Auricle*	*34*	*0.3*
74402-9	*Other*	*20*	*0.2*
7441-3	Other Ear	735	7.2
7444-9	Face & Neck	226	
745	**Bulbus Cordis & Cardiac Septal Closure**	**6,021**	**59.3**
7450	Common Truncus	101	1.0
7451*	Transposition of Great Vessels	543	5.4
7452	Tetralogy of Fallot	363	3.6
7453	Common Ventricle	103	1.0
7454*	Ventricular Septal Defect	2,785	27.4
7455	Atrial Septal Defect	1,624	16.0
7456	Endocardial Cushion	483	4.8
7457-9	Other	19	0.2

* It is possible for one case to have two or more conditions within this code range. Therefore the number of birth defects may exceed the number of cases.

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

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Table 2.6 Birth Defects by Four Digit Code, 1983–1998 (cont.)

Codes	Defects	Number	N/10,000 Pregnancies
746	**Other Heart**	**3,574**	**35.2**
425	Cardiomyopathy	136	1.3
426	Conduction Disorder	27	0.3
7460	Pulmonary Valve	927	9.1
74600	*Atresia*	*163*	*1.6*
74601	*Stenosis*	*651*	*6.4*
74602-9	*Other*	*113*	*1.1*
7461	Tricuspid Atresia/Stenosis	317	3.1
7462	Ebstein Anomaly	51	0.5
7463-4	Aortic Valve Stenosis/Insufficiency	504	5.0
7465-6	Mitral Stenosis/Insufficiency	335	3.3
7467	Hypoplastic Left Heart Syndrome	268	2.6
7468	Other Specified	887	8.7
7469	Unspecified	122	1.2
747	**Circulatory**	**4,348**	**42.8**
7470	Patent Ductus Arteriosus	2,015	19.9
7471	Coarctation of Aorta	553	5.4
7472	Other Aorta	443	4.4
7473	Pulmonary Artery	409	4.0
7474	Great Veins	338	3.3
7475	Single Umbilical Artery	130	1.3
7476	Peripheral Vascular	76	0.7
7478	Other Specified	234	2.3
7479	Unspecified	150	1.5
748	**Respiratory**	**1,584**	**15.6**
7480	Choanal Atresia	193	1.9
7481	Other Nose	95	0.9
7482-3	Larynx/Trachea/Bronchus	347	3.4
7484-6	Lung	925	9.1
7488-9	Other Respiratory	24	0.2
749*	**Cleft Palate/Lip**	**1,810**	**17.8**
7490	Cleft Palate	768	7.6
7491	Cleft Lip	381	3.8
7492	Cleft Lip & Palate	661	6.5
750	**Upper Alimentary Tract**	**1,253**	**12.3**
7503	Oesophageal Atresia/Stenosis with/without fistula	378	3.7
7501-2/4-9	Other	875	8.6
751	**Other Digestive**	**2,222**	**21.9**
7510	Meckels Diverticulum	83	0.8
7511	Small Intestine Atresia/Stenosis	263	2.6
75110	*Duodenum Atresia/Stenosis*	*146*	*1.4*
75111	*Jejeunum Atresia/Stenosis*	*38*	*0.4*
75112	*Ileum Atresia/Stenosis*	*31*	*0.3*
75119	*Unspecified Atresia/Stenosis*	*48*	*0.5*
7512	Large Intestine Rectum Anal Atresia/Stenosis	454	4.5
75120	*Large Intestine Atresia/Stenosis*	*36*	*0.4*
75121-2	*Rectum Atresia/Stenosis*	*48*	*0.5*
75123-4	*Anus Atresia/Stenosis*	*370*	*3.6*

* It is possible for one case to have two or more conditions within this code range. Therefore the number of birth defects may exceed the number of cases.

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Table 2.6 Birth Defects by Four Digit Code, 1983–1998 (cont.)

Codes	Defects	Number	N/10,000 Pregnancies
7513	Hirschsprungs	239	2.4
7514	Intestinal Fixation	208	2.0
7515-9	Other Digestive	511	5.0
524-579	Other	464	4.6
752	**Genital Organs**	**3,992**	**39.3**
7520-1	Ovaries/Fallopian etc	54	0.5
7522-3	Uterus	62	0.6
7524	Cervix/Vagina/ External Genitalia	175	1.7
7525*	Undescended Testes	320	3.2
75260/3-5	Hypospadias	2,657	26.2
75261	Epispadias	34	0.3
75262	Chordee	138	1.4
7527	Indeterminate Sex	229	2.3
7528	Other Specified	312	3.1
7529	Unspecified	11	0.1
753	**Urinary**	**3,928**	**38.7**
7530	Renal Agenesis/Dysgenesis	537	5.3
75300	*Bilateral*	*293*	*2.9*
75301	*Unilateral*	*244*	*2.4*
7531#	Cystic Kidney Disease	476	4.7
75311-3	*Polycystic*	*154*	*1.5*
75316	*Multicystic*	*242*	*2.4*
75310/4/8	*Other*	*80*	*0.8*
7532#	Obstructive Defects Renal Pelvis/Ureter	2,093	20.6
75320	Hydronephrosis	1,194	11.8
75321-9	*Other*	*507*	*5.0*
75330/1/3-9	*Other Specified Kidney Disorders*	*243*	*2.4*
75332	*Horseshoe Kidney*	*149*	*1.5*
7534	Other Specified Disorders of Ureter	352	3.5
7535	Exstrophy of Bladder	39	0.4
7536	Urethra Bladder Neck Atresia/Stenosis	162	1.6
7537	Urachus	18	0.2
7538	Other Bladder/Urethra	172	1.7
7539	Unspecified	27	0.3
592-608	Other	52	0.5
754	**Certain Musculoskeletal**	**6,036**	**59.5**
7540	Of Skull, Face & Jaw	230	2.3
7541	Of Sternocleidomastoid	55	0.5
7542	Of Spine	103	1.0
75430	Congenital Dislocation of Hip	2,902	28.6
75431-2	Other Hip	137	1.3
7544	Genu Recurvatum/Bowing	59	0.6
7545-7	Of Feet	2477	24.4
7548	Other	73	0.7

* Undescended testes in term babies is included in BDR, 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 malformations.

\# It is possible for one case to have two or more conditions within this code range. Therefore the number of birth defects may exceed the number of cases.

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Table 2.6 Birth Defects by Four Digit Code, 1983–1998 (cont.)

Codes	Defects	Number	N/10,000 Pregnancies
755*	**Limbs**	**3,711**	**36.6**
7550	Polydactyly	1,001	9.9
7551	Syndactyly	801	7.9
7552	Reduction Deformities Upper Limb	480	4.7
7553	Reduction Deformities Lower Limb	293	2.9
7554	Reduction Deformities Unspecified Limb	44	0.4
7555	Other Upper Limb	493	4.9
7556	Other Lower Limb	468	4.6
75580	Arthropgryposis Multiplex Congenita	82	0.8
75581-8	Other Specified	85	0.8
7559	Unspecified	12	0.1
756	**Other Musculoskeletal**	**2,527**	**24.9**
7560	Skull, Face & Bones	681	6.7
75600	*Craniosynostosis*	*221*	*2.2*
75601-2/4-9	*Other*	*345*	*3.4*
75603	*Pierre Robin Syndrome*	*115*	*1.1*
7561	Spine	316	3.1
7562-3	Ribs & Sternum	138	1.4
7564	Chondrodystrophy	143	1.4
75643	*Achondroplasia*	*37*	*0.4*
75644	*Other Dwarfing*	*33*	*0.3*
75640-2/5-9	*Other*	*73*	*0.7*
7565	Osteodystrophies	122	1.2
75650	*Osteogenesis*	*75*	*0.7*
75651-9	*Other*	*47*	*0.5*
7566	Diaphragm	444	4.4
75660/2-9	*Other*	*87*	*0.9*
75661	*Diaphragmatic Hernia*	*357*	*3.5*
7567	Abdominal Wall	549	5.4
75670	*Exomphalos*	*315*	*3.1*
75671	*Gastroschisis*	*165*	*1.6*
75672-9	*Other*	*69*	*0.7*
7568	Other Specified of Muscle/ Tendon/ Fasica/ Connective Tissue	116	1.1
7569	Unspecified	18	0.2
757	**Integument**	**467**	**4.6**
7570	Hereditary Oedema of Legs	5	0.0
7571	Icthyosis Congenita	32	0.3
7572	Dermatoglyphic Anomalies	41	0.4
7573	Other Specified Anomalies Skin	342	3.4
7574	Specified Anomalies of Hair	5	0.0
7575	Specified Anomalies of Nails	10	0.1
7576	Specified Anomalies of Breast	18	0.2
7578	Other Specified Anomalies of Integument	8	0.1
7579	Unspecified	6	0.1

It is possible for one case to have two or more conditions within this code range. Therefore the number of birth defects may exceed the number of cases.

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Table 2.6 Birth Defects by Four Digit Code, 1983–1998 (cont.)

Codes	Defects	Number	N/10,000 Pregnancies
758	**Chromosomal**	**3,746**	**36.9**
7580	Trisomy 21	1,720	16.9
7581	Trisomy 13	171	1.7
7582	Trisomy 18	423	4.2
7583	Autosomal Deletion	209	2.1
7584	Balanced Autosomal Translocation	129	1.3
7585	Other Autosomal	561	5.5
7586	Turner	213	2.1
7587	Klinefelter	77	0.8
7588	Other Sex Chromosomes	189	1.9
7589	Unspecified	54	0.5
759	**Other**	**1,289**	**12.7**
7590	Spleen	125	1.2
7591	Adrenal Gland	137	1.3
7592	Other Endocrine Gland	174	1.7
7593	Situs Inversus	73	0.7
7594	Conjoined Twins	25	0.2
7595	Tuberous Sclerosus	46	0.5
7596	Harmartoses NEC	30	0.3
7597	Multiple So Described	64	0.6
7598	Other Specified	559	5.5
7599	Unspecified	56	0.6
	Other	**493**	**4.9**
7602-7611/2	Maternal Conditions	6	0.0
76070	Fetal Hydantoin Syndrome	2	0.0
76076	Fetal Alcohol Syndrome	3	0.0
7710-2	Congenital Infection	87	0.9
7780	Hydrops Fetalis (Non-Immune)	326	3.2
7786	Hydrocele	70	0.7
140-239	**Neoplasms**	**448**	**4.4**
140-208	Malignant	45	0.4
210-229	Benign	327	3.2
22809	*Haemangioma*	*82*	*0.8*
22819	*Cystic Hygroma*	*192*	*1.9*
2100-227/229	*Other*	*53*	*0.5*
235-9	Uncertain Behaviour	76	0.7
240-279	**Endocrine/Nutritional/Metabolic**	**1,375**	**13.5**
2439	Congenital Hypothyroidism	246	2.4
240-2/4-6	Other Thyroid Gland	3	0.0
250-9	Other Endocrine Glands	59	0.6
260-9	Nutritional Deficiencies	6	0.1
2701	Phenylketonuria	72	0.7
2700/2-9	Other Disorders of Amino-Acid Metabolism	84	0.8
271	Of Carbohydrate Metabolism	164	1.6
272	Of Lipid Metabolism	27	0.3
273	Of Plasma Protein Metabolism	38	0.4
275	Of Mineral Metabolism	108	1.1
276	Of Fluid Electrolyte/Acid Base	16	0.2
2770	Cystic Fibrosis	338	3.3
2771-9	Other Metabolism	109	1.1
279	Of Immunity	105	1.0

continued next page

Table 2.6 Birth Defects by Four Digit Code, 1983–1998 (cont.)

Codes	Defects	Number	N/10,000 Pregnancies
280-9	**Diseases of Blood**	**284**	**2.8**
282	Hereditary Haemolytic Anaemia	237	2.3
281/3-5	Other Anaemias	9	0.1
286	Coagulation Defects	25	0.2
287-9	Other	131	1.3
315/8 (also 783-786)	**Developmental Delay/Problems***	**222**	**2.2**

* Very incomplete ascertainment of developmental delay.

2.7 Indications for Terminations of Pregnancy for a Birth Defect Before 20 weeks Gestation, 1989–1998

Between 1989–1998 there were 2,119 terminations of pregnancy for birth defects before 20 weeks reported to the BDR. Of these cases, almost 50% of pregnancies were terminated for a chromosomal anomaly and another 24% for Central Nervous System Defects (mainly Anencephaly or Spina Bifida).

Table 2.7 Indications for Terminations for Pregnancy for a Birth Defect Pre 20 weeks Gestation, 1989–1998

Anomaly	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998	Total
Chromosomal											
Down Syndrome (inc. tris/mosaic/transloc.)	20	27	24	28	40	48	54	51	76	71	439
Patau Syndrome (inc. tris/mosaic/transloc.)	2	2	7	2	12	7	11	8	11	7	69
Edward Syndrome (inc. tris/mosaic/transloc.)	9	7	17	13	14	23	18	28	20	26	175
Conditions due to Other Autosomal Anomalies	6	8	12	12	22	21	14	15	17	15	142
Turner's Syndrome	8	6	8	9	10	11	7	13	12	14	98
Klinefelter Syndrome	0	3	1	1	2	3	4	5	1	8	28
Conditions due to Other Sex Chromosome Anomalies	2	3	2	2	4	4	5	6	4	7	39
Conditions due to Structural Chromosome Anomalies	0	1	0	1	0	0	0	2	4	4	12
											1002
Central Nervous System (CNS)											
Anencephaly	14	20	14	27	14	18	30	28	32	20	217
Spina Bifida with or without Hydrocephalus	13	16	14	9	19	17	22	16	23	13	162
Encephalocele	0	3	1	2	3	3	0	1	4	1	18
Mutiple Neural Tube Defects	2	0	3	2	7	10	5	3	5	1	38
Hydrocephalus	4	7	4	7	4	8	6	3	10	9	62
Other CNS Defects	2	2	0	2	3	2	0	2	1	1	15
											512
Cardiac System											
Fallot's Tetralogy/Pentalogy	0	0	0	1	0	0	0	0	0	2	3
Hypoplastic Left Heart Syndrome	0	0	0	2	0	1	1	0	1	1	6
Multiple Cardiac	2	2	2	0	2	2	5	0	1	3	19
Other Cardiac Conditions (Single)	1	1	0	0	2	0	2	1	0	3	10
Cardiac System NOS	2	0	0	1	2	1	0	1	0	2	9
											57
Respiratory System											
Cystic Lung	0	0	0	0	1	0	1	0	0	2	4
Other Respiratory	1	0	0	0	0	3	0	0	0	0	4
											8

continued next page

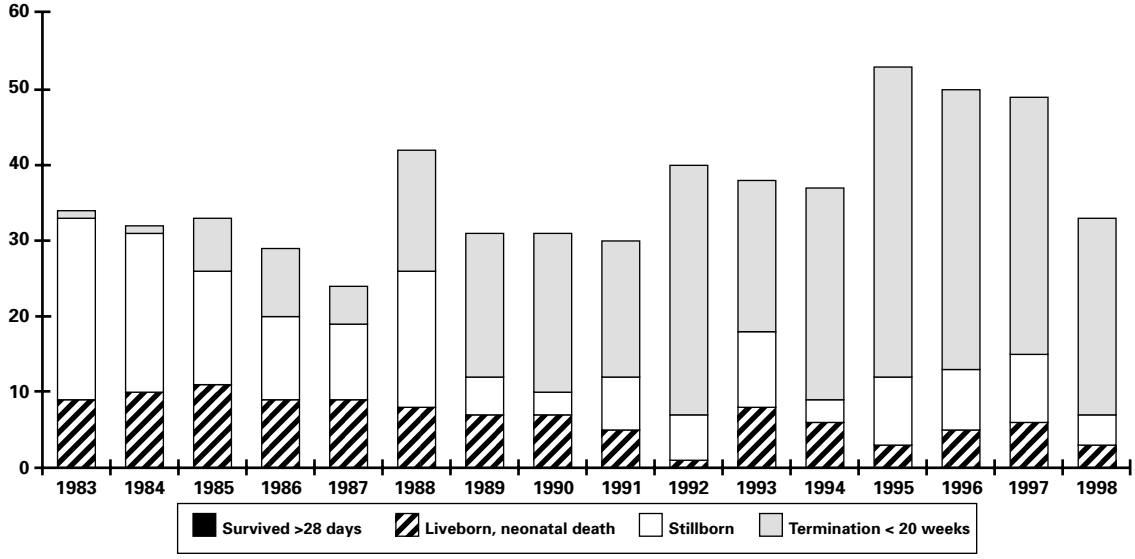
Table 2.7 Indications for Terminations of Pregnancy for a Birth Defect Pre 20 weeks Gestation, 1989–1998 (cont.)

Anomaly	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998	Total
Digestive System	**0**	**1**	**0**	**0**	**0**	**0**	**0**	**1**	**1**	**1**	**4**
Urinary System											
Potter's Syndrome	1	0	2	0	1	5	1	0	1	3	14
Cystic Kidney	1	0	0	4	0	0	0	1	4	1	11
Multiple Anomalies of the Urinary System	0	0	0	0	1	0	3	2	0	0	6
Other Anomalies of the Urinary System	0	1	0	0	0	1	0	1	1	0	4
											35
Musculoskeletal System											
Arthrogryposis	1	0	0	0	0	0	0	0	1	1	3
Chondrodystrophy	1	0	1	2	1	2	6	4	0	0	17
Skeletal Dysplasia (Osteodystrophy)	2	0	0	0	2	1	0	1	0	2	8
Diaphragmatic Hernia	0	1	0	2	1	1	1	0	2	0	8
Exomphalos	2	0	2	0	4	2	0	1	1	0	12
Gastroschisis	0	0	0	0	0	0	1	0	0	1	2
Other Musculoskeletal	1	3	2	0	1	4	1	3	3	4	22
Multiple Musculoskeletal	2	0	0	2	3	2	3	2	3	5	22
Musculoskeletal NOS	2	1	0	0	0	0	0	0	0	0	3
											97
Other Specified Congenital Anomaly	**0**	**0**	**0**	**0**	**0**	**1**	**1**	**0**	**0**	**0**	**2**
Thalassaemia	2	2	3	3	3	8	4	5	3	2	**35**
Haemophilia	0	1	0	0	0	1	0	0	0	0	**2**
Cystic Fibrosis	2	3	1	1	1	1	1	2	2	1	**15**
Cystic Hygroma	0	2	1	0	0	0	5	1	3	1	**13**
Phenylketonuria	1	1	0	0	0	0	0	0	0	0	**2**
Other Disorders of Metabolic & Immune System	0	1	0	0	1	0	1	1	1	0	**5**
Huntington's Disease	0	0	1	0	0	1	1	1	2	0	**6**
Werdnig-Hoffman	0	0	0	1	0	0	0	0	3	0	**4**
Muscular Dystrophy	0	0	0	0	1	0	5	2	0	1	**9**
Hydrops Fetalis	1	0	3	0	1	2	1	4	3	1	**16**
Conjoined Twins	0	1	0	1	0	0	0	0	4	0	**6**
Congenital Infections	0	0	1	0	0	0	0	0	0	0	**1**
Maternal Complications	0	2	0	1	0	0	0	1	0	0	**4**
Other	0	0	0	0	1	1	3	0	0	0	**5**
Multiple System Defects	18	15	16	16	21	35	35	45	29	29	**259**
Congenital Anomalies NOS	1	0	0	3	3	3	1	8	5	6	**30**
Total	**126**	**143**	**142**	**157**	**207**	**253**	**259**	**269**	**294**	**269**	**2119**

3. Major Birth Defects

3.1 Anencephaly

Figure 3.1 Anencephaly, Number of Cases by Year



	1983	1984	1985	1986	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998
Number	34	32	33	29	24	42	31	31	30	40	38	37	53	50	49	33
N/10,000	5.6	5.3	5.4	4.7	3.9	6.6	4.8	4.6	4.6	6.0	5.9	5.7	8.3	7.9	7.9	5.3

Anencephaly

British Paediatric Association code 740.02

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

- By 1989 the number of terminations of pregnancy exceeded the number of stillbirths and neonatal deaths, due to the relative ease with which this malformation is detected *in utero* by ultrasound. Before 1992 information about terminations was not readily available and the numbers presented for the earlier time period are almost certainly an underestimate of the true overall prevalence. Between 1995 and 1998, 75% of fetuses with anencephaly were detected during pregnancy and followed by a termination. `
- The number of births reported to the BDR (including terminations) has shown annual fluctuations with no evidence of a significant decline in the actual number of affected pregnancies. An increase was observed in 1995 and a substantial decline in 1998. This decline (which includes terminations) may reflect greater nutritional intake of periconceptional folate and thus a reduction in incidence of neural tube defects. There was also a decline in apparent incidence of spina bifida in 1998 (see Figure 3.2). These data will be monitored carefully to ensure they do not represent a chance fluctuation.
- Almost two-thirds of babies with anencephaly are female, a statistically significant difference from the sex ratio in all births.
- Five percent of anencephalic babies are one of a twin. A slight decline in twin pregnancies associated with anencephaly was observed in 1995–1998 (4.9% compared with 5.2%). The birth prevalence amongst twins, 11.5 / 10,000, is twice that amongst singletons, 5.6 / 10,000, and this difference is significant.
- The decrease in proportion of babies born with birth weight >2,500 gm in 1995–1998 is due to the increased proportion of pregnancy terminations.
- There are no significant associations of anencephaly with the maternal characteristics, age country of birth and region of residence.

Table 3.1.1 Anencephaly, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**586**		**401**		**185**		**5.8**	***5.31***	***6.24***
Survived > 28 days	1,000,776	*98.6*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Neonatal death	4,288	*0.4*	107	*18.3*	90	*22.4*	17	*9.2*			
Stillbirth	7,397	*0.7*	163	*27.8*	133	*33.2*	30	*16.2*			
Termination < 20 wks	2,402	*0.2*	316	*53.9*	178	*44.4*	138	*74.6*			
Sex											
Male	521,553	*51.4*	199	*34.0*	135	*33.7*	64	*34.6*	3.8	*3.3*	*4.40*
Female	492,558	*48.5*	272	*46.4*	212	*52.9*	60	*32.4*	5.5	*4.9*	*6.23*
Indeterminate	298	*0.0*	25	*4.3*	19	*4.7*	6	*3.2*			
Unknown#	454	*0.0*	90	*15.4*	35	*8.7*	55	*29.7*			
Plurality											
Singleton	987,675	*97.3*	554	*94.5*	379	*94.5*	175	*94.6*	5.6	*5.1*	*6.08*
Twin	26,192	*2.6*	30	*5.1*	21	*5.2*	9	*4.9*	11.5	*7.7*	*16.38*
Triplet	939	*0.1*	2	*0.3*	1	*0.2*	1	*0.5*	21.3	*2.6*	*76.89*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**270**		**223**		**47**		**2.7**		
Birthweight											
< 1,000	6,784	*0.7*	103	*38.1*	76	*34.1*	27	*57.4*	151.8		
1,000–2,499	54,688	*5.4*	120	*44.4*	104	*46.6*	16	*34.0*	21.9		
2,500+	949,958	*93.8*	29	*10.7*	27	*12.1*	2	*4.3*	0.3		
Unknown	1,067	*0.1*	18	*6.7*	16	*7.2*	2	*4.3*	N/A		

95% of unknowns were in terminations of pregnancy before 20 weeks

Table 3.1.2 Patterns of Birth Defects, Anencephaly, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	426	72.7
Other Associations:		
• Chromosomal	7	1.2
• Multiple NTD	46	7.8
• Other Same System (CNS)	1	0.2
• Other Different Systems	106	18.1
Total	**586**	**100.0**

* Isolated cases may include cases with one of the following minor conditions or conditions commonly associated with anencephaly: absence of adrenal gland or adrenal hypoplasia, talipes, undescended testes, misplaced ears. If a case has two or more of these conditions it is classified as a multiple system defect.

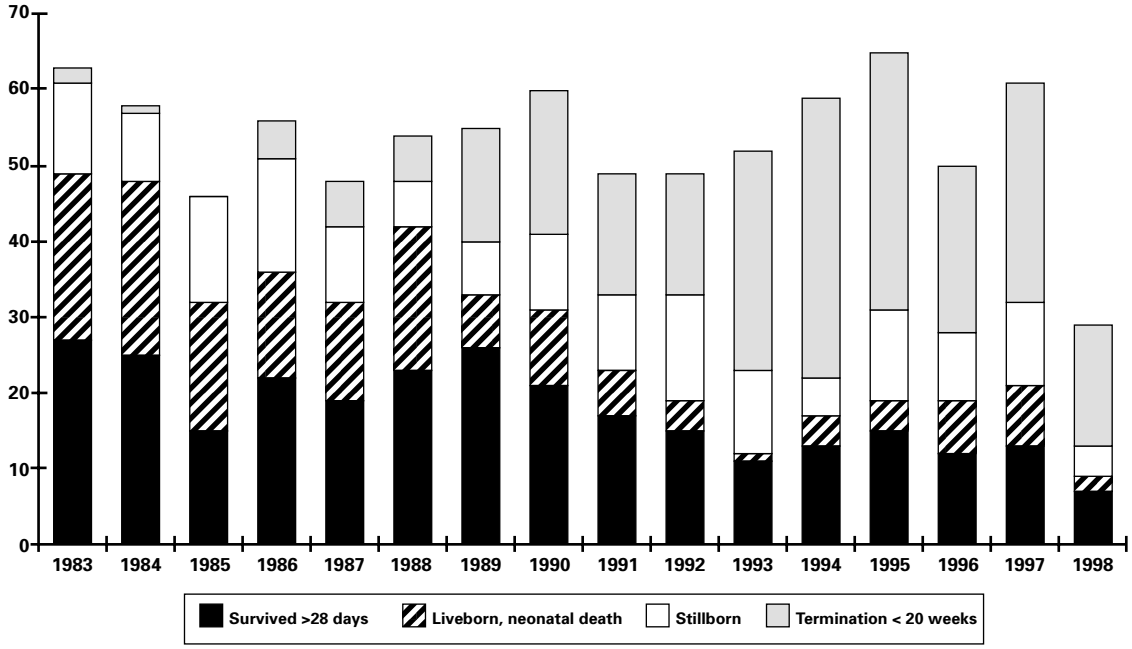
Table 3.1.3 Anencephaly, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**585**		**401**		**184**		**5.8**	***5.40***	***6.29***
Maternal Age											
<20	40,400	*4.0*	21	*3.6*	14	*3.5*	7	*3.8*	5.2	*3.22*	*7.95*
20–24	185,901	*18.6*	106	*18.1*	79	*19.7*	27	*14.7*	5.7	*4.69*	*6.93*
25–29	367,374	*36.7*	213	*36.4*	157	*39.2*	56	*30.4*	5.8	*5.06*	*6.64*
30–34	289,879	*29.0*	150	*25.6*	88	*21.9*	62	*33.7*	5.2	*4.39*	*6.09*
35–39	101,575	*10.1*	60	*10.3*	34	*8.5*	26	*14.1*	5.9	*4.55*	*7.66*
40+	15,718	*1.6*	6	*1.0*	2	*0.5*	4	*2.2*	3.8	*1.40*	*8.32*
Unknown#	273	*0.0*	29	*5.0*	27	*6.7*	2	*1.1*	N/A		
Country of birth											
Australia	753,424	*75.3*	322	*55.0*	194	*48.4*	128	*69.6*	4.3	*3.83*	*4.77*
Oceania inc NZ	20,620	*2.1*	8	*1.4*	6	*1.5*	2	*1.1*	3.9	*1.67*	*7.64*
UK inc Eire	50,596	*5.1*	25	*4.3*	15	*3.7*	10	*5.4*	4.9	*3.20*	*7.31*
Europe	58,931	*5.9*	29	*5.0*	19	*4.7*	10	*5.4*	4.9	*3.30*	*7.09*
Middle East	22,818	*2.3*	9	*1.5*	3	*0.7*	6	*3.3*	3.9	*1.81*	*7.49*
Asia	71,685	*7.2*	38	*6.5*	18	*4.5*	20	*10.9*	5.3	*3.75*	*7.27*
Nth America	5,003	*0.5*	2	*0.3*	0	*0.0*	2	*1.1*	4.0	*0.48*	*14.43*
Sth America	4,689	*0.5*	3	*0.5*	2	*0.5*	1	*0.5*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	5	*0.9*	5	*1.2*	0	*0.0*	4.6	*1.48*	*10.65*
Unknown#	2,950	*0.3*	144	*24.6*	139	*34.7*	5	*2.7*	N/A		
Region											
Barwon S W	70,984	*7.1*	41	*7.0*	23	*5.7*	18	*9.8*		*4.14*	*7.83*
Grampians	44,084	*4.4*	21	*3.6*	14	*3.5*	7	3.8	4.8	*2.95*	*7.29*
Loddon Mallee	63,104	*6.3*	40	*6.8*	29	*7.2*	11	*6.0*	6.3	*4.53*	*8.62*
Hume	51,348	*5.1*	33	*5.6*	23	*5.7*	10	*5.4*	6.4	*4.42*	*9.04*
Gippsland	56,679	*5.7*	31	*5.3*	21	*5.2*	10	*5.4*	5.5	*3.71*	*7.78*
Western Metro	126,711	*12.7*	88	*15.0*	54	*13.5*	34	*18.5*	6.9	*5.60*	*8.60*
Northern Metro	171,545	*17.1*	97	*16.6*	69	*17.2*	28	*15.2*	5.7	*4.61*	*6.93*
Eastern Metro	192,893	*19.3*	106	*18.1*	78	*19.5*	28	*15.2*	5.5	*4.52*	*6.68*
Southern Metro	214,947	*21.5*	121	*20.7*	87	*21.7*	34	*18.5*	5.6	*4.69*	*6.75*
Other	8,777	*0.9*	6	*1.0*	3	*0.7*	3	*1.6*	6.8	*2.51*	*14.90*
Unknown#	48	*0.0*	1	*0.2*	1	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.2 Spina Bifida

Figure 3.2 Spina Bifida, Number of Cases by Year



Number	63	58	46	56	48	54	55	60	49	49	52	59	65	50	61	29
N/10,000	10.4	9.5	7.5	9.1	7.8	8.5	8.6	9.0	7.5	7.4	8.0	9.1	10.2	7.9	9.8	4.7

Spina Bifida

British Paediatric Association code 741.00–741.99

Non-closure of the spine resulting in herniation or exposure of the spinal cord and/or meninges. Hydrocephalus may or may not be present.

- This is a more common neural tube malformation than anencephaly. There have been 854 reported in the 16 year period (8.4 /10,000) compared with 586 anencephaly (5.8 /10,000). This may reflect ascertainment differences in the 1980s when information on pregnancy terminations associated with anencephaly was often missing. In more recent years, the prevalence was similar—7.3/10.000 for anencephaly and 8.1/10,000 for spina bifida.
- Until 1998, there had been no marked change in the annual number of babies with spina bifida, with fluctuations of between 50 to 60 per year (approximately 1/1,200). However, there has been a significant increase in the number that are detected *in utero* and a subsequent decline in the number of livebirths (less than 20 per year). In 1998 there was a substantial drop in prevalence and incidence (as indicated by the decline in terminations of pregnancies). As with anencephaly, this may be a chance fluctuation or it may indicate effective use of periconceptional folate.
- Since 1993 the number of terminations has exceeded the number of births
- Amongst babies with spina bifida, 33% survived the neonatal period, 48% were stillborn or terminations and 19% died within 28 days. The proportion of babies surviving beyond 28 days has decreased from 36% in 1983–1994 to 23% in 1995–1998.
- There is a non-significant excess of females and twin gestations.
- There is an inverse linear trend with maternal age with women in younger age groups at greater risk than older women, and with teenagers having a particularly high risk. However, from 1995–1998, there were fewer births to teenage mothers and more to older mothers than in the earlier years.
- This neural tube defect is not as common in births to Asian-born women and the prevalence is significantly different from that of several other ethnic groups, including Australian-born women.
- There is a significantly lower birth prevalence in Eastern Metro region compared with Gippsland, Western and Southern Metro regions.

Table 3.2.1 Spina Bifida, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**854**		**649**		**205**		**8.4**	***7.85***	***8.98***
Survived > 28 days	1,000,776	*98.6*	281	*32.9*	234	*36.1*	47	*22.9*	2.8		
Neonatal death	4,288	*0.4*	161	*18.9*	140	*21.6*	21	*10.2*			
Stillbirth	7,397	*0.7*	159	*18.6*	123	*19.0*	36	*17.6*			
Termination < 20 wks	2,402	*0.2*	253	*29.6*	152	*23.4*	101	*49.3*			
Sex											
Male	521,553	*51.4*	371	*43.4*	285	*43.9*	86	*42.0*	7.1	*6.4*	*7.88*
Female	492,558	*48.5*	409	*47.9*	315	*48.5*	94	*45.9*	8.3	*7.5*	*9.11*
Indeterminate	298	*0.0*	28	*3.3*	24	*3.7*	4	*2.0*			
Unknown#	454	*0.0*	46	*5.4*	25	*3.9*	21	*10.2*			
Plurality											
Singleton	987,675	*97.3*	829	*97.1*	628	*96.8*	201	*98.0*	8.4	*7.8*	*8.96*
Twin	26,192	*2.6*	24	*2.8*	20	*3.1*	4	*2.0*	9.2	*5.9*	*13.65*
Triplet	939	*0.1*	1	*0.1*	1	*0.2*	0	*0.0*	10.6	*0.3*	*59.32*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**601**		**497**		**104**		**5.9**		
Birthweight											
< 1,000	6,784	*0.7*	121	*20.1*	82	*16.5*	39	*37.5*	178.4		
1,000–2,499	54,688	*5.4*	111	*18.5*	94	*18.9*	17	*16.3*	20.3		
2,500+	949,958	*93.8*	342	*56.9*	297	*59.8*	45	*43.3*	3.6		
Unknown	1,067	*0.1*	27	*4.5*	24	*4.8*	3	*2.9*			

95% of unknowns were in terminations before 20 weeks

Table 3.2.2 Patterns of Birth Defects, Spina Bifida,1983–1998

Type	Number	Per Cent
Isolated anomaly*	540	63.2
Other Associations:		
• Chromosomal	30	3.5
• Multiple NTD	39	4.6
• Other Same System (CNS)	19	2.2
• Other Different Systems	226	26.5
Total	**854**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, pyloric stenosis, PDA < 37 weeks, talipes, low set ears. If a case has two or more of these conditions it is classified as a multiple system defect.

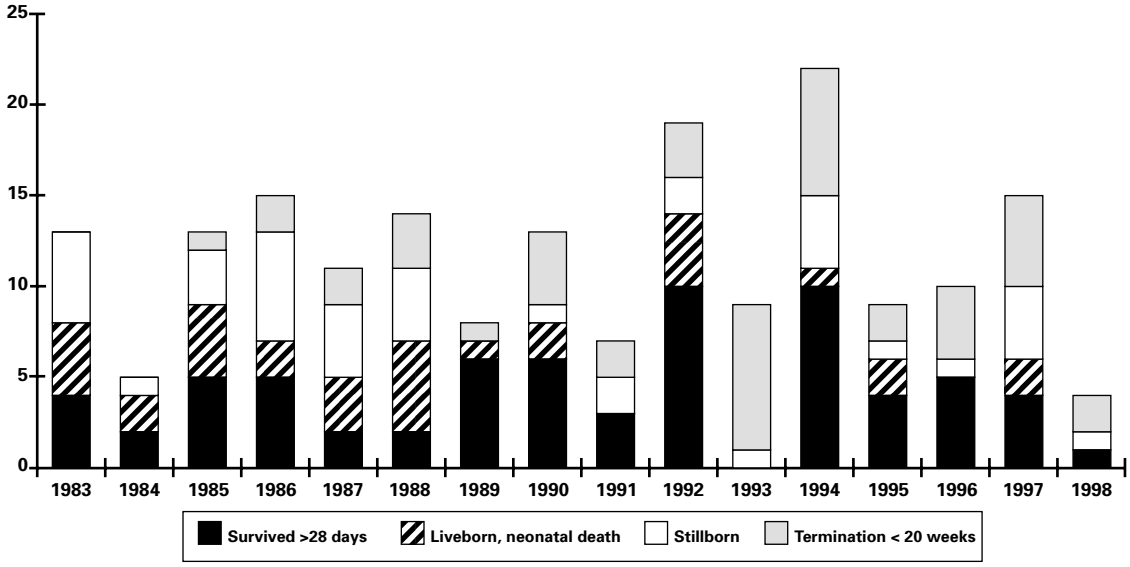
Table 3.2.3 Spina Bifida, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**852**		**647**		**205**		**8.5**	***7.96***	***9.06***
Maternal Age											
<20	40,400	*4.0*	45	*5.3*	40	*6.2*	5	*2.4*	11.1	*8.12*	*14.93*
20–24	185,901	*18.6*	178	*20.9*	139	*21.5*	39	*19.0*	9.6	*8.24*	*11.12*
25–29	367,374	*36.7*	291	*34.2*	217	*33.5*	74	*36.1*	7.9	*7.05*	*8.90*
30–34	289,879	*29.0*	236	*27.7*	178	*27.5*	58	*28.3*	8.1	*7.15*	*9.26*
35–39	101,575	*10.1*	71	*8.3*	47	*7.3*	24	*11.7*	7.0	*5.50*	*8.87*
40+	15,718	*1.6*	9	*1.1*	6	*0.9*	3	*1.5*	5.7	*2.62*	*10.88*
Unknown#	273	*0.0*	22	*2.6*	22	*3.4*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	589	*69.1*	428	*66.2*	161	*78.5*	7.8	*7.19*	*8.45*
Oceania inc NZ	20,620	*2.1*	24	*2.8*	12	*1.9*	12	*5.9*	11.6	*7.46*	*17.34*
UK inc Eire	50,596	*5.1*	40	*4.7*	34	*5.3*	6	*2.9*	7.9	*5.64*	*10.75*
Europe	58,931	*5.9*	40	*4.7*	35	*5.4*	5	*2.4*	6.8	*4.85*	*9.23*
Middle East	22,818	*2.3*	15	*1.8*	10	*1.5*	5	*2.4*	6.6	*3.68*	*10.85*
Asia	71,685	*7.2*	27	*3.2*	21	*3.2*	6	*2.9*	3.8	*2.48*	*5.50*
Nth America	5,003	*0.5*	7	*0.8*	6	*0.9*	1	*0.5*	14.0	*5.61*	*28.82*
Sth America	4,689	*0.5*	1	*0.1*	0	*0.0*	1	*0.5*	2.1	*0.05*	*11.88*
Africa	10,944	*1.1*	3	*0.4*	2	*0.3*	1	*0.5*	2.7	*0.56*	*8.00*
Unknown#	2,950	*0.3*	106	*12.4*	106	*16.4*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	49	*5.8*	40	*6.2*	9	*4.4*	6.9	*5.10*	*9.14*
Grampians	44,084	*4.4*	30	*3.5*	24	*3.7*	6	*2.9*	6.8	*4.59*	*9.73*
Loddon Mallee	63,104	*6.3*	60	*7.0*	47	*7.3*	13	*6.3*	9.5	*7.32*	*12.32*
Hume	51,348	*5.1*	46	*5.4*	38	*5.9*	8	*3.9*	9.0	*6.56*	*11.97*
Gippsland	56,679	*5.7*	58	*6.8*	48	*7.4*	10	*4.9*	10.2	*7.84*	*13.32*
Western Metro	126,711	*12.7*	129	*15.1*	101	*15.6*	28	*13.7*	10.2	*8.53*	*12.14*
Northern Metro	171,545	*17.1*	145	*17.0*	112	*17.3*	33	*16.1*	8.5	*7.16*	*9.97*
Eastern Metro	192,893	*19.3*	126	*14.8*	86	*13.3*	40	*19.5*	6.5	*5.46*	*7.81*
Southern Metro	214,947	*21.5*	194	*22.8*	139	*21.5*	55	*26.8*	9.0	*7.82*	*10.42*
Other	8,777	*0.9*	14	*1.6*	11	*1.7*	3	*1.5*	16.0	*8.71*	*26.80*
Unknown#	48	*0.0*	1	*0.1*	1	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.3 Encephalocele

Figure 3.3 Encephalocele, Number of Cases by Year



Number	13	5	13	15	11	14	8	13	7	19	9	22	9	10	15	4
N/10,000	2.1	0.8	2.1	2.4	1.8	2.2	1.2	1.9	1.1	2.9	1.4	3.4	1.4	1.6	2.4	0.6

Encephalocele

British Paediatric Association code 742.00–742.09

Cystic expansion (herniation) of meninges and brain tissue outside the cranium, covered by normal or atrophic skin

- This is the rarest of the neural tube defects with only 14 cases reported from 1995–1998 and an overall prevalence of 1.8/10,000. The decline in overall prevalence that was observed for other neural tube defects was also observed for encephalocele in 1998.
- Fluctuations in reported number of cases are seen, with 1994 having the highest number.
- Many babies are stillborn or die in the newborn period, this malformation often being associated with others.
- There is a non-significant excess of females.
- As with anencephaly, this neural tube defect is present in twin babies twice as often as amongst all births. However, the numbers are small and this is not a significant difference.
- There is no association with maternal age
- There is a borderline significant two-fold increased risk of encephalocele in the Asian-born women, compared with Australian-born women, especially apparent in the last four year period.
- Regional variations exist but the only significant difference is that Western Metro has significantly more babies born with this neural tube defect compared with Southern Metro.

Table 3.3.1 Encephalocele, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**187**		**149**		**38**		**1.8**	***1.59***	***2.13***
Survived > 28 days	1,000,776	*98.6*	69	*36.9*	55	*36.9*	14	*36.8*	0.7		
Neonatal death	4,288	*0.4*	32	*17.1*	28	*18.8*	4	*10.5*			
Stillbirth	7,397	*0.7*	40	*21.4*	33	*22.1*	7	*18.4*			
Termination < 20 wks	2,402	*0.2*	46	*24.6*	33	*22.1*	13	*34.2*			
Sex											
Male	521,553	*51.4*	76	*40.6*	63	*42.3*	13	*34.2*	1.5	*1.2*	*1.83*
Female	492,558	*48.5*	100	*53.5*	78	*52.3*	22	*57.9*	2.0	*1.7*	*2.48*
Indeterminate	298	*0.0*	2	*1.1*	2	*1.3*	0	*0.0*	67.1	*8.1*	*242.28*
Unknown#	454	*0.0*	9	*4.8*	6	*4.0*	3	*7.9*			
Plurality											
Singleton	987,675	*97.3*	177	*94.7*	141	*94.6*	36	*94.7*	1.8	*1.5*	*2.08*
Twin	26,192	*2.6*	10	*5.3*	8	*5.4*	2	*5.3*	3.8	*1.8*	*7.03*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**141**		**116**		**25**		**1.4**		
Birthweight											
< 1,000	6,784	*0.7*	27	*19.1*	18	*15.5*	9	*36.0*	39.8		
1,000–2,499	54,688	*5.4*	38	*27.0*	35	*30.2*	3	*12.0*	6.9		
2,500+	949,958	*93.8*	70	*49.6*	57	*49.1*	13	*52.0*	0.7		
Unknown	1,067	*0.1*	6	*4.3*	6	*5.2*	0	*0.0*			

95% of unknowns were terminations of pregnancy before 20 weeks

Table 3.3.2 Patterns of Birth Defects, Encephalocele, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	85	45.5
Other Associations:		
• Chromosomal	5	2.7
• Multiple NTD	9	1.5
• Other Same System (CNS)	28	15.0
• Other Different Systems	60	32.1
Total	**187**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: talipes and adrenal hypoplasia. If a case has two or more of these conditions it is classified as a multiple system defect.

Table 3.3.3 Encephalocele, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**187**		**149**		**38**		**1.9**	***1.61***	***2.16***
Maternal Age											
<20	40,400	*4.0*	11	*5.9*	6	*4.0*	5	*13.2*	2.7	*1.36*	*4.87*
20–24	185,901	*18.6*	31	*16.6*	24	*16.1*	7	*18.4*	1.7	*1.13*	*2.37*
25–29	367,374	*36.7*	64	*34.2*	58	*38.9*	6	*15.8*	1.7	*1.35*	*2.24*
30–34	289,879	*29.0*	57	*30.5*	44	*29.5*	13	*34.2*	2.0	*1.50*	*2.57*
35–39	101,575	*10.1*	19	*10.2*	15	*10.1*	4	*10.5*	1.9	*1.13*	*2.92*
40+	15,718	*1.6*	2	*1.1*	0	*0.0*	2	*5.3*	1.3	*0.15*	*4.59*
Unknown#	273	*0.0*	3	*1.6*	2	*1.3*	1	*2.6*	N/A		
Country of birth											
Australia	753,424	*75.3*	122	*65.2*	97	*65.1*	25	*65.8*	1.6	*1.35*	*1.94*
Oceania inc NZ	20,620	*2.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
UK inc Eire	50,596	*5.1*	6	*3.2*	6	*4.0*	0	*0.0*	1.2	*0.44*	*2.59*
Europe	58,931	*5.9*	11	*5.9*	9	*6.0*	2	*5.3*	1.9	*0.93*	*3.34*
Middle East	22,818	*2.3*	3	*1.6*	2	*1.3*	1	*2.6*	1.3	*0.27*	*3.84*
Asia	71,685	*7.2*	22	*11.8*	14	*9.4*	8	*21.1*	3.1	*1.92*	*4.63*
Nth America	5,003	*0.5*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Sth America	4,689	*0.5*	1	*0.5*	1	*0.7*	0	*0.0*	2.1	*0.05*	*11.88*
Africa	10,944	*1.1*	1	*0.5*	1	*0.7*	0	*0.0*	0.9	*0.02*	*5.09*
Unknown#	2,950	*0.3*	21	*11.2*	19	*12.8*	2	*5.3*	N/A		
Region											
Barwon S W	70,984	*7.1*	8	*4.3*	5	*3.4*	3	*7.9*	1.1	*0.49*	*2.22*
Grampians	44,084	*4.4*	10	*5.3*	9	*6.0*	1	*2.6*	2.3	*1.09*	*4.17*
Loddon Mallee	63,104	*6.3*	16	*8.6*	13	*8.7*	3	*7.9*	2.5	*1.45*	*4.11*
Hume	51,348	*5.1*	5	*2.7*	4	*2.7*	1	*2.6*	1.0	*0.32*	*2.27*
Gippsland	56,679	*5.7*	10	*5.3*	10	*6.7*	0	*0.0*	1.8	*0.85*	*3.25*
Western Metro	126,711	*12.7*	38	*20.3*	30	*20.1*	8	*21.1*	3.0	*2.12*	*4.11*
Northern Metro	171,545	*17.1*	31	*16.6*	22	*14.8*	9	*23.7*	1.8	*1.23*	*2.57*
Eastern Metro	192,893	*19.3*	40	*21.4*	34	*22.8*	6	*15.8*	2.1	*1.48*	*2.82*
Southern Metro	214,947	*21.5*	25	*13.4*	19	*12.8*	6	*15.8*	1.2	*0.75*	*1.72*
Other	8,777	*0.9*	4	*2.1*	3	*2.0*	1	*2.6*	4.6	*1.24*	*11.67*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.4 Microcephaly

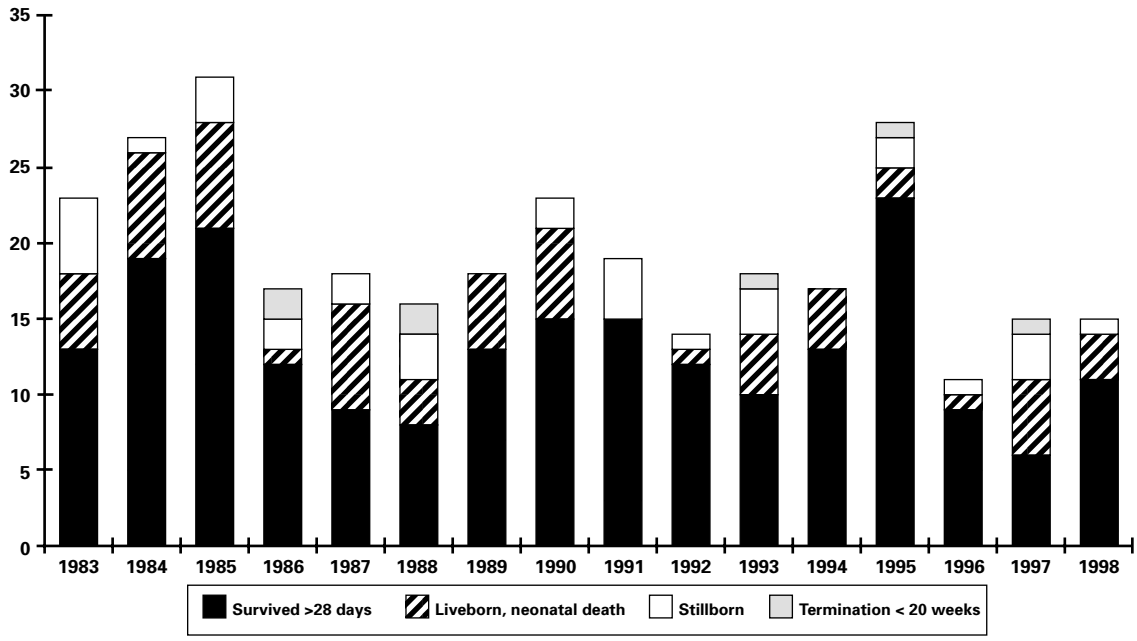


Figure 3.4 Microcephaly, Number of Cases by Year

Number	23	27	31	17	18	16	18	23	19	14	18	17	28	11	15	15
N/10,000	3.8	4.4	5.1	2.8	2.9	2.5	2.8	3.4	2.9	2.1	2.8	2.6	4.4	1.7	2.4	2.4

Microcephalus

British Paediatric Association code 742.19

Reduction in the size of the brain, with head circumference less than three standard deviations below the mean measurement for the same gestation or age. There are multiple known and unknown causes of microcephaly and it therefore cannot be considered as a single condition.

- The significant decline in the overall prevalence of microcephaly in the 12 year study period to 1994 (χ^2 for linear trend = 11.71, $p < 0.001$) did not continue in 1995. In that year there was a large increase, but since then, the numbers have remained stable. This demonstrates clearly the marked annual fluctuations that are sometimes observed. We do not know whether there may have existed some contributing unidentified causal agent that year or whether this was just a chance fluctuation.

  The original decline is thought to be the result of reporting inconsistencies—in the earlier years absolute microcephaly may have been reported to the BDR i.e. head circumference below the third standard deviation from the mean, no matter whether the baby was also of low birth weight (due to IUGR or prematurity). Such practice has changed, with relative microcephaly being the usual criterion for reporting in more recent years.

- There are almost no terminations of pregnancy reported for this condition.
- Babies with microcephaly are seldom stillborn (9%), and the neonatal death rate has dropped from 24% to 16%.
- There is a non-significant female excess .
- There is no significant maternal age effect.
- There is an increased risk for women of Middle Eastern background, compared with Australian-born women.
- There are no significant regional differences in birth prevalence.

Table 3.4.1 Microcephaly, 1983–1998 by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**310**		**241**		**69**		**3.1**	***2.73***	***3.42***
Survived > 28 days	1,000,776	*98.6*	209	*67.4*	162	*67.2*	47	*68.1*	2.1		
Neonatal death	4,288	*0.4*	68	*21.9*	57	*23.7*	11	*15.9*			
Stillbirth	7,397	0.7	28	*9.0*	21	*8.7*	7	*10.1*			
Termination < 20 wks	2,402	*0.2*	5	*1.6*	3	*1.2*	2	*2.9*			
Sex											
Male	521,553	*51.4*	143	*46.1*	109	*45.2*	34	*49.3*	2.7	*2.3*	*3.24*
Female	492,558	*48.5*	164	*52.9*	129	*53.5*	35	*50.7*	3.3	*2.8*	*3.89*
Indeterminate	298	*0.0*	3	*1.0*	3	*1.2*	0	*0.0*			
Unknown#	454	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	292	*94.2*	226	*93.8*	66	*95.7*	3.0	*2.6*	*3.32*
Twin	26,192	*2.6*	18	*5.8*	15	*6.2*	3	*4.3*	6.9	*4.1*	*10.86*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**305**		**238**		**67**	**3.0**			
Birthweight											
< 1,000	6,784	*0.7*	18	*5.9*	11	*4.6*	7	*10.4*	26.5		
1,000–2,499	54,688	*5.4*	108	*35.4*	88	*37.0*	20	*29.9*	19.7		
2,500+	949,958	*93.8*	171	*56.1*	131	*55.0*	40	*59.7*	1.8		
Unknown	1,067	*0.1*	8	*2.6*	8	*3.4*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.4.2 Patterns of Birth Defects, Microcephaly, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	91	29.4
Other Associations:		
• Chromosomal	44	14.2
• Other Same System (CNS)	27	8.7
• With cerebral palsy developmental delay	22	7.1
• Other Different Systems	126	40.7
Total	**310**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes and talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

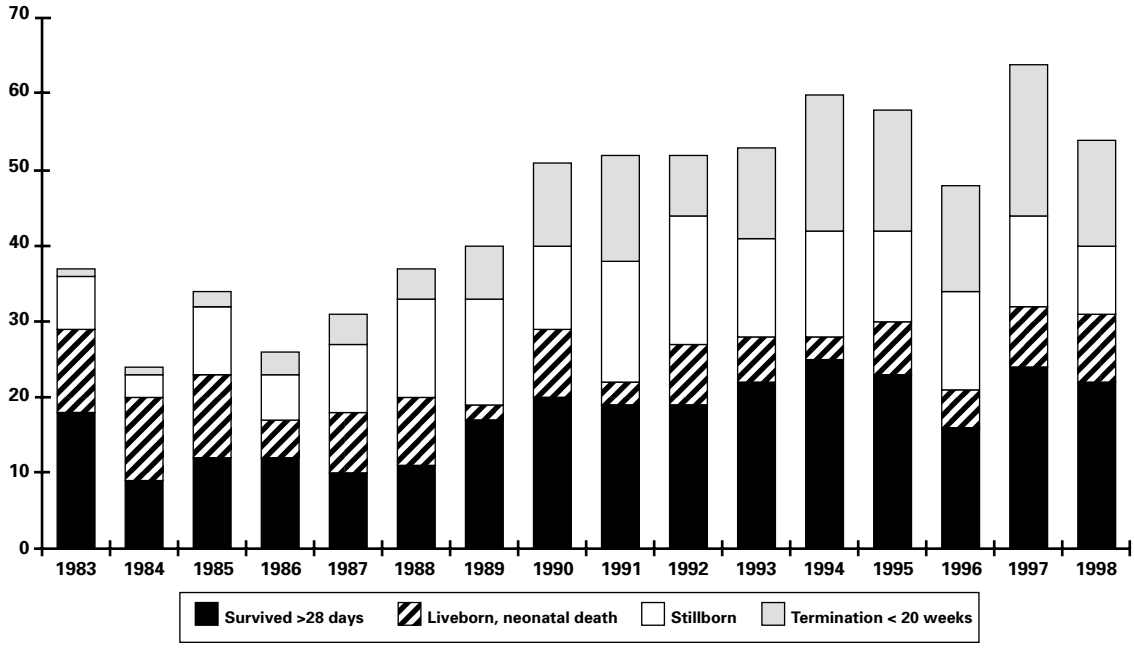
Table 3.4.3 Microcephaly, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**308**		**239**		**69**		**3.1**	***2.75***	***3.45***
Maternal Age											
<20	40,400	*4.0*	16	*5.2*	8	*3.3*	8	*11.6*	4.0	*2.27*	*6.42*
20–24	185,901	*18.6*	53	*17.2*	50	*20.9*	3	*4.3*	2.9	*2.16*	*3.76*
25–29	367,374	*36.7*	117	*38.0*	89	*37.2*	28	*40.6*	3.2	*2.65*	*3.83*
30–34	289,879	*29.0*	85	*27.6*	66	*27.6*	19	*27.5*	2.9	*2.36*	*3.64*
35–39	101,575	*10.1*	26	*8.4*	18	*7.5*	8	*11.6*	2.6	*1.67*	*3.76*
40+	15,718	*1.6*	10	*3.2*	8	*3.3*	2	*2.9*	6.4	*3.05*	*11.71*
Unknown#	273	*0.0*	1	*0.3*	0	*0.0*	1	*1.4*	N/A		
Country of birth											
Australia	753,424	*75.3*	213	*69.2*	163	*68.2*	50	*72.5*	2.8	*2.47*	*3.24*
Oceania inc NZ	20,620	*2.1*	5	*1.6*	3	*1.3*	2	*2.9*	2.4	*0.79*	*5.65*
UK inc Eire	50,596	*5.1*	16	*5.2*	14	*5.9*	2	*2.9*	3.2	*1.81*	*5.12*
Europe	58,931	*5.9*	19	*6.2*	17	*7.1*	2	*2.9*	3.2	*1.94*	*5.03*
Middle East	22,818	*2.3*	17	*5.5*	15	*6.3*	2	*2.9*	7.5	*4.34*	*11.92*
Asia	71,685	*7.2*	27	*8.8*	18	*7.5*	9	*13.0*	3.8	*2.48*	*5.50*
Nth America	5,003	*0.5*	1	*0.3*	1	*0.4*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	3	*1.0*	1	*0.4*	2	*2.9*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	4	*1.3*	4	*1.7*	0	*0.0*	3.7	*0.99*	*9.36*
Unknown#	2,950	*0.3*	3	*1.0*	3	*1.3*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	20	*6.5*	13	*5.4*	7	*10.1*	2.8	*1.72*	*4.34*
Grampians	44,084	*4.4*	7	*2.3*	5	*2.1*	2	*2.9*	1.6	*0.64*	*3.27*
Loddon Mallee	63,104	*6.3*	16	*5.2*	13	*5.4*	3	*4.3*	2.5	*1.45*	*4.11*
Hume	51,348	*5.1*	16	*5.2*	14	*5.9*	2	*2.9*	3.1	*1.78*	*5.05*
Gippsland	56,679	*5.7*	21	*6.8*	18	*7.5*	3	*4.3*	3.7	*2.29*	*5.67*
Western Metro	126,711	*12.7*	36	*11.7*	26	*10.9*	10	*14.5*	2.8	*1.99*	*3.93*
Northern Metro	171,545	*17.1*	54	*17.5*	39	*16.3*	15	*21.7*	3.1	*2.39*	*4.14*
Eastern Metro	192,893	*19.3*	65	*21.1*	48	*20.1*	17	*24.6*	3.4	*2.62*	*4.32*
Southern Metro	214,947	*21.5*	72	*23.4*	62	*25.9*	10	*14.5*	3.3	*2.64*	*4.24*
Other	8,777	*0.9*	1	*0.3*	1	*0.4*	0	*0.0*	1.1	*0.03*	*6.35*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.5 Hydrocephalus

Figure 3.5 Hydrocephalus, Number of Cases by Year



Number	37	24	34	26	31	37	40	51	52	52	53	60	58	48	64	54
N/10,000	6.1	4.0	5.6	4.2	5.0	5.8	6.2	7.6	8.0	7.8	8.2	9.2	9.1	7.6	10.3	8.7

Hydrocephalus

British Paediatric Association code 742.30–742.39

Dilatation of the ventricular system, not due to primary atrophy of brain and not necessarily associated with enlargement of the skull. These cases exclude hydrocephalus associated with spina bifida.

- There has been an increase in reported numbers since 1990, from 30–35 per year to around 50 per year.

  This increase reflected better ascertainment when new technologies enabled recognition of dilated ventricles otherwise missed.
  - Mild cases of hydrocephaly are diagnosed by CAT scanning which is used more frequently than in the early and mid 1980s. Then it was used only when there was a strong indication to do so.
  - Affected fetuses are being diagnosed prenatally by routine ultrasound leading to an increased number of pregnancy terminations. There has been no increase in prenatal detection and pregnancy termination in the last study period.
- There is a high stillbirth rate of 25% and another 20% of pregnancies are terminated before 20 weeks. There are about 20 babies surviving the neonatal period (39%) each year.
- There is a non-significant excess of males with hydrocephalus.
- This malformation is overrepresented in twin pregnancies, being found in 6% of cases.
- There is a strong U-shaped association with maternal age: a significant increased risk for teenage women and those over 40 years of age, compared to women of all other ages.
- There are no significant associations with maternal country of birth nor region of residence.

Table 3.5.1 Hydrocephalus, 1983–1988, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**721**		**497**		**224**		**7.1**	***6.59***	***7.62***
Survived > 28 days	1,000,776	*98.6*	279	*38.7*	194	*39.0*	85	*37.9*	2.8		
Neonatal death	4,288	*0.4*	115	*16.0*	86	*17.3*	29	*12.9*			
Stillbirth	7,397	*0.7*	178	*24.7*	132	*26.6*	46	*20.5*			
Termination < 20 wks	2,402	*0.2*	149	*20.7*	85	*17.1*	64	*28.6*			
Sex											
Male	521,553	*51.4*	391	*54.2*	273	*54.9*	118	*52.7*	7.5	*6.8*	*8.28*
Female	492,558	*48.5*	305	*42.3*	207	*41.6*	98	*43.8*	6.2	*5.5*	*6.94*
Indeterminate	298	*0.0*	11	*1.5*	9	*1.8*	2	*0.9*			
Unknown#	454	*0.0*	14	*1.9*	8	*1.6*	6	*2.7*			
Plurality											
Singleton	987,675	*97.3*	674	*93.5*	462	*93.0*	212	*94.6*	6.8	*6.3*	*7.34*
Twin	26,192	*2.6*	44	*6.1*	32	*6.4*	12	*5.4*	16.8	*12.2*	*22.58*
Triplet	939	*0.1*	3	*0.4*	3	*0.6*	0	*0.0*	31.9	*6.6*	*93.29*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**572**		**412**		**160**		**5.6**		
Birthweight											
< 1,000	6,784	*0.7*	145	*25.3*	84	*20.4*	61	*38.1*	213.7		
1,000–2,499	54,688	*5.4*	159	*27.8*	127	*30.8*	32	*20.0*	29.1		
2,500+	949,958	*93.8*	246	*43.0*	180	*43.7*	66	*41.3*	2.6		
Unknown	1,067	*0.1*	22	*3.8*	21	*5.1*	1	*0.6*			

95% of unknowns were in terminations before 20 weeks

Table 3.5.2 Patterns of Birth Defects, Hydrocephalus, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	308	42.7
Other Associations:		
• Chromosomal	88	12.2
• Other Same System (CNS)	62	8.6
• With cerebral palsy & developmental delay	4	0.6
• Other Different Systems	259	35.9
Total	**721**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes, PDA < 37 weeks and micrognathia. If a case has two or more of these conditions it is classified as a multiple system defect.

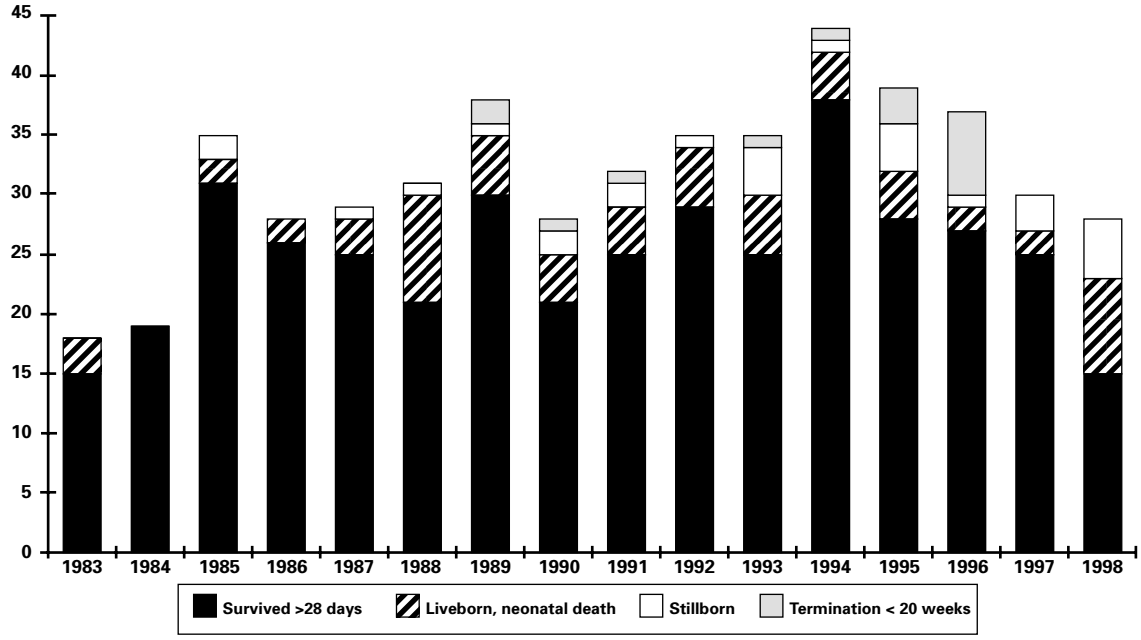
Table 3.5.3 Hydrocephalus, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**720**		**496**		**224**		**7.2**	***6.68***	***7.70***
Maternal Age											
<20	40,400	*4.0*	44	*6.1*	37	*7.5*	7	*3.1*	10.9	*7.91*	*14.64*
20–24	185,901	*18.6*	129	*17.9*	86	*17.3*	43	*19.2*	6.9	*5.82*	*8.27*
25–29	367,374	*36.7*	224	*31.1*	168	*33.9*	56	*25.0*	6.1	*5.34*	*6.96*
30–34	289,879	*29.0*	203	*28.2*	135	*27.2*	68	*30.4*	7.0	*6.09*	*8.05*
35–39	101,575	*10.1*	87	*12.1*	48	*9.7*	39	*17.4*	8.6	*6.90*	*10.62*
40+	15,718	*1.6*	20	*2.8*	12	*2.4*	8	*3.6*	12.7	*7.77*	*19.60*
Unknown#	273	*0.0*	13	*1.8*	10	*2.0*	3	*1.3*	N/A		
Country of birth											
Australia	753,424	*75.3*	486	*67.5*	319	*64.3*	167	*74.6*	6.5	*5.88*	*7.02*
Oceania inc NZ	20,620	*2.1*	17	*2.4*	8	*1.6*	9	*4.0*	8.2	*4.81*	*13.19*
UK inc Eire	50,596	*5.1*	40	*5.6*	35	*7.1*	5	*2.2*	7.9	*5.64*	*10.75*
Europe	58,931	*5.9*	34	*4.7*	23	*4.6*	11	*4.9*	5.8	*4.00*	*8.07*
Middle East	22,818	*2.3*	24	*3.3*	17	*3.4*	7	*3.1*	10.5	*6.74*	*15.67*
Asia	71,685	*7.2*	44	*6.1*	27	*5.4*	17	*7.6*	6.1	*4.46*	*8.25*
Nth America	5,003	*0.5*	3	*0.4*	2	*0.4*	1	*0.4*	6.0	*1.24*	*17.51*
Sth America	4,689	*0.5*	4	*0.6*	3	*0.6*	1	*0.4*	8.5	*2.32*	*21.84*
Africa	10,944	*1.1*	8	*1.1*	4	*0.8*	4	*1.8*	7.3	*3.15*	*14.40*
Unknown#	2,950	*0.3*	60	*8.3*	58	*11.7*	2	*0.9*	N/A		
Region											
Barwon S W	70,984	*7.1*	47	*6.5*	30	*6.0*	17	*7.6*	6.6	*4.86*	*8.82*
Grampians	44,084	*4.4*	31	*4.3*	20	*4.0*	11	*4.9*	7.0	*4.77*	*10.00*
Loddon Mallee	63,104	*6.3*	30	*4.2*	22	*4.4*	8	*3.6*	4.8	*3.21*	*6.80*
Hume	51,348	*5.1*	40	*5.6*	25	*5.0*	15	*6.7*	7.8	*5.56*	*10.59*
Gippsland	56,679	*5.7*	44	*6.1*	30	*6.0*	14	*6.3*	7.8	*5.64*	*10.43*
Western Metro	126,711	*12.7*	102	*14.2*	75	*15.1*	27	*12.1*	8.0	*6.59*	*9.81*
Northern Metro	171,545	*17.1*	117	*16.3*	76	*15.3*	41	*18.3*	6.8	*5.67*	*8.20*
Eastern Metro	192,893	*19.3*	135	*18.8*	100	*20.2*	35	*15.6*	7.0	*5.89*	*8.31*
Southern Metro	214,947	*21.5*	157	*21.8*	107	*21.6*	50	*22.3*	7.3	*6.22*	*8.57*
Other	8,777	*0.9*	13	*1.8*	8	*1.6*	5	*2.2*	14.8	*7.88*	*25.33*
Unknown#	48	*0.0*	4	*0.6*	3	*0.6*	1	*0.4*	N/A		

95% of unknowns were in terminations before 20 weeks

3.6 Transposition of Great Vessels

Figure 3.6 Transposition of Great Vessels, Number of Cases by Year



Number	18	19	35	28	29	31	38	28	32	35	35	44	39	37	30	28
N/10,000	3.0	3.1	5.7	4.6	4.7	4.9	5.9	4.2	4.9	5.3	5.4	6.8	6.1	5.9	4.8	4.5

Transposition of Great Vessels

British Paediatric Association code 745.10–745.19

Maldivision of the arterial truncus causes the aorta to emerge from the right ventricle and the pulmonary truncus from the left ventricle.

- The relatively high overall prevalence rate (compared with the Australian data ref: Congenital Malformations Australia, 1995 and 1996(3) of 5.0/10,000 probably reflects good ascertainment in Victoria, due primarily to our receiving the cardiology inpatient listings from the Royal Childrens Hospital.
- There has been a decline in birth prevalence in the last five years, despite no pregnancy terminations less than 20 weeks for this malformation since 1996. There has however been an increase in the number of stillbirths in this period.
- Almost 80% survive the neonatal period—this is not a lethal malformation if a successful operation is performed.
- There is a significant excess of males with this condition.
- There are no significant associations with mother's age, country of birth nor region of residence.

Table 3.6.1 Transposition of Great Vessels, 1983–1988, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**506**		**372**		**134**		**5.0**	***4.55***	***5.42***
Survived > 28 days	1,000,776	*98.6*	400	*79.1*	305	*82.0*	95	*70.9*	4.0		
Neonatal death	4,288	*0.4*	62	*12.3*	46	*12.4*	16	*11.9*			
Stillbirth	7,397	*0.7*	28	*5.5*	15	*4.0*	13	*9.7*			
Termination < 20 wks	2,402	*0.2*	16	*3.2*	6	*1.6*	10	*7.5*			
Sex											
Male	521,553	*51.4*	319	*63.0*	241	*64.8*	78	*58.2*	6.1	*5.5*	*6.84*
Female	492,558	*48.5*	182	*36.0*	126	*33.9*	56	*41.8*	3.7	*3.2*	*4.28*
Indeterminate	298	*0.0*	4	*0.8*	4	*1.1*	0	*0.0*			
Unknown#	454	*0.0*	1	*0.2*	1	*0.3*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	482	*95.3*	351	*94.4*	131	*97.8*	4.9	*4.4*	*5.32*
Twin	26,192	*2.6*	21	*4.2*	19	*5.1*	2	*1.5*	8.0	*5.0*	*12.27*
Triplet	939	*0.1*	3	*0.6*	2	*0.5*	1	*0.7*	31.9	*6.6*	*93.29*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**490**		**366**		**124**	**4.8**			
Birthweight											
< 1,000	6,784	*0.7*	25	*5.1*	11	*3.0*	14	*11.3*	36.9		
1,000–2,499	54,688	*5.4*	68	*13.9*	52	*14.2*	16	*12.9*	12.4		
2,500+	949,958	*93.8*	389	*79.4*	295	*80.6*	94	*75.8*	4.1		
Unknown	1,067	*0.1*	8	*1.6*	8	*2.2*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.6.2 Patterns of Birth Defects, Transposition of Great Vessels, 1983–1998

Type	Number	Per Cent
Isolated anomaly	46	9.1
Other Associations:		
• Chromosomal	28	5.5
• Other Same System (Cardiac)	338	66.8
• Other Different Systems	94	15.6
Total	**506**	**100.0**

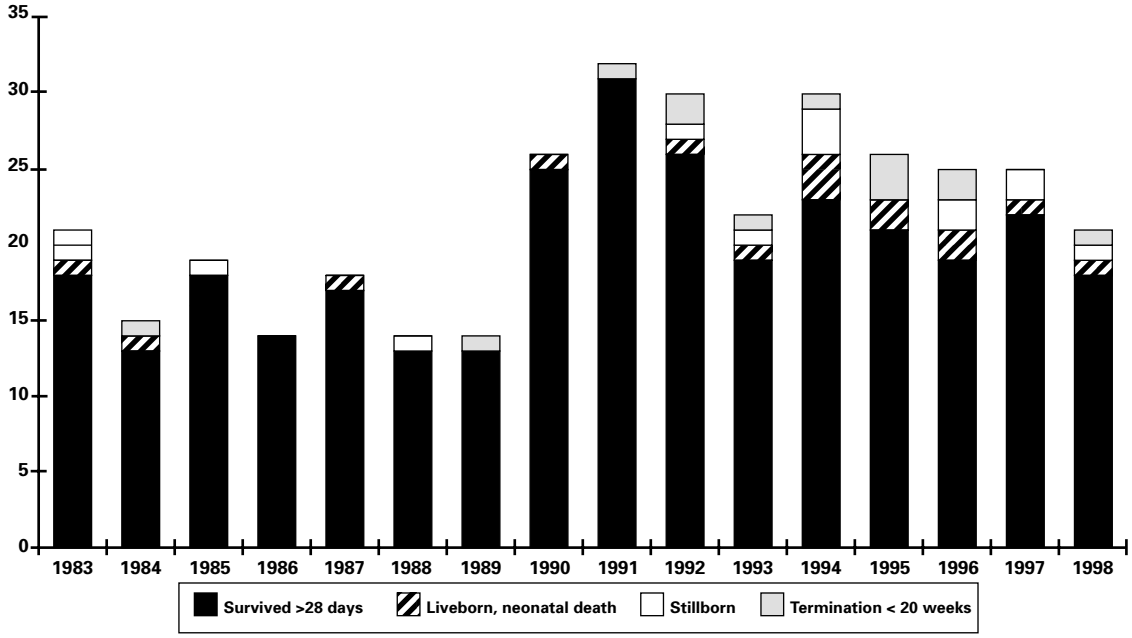
Table 3.6.3 Transposition of Great Vessels, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**506**		**372**		**134**		**5.1**	***4.62***	***5.49***
Maternal Age											
<20	40,400	*4.0*	19	*3.8*	13	*3.5*	6	*4.5*	4.7	*2.83*	*7.34*
20–24	185,901	*18.6*	83	*16.4*	53	*14.2*	30	*22.4*	4.5	*3.58*	*5.56*
25–29	367,374	*36.7*	194	*38.3*	153	*41.1*	41	*30.6*	5.3	*4.57*	*6.09*
30–34	289,879	*29.0*	137	*27.1*	104	*28.0*	33	*24.6*	4.7	*3.98*	*5.61*
35–39	101,575	*10.1*	62	*12.3*	41	*11.0*	21	*15.7*	6.1	*4.72*	*7.88*
40+	15,718	*1.6*	9	*1.8*	7	*1.9*	2	*1.5*	5.7	*2.62*	*10.88*
Unknown#	273	*0.0*	2	*0.4*	1	*0.3*	1	*0.7*	N/A		
Country of birth											
Australia	753,424	*75.3*	372	*73.5*	275	*73.9*	97	*72.4*	4.9	*4.45*	*5.47*
Oceania inc NZ	20,620	*2.1*	10	*2.0*	8	*2.2*	2	*1.5*	4.8	*2.33*	*8.92*
UK inc Eire	50,596	*5.1*	28	*5.5*	19	*5.1*	9	*6.7*	5.5	*3.68*	*8.02*
Europe	58,931	*5.9*	36	*7.1*	28	*7.5*	8	*6.0*	6.1	*4.28*	*8.45*
Middle East	22,818	*2.3*	14	*2.8*	12	*3.2*	2	*1.5*	6.1	*3.35*	*10.31*
Asia	71,685	*7.2*	29	*5.7*	20	*5.4*	9	*6.7*	4.0	*2.71*	*5.83*
Nth America	5,003	*0.5*	3	*0.6*	2	*0.5*	1	*0.7*	6.0	*1.24*	*17.51*
Sth America	4,689	*0.5*	4	*0.8*	3	*0.7*	1	*0.8*	8.5	*2.32*	*21.84*
Africa	10,944	*1.1*	4	*0.8*	1	*0.3*	3	*2.2*	3.7	*0.99*	*9.36*
Unknown#	2,950	*0.3*	6	*1.2*	5	*1.3*	1	*0.7*	N/A		
Region											
Barwon S W	70,984	*7.1*	38	*7.5*	33	*8.9*	5	*3.7*	5.4	*3.78*	*7.34*
Grampians	44,084	*4.4*	21	*4.2*	13	*3.5*	8	*6.0*	4.8	*2.95*	*7.29*
Loddon Mallee	63,104	*6.3*	32	*6.3*	22	*5.9*	10	*7.5*	5.1	*3.47*	*7.17*
Hume	51,348	*5.1*	21	*4.2*	17	*4.6*	4	*3.0*	4.1	*2.53*	*6.26*
Gippsland	56,679	*5.7*	30	*5.9*	25	*6.7*	5	*3.7*	5.3	*3.57*	*7.57*
Western Metro	126,711	*12.7*	63	*12.5*	45	*12.1*	18	*13.4*	5.0	*3.85*	*6.40*
Northern Metro	171,545	*17.1*	84	*16.6*	58	*15.6*	26	*19.4*	4.9	*3.93*	*6.10*
Eastern Metro	192,893	*19.3*	90	*17.8*	66	*17.7*	24	*17.9*	4.7	*3.77*	*5.76*
Southern Metro	214,947	*21.5*	121	*23.9*	90	*24.2*	31	*23.1*	5.6	*4.69*	*6.75*
Other	8,777	*0.9*	4	*0.8*	1	*0.3*	3	*2.2*	4.6	*1.24*	*11.67*
Unknown#	48	*0.0*	2	*0.4*	2	*0.5*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.7 Tetralogy of Fallot

Figure 3.7 Tetralogy of Fallot, Number of Cases by Year



	1983	1984	1985	1986	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998
Number	21	15	19	14	18	14	14	27	32	30	22	30	26	25	25	21
N/10,000	3.5	2.5	3.1	2.3	2.9	2.2	2.2	4.0	4.9	4.5	3.4	4.6	4.1	4.0	4.0	3.4

Tetralogy of Fallot

British Paediatric Association code 745.20

An anomaly of the heart consisting of pulmonary stenosis, interventricular septal defect, dextraposed aorta that receives blood from both ventricles and hypertrophy of the right ventricle.

- The overall birth prevalence before 1990 was generally lower than in later years, but this may reflect the method of reporting only. This condition used to be reported as VSD plus pulmonary stenosis or atresia and was not necessarily called Tetralogy of Fallot. Reporting changed in about 1990 at the Royal Children's Hospital so that nomenclature of cases included Tetralogy of Fallot. There is continuing lower overall prevalence of Tetralogy of Fallot (3.5 / 10,000) compared with Transposition of the Great Arteries (5.0 / 10,000).
- There has been a slight decline in the proportion of cases that survive the neonatal period: 90% in 1983–94 to 82% in 1995–98. This is associated with an increase in the proportion of terminations of pregnancy and stillbirths in the presence of this condition.
- There is a significant excess of males with this condition.
- There is no significant linear trend with advancing maternal age.
- Women born in Asia have a significantly greater risk of having a baby with this condition compared to Australian-born women.
- There are no significant regional associations.

Table 3.7.1 Tetralogy of Fallot, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**353**		**256**		**97**		**3.5**	***3.13***	***3.86***
Survived > 28 days	1,000,776	*98.6*	310	*87.8*	230	*89.8*	80	*82.5*	3.1		
Neonatal death	4,288	*0.4*	15	*4.2*	9	*3.5*	6	*6.2*			
Stillbirth	7,397	*0.7*	15	*4.2*	10	*3.9*	5	*5.2*			
Termination < 20 wks	2,402	*0.2*	13	*3.7*	7	*2.7*	6	*6.2*			
Sex											
Male	521,553	*51.4*	209	*59.2*	153	*59.8*	56	*57.7*	4.0	*3.5*	*4.60*
Female	492,558	*48.5*	143	*40.5*	102	*39.8*	41	*42.3*	2.9	*2.5*	*3.43*
Indeterminate	298	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
Unknown#	454	*0.0*	1	*0.3*	1	*0.4*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	338	*95.8*	246	*96.1*	92	*94.8*	3.4	*3.1*	*3.81*
Twin	26,192	*2.6*	15	*4.2*	10	*3.9*	5	*5.2*	5.7	*3.2*	*9.45*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**340**		**249**		**91**		**3.4**		
Birthweight											
< 1,000	6,784	*0.7*	19	*5.6*	11	*4.4*	8	*8.8*	28.0		
1,000–2,499	54,688	*5.4*	76	*22.4*	53	*21.3*	23	*25.3*	13.9		
2,500+	949,958	*93.8*	244	*71.8*	184	*73.9*	60	*65.9*	2.6		
Unknown	1,067	*0.1*	1	*0.3*	1	*0.4*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.7.2 Patterns of Birth Defects, Tetralogy of Fallot, 1983–1998

Type	Number	Per Cent
Isolated anomaly	75	21.2
Other Associations:		
• Chromosomal	49	13.9
• Other Same System (Cardiac)	143	40.5
• Other Different Systems	86	24.4
Total	**353**	**100.0**

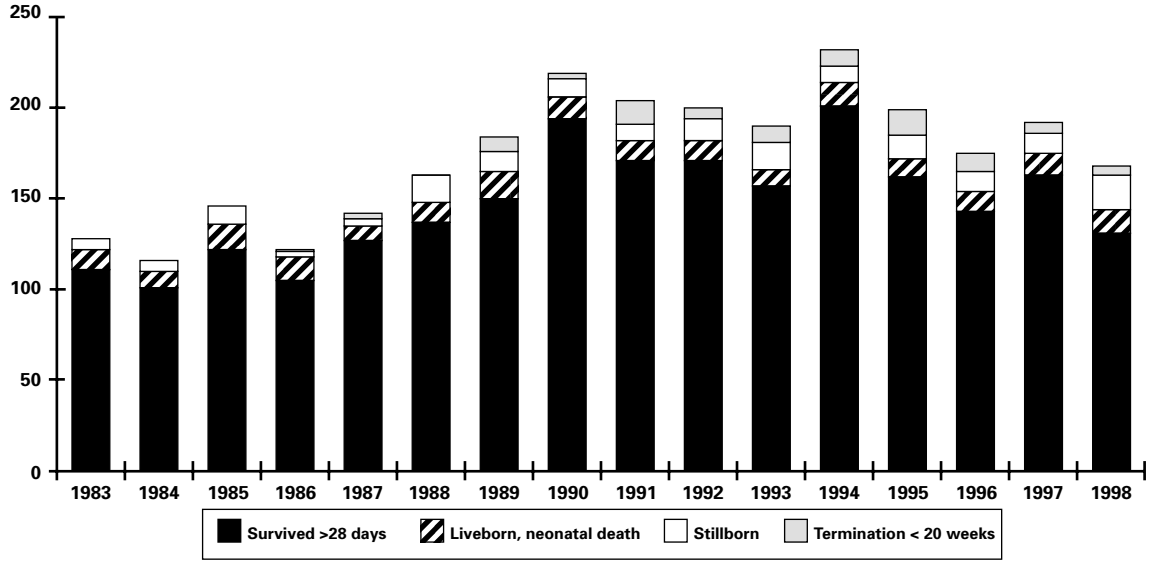
Table 3.7.3 Tetralogy of Fallot, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**353**		**256**		**97**		**3.5**	***3.17***	***3.92***
Maternal Age											
<20	40,400	*4.0*	14	*4.0*	9	*3.5*	5	*5.2*	3.5	*1.89*	*5.82*
20–24	185,901	*18.6*	66	*18.7*	49	19.1	17	*17.5*	3.6	*2.77*	*4.55*
25–29	367,374	*36.7*	113	*32.0*	86	33.6	27	*27.8*	3.1	*2.55*	*3.71*
30–34	289,879	*29.0*	105	*29.7*	74	28.9	31	*32.0*	3.6	*2.98*	*4.40*
35–39	101,575	*10.1*	48	*13.6*	33	12.9	15	*15.5*	4.7	*3.48*	*6.28*
40+	15,718	*1.6*	5	*1.4*	4	1.6	1	*1.0*	3.2	*1.03*	*7.41*
Unknown#	273	*0.0*	2	*0.6*	1	0.4	1	*1.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	251	*71.1*	186	72.7	65	*67.0*	3.3	*2.94*	*3.78*
Oceania inc NZ	20,620	*2.1*	3	*0.8*	2	0.8	1	*1.0*	1.5	*0.30*	*4.25*
UK inc Eire	50,596	*5.1*	19	*5.4*	17	6.6	2	*2.1*	3.8	*2.26*	*5.86*
Europe	58,931	*5.9*	18	*5.1*	12	4.7	6	*6.2*	3.1	*1.81*	*4.83*
Middle East	22,818	*2.3*	6	*1.7*	3	1.2	3	*3.1*	2.6	*0.97*	*5.73*
Asia	71,685	*7.2*	43	*12.2*	29	11.3	14	*14.4*	6.0	*4.34*	*8.09*
Nth America	5,003	*0.5*	2	*0.6*	0	0.0	2	*2.1*	4.0	*0.48*	*14.43*
Sth America	4,689	*0.5*	0	*0.0*	0	0.0	0	*0.0*	0.0		
Africa	10,944	*1.1*	5	*1.4*	2	0.8	3	*3.1*	4.6	*1.48*	*10.65*
Unknown#	2,950	*0.3*	6	*1.7*	5	2.0	1	*1.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	24	*6.8*	15	5.9	9	*9.3*	3.4	*2.17*	*5.04*
Grampians	44,084	*4.4*	11	*3.1*	9	3.5	2	*2.1*	2.5	*1.25*	*4.47*
Loddon Mallee	63,104	*6.3*	23	*6.5*	22	8.6	1	*1.0*	3.6	*2.31*	*5.47*
Hume	51,348	*5.1*	17	*4.8*	11	4.3	6	*6.2*	3.3	*1.93*	*5.30*
Gippsland	56,679	*5.7*	12	*3.4*	11	4.3	1	*1.0*	2.1	*1.09*	*3.71*
Western Metro	126,711	*12.7*	54	*15.3*	37	14.5	17	*17.5*	4.3	*3.23*	*5.60*
Northern Metro	171,545	*17.1*	72	*20.4*	50	19.5	22	*22.7*	4.2	*3.31*	*5.32*
Eastern Metro	192,893	*19.3*	66	*18.7*	50	19.5	16	*16.5*	3.4	*2.67*	*4.38*
Southern Metro	214,947	*21.5*	70	*19.8*	49	19.1	21	*21.6*	3.3	*2.56*	*4.14*
Other	8,777	*0.9*	2	*0.6*	1	0.4	1	*1.0*	2.3	*0.28*	*8.23*
Unknown#	48	*0.0*	2	*0.6*	1	0.4	1	*1.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.8 Ventricular Septal Defect

Figure 3.8 Ventricular Septal Defect, Number of Cases by Year



Number	128	119	146	122	142	163	184	219	204	200	190	232	199	175	192	168
N/10,000	21.1	19.6	23.9	19.9	23.1	25.6	28.6	32.7	31.3	30.2	29.3	35.7	31.2	27.8	30.8	27.1

Ventricular Septal Defect

British Paediatric Association code 745.40–745.49

A defect in the septum between the left and right ventricles of the heart, which permits blood to be shunted between them.

- This is a one of the most common defects reported and is seen with other cardiac malformations in 32% of cases and as an isolated abnormality in 37% of cases.
- There was an increased overall prevalence recorded up to 1994, but in the last four years there has been a decline. There are now over 160 cases per year and a prevalence of 29.1/10,000 births in the last four years.
- This defect is not often associated with death in the perinatal period.
- There is a non-significant linear trend with advancing maternal age. The relatively high frequency of this malformation in babies born to women 40 years and over may be related to the association of this malformation with Down syndrome and other chromosomal disorders.
- There are significantly more babies with VSD born to women of Middle Eastern origin compared to Australian-born women, but no other significant associations with mother's country of birth nor region of residence.

Table 3.8.1 Ventricular Septal Defect, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**2,783**		**2,049**		**734**		**27.4**	***26.40***	***28.44***
Survived > 28 days	1,000,776	*98.6*	2,352	*84.5*	1753	*85.6*	599	*81.6*	23.5		
Neonatal death	4,288	*0.4*	180	*6.5*	134	*6.5*	46	*6.3*			
Stillbirth	7,397	*0.7*	164	*5.9*	110	*5.4*	54	*7.4*			
Termination < 20 wks	2,402	*0.2*	87	*3.1*	52	*2.5*	35	*4.8*			
Sex											
Male	521,553	*51.4*	1,366	*49.1*	998	*48.7*	368	*50.1*	26.2	*24.8*	*27.58*
Female	492,558	*48.5*	1,393	*50.1*	1,035	*50.5*	358	*48.8*	28.3	*26.8*	*29.76*
Indeterminate	298	*0.0*	14	*0.5*	12	*0.6*	2	*0.3*			
Unknown#	454	*0.0*	10	*0.4*	4	*0.2*	6	*0.8*			
Plurality											
Singleton	987,675	*97.3*	2,643	*95.0*	1,959	*95.6*	684	*93.2*	26.8	*25.7*	*27.78*
Twin	26,192	*2.6*	130	*4.7*	88	*4.3*	42	*5.7*	49.6	*41.6*	*59.11*
Triplet	939	*0.1*	8	*0.3*	2	*0.1*	6	*0.8*	85.2	*36.7*	*167.84*
Other	48	*0.0*	2	*0.1*	0	*0.0*	2	*0.3*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**2,696**		**1997**		**699**		**26.6**		
Birthweight											
< 1,000	6,784	*0.7*	128	*4.7*	74	*3.7*	54	*7.7*	188.7		
1,000–2,499	54,688	*5.4*	519	*19.3*	364	*18.2*	155	*22.2*	94.9		
2,500+	949,958	*93.8*	2,023	*75.0*	1,535	*76.9*	488	*69.8*	21.3		
Unknown	1,067	*0.1*	26	*1.0*	24	*1.2*	2	*0.3*			

95% of unknowns were in terminations before 20 weeks

Table 3.8.2 Patterns of Birth Defects, Ventricular Septal Defect, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	1,040	37.4
Other Associations:		
• Chromosomal	409	14.7
• Other Same System (Cardiac)	894	32.1
• Other Different Systems	440	15.8
Total	**2,783**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: clicky hip, undescended testes, inguinal hernia, talipes, pyloric stenosis or PDA < 37 weeks. If a case has two or more of these conditions it is classified as a multiple system defect.

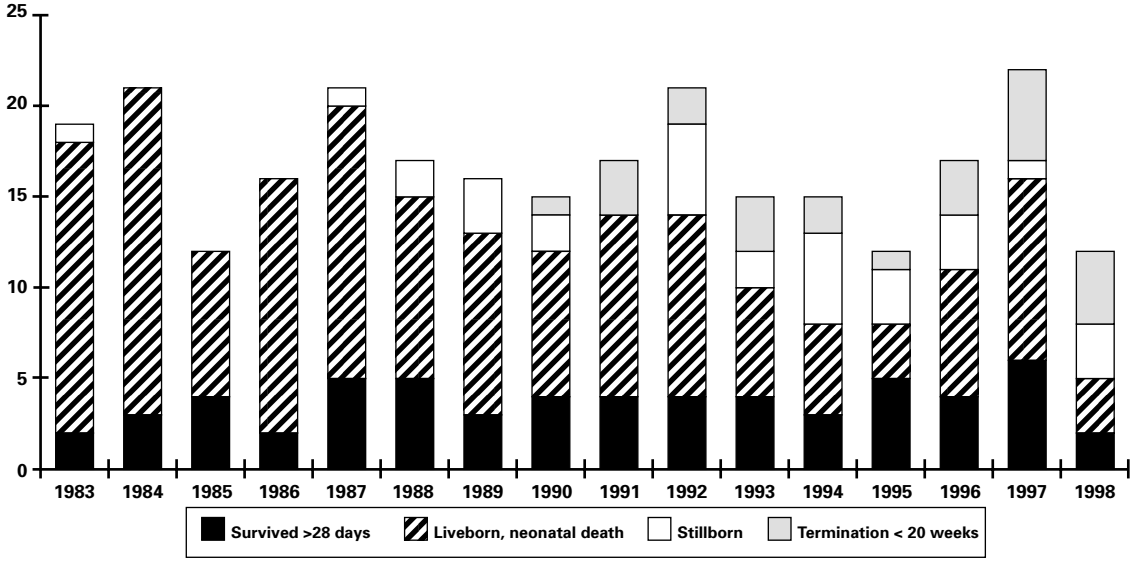
Table 3.8.3 Ventricular Septal Defect, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**2,776**		**2044**		**732**		**27.7**	***26.71***	***28.75***
Maternal Age											
<20	40,400	*4.0*	100	*3.6*	80	*3.9*	20	*2.7*	24.8	*20.25*	*30.25*
20–24	185,901	*18.6*	457	*16.5*	343	*16.8*	114	*15.6*	24.6	*22.33*	*26.83*
25–29	367,374	*36.7*	964	*34.7*	738	*36.1*	226	*30.9*	26.2	*24.58*	*27.89*
30–34	289,879	*29.0*	833	*30.0*	602	*29.5*	231	*31.6*	28.7	*26.79*	*30.68*
35–39	101,575	*10.1*	341	*12.3*	231	*11.3*	110	*15.0*	33.6	*30.15*	*37.36*
40+	15,718	*1.6*	74	*2.7*	47	*2.3*	27	*3.7*	47.1	*37.24*	*59.46*
Unknown#	273	*0.0*	7	*0.3*	3	*0.1*	4	*0.5*	N/A		
Country of birth											
Australia	753,424	*75.3*	2,011	*72.4*	1,499	*73.3*	512	*69.9*	26.7	*25.53*	*27.86*
Oceania inc NZ	20,620	*2.1*	56	*2.0*	38	*1.9*	18	*2.5*	27.2	*20.69*	*35.55*
UK inc Eire	50,596	*5.1*	148	*5.3*	113	*5.5*	35	*4.8*	29.3	*24.81*	*34.46*
Europe	58,931	*5.9*	148	*5.3*	118	*5.8*	30	*4.1*	25.1	*21.30*	*29.58*
Middle East	22,818	*2.3*	81	*2.9*	58	*2.8*	23	*3.1*	35.5	*28.36*	*44.37*
Asia	71,685	*7.2*	220	*7.9*	133	*6.5*	87	*11.9*	30.7	*26.82*	*35.11*
Nth America	5,003	*0.5*	14	*0.5*	12	*0.6*	2	*0.3*	28.0	*15.28*	*47.01*
Sth America	4,689	*0.5*	15	*0.5*	10	*0.5*	5	*0.7*	32.0	*17.91*	*52.78*
Africa	10,944	*1.1*	29	*1.0*	18	*0.9*	11	*1.5*	26.5	*17.75*	*38.16*
Unknown#	2,950	*0.3*	54	*1.9*	45	*2.2*	9	*1.2*	N/A		
Region											
Barwon S W	70,984	*7.1*	182	*6.6*	106	*5.2*	76	*10.4*	25.6	*22.10*	*29.72*
Grampians	44,084	*4.4*	133	*4.8*	97	*4.7*	36	*4.9*	30.2	*25.34*	*35.87*
Loddon Mallee	63,104	*6.3*	176	*6.3*	144	*7.0*	32	*4.4*	27.9	*23.99*	*32.41*
Hume	51,348	*5.1*	145	*5.2*	104	*5.1*	41	*5.6*	28.2	*23.92*	*33.32*
Gippsland	56,679	*5.7*	143	*5.2*	106	*5.2*	37	*5.1*	25.2	*21.34*	*29.82*
Western Metro	126,711	*12.7*	364	*13.1*	273	*13.4*	91	*12.4*	28.7	*25.88*	*31.89*
Northern Metro	171,545	*17.1*	485	*17.5*	332	*16.2*	153	*20.9*	28.3	*25.76*	*30.79*
Eastern Metro	192,893	*19.3*	532	*19.2*	412	*20.2*	120	*16.4*	27.6	*25.24*	*29.92*
Southern Metro	214,947	*21.5*	585	*21.1*	423	*20.7*	162	*22.1*	27.2	*25.01*	*29.42*
Other	8,777	*0.9*	26	*0.9*	12	*0.6*	14	*1.9*	29.6	*19.34*	*43.55*
Unknown#	48	*0.0*	5	*0.2*	5	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.9 Hypoplastic Left Heart Syndrome

Figure 3.9 Hypoplastic Left Heart Syndrome, Number of Cases by Year



Number	19	21	12	16	21	17	16	15	17	21	15	15	12	17	22	12
N/10,000	3.1	3.5	2.0	2.6	3.4	2.7	2.5	2.2	2.6	3.2	2.3	2.3	1.9	2.7	3.5	1.9

Hypoplastic Left Heart Syndrome

British Paediatric Association code 746.79

Severely underdeveloped left side of the heart as a result of aortic valve atresia, mitral valve atresia, or a combination of both.

- There has been no change in overall prevalence with numbers fluctuating each year.
- This has a lower prevalence than the other major cardiac malformations (2.6 / 10,000), but is considerably more likely to be lethal, with 62% dying in the newborn period. Neonatal death rates were higher in 1983–1994 than in 1995–1998, but the proportion surviving beyond 28 days has not increased substantially. The proportion of terminations has increased.
- There is no significant difference in the prevalence of this defect in male and female babies.
- There is no significant association with maternal age, country of birth nor region of residence. The increased prevalence in babies born to Middle Eastern women and to women living in Hume region is not significant.

Table 3.9.1 Hypoplastic Left Heart Syndrome, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**268**		**205**		**63**		**2.6**	***2.34***	***2.98***
Survived > 28 days	1,000,776	*98.6*	60	*22.4*	43	*21.0*	17	*27.0*	0.6		
Neonatal death	4,288	*0.4*	153	*57.1*	130	*63.4*	23	*36.5*			
Stillbirth	7,397	*0.7*	31	*11.6*	21	*10.2*	10	*15.9*			
Termination < 20 wks	2,402	*0.2*	24	*9.0*	11	*5.4*	13	*20.6*			
Sex											
Male	521,553	*51.4*	131	*48.9*	99	*48.3*	32	*50.8*	2.5	*2.1*	*2.99*
Female	492,558	*48.5*	130	*48.5*	101	*49.3*	29	*46.0*	2.6	*2.2*	*3.14*
Indeterminate	298	*0.0*	5	*1.9*	4	*2.0*	1	*1.6*			
Unknown#	454	*0.0*	2	*0.7*	1	*0.5*	1	*1.6*			
Plurality											
Singleton	987,675	*97.3*	265	*98.9*	202	*98.5*	63	*100.0*	2.7	*2.4*	*3.03*
Twin	26,192	*2.6*	3	*1.1*	3	*1.5*	0	*0.0*	1.1	*0.2*	*3.34*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**244**		**194**		**50**		**2.4**		
Birthweight											
< 1,000	6,784	*0.7*	22	*9.0*	12	*6.2*	10	*20.0*	32.4		
1,000–2,499	54,688	*5.4*	33	*13.5*	27	*13.9*	6	*12.0*	6.0		
2,500+	949,958	*93.8*	186	*76.2*	152	*78.4*	34	*68.0*	2.0		
Unknown	1,067	*0.1*	3	*1.2*	3	*1.5*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.9.2 Patterns of Birth Defects, Hypoplastic Left Heart Syndrome, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	98	36.6
Other Associations:		
• Chromosomal	29	10.8
• Other Same System (Cardiac)	98	36.6
• Other Different Systems	43	16.0
Total	**268**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes or micrognathia. If a case has two or more of these conditions it is classified as a multiple system defect.

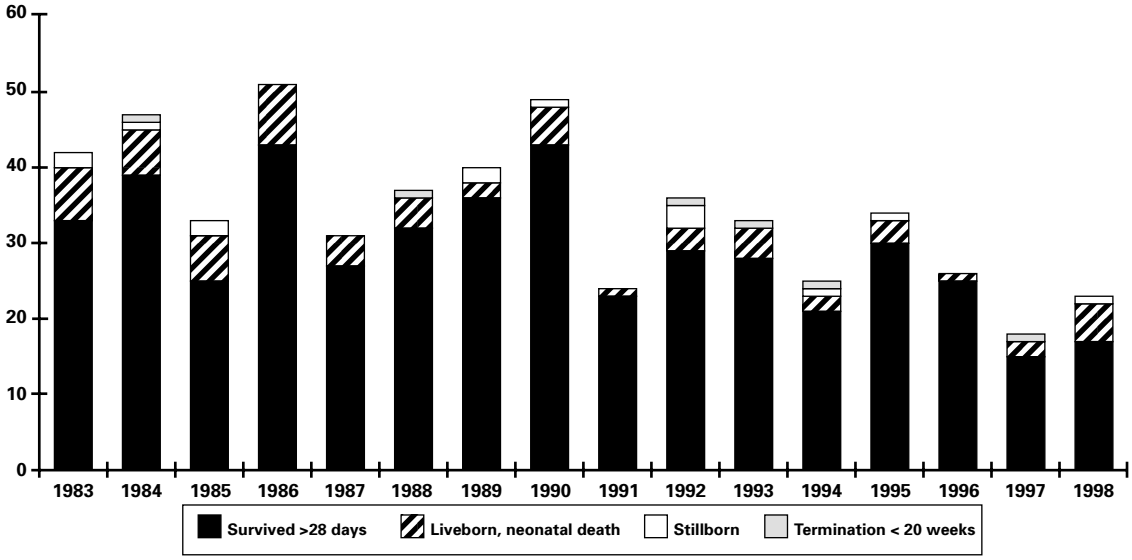
Table 3.9.3 Hypoplastic Left Heart Syndrome, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**268**		**205**		**63**		**2.7**	***2.37***	***3.02***
Maternal Age											
<20	40,400	*4.0*	11	*4.1*	10	*4.9*	1	*1.6*	2.7	*1.36*	*4.87*
20–24	185,901	*18.6*	53	*19.8*	39	*19.0*	14	*22.2*	2.9	*2.16*	*3.76*
25–29	367,374	*36.7*	100	*37.3*	83	*40.5*	17	*27.0*	2.7	*2.23*	*3.33*
30–34	289,879	*29.0*	66	*24.6*	53	*25.9*	13	*20.6*	2.3	*1.77*	*2.92*
35–39	101,575	*10.1*	30	*11.2*	15	*7.3*	15	*23.8*	3.0	*1.99*	*4.22*
40+	15,718	*1.6*	7	*2.6*	4	*2.0*	3	*4.8*	4.5	*1.79*	*9.17*
Unknown#	273	*0.0*	1	*0.4*	1	*0.5*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	210	*78.4*	158	*77.1*	52	*82.5*	2.8	*2.43*	*3.20*
Oceania inc NZ	20,620	*2.1*	7	*2.6*	6	*2.9*	1	*1.6*	3.4	*1.36*	*6.99*
UK inc Eire	50,596	*5.1*	8	*3.0*	6	*2.9*	2	*3.2*	1.6	*0.68*	*3.11*
Europe	58,931	*5.9*	9	*3.4*	7	*3.4*	2	*3.2*	1.5	*0.70*	*2.90*
Middle East	22,818	*2.3*	13	*4.9*	10	*4.9*	3	*4.8*	5.7	*3.03*	*9.74*
Asia	71,685	*7.2*	11	*4.1*	9	*4.4*	2	*3.2*	1.5	*0.77*	*2.75*
Nth America	5,003	*0.5*	1	*0.4*	1	*0.5*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	0.5	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Africa	10,944	*1.1*	2	*0.7*	1	*0.5*	1	*1.6*	1.8	*0.22*	*6.60*
Unknown#	2,950	*0.3*	7	*2.6*	7	*3.4*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	18	*6.7*	14	*6.8*	4	*6.3*	2.5	*1.50*	*4.01*
Grampians	44,084	*4.4*	14	*5.2*	10	*4.9*	4	*6.3*	3.2	*1.73*	*5.34*
Loddon Mallee	63,104	*6.3*	14	*5.2*	9	*4.4*	5	*7.9*	2.2	*1.21*	*3.73*
Hume	51,348	*5.1*	24	*9.0*	18	*8.8*	6	*9.5*	4.7	*3.00*	*6.96*
Gippsland	56,679	*5.7*	11	*4.1*	8	*3.9*	3	*4.8*	1.9	*0.97*	*3.47*
Western Metro	126,711	*12.7*	38	*14.2*	25	*12.2*	13	*20.6*	3.0	*2.12*	*4.11*
Northern Metro	171,545	*17.1*	42	*15.7*	32	*15.6*	10	*15.9*	2.4	*1.76*	*3.31*
Eastern Metro	192,893	*19.3*	46	*17.2*	38	*18.5*	8	*12.7*	2.4	*1.75*	*3.19*
Southern Metro	214,947	*21.5*	84	*31.3*	76	*37.1*	8	*12.7*	3.9	*3.13*	*4.87*
Other	8,777	*0.9*	2	*0.7*	1	*0.5*	1	*1.6*	2.3	*0.28*	*8.23*
Unknown#	48	*0.0*	1	*0.4*	0	*0.0*	1	*1.6*	N/A		

95% of unknowns were in terminations before 20 weeks

3.10 Coarctation of Aorta

Figure 3.10 Coarctation of Aorta, Number of Cases by Year



Number	41	47	33	51	31	37	40	49	24	36	33	25	34	26	18	23
N/10,000	6.8	7.7	5.4	8.3	5.0	5.8	6.2	7.3	3.7	5.4	5.1	3.9	5.3	4.1	2.9	3.7

Coarctation of Aorta

British Paediatric Association code 747.10–747.19

Narrowing of the aorta, either distal or proximal to the ductus arteriosus

- This cardiac malformation is twice as prevalent as Hypoplastic Left Heart syndrome and Tetralogy of Fallot, with a slight decline in prevalence in the last few years. It is often associated with other cardiac defects (58%).
- There is little association with neonatal death, 86% surviving this period.
- There is a significant excess in male babies.
- There is no association with maternal age.
- There is a significantly reduced prevalence in babies born to Asian-born women compared to Australian-born women.
- There are no significant associations with region of residence.

Table 3.10.1 Coarctation of Aorta, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**548**		**447**		**101**		**5.4**	***4.95***	***5.85***
Survived > 28 days	1,000,776	*98.6*	466	*85.0*	379	*84.8*	87	*86.1*	4.7		
Neonatal death	4,288	*0.4*	63	*11.5*	52	*11.6*	11	*10.9*			
Stillbirth	7,397	*0.7*	13	*2.4*	11	*2.5*	2	*2.0*			
Termination < 20 wks	2,402	*0.2*	6	*1.1*	5	*1.1*	1	*1.0*			
Sex											
Male	521,553	*51.4*	315	*57.5*	262	*58.6*	53	*52.5*	6.0	*5.4*	*6.75*
Female	492,558	*48.5*	229	*41.8*	181	*40.5*	48	*47.5*	4.6	*4.1*	*5.30*
Indeterminate	298	*0.0*	3	*0.5*	3	*0.7*	0	*0.0*			
Unknown#	454	*0.0*	1	*0.2*	1	*0.2*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	527	*96.2*	428	*95.7*	99	*98.0*	5.3	*4.9*	*5.79*
Twin	26,192	*2.6*	20	*3.6*	18	*4.0*	2	*2.0*	7.6	*4.7*	*11.76*
Triplet	939	*0.1*	1	*0.2*	1	*0.2*	0	*0.0*	10.6	*0.3*	*59.32*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**542**		**442**		**100**		**5.4**		
Birthweight											
< 1,000	6,784	*0.7*	12	*2.2*	10	*2.3*	2	*2.0*	17.7		
1,000–2,499	54,688	*5.4*	90	*16.6*	74	*16.7*	16	*16.0*	16.5		
2,500+	949,958	*93.8*	436	*80.4*	354	*80.1*	82	*82.0*	4.6		
Unknown	1,067	*0.1*	4	*0.7*	4	*0.9*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.10.2 Patterns of Birth Defects, Coarctation of Aorta, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	98	17.9
Other Associations:		
• Chromosomal	45	8.2
• Other Same System (Cardiac)	317	57.9
• Other Different Systems	88	16.1
Total	**548**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes. If a case has two or more of these conditions it is classified as a multiple system defect.

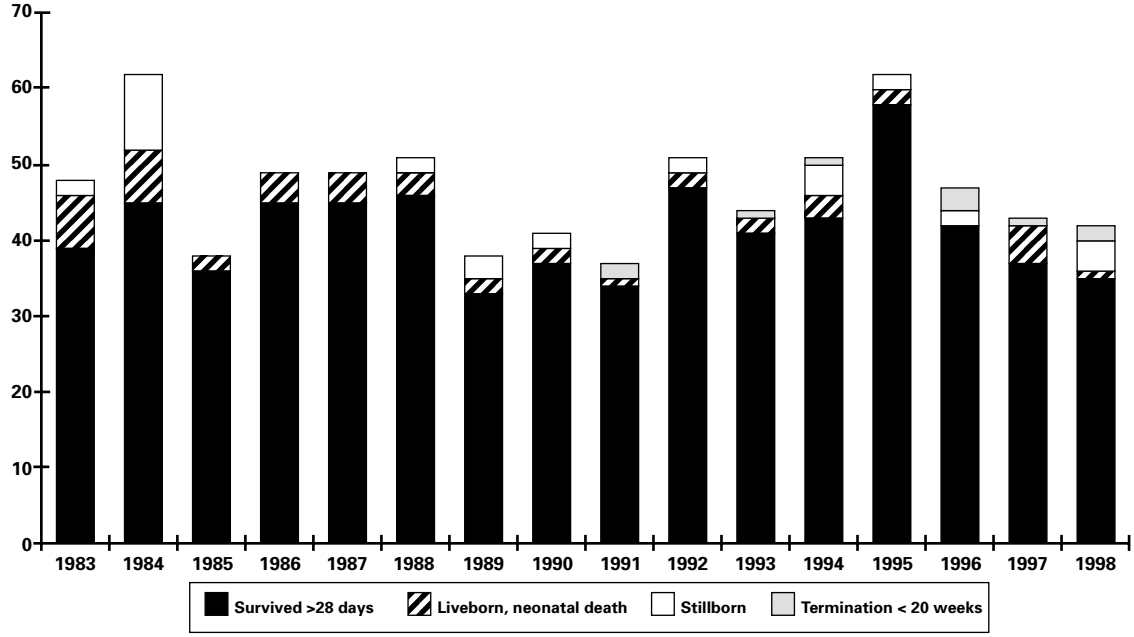
Table 3.10.3 Coarctation of Aorta, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**547**		**446**		**101**		**5.5**	***5.01***	***5.92***
Maternal Age											
<20	40,400	*4.0*	23	*4.2*	17	*3.8*	6	*5.9*	5.7	*3.61*	*8.54*
20–24	185,901	*18.6*	98	*17.9*	76	*17.0*	22	*21.8*	5.3	*4.30*	*6.45*
25–29	367,374	*36.7*	217	*39.7*	187	*41.9*	30	*29.7*	5.9	*5.16*	*6.76*
30–34	289,879	*29.0*	153	*28.0*	128	*28.7*	25	*24.8*	5.3	*4.49*	*6.20*
35–39	101,575	*10.1*	49	*9.0*	33	*7.4*	16	*15.8*	4.8	*3.56*	*6.39*
40+	15,718	*1.6*	5	*0.9*	4	*0.9*	1	*1.0*	3.2	*1.03*	*7.41*
Unknown#	273	*0.0*	2	*0.4*	1	*0.2*	1	*1.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	429	*78.4*	353	*79.1*	76	*75.2*	5.7	*5.16*	*6.23*
Oceania inc NZ	20,620	*2.1*	17	*3.1*	16	*3.6*	1	*1.0*	8.2	*4.81*	*13.19*
UK inc Eire	50,596	*5.1*	24	*4.4*	23	*5.2*	1	*1.0*	4.7	*3.04*	*7.07*
Europe	58,931	*5.9*	27	*4.9*	22	*4.9*	5	*5.0*	4.6	*3.02*	*6.69*
Middle East	22,818	*2.3*	11	*2.0*	7	*1.6*	4	*4.0*	4.8	*2.41*	*8.63*
Asia	71,685	*7.2*	24	*4.4*	16	*3.6*	8	*7.9*	3.3	*2.15*	*4.99*
Nth America	5,003	*0.5*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Sth America	4,689	*0.5*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Africa	10,944	*1.1*	10	*1.8*	6	*1.3*	4	*4.0*	9.1	*4.39*	*16.81*
Unknown#	2,950	*0.3*	5	*0.9*	3	*0.7*	2	*2.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	40	*7.3*	32	*7.2*	8	*7.9*	5.6	*4.02*	*7.66*
Grampians	44,084	*4.4*	28	*5.1*	22	*4.9*	6	*5.9*	6.4	*4.22*	*9.21*
Loddon Mallee	63,104	*6.3*	49	*9.0*	44	*9.9*	5	*5.0*	7.8	*5.74*	*10.28*
Hume	51,348	*5.1*	37	*6.8*	29	*6.5*	8	*7.9*	7.2	*5.07*	*9.93*
Gippsland	56,679	*5.7*	23	*4.2*	20	*4.5*	3	*3.0*	4.1	*2.57*	*6.09*
Western Metro	126,711	*12.7*	72	*13.2*	56	*12.6*	16	*15.8*	5.7	*4.48*	*7.20*
Northern Metro	171,545	*17.1*	76	*13.9*	55	*12.3*	21	*20.8*	4.4	*3.51*	*5.58*
Eastern Metro	192,893	*19.3*	105	*19.2*	91	*20.4*	14	*13.9*	5.4	*4.47*	*6.62*
Southern Metro	214,947	*21.5*	109	*19.9*	92	*20.6*	17	*16.8*	5.1	*4.18*	*6.14*
Other	8,777	*0.9*	6	*1.1*	4	*0.9*	2	*2.0*	6.8	*2.51*	*14.90*
Unknown#	48	*0.0*	2	*0.4*	1	*0.2*	1	*1.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.11 Cleft Palate

Figure 3.11 Cleft Palate, Number of Cases by Year



Number	48	62	38	49	44	51	38	41	37	51	44	51	62	47	43	42
N/10,000	7.9	10.2	6.2	8.0	7.1	8.0	5.9	6.1	5.7	7.7	6.8	7.9	9.7	7.5	6.9	6.8

Cleft Palate

British Paediatric Association code 749.00 – 749.09

Fissure defect of the hard and / or soft palate usually positioned in the mid-line, but without clefting of the lips. This is considered to be a distinct malformation, with different aetiology from cleft lip and palate together (see next section).

- The numbers of babies with cleft palate have fluctuated over the years, with no significant change in birth prevalence over the 16-year period (7.3 / 10,000 in 1983–94 and 7.8 / 10,000 in 1995–98).
- This is often associated with other malformations and thus the occurrence of terminations, stillbirths and neonatal deaths in 13% of cases.
- There is a significant excess of female babies with this condition.
- There is no significant association with maternal age, maternal country of birth nor region of residence.

Table 3.11.1 Cleft Palate, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**748**		**554**		**194**		**7.4**	***6.84***	***7.90***
Survived > 28 days	1,000,776	*98.6*	652	*87.2*	480	*86.6*	172	*88.7*	6.5		
Neonatal death	4,288	*0.4*	48	*6.4*	40	*7.2*	8	*4.1*			
Stillbirth	7,397	*0.7*	38	*5.1*	30	*5.4*	8	*4.1*			
Termination < 20 wks	2,402	*0.2*	10	*1.3*	4	*0.7*	6	*3.1*			
Sex											
Male	521,553	*51.4*	327	*43.7*	256	*46.2*	71	*36.6*	6.3	*5.6*	*7.00*
Female	492,558	*48.5*	419	*56.0*	297	*53.6*	122	*62.9*	8.5	*7.7*	*9.32*
Indeterminate	298	*0.0*	2	*0.3*	1	*0.2*	1	*0.5*			
Unknown#	454	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	728	*97.3*	535	*96.6*	193	*99.5*	7.4	*6.8*	*7.91*
Twin	26,192	*2.6*	17	*2.3*	16	*2.9*	1	*0.5*	6.5	*3.8*	*10.38*
Triplet	939	*0.1*	3	*0.4*	3	*0.5*	0	*0.0*	31.9	*6.6*	*93.29*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**738**		**548**		**190**		**7.3**		
Birthweight											
< 1,000	6,784	*0.7*	26	*3.5*	17	*3.1*	9	*4.7*	38.3		
1,000–2,499	54,688	*5.4*	115	*15.6*	86	*15.7*	29	*15.3*	21.0		
2,500+	949,958	*93.8*	592	*80.2*	442	*80.7*	150	*78.9*	6.2		
Unknown	1,067	*0.1*	5	*0.7*	5	*0.9*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

Table 3.11.2 Patterns of Birth Defects, Cleft Palate, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	441	59.0
Other Associations:		
• Chromosomal	67	9.0
• Other Same System (Digestive)	11	1.5
• Pierre Robin Syndrome only#	74	9.9
• Other Different Systems	155	20.7
Total	**748**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia and talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

There were a total of 109 cases with Pierre Robin Syndrome: 35 cases associated with malformations from other systems were classified as multiple system defects.

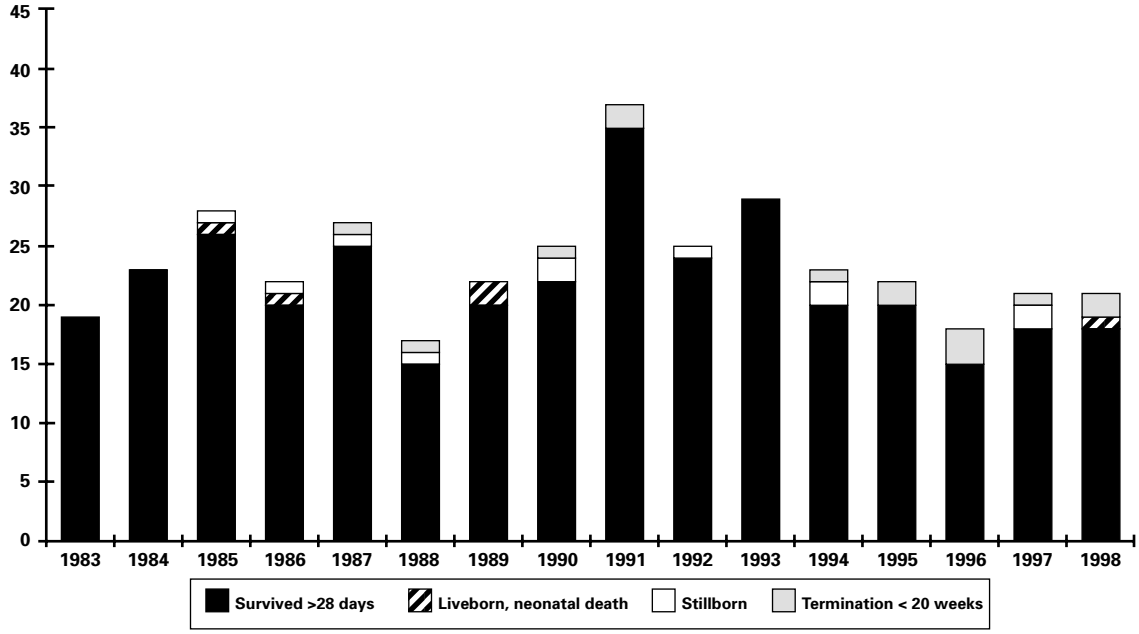
Table 3.11.3 Cleft Palate, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**745**		**551**		**194**		**7.4**	***6.91***	***7.98***
Maternal Age											
<20	40,400	*4.0*	26	*3.5*	21	*3.8*	5	*2.6*	6.4	*4.20*	*9.46*
20–24	185,901	*18.6*	130	*17.4*	104	*18.9*	26	*13.4*	7.0	*5.87*	*8.33*
25–29	367,374	*36.7*	268	*36.0*	198	*35.9*	70	*36.1*	7.3	*6.46*	*8.24*
30–34	289,879	*29.0*	207	*27.8*	153	*27.8*	54	*27.8*	7.1	*6.21*	*8.20*
35–39	101,575	*10.1*	96	*12.9*	61	*11.1*	35	*18.0*	9.5	*7.69*	*11.60*
40+	15,718	*1.6*	17	*2.3*	11	*2.0*	6	*3.1*	10.8	*6.31*	*17.31*
Unknown#	273	*0.0*	1	*0.1*	1	*0.2*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	547	*73.4*	406	*73.7*	141	*72.7*	7.3	*6.65*	*7.87*
Oceania inc NZ	20,620	*2.1*	16	*2.1*	10	*1.8*	6	*3.1*	7.8	*4.44*	*12.57*
UK inc Eire	50,596	*5.1*	38	*5.1*	34	*6.2*	4	*2.1*	7.5	*5.31*	*10.30*
Europe	58,931	*5.9*	43	*5.8*	38	*6.9*	5	*2.6*	7.3	*5.28*	*9.84*
Middle East	22,818	*2.3*	22	*3.0*	18	*3.3*	4	*2.1*	9.6	*6.05*	*14.56*
Asia	71,685	*7.2*	59	*7.9*	30	*5.4*	29	*14.9*	8.2	*6.32*	*10.69*
Nth America	5,003	*0.5*	7	*0.9*	6	*1.1*	1	*0.5*	14.0	*5.61*	*28.82*
Sth America	4,689	*0.5*	1	*0.1*	0	*0.0*	1	*0.5*	2.1	*0.05*	*11.88*
Africa	10,944	*1.1*	9	*1.2*	4	*0.7*	5	*2.6*	8.2	*3.77*	*15.63*
Unknown#	2,950	*0.3*	3	*0.4*	3	*0.5*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	45	*6.0*	36	*6.5*	9	*4.6*	6.3	*4.62*	*8.49*
Grampians	44,084	*4.4*	22	*3.0*	18	*3.3*	4	*2.1*	5.0	*3.13*	*7.54*
Loddon Mallee	63,104	*6.3*	53	*7.1*	42	*7.6*	11	*5.7*	8.4	*6.35*	*11.08*
Hume	51,348	*5.1*	39	*5.2*	31	*5.6*	8	*4.1*	7.6	*5.40*	*10.38*
Gippsland	56,679	*5.7*	49	*6.6*	39	*7.1*	10	*5.2*	8.6	*6.39*	*11.45*
Western Metro	126,711	*12.7*	108	*14.5*	76	*13.8*	32	*16.5*	8.5	*7.02*	*10.33*
Northern Metro	171,545	*17.1*	128	*17.2*	99	*18.0*	29	*14.9*	7.5	*6.25*	*8.90*
Eastern Metro	192,893	*19.3*	116	*15.6*	85	*15.4*	31	*16.0*	6.0	*4.99*	*7.24*
Southern Metro	214,947	*21.5*	175	*23.5*	116	*21.1*	59	*30.4*	8.1	*7.00*	*9.47*
Other	8,777	*0.9*	9	*1.2*	6	*1.1*	3	*1.5*	10.3	*4.70*	*19.48*
Unknown#	48	*0.0*	1	*0.1*	1	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.12 Cleft Lip

Figure 3.12 Cleft Lip, Number of Cases by Year



Number	19	24	28	22	27	17	22	25	37	25	29	23	22	18	21	21
N/10,000	3.1	4.0	4.6	3.6	4.4	2.7	3.4	3.7	5.7	3.8	4.5	3.5	3.5	2.9	3.4	3.4

Cleft Lip

British Paediatric Association code 749.10–749.19

Clefting of the upper lip, without clefting of the alveolar ridge and palate.

- Numbers have fluctuated each year with an overall prevalence of 3.8/10,000, or almost half as prevalent as cleft palate.
- This is an isolated birth defect in 85% of cases.
- Isolated cleft lip is not associated with stillbirth nor neonatal death as often as cleft palate.
- There are no significant associations with gender, maternal age, maternal country of birth, nor region of residence.

Table 3.12.1 Cleft Lip, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**380**		**298**		**82**		**3.7**	***3.38***	***4.14***
Survived > 28 days	1,000,776	*98.6*	349	*91.8*	278	*93.3*	71	*86.6*	3.5		
Neonatal death	4,288	*0.4*	5	*1.3*	4	*1.3*	1	*1.2*			
Stillbirth	7,397	*0.7*	12	*3.2*	10	*3.4*	2	*2.4*			
Termination < 20 wks	2,402	*0.2*	14	*3.7*	6	*2.0*	8	*9.8*			
Sex											
Male	521,553	*51.4*	228	*60.0*	176	*59.1*	52	*63.4*	4.4	*3.8*	*4.99*
Female	492,558	*48.5*	148	*38.9*	119	*39.9*	29	*35.4*	3.0	*2.5*	*3.54*
Indeterminate	298	*0.0*	3	*0.8*	2	*0.7*	1	*1.2*			
Unknown#	454	*0.0*	1	*0.3*	1	*0.3*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	372	*97.9*	291	*97.7*	81	*98.8*	3.8	*3.4*	*4.17*
Twin	26,192	*2.6*	8	*2.1*	7	*2.3*	1	*1.2*	3.1	*1.3*	*6.02*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**366**		**292**		**74**		**3.6**		
Birthweight											
< 1,000	6,784	*0.7*	8	*2.2*	4	*1.4*	4	*5.4*	11.8		
1,000–2,499	54,688	*5.4*	35	*9.6*	24	*8.2*	11	*14.9*	6.4		
2,500+	949,958	*93.8*	323	*88.3*	264	*90.4*	59	*79.7*	3.4		
Unknown	1,067	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.12.2 Patterns of Birth Defects, Cleft Lip, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	323	85.0
Other Associations:		
• Chromosomal	13	3.4
• Other Same System (Digestive)	3	0.8
• Other Different Systems	41	10.8
Total	**380**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia. If a case has two or more of these conditions it is classified as a multiple system defect.

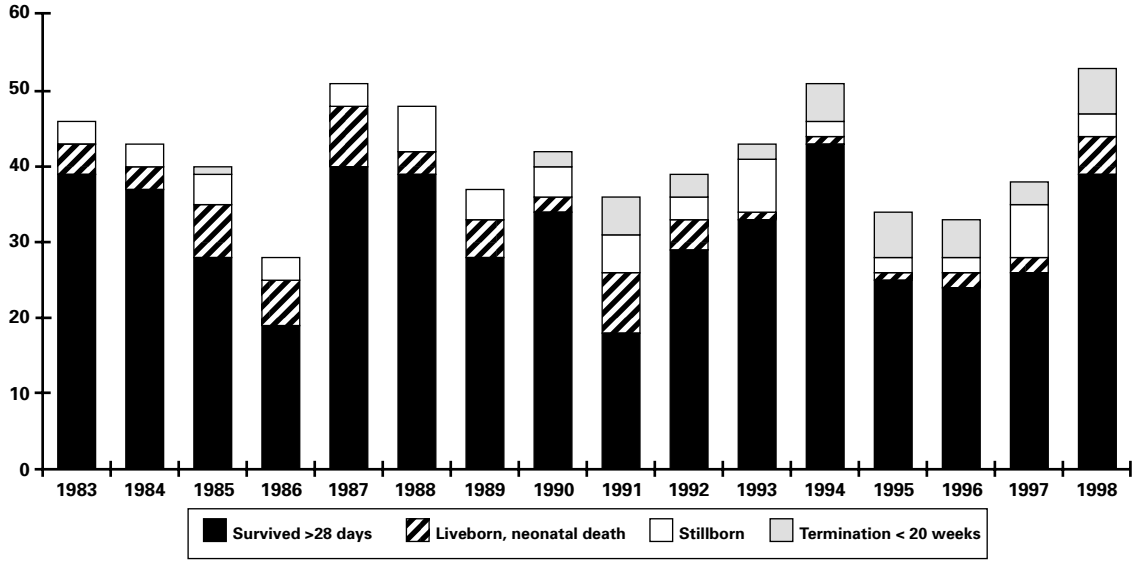
Table 3.12.3 Cleft Lip, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**380**		**298**		**82**		**3.8**	***3.43***	***4.20***
Maternal Age											
<20	40,400	*4.0*	16	*4.2*	13	*4.4*	3	*3.7*	4.0	*2.27*	*6.42*
20–24	185,901	*18.6*	64	*16.8*	52	*17.4*	12	*14.6*	3.4	*2.67*	*4.43*
25–29	367,374	*36.7*	133	*35.0*	109	*36.6*	24	*29.3*	3.6	*3.04*	*4.30*
30–34	289,879	*29.0*	114	*30.0*	89	*29.9*	25	*30.5*	3.9	*3.26*	*4.74*
35–39	101,575	*10.1*	40	*10.5*	33	*11.1*	7	*8.5*	3.9	*2.81*	*5.36*
40+	15,718	*1.6*	11	*2.9*	1	*0.3*	10	*12.2*	7.0	*3.49*	*12.53*
Unknown#	273	*0.0*	2	*0.5*	1	*0.3*	1	*1.2*	N/A		
Country of birth											
Australia	753,424	*75.3*	283	*74.5*	220	*73.8*	63	*76.8*	3.8	*3.34*	*4.23*
Oceania inc NZ	20,620	*2.1*	11	*2.9*	8	*2.7*	3	*3.7*	5.3	*2.66*	*9.55*
UK inc Eire	50,596	*5.1*	14	*3.7*	13	*4.4*	1	*1.2*	2.8	*1.51*	*4.65*
Europe	58,931	*5.9*	23	*6.1*	21	*7.0*	2	*2.4*	3.9	*2.47*	*5.85*
Middle East	22,818	*2.3*	9	*2.4*	7	*2.3*	2	*2.4*	3.9	*1.81*	*7.49*
Asia	71,685	*7.2*	23	*6.1*	18	*6.0*	5	*6.1*	3.2	*2.03*	*4.81*
Nth America	5,003	*0.5*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Sth America	4,689	*0.5*	2	*0.5*	1	*0.3*	1	*1.2*	4.3	*0.52*	*15.40*
Africa	10,944	*1.1*	7	*1.8*	4	*1.3*	3	*3.7*	6.4	*2.56*	*13.18*
Unknown#	2,950	*0.3*	8	*2.1*	6	*2.0*	2	*2.4*	N/A		
Region											
Barwon S W	70,984	*7.1*	28	*7.4*	25	*8.4*	3	*3.7*	3.9	*2.62*	*5.72*
Grampians	44,084	*4.4*	22	*5.8*	18	*6.0*	4	*4.9*	5.0	*3.13*	*7.54*
Loddon Mallee	63,104	*6.3*	20	*5.3*	14	*4.7*	6	*7.3*	3.2	*1.94*	*4.88*
Hume	51,348	*5.1*	29	*7.6*	24	*8.1*	5	*6.1*	5.6	*3.78*	*8.13*
Gippsland	56,679	*5.7*	21	*5.5*	15	*5.0*	6	*7.3*	3.7	*2.29*	*5.67*
Western Metro	126,711	*12.7*	46	*12.1*	31	*10.4*	15	*18.3*	3.6	*2.66*	*4.85*
Northern Metro	171,545	*17.1*	70	*18.4*	54	*18.1*	16	*19.5*	4.1	*3.20*	*5.19*
Eastern Metro	192,893	*19.3*	71	*18.7*	57	*19.1*	14	*17.1*	3.7	*2.90*	*4.67*
Southern Metro	214,947	*21.5*	68	*17.9*	56	*18.8*	12	*14.6*	3.2	*2.47*	*4.04*
Other	8,777	*0.9*	3	*0.8*	2	*0.7*	1	*1.2*	3.4	*0.70*	*9.98*
Unknown#	48	*0.0*	2	*0.5*	2	*0.7*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.13 Cleft Lip and Palate

Figure 3.13 Cleft Lip and Palate, Number of Cases by Year



Number	46	43	40	28	51	48	37	42	36	39	43	51	34	33	38	53
N/10,000	7.6	7.1	6.5	4.6	8.3	7.5	5.8	6.3	5.5	5.9	6.6	7.9	5.3	5.2	6.1	8.5

Cleft Lip and Palate

British Paediatric Association code 749.20–749.29

Clefting of the upper lip, with clefting of the alveolar ridge and palate.

- The numbers are fluctuating each year, with an overall prevalence of 6.5 / 10,000.
- This condition is associated with stillbirths and neonatal deaths in 19% of cases. In 1995–98, 13% of reports of this condition were associated with pregnancy termination, probably indicating the presence of other more serious malformations in the fetus.
- A large significant excess of male babies is seen.
- There is no significant association with maternal age.
- The increased prevalence in babies of Asian-born women is approaching significance.
- There are no significant regional variations.

Table 3.13.1 Cleft Lip and Palate, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**662**		**504**		**158**		**6.5**	***6.03***	***7.02***
Survived > 28 days	1,000,776	*98.6*	500	*75.5*	386	*76.6*	114	*72.2*	5.0		
Neonatal death	4,288	*0.4*	62	*9.4*	52	*10.3*	10	*6.3*			
Stillbirth	7,397	*0.7*	61	*9.2*	47	*9.3*	14	*8.9*			
Termination < 20 wks	2,402	*0.2*	38	*5.7*	18	*3.6*	20	*12.7*			
Sex											
Male	521,553	*51.4*	416	*62.8*	311	*61.7*	105	*66.5*	8.0	*7.2*	*8.74*
Female	492,558	*48.5*	239	*36.1*	188	*37.3*	51	*32.3*	4.9	*4.3*	*5.52*
Indeterminate	298	*0.0*	4	*0.6*	3	*0.6*	1	*0.6*			
Unknown#	454	*0.0*	3	*0.5*	2	*0.4*	1	*0.6*			
Plurality											
Singleton	987,675	*97.3*	646	*97.6*	494	*98.0*	152	*96.2*	6.5	*6.0*	*7.04*
Twin	26,192	*2.6*	16	*2.4*	10	*2.0*	6	*3.8*	6.1	*3.5*	*9.90*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**624**		**486**		**138**		**6.2**		
Birthweight											
< 1,000	6,784	*0.7*	37	*5.9*	23	*4.7*	14	*10.1*	54.5		
1,000–2,499	54,688	*5.4*	94	*15.1*	73	*15.0*	21	*15.2*	17.2		
2,500+	949,958	*93.8*	482	*77.2*	380	*78.2*	102	*73.9*	5.1		
Unknown	1,067	*0.1*	11	*1.8*	10	*2.1*	1	*0.7*			

95% of unknowns were in terminations before 20 weeks

Table 3.13.2 Patterns of Birth Defects, Cleft Lip and Palate, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	451	68.1
Other Associations:		
• Chromosomal	75	11.3
• Other Same System (Digestive)	2	0.3
• Other Different Systems	134	20.2
Total	**662**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes, umbilical hernia or pyloric stenosis. If a case has two or more of these conditions it is classified as a multiple system defect.

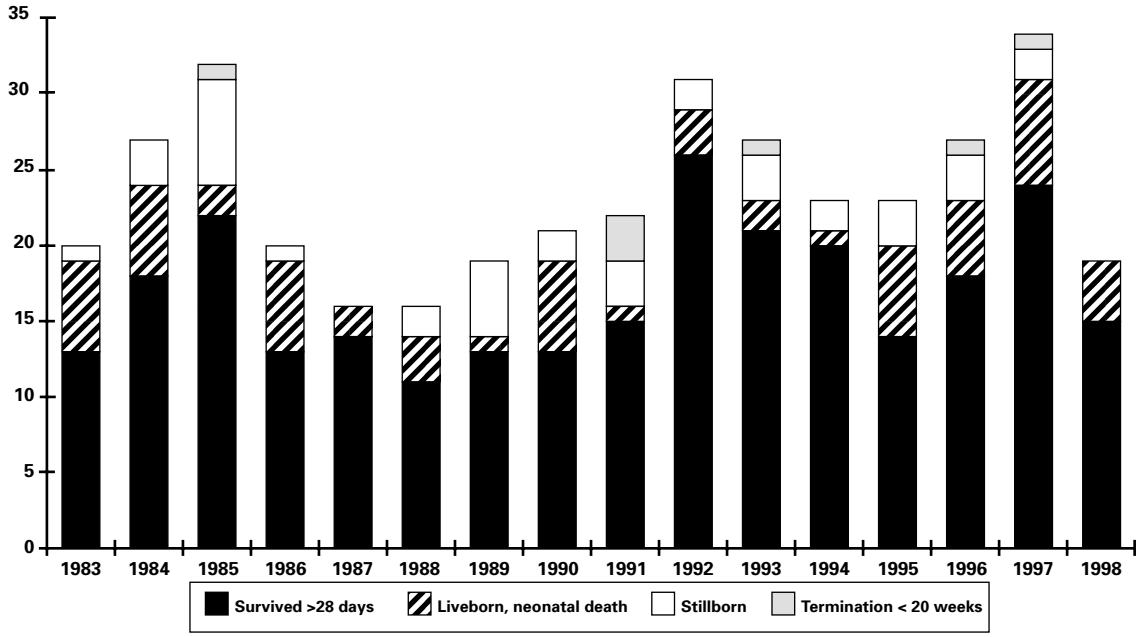
Table 3.13.3 Cleft Lip and Palate, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**661**		**503**		**158**		**6.6**	***6.10***	***7.10***
Maternal Age											
<20	40,400	*4.0*	31	*4.7*	24	*4.8*	7	*4.4*	7.7	*5.21*	*10.91*
20–24	185,901	*18.6*	123	*18.6*	104	*20.7*	19	*12.0*	6.6	*5.52*	*7.92*
25–29	367,374	*36.7*	222	*33.6*	171	*34.0*	51	*32.3*	6.0	*5.29*	*6.91*
30–34	289,879	*29.0*	189	*28.6*	138	*27.4*	51	*32.3*	6.5	*5.64*	*7.54*
35–39	101,575	*10.1*	77	*11.6*	55	*10.9*	22	*13.9*	7.6	*6.02*	*9.53*
40+	15,718	*1.6*	16	*2.4*	8	*1.6*	8	*5.1*	10.2	*5.82*	*16.49*
Unknown#	273	*0.0*	3	*0.5*	3	*0.6*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	479	*72.5*	347	*69.0*	132	*83.5*	6.4	*5.79*	*6.93*
Oceania inc NZ	20,620	*2.1*	13	*2.0*	8	*1.6*	5	*3.2*	6.3	*3.35*	*10.78*
UK inc Eire	50,596	*5.1*	38	*5.7*	33	*6.6*	5	*3.2*	7.5	*5.31*	*10.30*
Europe	58,931	*5.9*	34	*5.1*	32	*6.4*	2	*1.3*	5.8	*4.00*	*8.07*
Middle East	22,818	*2.3*	11	*1.7*	10	*2.0*	1	*0.6*	4.8	*2.41*	*8.63*
Asia	71,685	*7.2*	61	*9.2*	52	*10.3*	9	*5.7*	8.5	*6.56*	*11.01*
Nth America	5,003	*0.5*	3	*0.5*	3	*0.6*	0	*0.0*	6.0	*1.24*	*17.51*
Sth America	4,689	*0.5*	3	*0.5*	1	*0.2*	2	*1.3*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	5	*0.8*	3	*0.6*	2	*1.3*	4.6	*1.48*	*10.65*
Unknown#	2,950	*0.3*	14	*2.1*	14	*2.8*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	41	*6.2*	26	*5.2*	15	*9.5*	5.8	*4.14*	*7.83*
Grampians	44,084	*4.4*	30	*4.5*	24	*4.8*	6	*3.8*	6.8	*4.59*	*9.73*
Loddon Mallee	63,104	*6.3*	45	*6.8*	35	*7.0*	10	*6.3*	7.1	*5.20*	*9.56*
Hume	51,348	*5.1*	30	*4.5*	20	*4.0*	10	*6.3*	5.8	*3.94*	*8.35*
Gippsland	56,679	*5.7*	36	*5.4*	26	*5.2*	10	*6.3*	6.4	*4.45*	*8.79*
Western Metro	126,711	*12.7*	94	*14.2*	74	*14.7*	20	*12.7*	7.4	*6.02*	*9.12*
Northern Metro	171,545	*17.1*	104	*15.7*	83	*16.5*	21	*13.3*	6.1	*4.98*	*7.38*
Eastern Metro	192,893	*19.3*	125	*18.9*	91	*18.1*	34	*21.5*	6.5	*5.42*	*7.75*
Southern Metro	214,947	*21.5*	151	*22.8*	120	*23.9*	31	*19.6*	7.0	*5.97*	*8.26*
Other	8,777	*0.9*	4	*0.6*	3	*0.6*	1	*0.6*	4.6	*1.24*	*11.67*
Unknown#	48	*0.0*	1	*0.2*	1	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.14 Oesophageal Atresia and/or Stenosis

Figure 3.14 Oesophageal Atresia and/or Stenosis, Number of Cases by Year



Number	20	27	32	20	16	16	19	21	22	31	27	23	23	27	34	19
N/10,000	3.3	4.4	5.2	3.3	2.6	2.5	3.0	3.1	3.4	4.7	4.2	3.5	3.6	4.3	5.5	3.1

Oesophageal Atresia and/or Stenosis

British Paediatric Association code 750.30–750.38

Occlusion or narrowing of the oesophagus, with or without tracheo-oesphageal fistula.

- The numbers are fluctuating each year with an overall prevalence in recent years of 4.1 / 10,000.
- In recent years, neonatal death occurred in 21% of cases and 8% were stillborn.
- There is a non-significant excess of males with this condition.
- Over 40% of these babies are born weighing between 1000 and 2499 gm. This high proportion of low birth weight babies is not seen for any other birth defect.
- A U-shaped maternal age effect (women aged <20 and >40 years have increased birth prevalence) is apparent, but not significant.
- There are no significant differences for women from different countries of birth nor are there any regional associations.

Table 3.14.1 Oesophageal Atresia and/or Stenosis, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**377**		**274**		**103**		**3.7**	***3.35***	***4.12***
Survived > 28 days	1,000,776	*98.6*	270	*71.6*	199	*72.6*	71	*68.9*	2.7		
Neonatal death	4,288	*0.4*	61	*16.2*	39	*14.2*	22	*21.4*			
Stillbirth	7,397	*0.7*	39	*10.3*	31	*11.3*	8	*7.8*			
Termination < 20 wks	2,402	*0.2*	7	*1.9*	5	*1.8*	2	*1.9*			
Sex											
Male	521,553	*51.4*	212	*56.2*	164	*59.9*	48	*46.6*	4.1	*3.5*	*4.66*
Female	492,558	*48.5*	161	*42.7*	108	*39.4*	53	*51.5*	3.3	*2.8*	*3.82*
Indeterminate	298	*0.0*	4	*1.1*	2	*0.7*	2	*1.9*			
Unknown#	454	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	345	*91.5*	257	*93.8*	88	*85.4*	3.5	*3.1*	*3.89*
Twin	26,192	*2.6*	30	*8.0*	16	*5.8*	14	*13.6*	11.5	*7.7*	*16.38*
Triplet	939	*0.1*	2	*0.5*	1	*0.4*	1	*1.0*	21.3	*2.6*	*76.89*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**370**		**269**		**101**		**3.7**		
Birthweight											
< 1,000	6,784	*0.7*	25	*6.8*	14	*5.2*	11	*10.9*	36.9		
1,000–2,499	54,688	*5.4*	158	*42.7*	113	*42.0*	45	*44.6*	28.9		
2,500+	949,958	*93.8*	184	*49.7*	140	*52.0*	44	*43.6*	1.9		
Unknown	1,067	*0.1*	3	*0.8*	2	*0.7*	1	*1.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.14.2 Patterns of Birth Defects, Oesophageal Atresia and/or Stenosis, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	135	35.8
Other Associations:		
• Chromosomal	45	11.9
• Other Same System (Digestive)	4	1.1
• Other Different Systems	193	51.2
Total	**377**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, single umbilical artery or talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

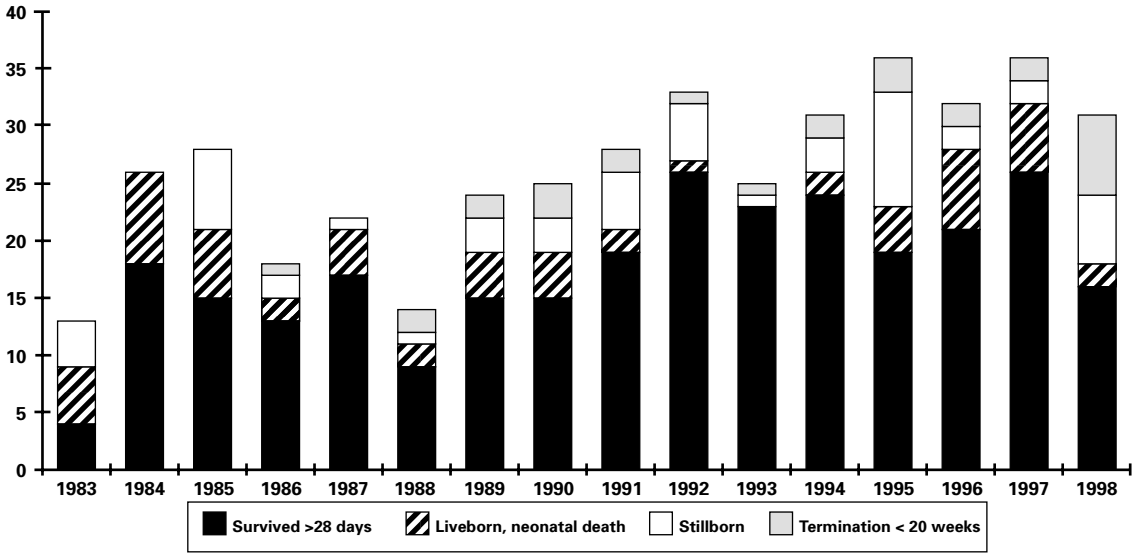
Table 3.14.3 Oesophageal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**375**		**274**		**101**		**3.7**	***3.38***	***4.15***
Maternal Age											
<20	40,400	*4.0*	20	*5.3*	15	*5.5*	5	*5.0*	5.0	*3.02*	*7.62*
20–24	185,901	*18.6*	54	*14.4*	36	*13.1*	18	*17.8*	2.9	*2.20*	*3.82*
25–29	367,374	*36.7*	154	*41.1*	121	*44.2*	33	*32.7*	4.2	*3.57*	*4.92*
30–34	289,879	*29.0*	91	*24.3*	68	*24.8*	23	*22.8*	3.1	*2.54*	*3.87*
35–39	101,575	*10.1*	46	*12.3*	27	*9.9*	19	*18.8*	4.5	*3.31*	*6.05*
40+	15,718	*1.6*	9	*2.4*	6	*2.2*	3	*3.0*	5.7	*2.62*	*10.88*
Unknown#	273	*0.0*	1	*0.3*	1	*0.4*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	278	*74.1*	203	*74.1*	75	*74.3*	3.7	*3.27*	*4.16*
Oceania inc NZ	20,620	*2.1*	5	*1.3*	2	*0.7*	3	*3.0*	2.4	*0.79*	*5.65*
UK inc Eire	50,596	*5.1*	26	*6.9*	22	*8.0*	4	*4.0*	5.1	*3.36*	*7.55*
Europe	58,931	*5.9*	22	*5.9*	15	*5.5*	7	*6.9*	3.7	*2.34*	*5.64*
Middle East	22,818	*2.3*	7	*1.9*	6	*2.2*	1	*1.0*	3.1	*1.23*	*6.32*
Asia	71,685	*7.2*	24	*6.4*	18	*6.6*	6	*5.9*	3.3	*2.15*	*4.99*
Nth America	5,003	*0.5*	1	*0.3*	1	*0.4*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	5	*1.3*	2	*0.7*	3	*3.0*	10.7	*3.45*	*24.85*
Africa	10,944	*1.1*	1	*0.3*	1	*0.4*	0	*0.0*	0.9	*0.02*	*5.09*
Unknown#	2,950	*0.3*	6	*1.6*	4	*1.5*	2	*2.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	21	5.6	17	*6.2*	4	*4.0*	3.0	*1.83*	*4.53*
Grampians	44,084	*4.4*	17	*4.5*	12	*4.4*	5	*5.0*	3.9	*2.25*	*6.17*
Loddon Mallee	63,104	*6.3*	22	*5.9*	21	*7.7*	1	*1.0*	3.5	*2.19*	*5.26*
Hume	51,348	*5.1*	10	*2.7*	8	*2.9*	2	*2.0*	1.9	*0.93*	*3.58*
Gippsland	56,679	*5.7*	20	*5.3*	17	*6.2*	3	*3.0*	3.5	*2.16*	*5.43*
Western Metro	126,711	*12.7*	46	*12.3*	29	*10.6*	17	*16.8*	3.6	*2.66*	*4.85*
Northern Metro	171,545	*17.1*	63	*16.8*	41	*15.0*	22	*21.8*	3.7	*2.85*	*4.73*
Eastern Metro	192,893	*19.3*	81	*21.6*	60	*21.9*	21	*20.8*	4.2	*3.36*	*5.25*
Southern Metro	214,947	*21.5*	91	*24.3*	66	*24.1*	25	*24.8*	4.2	*3.43*	*5.22*
Other	8,777	*0.9*	4	*1.1*	3	*1.1*	1	*1.0*	4.6	*1.24*	*11.67*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.15 Anorectal Atresia and/or Stenosis

Figure 3.15 Anorectal Atresia and/or Stenosis, Number of Cases by Year



Number	18	26	28	18	22	14	24	25	28	33	25	31	36	32	36	31
N/10,000	3.0	4.3	4.6	2.9	3.6	2.2	3.7	3.7	4.3	5.0	3.9	4.8	5.6	5.1	5.8	5.0

Anorectal Atresia and/or Stenosis

British Paediatric Association code 751.21–751.24

Imperforate anus or absence or narrowing of the communication between rectum and anus, with or without fistula to neighbouring organs.

- There has been an increase in the overall birth prevalence of this condition from 3.8/1000 births in 1983–1994 to 5.4/1000 in 1995–1998. It is present with another system defect in 57% of cases.
- There is a significant two-fold increased birth prevalence in males.
- There is a significant excess of twins.
- There are no associations with maternal age, country of birth nor region of residence.

Table 3.15.1 Anorectal Atresia and/or Stenosis, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**427**		**292**		**135**		**4.2**	***3.81***	***4.61***
Survived > 28 days	1,000,776	*98.6*	280	*65.6*	198	*67.8*	82	*60.7*	2.8		
Neonatal death	4,288	*0.4*	64	*15.0*	45	*15.4*	19	*14.1*			
Stillbirth	7,397	*0.7*	55	*12.9*	35	*12.0*	20	*14.8*			
Termination < 20wks	2,402	*0.2*	28	*6.6*	14	*4.8*	14	*10.4*			
Sex											
Male	521,553	*51.4*	262	*61.4*	180	*61.6*	82	*60.7*	5.0	*4.4*	*5.68*
Female	492,558	*48.5*	133	*31.1*	90	*30.8*	43	*31.9*	2.7	*2.3*	*3.21*
Indeterminate	298	*0.0*	31	*7.3*	22	*7.5*	9	*6.7*			
Unknown#	454	*0.0*	1	*0.2*	0	*0.0*	1	*0.7*			
Plurality											
Singleton	987,675	*97.3*	398	*93.2*	273	*93.5*	125	*92.6*	4.0	*3.6*	*4.45*
Twin	26,192	*2.6*	29	*6.8*	19	*6.5*	10	*7.4*	11.1	*7.4*	*15.94*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**399**		**278**		**121**		**3.9**		
Birthweight											
< 1,000	6,784	*0.7*	34	*8.5*	17	*6.1*	17	*14.0*	50.1		
1,000–2,499	54,688	*5.4*	99	*24.8*	67	*24.1*	32	*26.4*	18.1		
2,500+	949,958	*93.8*	259	*64.9*	189	*68.0*	70	*57.9*	2.7		
Unknown	1,067	*0.1*	7	*1.8*	5	*1.8*	2	*1.7*			

95% of unknowns were in terminations before 20 weeks

Table 3.15.2 Patterns of Birth Defects, Anorectal Atresia and/or Stenosis, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	135	31.6
Other Associations:		
• Chromosomal	31	7.3
• Other Same System (Digestive)	16	3.7
• Other Different Systems	245	57.4
Total	**427**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: undescended testes or talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

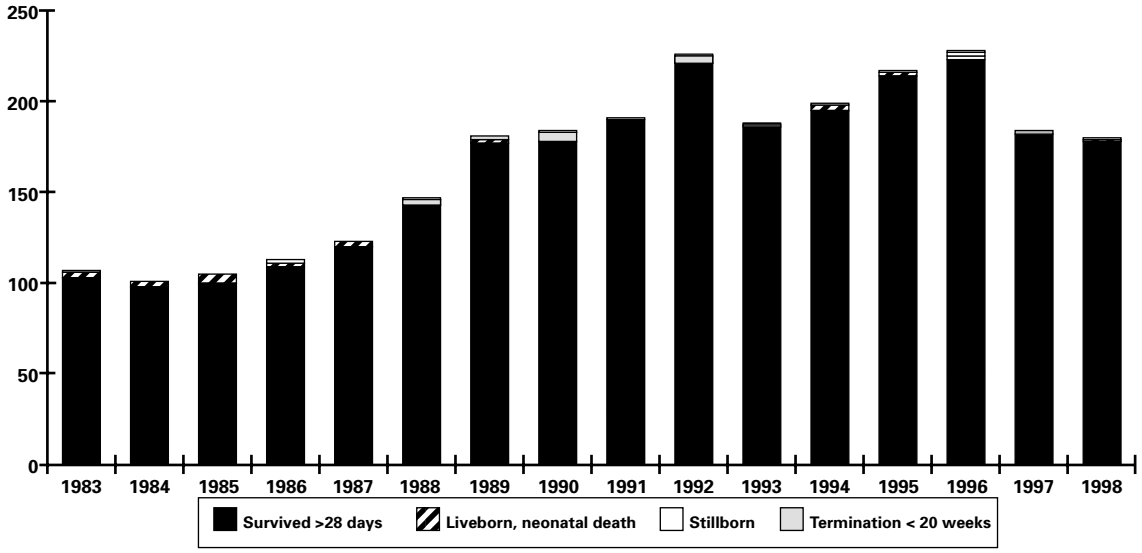
Table 3.15.3 Anorectal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**424**		**290**		**134**		**4.2**	***3.83***	***4.64***
Maternal Age											
<20	40,400	*4.0*	29	*6.8*	18	*6.2*	11	*8.2*	7.2	*4.81*	*10.34*
20–24	185,901	*18.6*	76	*17.9*	53	*18.3*	23	*17.2*	4.1	*3.24*	*5.15*
25–29	367,374	*36.7*	157	*37.0*	116	*40.0*	41	*30.6*	4.3	*3.64*	*5.01*
30–34	289,879	*29.0*	115	*27.1*	79	*27.2*	36	*26.9*	4.0	*3.29*	*4.78*
35–39	101,575	*10.1*	37	*8.7*	18	*6.2*	19	*14.2*	3.6	*2.56*	*5.02*
40+	15,718	*1.6*	7	*1.7*	3	*1.0*	4	*3.0*	4.5	*1.79*	*9.17*
Unknown#	273	*0.0*	3	*0.7*	3	*1.0*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	307	*72.4*	214	*73.8*	93	*69.4*	4.1	*3.64*	*4.56*
Oceania inc NZ	20,620	*2.1*	14	*3.3*	7	*2.4*	7	*5.2*	6.8	*3.71*	*11.41*
UK inc Eire	50,596	*5.1*	18	*4.2*	12	*4.1*	6	*4.5*	3.6	*2.11*	*5.62*
Europe	58,931	*5.9*	20	*4.7*	14	*4.8*	6	*4.5*	3.4	*2.07*	*5.23*
Middle East	22,818	*2.3*	6	*1.4*	4	*1.4*	2	*1.5*	2.6	*0.97*	*5.73*
Asia	71,685	*7.2*	34	*8.0*	20	*6.9*	14	*10.4*	4.7	*3.29*	*6.63*
Nth America	5,003	*0.5*	4	*0.9*	3	*1.0*	1	*0.7*	8.0	*2.17*	*20.47*
Sth America	4,689	*0.5*	2	*0.5*	0	*0.0*	2	*1.5*	4.3	*0.52*	*15.40*
Africa	10,944	*1.1*	3	*0.7*	2	*0.7*	1	*0.7*	2.7	*0.56*	*8.00*
Unknown#	2,950	*0.3*	16	*3.8*	14	*4.8*	2	*1.5*	N/A		
Region											
Barwon S W	70,984	*7.1*	26	*6.1*	16	*5.5*	10	*7.5*	3.7	*2.39*	*5.38*
Grampians	44,084	*4.4*	19	*4.5*	10	*3.4*	9	*6.7*	4.3	*2.59*	*6.72*
Loddon Mallee	63,104	*6.3*	29	*6.8*	22	*7.6*	7	*5.2*	4.6	*3.08*	*6.62*
Hume	51,348	*5.1*	21	*5.0*	17	*5.9*	4	*3.0*	4.1	*2.53*	*6.26*
Gippsland	56,679	*5.7*	30	*7.1*	24	*8.3*	6	*4.5*	5.3	*3.57*	*7.57*
Western Metro	126,711	*12.7*	65	*15.3*	38	*13.1*	27	*20.1*	5.1	*3.99*	*6.58*
Northern Metro	171,545	*17.1*	71	*16.7*	48	*16.6*	23	*17.2*	4.1	*3.26*	*5.25*
Eastern Metro	192,893	*19.3*	76	*17.9*	60	*20.7*	16	*11.9*	3.9	*3.12*	*4.96*
Southern Metro	214,947	*21.5*	81	*19.1*	51	*17.6*	30	*22.4*	3.8	*3.01*	*4.71*
Other	8,777	*0.9*	6	*1.4*	4	*1.4*	2	*1.5*	6.8	*2.51*	*14.90*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.16 Hypospadias

Figure 3.16 Hypospadias and/or Stenosis, Number of Cases by Year



Number	**107**	**101**	**105**	**113**	**105**	**147**	**181**	**184**	**191**	**226**	**189**	**199**	**217**	**228**	**184**	**180**
N/10,000	**17.6**	**16.6**	**17.2**	**18.4**	**17.1**	**23.1**	**28.2**	**27.5**	**29.3**	**34.1**	**29.2**	**30.6**	**34.1**	**36.2**	**29.5**	**29.0**

Hypospadias

British Paediatric Association code 752.60

Abnormal opening of the male urethra upon the undersurface of the penis.

- There was a marked increase in notifications of hypospadias in 1989. Since 1989 over 150 cases each year reported to the BDR, compared with 100 or so before 1988. Repair of this defect has been done at 6–18 months of age since the mid 1980s, however prior to that time repair was done at 3–4 years of age and notification may not have been made. The earlier management, plus our current system of receiving inpatient listings from the Royal Children's Hospital, may account for more notifications to the BDR in recent years. The birth prevalence for 1995–1998 is 32.1 / 1000.
- As there are few associated perinatal deaths (1.5% in 1995–1998), it is likely hypospadias is rarely associated with major (lethal) malformations. It occurs as an isolated defect in 86% of cases.
- There is no linear trend with maternal age.
- There is significant variation in prevalence with maternal country of birth. Women of Asian background have a significantly reduced risk compared to almost all other ethnic groups of women.
- There are no significant regional variations.

Table 3.16.1 Hypospadias, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**2,657**		**1848**		**809**		**26.2**	***25.19***	***27.18***
Survived > 28 days	1,000,776	*98.6*	2,599	*97.8*	1,802	*97.5*	797	*98.5*	26.0		
Neonatal death	4,288	*0.4*	41	*1.5*	34	*1.8*	7	*0.9*			
Stillbirth	7,397	*0.7*	14	*0.5*	10	*0.5*	4	*0.5*			
Termination < 20 wks	2,402	*0.2*	3	*0.1*	2	*0.1*	1	*0.1*			
Sex											
Male	521,553	*51.4*	2,648	*99.7*	1,840	*99.6*	808	*99.9*	50.8	*48.8*	*52.70*
Female	492,558	*48.5*									
Indeterminate	298	*0.0*	8	*0.3*	7	*0.4*	1 0.1				
Unknown#	454	*0.0*	1	*0.0*	1	*0.1*	0 0.0				
Plurality											
Singleton	987,675	*97.3*	2,553	*96.1*	1,786	*96.6*	767	*94.8*	25.8	*24.8*	*26.85*
Twin	26,192	*2.6*	97	*3.7*	58	*3.1*	39	*4.8*	37.0	*30.2*	*45.37*
Triplet	939	*0.1*	6	*0.2*	3	*0.2*	3	*0.4*	63.9	*23.5*	*139.3*
Other	48	*0.0*	1	*0.0*	1	*0.1*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**2,654**		**1846**		**808**		**26.2**		
Birthweight											
< 1,000	6,784	*0.7*	30	*1.1*	18	*1.0*	12	*1.5*	44.2		
1,000–2,499	54,688	*5.4*	315	*11.9*	207	*11.2*	108	*13.4*	57.6		
2,500+	949,958	*93.8*	2,306	*86.9*	1,618	*87.6*	688	*85.1*	24.3		
Unknown	1,067	*0.1*	3	*0.1*	3	*0.2*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

Table 3.16.2 Patterns of Birth Defects, Hypospadias, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	2,295	86.4
Other Associations:		
• Chromosomal	38	1.49
• Other Same System (Genital)	65	2.4
• Other Different Systems	259	9.8
Total	**2,657**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, misplaced ears, talipes hydrocoele, undescended testes or pyloric stenosis. If a case has two or more of these conditions it is classified as a multiple system defect.

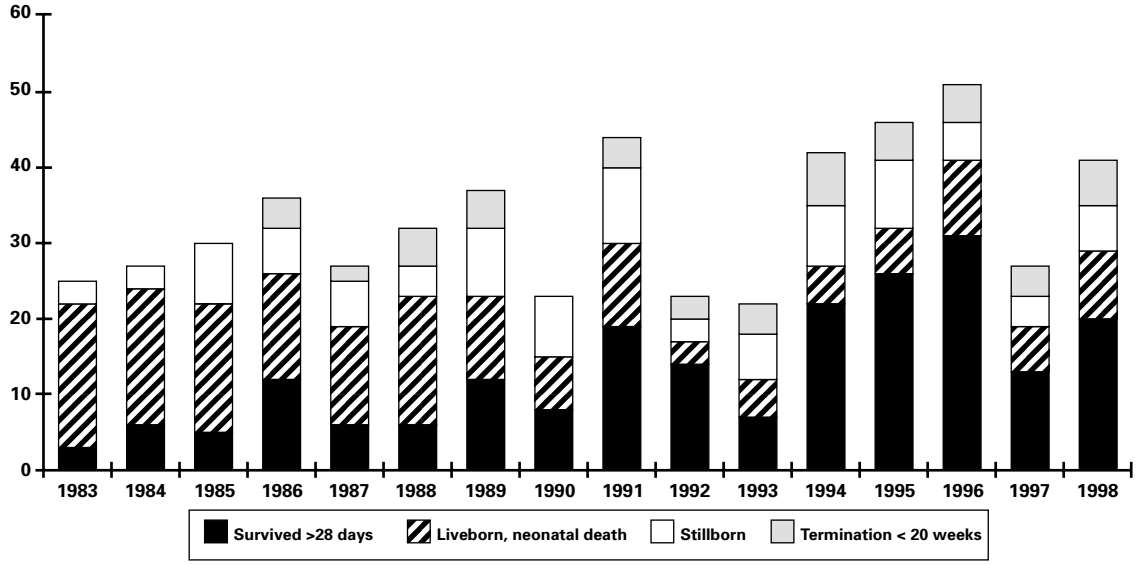
Table 3.16.3 Hypospadias, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**2,649**		**1,842**		**807**		**26.5**	***25.48***	***27.44***
Maternal Age											
<20	40,400	*4.0*	123	*4.6*	94	*5.1*	29	*3.6*	30.4	*25.39*	*36.44*
20–24	185,901	*18.6*	516	*19.5*	380	*20.6*	136	*16.9*	27.8	*25.37*	*30.15*
25–29	367,374	*36.7*	958	*36.2*	699	*37.9*	259	*32.1*	26.1	*24.43*	*27.73*
30–34	289,879	*29.0*	729	*27.5*	491	*26.7*	238	*29.5*	25.1	*23.33*	*26.97*
35–39	101,575	*10.1*	267	*10.1*	154	*8.4*	113	*14.0*	26.3	*23.26*	*29.68*
40+	15,718	*1.6*	43	*1.6*	23	*1.2*	20	*2.5*	27.4	*19.78*	*36.88*
Unknown#	273	*0.0*	13	*0.5*	1	*0.1*	12	*1.5*	N/A		
Country of birth											
Australia	753,424	*75.3*	2,089	*78.9*	1,437	*78.0*	652	*80.8*	27.7	*26.54*	*28.91*
Oceania inc NZ	20,620	*2.1*	52	*2.0*	38	*2.1*	14	*1.7*	25.2	*19.01*	*33.34*
UK inc Eire	50,596	*5.1*	129	*4.9*	95	*5.2*	34	*4.2*	25.5	*21.37*	*30.39*
Europe	58,931	*5.9*	155	*5.9*	129	*7.0*	26	*3.2*	26.3	*22.41*	*30.88*
Middle East	22,818	*2.3*	76	*2.9*	50	*2.7*	26	*3.2*	33.3	*26.41*	*41.93*
Asia	71,685	*7.2*	94	*3.5*	57	*3.1*	37	*4.6*	13.1	*10.65*	*16.12*
Nth America	5,003	*0.5*	11	*0.4*	6	*0.3*	5	*0.6*	22.0	*10.97*	*39.36*
SthAmerica	4,689	*0.5*	3	*0.1*	3	*0.2*	0	*0.0*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	36	*1.4*	23	*1.2*	13	*1.6*	32.9	*23.03*	*45.53*
Unknown#	2,950	*0.3*	4	*0.2*	4	*0.2*	0	*0.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	197	*7.4*	138	*7.5*	59	*7.3*	27.8	*24.06*	*31.97*
Grampians	44,084	*4.4*	130	*4.9*	85	*4.6*	45	*5.6*	29.5	*24.74*	*35.12*
Loddon Mallee	63,104	*6.3*	154	*5.8*	107	*5.8*	47	*5.8*	24.4	*20.77*	*28.65*
Hume	51,348	*5.1*	149	*5.6*	116	*6.3*	33	*4.1*	29.0	*24.64*	*34.18*
Gippsland	56,679	*5.7*	149	*5.6*	110	*6.0*	39	*4.8*	26.3	*22.32*	*30.97*
Western Metro	126,711	*12.7*	350	*13.2*	251	*13.6*	99	*12.3*	27.6	*24.83*	*30.72*
Northern Metro	171,545	*17.1*	501	*18.9*	325	*17.6*	176	*21.8*	29.2	*26.65*	*31.76*
Eastern Metro	192,893	*19.3*	468	*17.7*	329	*17.9*	139	*17.2*	24.3	*22.06*	*26.46*
Southern Metro	214,947	*21.5*	523	*19.7*	360	*19.5*	163	*20.2*	24.3	*22.25*	*26.41*
Other	8,777	*0.9*	27	*1.0*	20	*1.1*	7	*0.9*	30.8	*20.27*	*44.91*
Unknown#	48	*0.0*	2	*0.1*	2	*0.1*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.17 Renal Agenesis and Dysgenesis

Figure 3.17 Renal Agenesis and Dysgenesis, Number of Cases by Year



	1983	1984	1985	1986	1987	1988	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998
Number	25	27	30	36	27	32	37	23	44	23	22	42	46	51	27	41
N/10,000	4.1	4.4	4.9	5.9	4.4	5.0	5.8	3.4	6.7	3.5	3.4	6.5	7.2	8.1	4.3	6.6

Renal Agenesis and Dysgenesis

British Paediatric Association code 753.00–753.01

Bilateral or unilateral absence of the kidneys or severe dysplasia.

Unilateral disease is very different from bilateral, but the figures here include both conditions.

- There are marked annual fluctuations in numbers of babies with renal agenesis, from over 40 to 20 cases per year. The overall birth prevalence has increased to 6.5 / 1000 in 1995–1998 from 4.8 / 1000 in 1983–1994.
- Some of the marked reduction in neonatal deaths and stillbirths may be due to the more severe cases being identified *in utero* and being represented amongst the increased proportion of terminations. Also, unilateral disease, compatible with life, may be more prevalent and account for the 54% surviving.
- There is a significant two-fold increased birth prevalence in males.
- In the earlier years, many of these babies were low birth weight, but in the recent study years, more than 50% had normal birth weights.
- There is no significant association with maternal age.
- Asian-born women have a significantly lower prevalence of babies with this condition, compared to Australian-born and Middle Eastern-born women.
- The differences in regional variation are not significant.

Table 3.17.1 Renal Agenesis and Dysgenesis, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**533**		**368**		**165**		**5.3**	***4.81***	***5.70***
Survived > 28 days	1,000,776	*98.6*	210	*39.4*	120	*32.6*	90	*54.5*	2.1		
Neonatal death	4,288	*0.4*	171	*32.1*	140	*38.0*	31	*18.8*			
Stillbirth	7,397	*0.7*	98	*18.4*	74	*20.1*	24	*14.5*			
Termination < 20 wks	2,402	*0.2*	54	*10.1*	34	*9.2*	20	*12.1*			
Sex											
Male	521,553	*51.4*	352	*66.0*	236	*64.1*	116	*70.3*	6.7	*6.1*	*7.50*
Female	492,558	*48.5*	152	*28.5*	111	*30.2*	41	*24.8*	3.1	*2.6*	*3.63*
Indeterminate	298	*0.0*	23	*4.3*	17	*4.6*	6	*3.6*			
Unknown#	454	*0.0*	6	*1.1*	4	*1.1*	2	*1.2*			
Plurality											
Singleton	987,675	*97.3*	505	*94.7*	349	*94.8*	156	*94.5*	5.1	*4.7*	*5.56*
Twin	26,192	*2.6*	28	*5.3*	19	*5.2*	9	*5.5*	10.7	*7.1*	*15.50*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**479**		**334**		**145**		**4.7**		
Birthweight											
< 1,000	6,784	*0.7*	64	*13.4*	39	*11.7*	25	*17.2*	94.3		
1,000–2,499	54,688	*5.4*	175	*36.5*	145	*43.4*	30	*20.7*	32.0		
2,500+	949,958	*93.8*	222	*46.3*	133	*39.8*	89	*61.4*	2.3		
Unknown	1,067	*0.1*	18	*3.8*	17	*5.1*	1	*0.7*			

95% of unknowns were in terminations before 20 weeks

Table 3.17.2 Patterns of Birth Defects, Renal Agenesis/Dysgenesis, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	166	31.1
Other Associations:		
• Chromosomal	32	6.0
• Other Same System (Urninary)	60	11.3
• Other Different Systems	275	51.6
Total	**533**	**100.0**

* Isolated cases may include cases with one of the following minor conditions or other minor conditions commonly associated with renal agenesis: pyloric stenosis, pulmonary hypoplasia, undescended testes or talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

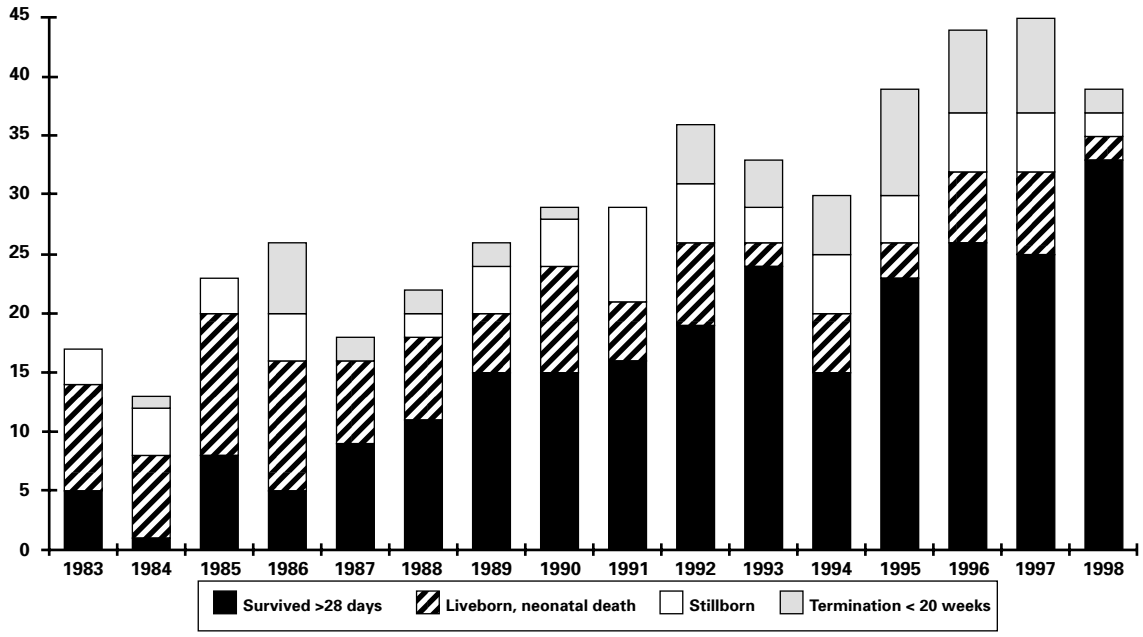
Table 3.17.3 Renal Agenesis and Dysgenesis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**533**		**368**		**165**		**5.3**	***4.87***	***5.77***
Maternal Age											
<20	40,400	*4.0*	30	*5.6*	23	*6.3*	7	*4.2*	7.4	5.01	*10.62*
20–24	185,901	*18.6*	112	*21.0*	83	*22.6*	29	*17.6*	6.0	4.98	*7.28*
25–29	367,374	*36.7*	175	*32.8*	122	*33.2*	53	*32.1*	4.8	4.10	*5.54*
30–34	289,879	*29.0*	156	*29.3*	114	*31.0*	42	*25.5*	5.4	4.59	*6.31*
35–39	101,575	*10.1*	46	*8.6*	18	*4.9*	28	*17.0*	4.5	3.31	*6.05*
40+	15,718	*1.6*	11	*2.1*	5	*1.4*	6	*3.6*	7.0	3.49	*12.53*
Unknown#	273	*0.0*	3	*0.6*	3	*0.8*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	386	*72.4*	256	*69.6*	130	*78.8*	5.1	4.63	*5.67*
Oceania inc NZ	20,620	*2.1*	9	*1.7*	4	*1.1*	5	*3.0*	4.4	2.00	*8.29*
UK inc Eire	50,596	*5.1*	23	*4.3*	16	*4.3*	7	*4.2*	4.5	2.88	*6.82*
Europe	58,931	*5.9*	36	*6.8*	33	*9.0*	3	*1.8*	6.1	4.28	*8.45*
Middle East	22,818	*2.3*	18	*3.4*	11	*3.0*	7	*4.2*	7.9	4.68	*12.46*
Asia	71,685	*7.2*	20	*3.8*	14	*3.8*	6	*3.6*	2.8	1.70	*4.30*
Nth America	5,003	*0.5*	4	*0.8*	1	*0.3*	3	*1.8*	8.0	2.17	*20.47*
Sth America	4,689	*0.5*	2	*0.4*	1	*0.3*	1	*0.6*	4.3	0.52	*15.40*
Africa	10,944	*1.1*	6	*1.1*	5	*1.4*	1	*0.6*	5.5	2.01	*11.95*
Unknown#	2,950	*0.3*	29	*5.4*	27	*7.3*	2	*1.2*	N/A		
Region											
Barwon S W	70,984	*7.1*	39	*7.3*	20	*5.4*	19	*11.5*	5.5	3.91	*7.51*
Grampians	44,084	*4.4*	20	*3.8*	10	*2.7*	10	*6.1*	4.5	2.77	*6.99*
Loddon Mallee	63,104	*6.3*	29	*5.4*	18	*4.9*	11	*6.7*	4.6	3.08	*6.62*
Hume	51,348	*5.1*	28	*5.3*	21	*5.7*	7	*4.2*	5.5	3.63	*7.91*
Gippsland	56,679	*5.7*	28	*5.3*	19	*5.2*	9	*5.5*	4.9	3.29	*7.16*
Western Metro	126,711	*12.7*	76	*14.3*	52	*14.1*	24	*14.5*	6.0	4.76	*7.55*
Northern Metro	171,545	*17.1*	102	*19.1*	75	*20.4*	27	*16.4*	5.9	4.87	*7.25*
Eastern Metro	192,893	*19.3*	89	*16.7*	69	*18.8*	20	*12.1*	4.6	3.73	*5.71*
Southern Metro	214,947	*21.5*	109	*20.5*	75	*20.4*	34	*20.6*	5.1	4.18	*6.14*
Other	8,777	*0.9*	11	*2.1*	8	*2.2*	3	*1.8*	12.5	6.25	*22.43*
Unknown#	48	*0.0*	2	*0.4*	1	*0.3*	1	*0.6*	N/A		

95% of unknowns were in terminations before 20 weeks

3.18 Cystic Kidney Disease

Figure 3.18 Cystic Kidney Disease, Number of Cases by Year



Number	17	13	23	26	18	22	26	29	29	36	33	30	39	44	45	39
N/10,000	2.8	2.1	3.8	4.2	2.9	3.5	4.0	4.3	4.4	5.4	5.1	4.6	6.1	7.0	7.2	6.3

Cystic Kidney Disease

British Paediatric Association code 753.10–753.18

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

- There has been a linear increase in overall birth prevalence of babies with this condition, from 4.0 / 1000 in 1983–1994 to 6.5 / 1000 in 1995–1998. The number of liveborn babies that survive at least 28 days has increased from 47% to 64%. This may be due to increased ascertainment of milder cases. There are now over 35 cases reported each year.
- Again there is an excess of males with renal disease, but in this condition, it is not significant.
- There is no significant association with maternal age, country of birth nor region of residence.

Table 3.18.1 Cystic Kidney Disease, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**469**		**302**		**167**		**4.6**	***4.20***	***5.04***
Survived > 28 days	1,000,776	*98.6*	250	*53.3*	143	*47.4*	107	*64.1*	2.5		
Neonatal death	4,288	*0.4*	104	*22.2*	86	*28.5*	18	*10.8*			
Stillbirth	7,397	*0.7*	61	*13.0*	45	*14.9*	16	*9.6*			
Termination < 20 wks	2,402	*0.2*	54	*11.5*	28	*9.3*	26	*15.6*			
Sex											
Male	521,553	*51.4*	283	*60.3*	179	*59.3*	104	*62.3*	5.4	*4.8*	*6.10*
Female	492,558	*48.5*	178	*38.0*	118	*39.1*	60	*35.9*	3.6	*3.1*	*4.20*
Indeterminate	298	*0.0*	3	*0.6*	0	*0.0*	3	*1.8*			
Unknown#	454	*0.0*	5	*1.1*	5	*1.7*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	445	*94.9*	286	*94.7*	159	*95.2*	4.5	*4.1*	*4.92*
Twin	26,192	*2.6*	23	*4.9*	15	*5.0*	8	*4.8*	8.8	*5.6*	*13.17*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	1	*0.2*	1	*0.3*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**415**		**274**		**141**		**4.1**		
Birthweight											
< 1,000	6,784	*0.7*	45	*10.8*	28	*10.2*	17	*12.1*	66.3		
1,000–2,499	54,688	*5.4*	95	*22.9*	68	*24.8*	27	*19.1*	17.4		
2,500+	949,958	*93.8*	259	*62.4*	162	*59.1*	97	*68.8*	2.7		
Unknown	1,067	*0.1*	16	*3.9*	16	*5.8*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.18.2 Patterns of Birth Defects, Cystic Kidney, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	179	38.2
Other Associations:		
• Chromosomal	35	7.5
• Other Same System (Urninary)	71	15.1
• Other Different Systems	184	39.2
Total	**469**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, pulmonary hypoplasia or talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

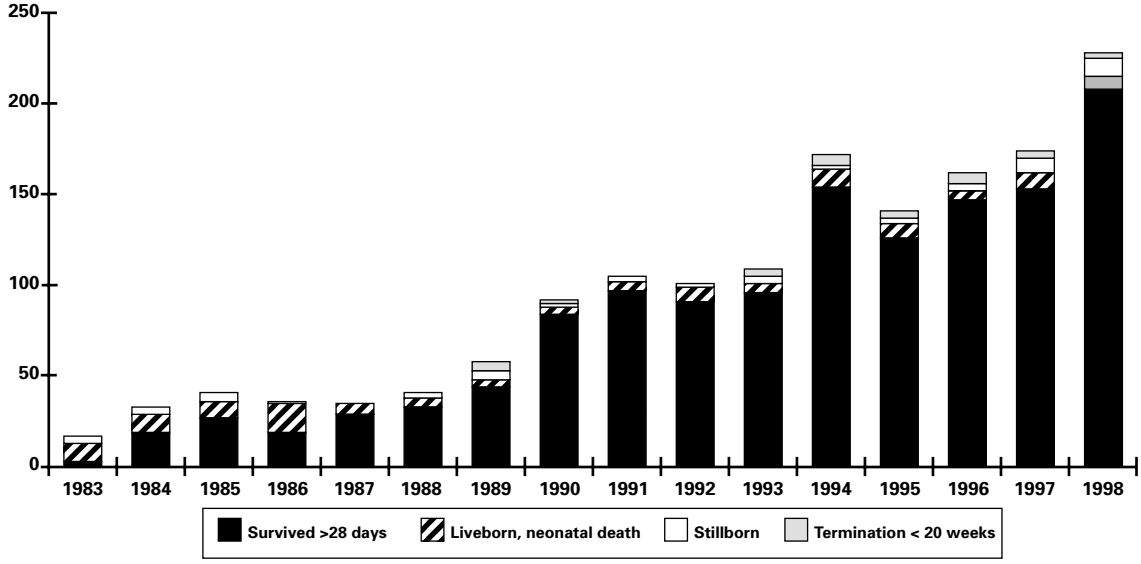
Table 3.18.3 Cystic Kidney Disease, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**468**		**301**		**167**		**4.7**	***4.26***	***5.09***
Maternal Age											
<20	40,400	*4.0*	21	*4.5*	16	*5.3*	5	*3.0*	5.2	*3.22*	*7.95*
20–24	185,901	*18.6*	79	*16.9*	50	*16.6*	29	*17.4*	4.2	*3.39*	*5.32*
25–29	367,374	*36.7*	171	*36.5*	121	*40.2*	50	*29.9*	4.7	*3.99*	*5.42*
30–34	289,879	*29.0*	134	*28.6*	110	*36.5*	24	*14.4*	4.6	*3.89*	*5.49*
35–39	101,575	*10.1*	50	*10.7*	45	*15.0*	5	*3.0*	4.9	*3.65*	*6.50*
40+	15,718	*1.6*	6	*1.3*	6	*2.0*	0	*0.0*	3.8	*1.40*	*8.32*
Unknown#	273	*0.0*	7	*1.5*	7	*2.3*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	353	*75.4*	227	*75.4*	126	*75.4*	4.7	*4.22*	*5.21*
Oceania Inc NZ	20,620	*2.1*	8	*1.7*	4	*1.3*	4	*2.4*	3.9	*1.67*	*7.64*
UK inc Eire	50,596	*5.1*	22	*4.7*	13	*4.3*	9	*5.4*	4.3	*2.73*	*6.57*
Europe	58,931	*5.9*	24	*5.1*	17	*5.6*	7	*4.2*	4.1	*2.61*	*6.07*
Middle East	22,818	*2.3*	8	*1.7*	7	*2.3*	1	*0.6*	3.5	*1.51*	*6.91*
Asia	71,685	*7.2*	22	*4.7*	11	*3.7*	11	*6.6*	3.1	*1.92*	*4.63*
Nth America	5,003	*0.5*	1	*0.2*	0	*0.0*	1	*0.6*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	3	*0.6*	1	*0.3*	2	*1.2*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	3	*0.6*	1	*0.3*	2	*1.2*	2.7	*0.56*	*8.00*
Unknown#	2,950	*0.3*	24	*5.1*	20	*6.6*	4	*2.4*	N/A		
Region											
Barwon S W	70,984	*7.1*	26	*5.6*	21	*7.0*	5	*3.0*	3.7	*2.39*	*5.38*
Grampians	44,084	*4.4*	23	*4.9*	15	*5.0*	8	*4.8*	5.2	*3.31*	*7.83*
Loddon Mallee	63,104	*6.3*	35	*7.5*	19	*6.3*	16	*9.6*	5.5	*3.87*	*7.71*
Hume	51,348	*5.1*	27	*5.8*	19	*6.3*	8	*4.8*	5.3	*3.47*	*7.68*
Gippsland	56,679	*5.7*	20	*4.3*	12	*4.0*	8	*4.8*	3.5	*2.16*	*5.43*
Western Metro	126,711	*12.7*	73	*15.6*	51	*16.9*	22	*13.2*	5.8	*4.55*	*7.29*
Northern Metro	171,545	*17.1*	95	*20.3*	56	*18.6*	39	*23.4*	5.5	*4.50*	*6.80*
Eastern Metro	192,893	*19.3*	84	*17.9*	58	*19.3*	26	*15.6*	4.4	*3.49*	*5.42*
Southern Metro	214,947	*21.5*	78	*16.7*	45	*15.0*	33	*19.8*	3.6	*2.89*	*4.55*
Other	8,777	*0.9*	6	*1.3*	4	*1.3*	2	*1.2*	6.8	*2.51*	*14.90*
Unknown#	48	*0.0*	1	*0.2*	1	*0.3*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.19 Obstructive Defects of Renal Pelvis

Figure 3.19 Obstructive Defects of Renal Pelvis, Number of Cases by Year



Number	17	33	41	36	35	41	58	92	105	101	109	172	141	162	174	228
N/10,000	2.8	5.4	6.7	5.9	5.7	6.4	9.0	13.8	16.1	15.2	16.8	26.5	22.1	25.7	27.9	36.7

Obstructive Defects of Renal Pelvis

British Paediatric Association code 753.20 – 753.29

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

- Ultrasound in pregnancy has identified a large number of babies with this condition. Where possible, this diagnosis is confirmed postnatally, before inclusion in the BDR. A huge increase was seen in 1994 and again in 1998, when over 210 cases were reported. The overall birth prevalence has increased three-fold from 10.5 / 1000 in 1983–1994 to 28.0 / 1000 in 1995–1998, and, specifically 36.7 / 1000 in 1998.
- There is nearly a 3-fold increased risk for males compared with females. This difference is apparent even when analysing the data year by year. The Table below shows that an excess of males was not caused by recent diagnoses in male babies less than one year of age; males are often investigated for this condition after only one urinary tract infection, while females may have several before such investigation.

Year	Male	Female
1983	11	5
1984	25	7
1985	28	10
1986	27	8
1987	23	12
1988	32	9
1989	35	15
1990	66	18
1991	69	29
1992	61	25
1993	68	27
1994	113	35
1995	101	39
1996	124	38
1997	130	44
1998	**167**	**60**

- In 1995–1998 only 8% died in the perinatal period, suggesting that association with lethal malformations is uncommon.
- Most babies (83.4%) are normal birth weight.
- The higher rate in babies of women aged 40 years and over, compared with all other ages, is not significant.
- Women born in the Middle East have a significantly higher prevalence of babies with this condition, compared to Australian-, UK- , European and Asian-born women.
- There are significantly more births of babies with this condition to women living in Northern Metro region compared to those living in Eastern Metro. Other differences are not significant.

Table 3.19.1 Obstructive Defects of Renal Pelvis, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**1,545**		**840**		**705**		**15.2**	***14.47***	***15.98***
Survived > 28 days	1,000,776	*98.6*	1,330	*86.1*	696	*82.9*	634	*89.9*	13.3		
Neonatal death	4,288	*0.4*	121	*7.8*	92	*11.0*	29	*4.1*			
Stillbirth	7,397	*0.7*	60	*3.9*	35	*4.2*	25	*3.5*			
Termination < 20 wks	2,402	*0.2*	34	*2.2*	17	*2.0*	17	*2.4*			
Sex											
Male	521,553	*51.4*	1,124	*72.8*	602	*71.7*	522	*74.0*	21.6	*20.3*	*22.81*
Female	492,558	*48.5*	411	*26.6*	230	*27.4*	181	*25.7*	8.3	*7.5*	*9.15*
Indeterminate	298	*0.0*	7	*0.5*	5	*0.6*	2	*0.3*			
Unknown#	454	*0.0*	3	*0.2*	3	*0.4*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	1,479	*95.7*	797	*94.9*	682	*96.7*	15.0	*14.2*	*15.74*
Twin	26,192	*2.6*	65	*4.2*	43	*5.1*	22	*3.1*	24.8	*19.3*	*31.84*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	1	*0.1*	0	*0.0*	1	*0.1*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**1,511**		**823**		**688**		**14.9**		
Birthweight											
< 1,000	6,784	*0.7*	56	*3.7*	21	*2.6*	35	*5.1*	82.5		
1,000–2,499	54,688	*5.4*	177	*11.7*	114	*13.9*	63	*9.2*	32.4		
2,500+	949,958	*93.8*	1,267	*83.9*	679	*82.5*	588	*85.5*	13.3		
Unknown	1,067	*0.1*	11	*0.7*	9	*1.1*	2	*0.3*			

95% of unknowns were in terminations before 20 weeks

Table 3.19.2 Patterns of Birth Defects, Obstructive Defects of Renal Pelvis, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	950	61.5
Other Associations:		
• Chromosomal	79	5.1
• Other Same System (Urinary)	208	13.5
• Other Different Systems	308	19.9
Total	**1,545**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, single umbilical artery or talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

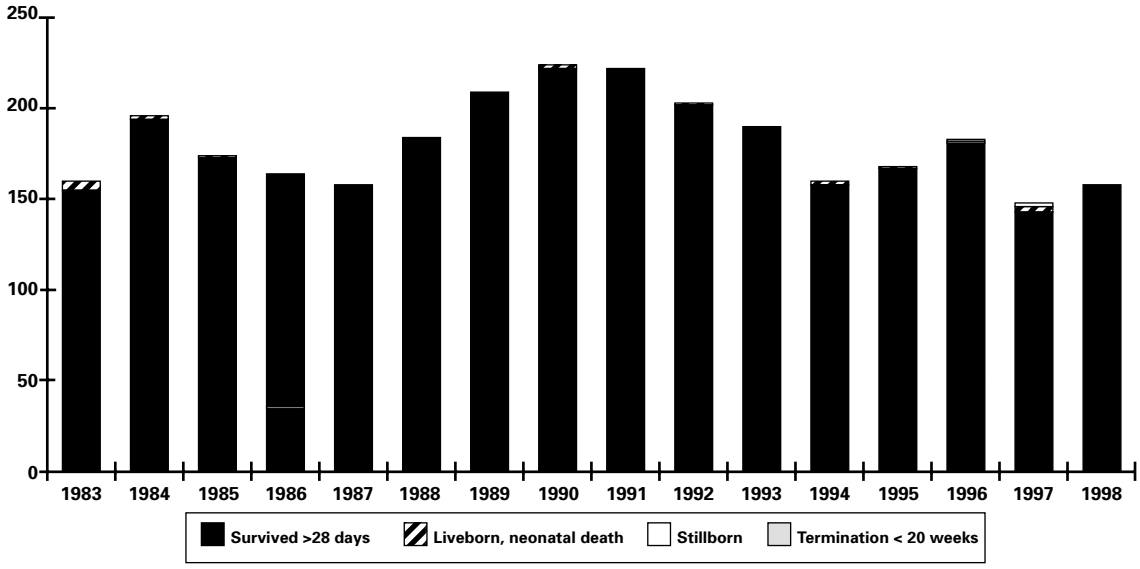
Table 3.19.3 Obstructive Defects of Renal Pelvis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**1,538**		**835**		**703**		**15.4**	***14.61***	***16.12***
Maternal Age											
<20	40,400	*4.0*	63	*4.1*	33	*4.0*	30	*4.3*	15.6	*12.09*	*20.09*
20–24	185,901	*18.6*	254	*16.5*	148	*17.7*	106	*15.1*	13.7	*12.06*	*15.48*
25–29	367,374	*36.7*	557	*36.2*	319	*38.2*	238	*33.9*	15.2	*13.90*	*16.42*
30–34	289,879	*29.0*	445	*28.9*	238	*28.5*	207	*29.4*	15.4	*13.93*	*16.78*
35–39	101,575	*10.1*	178	*11.6*	75	*9.0*	103	*14.7*	17.5	*15.09*	*20.35*
40+	15,718	*1.6*	33	*2.1*	20	*2.4*	13	*1.8*	21.0	*14.44*	*29.52*
Unknown#	273	*0.0*	8	*0.5*	2	*0.2*	6	*0.9*	N/A		
Country of birth											
Australia	753,424	*75.3*	1,120	*72.8*	620	*74.3*	500	*71.1*	14.9	*14.00*	*15.74*
Oceania inc NZ	20,620	*2.1*	44	*2.9*	21	*2.5*	23	*3.3*	21.3	*15.49*	*28.68*
UK inc Eire	50,596	*5.1*	72	*4.7*	47	*5.6*	25	*3.6*	14.2	*11.21*	*18.03*
Europe	58,931	*5.9*	80	*5.2*	44	*5.3*	36	*5.1*	13.6	*10.83*	*16.98*
Middle East	22,818	*2.3*	58	*3.8*	24	*2.9*	34	*4.8*	25.4	*19.47*	*33.09*
Asia	71,685	*7.2*	109	*7.1*	46	*5.5*	63	*9.0*	15.2	*12.54*	*18.41*
Nth America	5,003	*0.5*	8	*0.5*	3	*0.4*	5	*0.7*	16.0	*6.89*	*31.50*
Sth America	4,689	*0.5*	11	*0.7*	5	*0.6*	6	*0.9*	23.5	*11.71*	*41.99*
Africa	10,944	*1.1*	21	*1.4*	13	*1.6*	8	*1.1*	19.2	*11.88*	*29.36*
Unknown#	2,950	*0.3*	15	*1.0*	13	*1.6*	2	*0.3*	N/A		
Region											
Barwon S W	70,984	*7.1*	122	*7.9*	68	*8.1*	54	*7.7*	17.2	*14.33*	*20.59*
Grampians	44,084	*4.4*	70	*4.6*	49	*5.9*	21	*3.0*	15.9	*12.46*	*20.18*
Loddon Mallee	63,104	*6.3*	87	*5.7*	43	*5.1*	44	*6.3*	13.8	*11.11*	*17.10*
Hume	51,348	*5.1*	81	*5.3*	46	*5.5*	35	*5.0*	15.8	*12.60*	*19.72*
Gippsland	56,679	*5.7*	67	*4.4*	46	*5.5*	21	*3.0*	11.8	*9.23*	*15.11*
Western Metro	126,711	*12.7*	191	*12.4*	103	*12.3*	88	*12.5*	15.1	*13.04*	*17.41*
Northern Metro	171,545	*17.1*	302	*19.6*	149	*17.8*	153	*21.8*	17.6	*15.70*	*19.73*
Eastern Metro	192,893	*19.3*	256	*16.6*	153	*18.3*	103	*14.7*	13.3	*11.72*	*15.02*
Southern Metro	214,947	*21.5*	333	*21.7*	165	*19.8*	168	*23.9*	15.5	*13.90*	*17.27*
Other	8,777	*0.9*	28	*1.8*	12	*1.4*	16	*2.3*	31.9	*21.21*	*46.26*
Unknown#	48	0.0	1	*0.1*	1	*0.1*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.20 Congenital Dislocation of Hip

Figure 3.20 Congenital Dislocation of Hip, Number of Cases by Year



Number	160	196	174	164	158	185	209	224	222	203	190	160	168	183	148	158
N/10,000	26.4	32.3	28.4	26.8	25.7	29.1	32.5	33.5	34.0	30.6	29.3	24.6	26.4	29.1	23.8	25.4

Congenital Dislocation of the Hip

British Paediatric Association code 754.30

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 BDR.
- This is one of the more commonly reported conditions, the birth prevalence of which has remained at over 150 cases per year since 1983 (except for 1997 when there were 148 cases). There has been a decline in prevalence from 29.4 / 1000 in 1983–1994 to 26.1 / 1000 in 1995–1998.
- There is almost no associated stillbirth or neonatal death.
- Female babies have a four-fold increased risk and a prevalence of 46.0 / 1000 births.
- This condition is very seldom seen in twins.
- This condition is seldom seen in babies weighing less than 2500 gram which suggests that CDH occurs late in pregnancy secondary to reduction in space.
- There are some significant associations with maternal age:
  – There is a reduced risk for women aged 20 –24 years compared to older women up to 40 years
  – There are significantly fewer babies with CDH born to women 40 years and over compared to women 25–34 years.
- A significantly lower birth prevalence of babies with CDH was seen in Oceanic-, Middle Eastern-, Asian-, European-, South American-born women, compared to Australian- and UK-born women.
- Women living in Western Metro and Northern Metro regions have the lowest birth prevalences of babies with CDH and these figures are both significantly lower than those of women from all other regions. Women living in Eastern Metro have significantly fewer babies with this condition than women from Southern Metro and Grampians region (the two regions with the highest prevalence).

Table 3.20.1 Congenital Dislocation of Hip, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**2,902**		**2245**		**657**		**28.6**	***27.56***	***29.63***
Survived > 28 days	1,000,776	*98.6*	2,879	*99.2*	2,230	*99.3*	649	*98.8*	28.8		
Neonatal death	4,288	*0.4*	18	*0.6*	14	*0.6*	4	*0.6*			
Stillbirth	7,397	*0.7*	4	*0.1*	1	*0.0*	3	*0.5*			
Termination < 20 wks	2,402	*0.2*	1	*0.0*	0	*0.0*	1	*0.2*			
Sex											
Male	521,553	*51.4*	636	*21.9*	475	*21.2*	161	*24.5*	12.2	*11.2*	*13.14*
Female	492,558	*48.5*	2,264	*78.0*	1,768	*78.8*	496	*75.5*	46.0	*44.1*	*47.85*
Indeterminate	298	*0.0*	2	*0.1*	2	*0.1*	0	*0.0*			
Unknown#	454	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
Plurality											
Singleton	987,675	*97.3*	2,882	*99.3*	2,230	*99.3*	652	*99.2*	29.2	*28.1*	*30.24*
Twin	26,192	*2.6*	20	*0.7*	15	*0.7*	5	*0.8*	7.6	*4.7*	*11.76*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**2,901**		**2245**		**656**		**28.7**		
Birthweight											
< 1,000	6,784	*0.7*	6	*0.2*	3	*0.1*	3	*0.5*	8.8		
1,000–2,499	54,688	*5.4*	111	*3.8*	73	*3.3*	38	*5.8*	20.3		
2,500+	949,958	*93.8*	2,783	*95.9*	2,168	*96.6*	615	*93.8*	29.3		
Unknown	1,067	*0.1*	1	*0.0*	1	*0.0*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.20.2 Patterns of Birth Defects, Congenital Dislocation of Hip, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	2,684	92.5
Other Associations:		
• Chromosomal	24	0.8
• Other Same System (Musculoskeletal)	55	1.9
• Other Different Systems	139	4.8
Total	**2,902**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: talipes or inguinal hernia. If a case has two or more of these conditions it is classified as a multiple system defect.

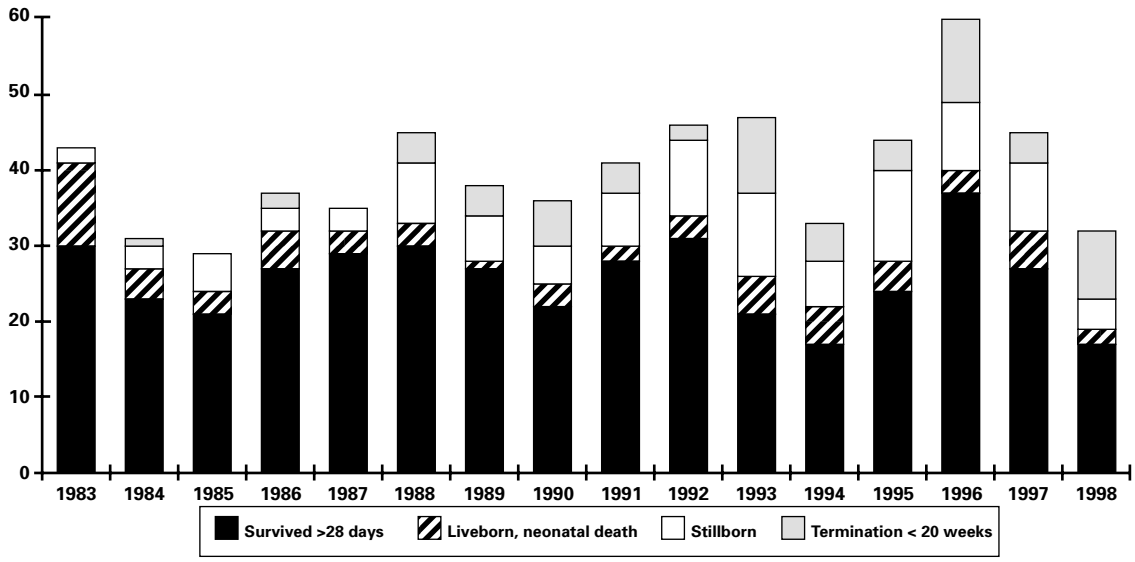
Table 3.20.3 Congenital Dislocation of Hip, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**2,901**		**2,244**		**657**		**29.0**	***27.94***	***30.02***
Maternal Age											
<20	40,400	*4.0*	99	*3.4*	78	*3.5*	21	*3.2*	24.5	*20.02*	*29.97*
20–24	185,901	*18.6*	447	*15.4*	371	*16.5*	76	*11.6*	24.0	*21.82*	*26.27*
25–29	367,374	*36.7*	1,129	*38.9*	915	*40.8*	214	*32.6*	30.7	*28.94*	*32.52*
30–34	289,879	*29.0*	880	*30.3*	645	*28.7*	235	*35.8*	30.4	*28.36*	*32.36*
35–39	101,575	*10.1*	310	*10.7*	215	*9.6*	95	*14.5*	30.5	*27.25*	*34.15*
40+	15,718	*1.6*	29	*1.0*	20	*0.9*	9	*1.4*	18.5	*12.36*	*26.57*
Unknown#	273	*0.0*	7	*0.2*	0	*0.0*	7	*1.1*	N/A		
Country of birth											
Australia	753,424	*75.3*	2,354	*81.1*	1,838	*81.9*	516	*78.5*	31.2	*29.98*	*32.50*
Oceania inc NZ	20,620	*2.1*	42	*1.4*	30	*1.3*	12	*1.8*	20.4	*14.67*	*27.54*
UK inc Eire	50,596	*5.1*	174	*6.0*	139	*6.2*	35	*5.3*	34.4	*29.54*	*40.00*
Europe	58,931	*5.9*	124	*4.3*	103	*4.6*	21	*3.2*	21.0	*17.57*	*25.19*
Middle East	22,818	*2.3*	42	*1.4*	31	*1.4*	11	*1.7*	18.4	*13.25*	*24.89*
Asia	71,685	*7.2*	114	*3.9*	69	*3.1*	45	*6.8*	15.9	*13.18*	*19.18*
Nth America	5,003	*0.5*	18	*0.6*	12	*0.5*	6	*0.9*	36.0	*21.34*	*56.85*
Sth America	4,689	*0.5*	4	*0.1*	3	*0.1*	1	*0.2*	8.5	*2.32*	*21.84*
Africa	10,944	*1.1*	23	*0.8*	15	*0.7*	8	*1.2*	21.0	*13.32*	*31.52*
Unknown#	2,950	*0.3*	6	*0.2*	4	*0.2*	2	*0.3*	N/A		
Region											
Barwon S W	70,984	*7.1*	237	*8.2*	201	*9.0*	36	*5.5*	33.4	*29.35*	*38.00*
Grampians	44,084	*4.4*	157	*5.4*	137	*6.1*	20	*3.0*	35.6	*30.34*	*41.78*
Loddon Mallee	63,104	*6.3*	203	*7.0*	169	*7.5*	34	*5.2*	32.2	*27.95*	*36.99*
Hume	51,348	*5.1*	146	*5.0*	112	*5.0*	34	*5.2*	28.4	*24.08*	*33.55*
Gippsland	56,679	*5.7*	188	*6.5*	145	*6.5*	43	*6.5*	33.2	*28.66*	*38.34*
Western Metro	126,711	*12.7*	269	*9.3*	186	*8.3*	83	*12.6*	21.2	*18.81*	*23.97*
Northern Metro	171,545	*17.1*	382	*13.2*	293	*13.1*	89	*13.5*	22.3	*20.11*	*24.65*
Eastern Metro	192,893	*19.3*	523	*18.0*	397	*17.7*	126	*19.2*	27.1	*24.79*	*29.43*
Southern Metro	214,947	*21.5*	759	*26.2*	579	*25.8*	180	*27.4*	35.3	*32.80*	*37.82*
Other	8,777	*0.9*	36	*1.2*	24	*1.1*	12	*1.8*	41.0	*28.71*	*56.77*
Unknown#	48	*0.0*	1	*0.0*	1	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.21 Limb Reduction Defects

Figure 3.21 Limb Reduction Defects, Number of Cases by Year



Number	43	31	29	37	35	45	38	36	41	46	47	33	44	60	45	32
N/10,000	7.1	5.1	4.7	6.0	5.7	7.1	5.9	5.4	6.3	6.9	7.3	5.1	6.9	9.5	7.2	5.2

Limb Reduction Defects

British Paediatric Association code 755.20–755.49

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.

- 30–40 cases of limb reduction defects are reported to the BDR per year, except for a peak in 1996 when 60 cases were notified. There are an increasing number of pregnancies terminated when this defect is recognised in a fetus. It is often associated with other malformations as suggested by the figure of approximately 26% dying in the perinatal period.
- There is no excess of one or other sex.
- There is a significantly increased prevalence in twins.
- There are no significant associations with maternal age, country of birth nor region of residence.

Table 3.21.1 Limb Reduction Defects, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**642**		**461**		**181**		**6.3**	***5.84***	***6.82***
Survived > 28 days	1,000,776	*98.6*	411	*64.0*	306	*66.4*	105	*58.0*	4.1		
Neonatal death	4,288	*0.4*	62	*9.7*	48	*10.4*	14	*7.7*			
Stillbirth	7,397	*0.7*	103	*16.0*	69	*15.0*	34	*18.8*			
Termination < 20 wks	2,402	*0.2*	66	*10.3*	38	*8.2*	28	*15.5*			
Sex											
Male	521,553	*51.4*	344	*53.6*	243	*52.7*	101	*55.8*	6.6	*5.9*	*7.34*
Female	492,558	*48.5*	278	*43.3*	207	*44.9*	71	*39.2*	5.6	*5.0*	*6.36*
Indeterminate	298	*0.0*	14	*2.2*	9	*2.0*	5	*2.8*			
Unknown#	454	*0.0*	6	*0.9*	2	*0.4*	4	*2.2*			
Plurality											
Singleton	987,675	*97.3*	611	*95.2*	444	*96.3*	167	*92.3*	6.2	*5.7*	*6.68*
Twin	26,192	*2.6*	28	*4.4*	16	*3.5*	12	*6.6*	10.7	*7.1*	*15.50*
Triplet	939	*0.1*	3	*0.5*	1	*0.2*	2	*1.1*	31.9	*6.6*	*93.29*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**576**		**423**		**153**		**5.7**		
Birthweight											
< 1,000	6,784	*0.7*	77	*13.4*	44	*10.4*	33	*21.6*	113.5		
1,000–2,499	54,688	*5.4*	122	*21.2*	85	*20.1*	37	*24.2*	22.3		
2,500+	949,958	*93.8*	370	*64.2*	288	*68.1*	82	*53.6*	3.9		
Unknown	1,067	*0.1*	7	*1.2*	6	*1.4*	1	*0.7*	N/A		

95% of unknowns were in terminations before 20 weeks

Table 3.21.2 Patterns of Birth Defects, Limb Reduction Defects, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	262	40.8
Other Associations:		
• Chromosomal	56	8.7
• Other Same System (Musculoskeletal)	92	14.3
• Other Different Systems	232	36.1
Total	**642**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: talipes. If a case has two or more of these conditions it is classified as a multiple system defect.

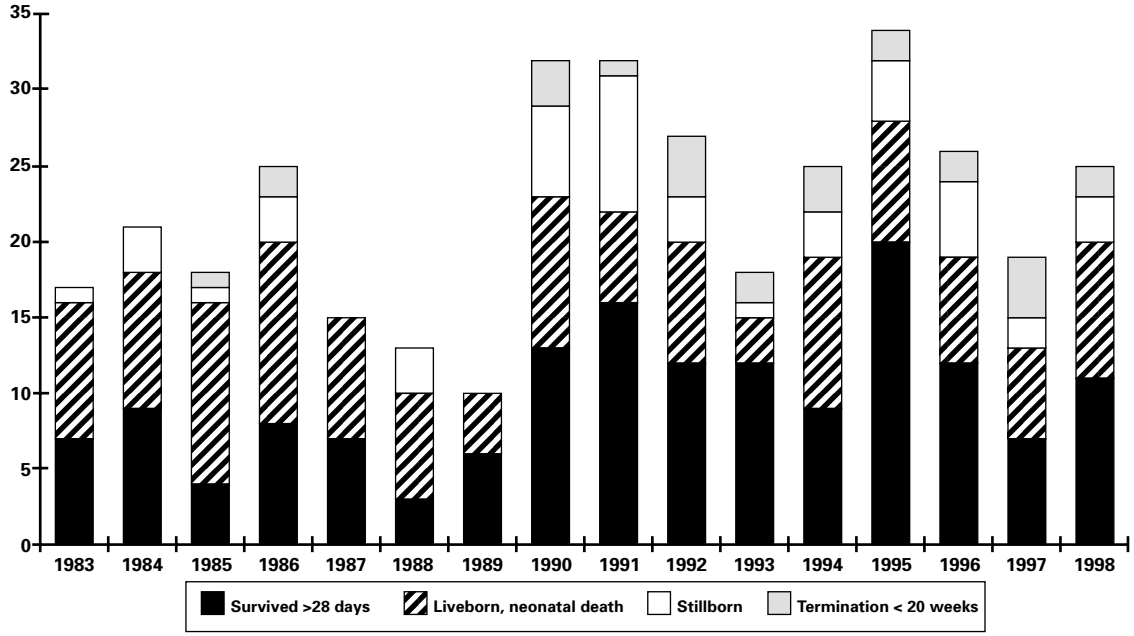
Table 3.21.3 Limb Reduction Defects, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**642**		**461**		**181**		**6.4**	***5.92***	***6.90***
Maternal Age											
<20	40,400	*4.0*	30	*4.7*	22	*4.8*	8	*4.4*	7.4	*5.01*	*10.62*
20–24	185,901	*18.6*	115	*17.9*	90	*19.5*	25	*13.8*	6.2	*5.13*	*7.45*
25–29	367,374	*36.7*	230	*35.8*	178	*38.6*	52	*28.7*	6.3	*5.49*	*7.14*
30–34	289,879	*29.0*	177	*27.6*	122	*26.5*	55	*30.4*	6.1	*5.26*	*7.10*
35–39	101,575	*10.1*	67	*10.4*	35	*7.6*	32	*17.7*	6.6	*5.15*	*8.43*
40+	15,718	*1.6*	18	*2.8*	11	*2.4*	7	*3.9*	11.5	*6.79*	*18.09*
Unknown#	273	*0.0*	5	*0.8*	3	*0.7*	2	*1.1*	N/A		
Country of birth											
Australia	753,424	*75.3*	455	*70.9*	333	*72.2*	122	*67.4*	6.0	*5.48*	*6.59*
Oceania inc NZ	20,620	*2.1*	11	*1.7*	7	*1.5*	4	*2.2*	5.3	*2.66*	*9.55*
UK inc Eire	50,596	*5.1*	40	*6.2*	25	*5.4*	15	*8.3*	7.9	*5.64*	*10.75*
Europe	58,931	*5.9*	42	*6.5*	33	*7.2*	9	*5.0*	7.1	*5.13*	*9.64*
Middle East	22,818	*2.3*	15	*2.3*	8	*1.7*	7	*3.9*	6.6	*3.68*	*10.85*
Asia	71,685	*7.2*	42	*6.5*	25	*5.4*	17	*9.4*	5.9	*4.22*	*7.92*
Nth America	5,003	*0.5*	1	*0.2*	1	*0.2*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	1	*0.2*	0	*0.0*	1	*0.6*	2.1	*0.05*	*11.88*
Africa	10,944	*1.1*	3	*0.5*	2	*0.4*	1	*0.6*	2.7	*0.56*	*8.00*
Unknown#	2,950	*0.3*	32	*5.0*	27	*5.9*	5	*2.8*	N/A		
Region											
Barwon S W	70,984	*7.1*	51	*7.9*	36	*7.8*	15	*8.3*	7.2	*5.40*	*9.53*
Grampians	44,084	*4.4*	26	*4.0*	15	*3.3*	11	*6.1*	5.9	*3.85*	*8.67*
Loddon Mallee	63,104	*6.3*	37	*5.8*	25	*5.4*	12	*6.6*	5.9	*4.13*	*8.08*
Hume	51,348	*5.1*	32	*5.0*	28	*6.1*	4	*2.2*	6.2	*4.26*	*8.81*
Gippsland	56,679	*5.7*	26	*4.0*	19	*4.1*	7	*3.9*	4.6	*3.00*	*6.74*
Western Metro	126,711	*12.7*	87	*13.6*	62	*13.4*	25	*13.8*	6.9	*5.53*	*8.51*
Northern Metro	171,545	*17.1*	116	*18.1*	85	*18.4*	31	*17.1*	6.8	*5.61*	*8.14*
Eastern Metro	192,893	*19.3*	124	*19.3*	92	*20.0*	32	*17.7*	6.4	*5.37*	*7.69*
Southern Metro	214,947	*21.5*	128	*19.9*	90	*19.5*	38	*21.0*	6.0	*4.99*	*7.10*
Other	8,777	*0.9*	13	*2.0*	8	*1.7*	5	*2.8*	14.8	*7.88*	*25.33*
Unknown#	48	*0.0*	2	*0.3*	1	*0.2*	1	*0.6*	N/A		

95% of unknowns were in terminations before 20 weeks

3.22 Diaphragmatic Hernia

Figure 3.22 Diaphragmatic Hernia, Number of Cases by Year



Number	17	21	18	25	15	13	10	32	32	27	18	25	34	26	19	25
N/10,000	2.8	3.5	2.9	4.1	2.4	2.0	1.6	4.8	4.9	4.1	2.8	3.9	5.3	4.1	3.0	4.0

Diaphragmatic Hernia

British Paediatric Association code 756.61

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

- In 1990 and 1991 there was a marked increase in the number of births where this defect was present (primarily stillbirths and terminations). Another marked fluctuation was observed in 1995 when the birth prevalence was 5.3/1000. The overall birth prevalence for 1995–1998 is 4.1/1000 compared to 3.3/1000 for 1983–1994.
- The annual proportion that have been liveborn, but died as neonates, has declined from 39% to 29% in 1995–1998. This may reflect identification of the problem prenatally by ultrasound and preparation for acute management at the time of birth, resulting in better neonatal outcome.
- There is no difference between male and female prevalence.
- The apparent increasing birth prevalence with advancing maternal age is not significant.
- There is no significant maternal country of birth nor regional association.

Table 3.22.1 Congenital Diaphragmatic Hernia, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**357**		**253**		**104**		**3.5**	***3.17***	***3.91***
Survived > 28 days	1,000,776	*98.6*	156	*43.7*	106	*41.9*	50	*48.1*	1.6		
Neonatal death	4,288	*0.4*	128	*35.9*	98	*38.7*	30	*28.8*			
Stillbirth	7,397	*0.7*	47	*13.2*	33	*13.0*	14	*13.5*			
Termination < 20 wks	2,402	*0.2*	26	*7.3*	16	*6.3*	10	*9.6*			
Sex											
Male	521,553	*51.4*	183	*51.3*	128	*50.6*	55	*52.9*	3.5	*3.0*	*4.07*
Female	492,558	*48.5*	170	*47.6*	122	*48.2*	48	*46.2*	3.5	*3.0*	*4.02*
Indeterminate	298	*0.0*	3	*0.8*	3	*1.2*	0	*0.0*			
Unknown#	454	*0.0*	1	*0.3*	0	*0.0*	1	*1.0*			
Plurality											
Singleton	987,675	*97.3*	349	*97.8*	248	*98.0*	101	*97.1*	3.5	*3.2*	*3.93*
Twin	26,192	*2.6*	8	*2.2*	5	*2.0*	3	*2.9*	3.1	*1.3*	*6.02*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**331**		**237**		**94**		**3.3**		
Birthweight											
< 1,000	6,784	*0.7*	39	*11.8*	20	*8.4*	19	*20.2*	57.5		
1,000–2,499	54,688	*5.4*	75	*22.7*	52	*21.9*	23	*24.5*	13.7		
2,500+	949,958	*93.8*	184	*55.6*	135	*57.0*	49	*52.1*	1.9		
Unknown	1,067	*0.1*	33	*10.0*	30	*12.7*	3	*3.2*			

95% of unknowns were in terminations before 20 weeks

Table 3.22.2 Patterns of Birth Defects, Diaphragmatic Hernia, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	173	43.5
Other Associations:		
• Chromosomal	32	9.0
• Other Same System (Musculoskeletal)	5	1.4
• Other Different Systems	147	41.2
Total	**357**	**100.0**

* Isolated cases may include cases with one of the following minor conditions or conditions commonly associated with congenital diaphragmatic hernia: inguinal hernia or pulmonary hypoplasia. If a case has two or more of these conditions it is classified as a multiple system defect.

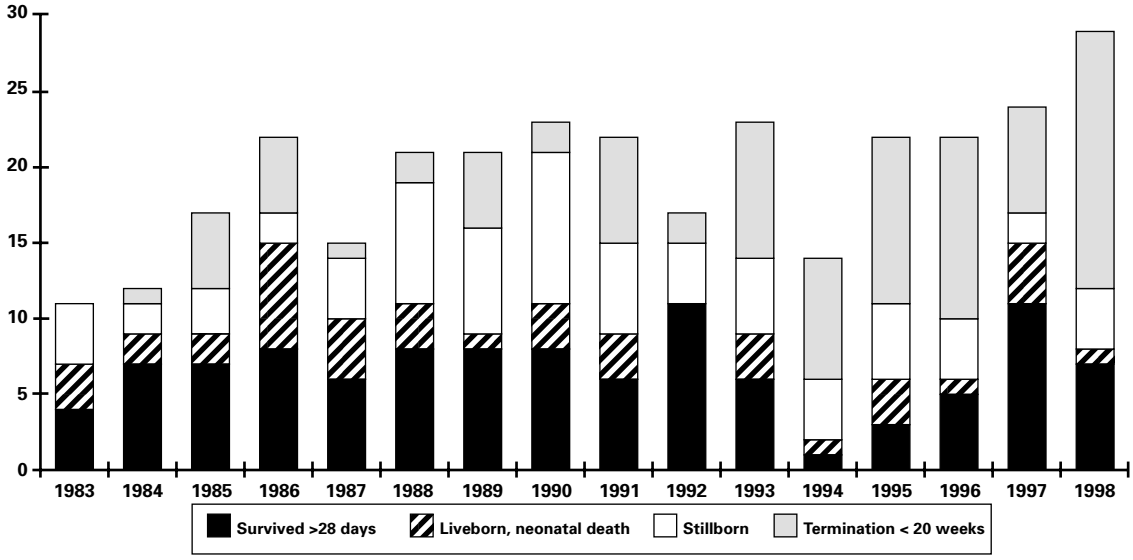
Table 3.22.3 Congenital Diaphragmatic Hernia, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**357**		**253**		**104**		**3.6**	***3.21***	***3.96***
Maternal Age											
<20	40,400	*4.0*	10	*2.8*	7	*2.8*	3	*2.9*	2.5	*1.19*	*4.55*
20–24	185,901	*18.6*	68	*19.0*	51	*20.2*	17	*16.3*	3.7	*2.86*	*4.67*
25–29	367,374	*36.7*	118	*33.1*	88	*34.8*	30	*28.8*	3.2	*2.67*	*3.86*
30–34	289,879	*29.0*	100	*28.0*	70	*27.7*	30	*28.8*	3.4	*2.82*	*4.22*
35–39	101,575	*10.1*	51	*14.3*	31	*12.3*	20	*19.2*	5.0	*3.78*	*6.66*
40+	15,718	*1.6*	8	*2.2*	4	*1.6*	4	*3.8*	5.1	*2.19*	*10.03*
Unknown#	273	*0.0*	2	*0.6*	2	*0.8*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	256	*71.7*	173	*68.4*	83	*79.8*	3.4	*3.00*	*3.85*
Oceania inc NZ	20,620	*2.1*	6	*1.7*	2	*0.8*	4	*3.8*	2.9	*1.07*	*6.34*
UK inc Eire	50,596	*5.1*	26	*7.3*	20	*7.9*	6	*5.8*	5.1	*3.36*	*7.55*
Europe	58,931	*5.9*	20	*5.6*	19	*7.5*	1	*1.0*	3.4	*2.07*	*5.23*
Middle East	22,818	*2.3*	6	*1.7*	3	*1.2*	3	*2.9*	2.6	*0.97*	*5.73*
Asia	71,685	*7.2*	18	*5.0*	13	*5.1*	5	*4.8*	2.5	*1.49*	*3.97*
Nth America	5,003	*0.5*	1	*0.3*	1	*0.4*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	4	*1.1*	3	*1.2*	1	*1.0*	8.5	*2.32*	*21.84*
Africa	10,944	*1.1*	7	*2.0*	7	*2.8*	0	*0.0*	6.4	*2.56*	*13.18*
Unknown#	2,950	*0.3*	13	*3.6*	12	*4.7*	1	*1.0*	N/A		
Region											
Barwon S W	70,984	*7.1*	21	*5.9*	14	*5.5*	7	*6.7*	3.0	*1.83*	*4.53*
Grampians	44,084	*4.4*	21	*5.9*	14	*5.5*	7	*6.7*	4.8	*2.95*	*7.29*
Loddon Mallee	63,104	*6.3*	26	*7.3*	21	*8.3*	5	*4.8*	4.1	*2.69*	*6.06*
Hume	51,348	*5.1*	20	*5.6*	16	*6.3*	4	*3.8*	3.9	*2.38*	*6.00*
Gippsland	56,679	*5.7*	21	*5.9*	16	*6.3*	5	*4.8*	3.7	*2.29*	*5.67*
Western Metro	126,711	*12.7*	50	*14.0*	36	*14.2*	14	*13.5*	3.9	*2.93*	*5.21*
Northern Metro	171,545	*17.1*	59	*16.5*	42	*16.6*	17	*16.3*	3.4	*2.64*	*4.47*
Eastern Metro	192,893	*19.3*	58	*16.2*	38	*15.0*	20	*19.2*	3.0	*2.30*	*3.91*
Southern Metro	214,947	*21.5*	74	*20.7*	51	*20.2*	23	*22.1*	3.4	*2.72*	*4.35*
Other	8,777	*0.9*	7	*2.0*	5	*2.0*	2	*1.9*	8.0	*3.20*	*16.43*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.23 Exomphalos

Figure 3.23 Exomphalos, Number of Cases by Year



Number	11	12	17	22	15	21	21	23	22	17	23	14	22	22	24	29
N/10,000	1.8	2.0	2.8	3.6	2.4	3.3	3.3	3.4	3.4	2.6	3.6	2.2	3.5	3.5	3.9	4.7

Exomphalos

British Paediatric Association code 756.70

Herniation of abdominal contents through umbilical insertion and covered by membrane which may or may not remain intact.

- Overall birth prevalence of exomphalos has gradually increased from 2.9 / 1000 to 3.8 / 1000 in 1995–1998, with prevalence in 1998 being 4.7 / 1000. In 1995–1998, there was a termination of pregnancy in 48.5% of cases.
- There is a 13% neonatal death rate, indicating the more severe nature of the condition compared with the other abdominal wall defect, gastroschisis, which has a 4% neonatal death rate.
- There is a significant increased prevalence in twin pregnancies.
- The significantly higher prevalence in women 35 years and older may reflect the association with trisomy 18. In 26% of cases there is a chromosomal abnormality.
- There are no significant associations with maternal country of birth nor region of residence.

Table 3.23.1 Exomphalos, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**315**		**218**		**97**		**3.1**	***2.77***	***3.47***
Survived > 28 days	1,000,776	*98.6*	105	*33.3*	80	*36.7*	25	*25.8*	1.0		
Neonatal death	4,288	*0.4*	42	*13.3*	32	*14.7*	10	*10.3*			
Stillbirth	7,397	*0.7*	74	*23.5*	59	*27.1*	15	*15.5*			
Termination < 20 wks	2,402	*0.2*	94	*29.8*	47	*21.6*	47	*48.5*			
Sex											
Male	521,553	*51.4*	154	*48.9*	115	*52.8*	39	*40.2*	3.0	*2.5*	*3.47*
Female	492,558	*48.5*	135	*42.9*	88	*40.4*	47	*48.5*	2.7	*2.3*	*3.26*
Indeterminate	298	*0.0*	13	*4.1*	10	*4.6*	3	*3.1*			
Unknown#	454	*0.0*	13	*4.1*	5	*2.3*	8	*8.2*			
Plurality											
Singleton	987,675	*97.3*	294	*93.3*	201	*92.2*	93	*95.9*	3.0	*2.6*	*3.34*
Twin	26,192	*2.6*	20	*6.3*	16	*7.3*	4	*4.1*	7.6	*4.7*	*11.76*
Triplet	939	*0.1*	1	*0.3*	1	*0.5*	0	*0.0*	10.6	*0.3*	*59.32*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**221**		**171**		**50**		**2.2**		
Birthweight											
< 1,000	6,784	*0.7*	49	*22.2*	36	*21.1*	13	*26.0*	72.2		
1,000–2,499	54,688	*5.4*	70	*31.7*	51	*29.8*	19	*38.0*	12.8		
2,500+	949,958	*93.8*	87	*39.4*	69	*40.1*	18	*36.0*	0.9		
Unknown	1,067	*0.1*	15	*6.8*	15	*8.8*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

Table 3.23.2 Patterns of Birth Defects, Exomphalos, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	85	27.0
Other Associations:		
• Chromosomal	81	25.7
• Other Same System (Musculoskeletal)	9	2.9
• Other Different Systems	140	44.4
Total	**315**	**100.0**

* Isolated cases may include cases with one of the following minor conditions: inguinal hernia, micrognathia or undescended testes. If a case has two or more of these conditions it is classified as a multiple system defect.

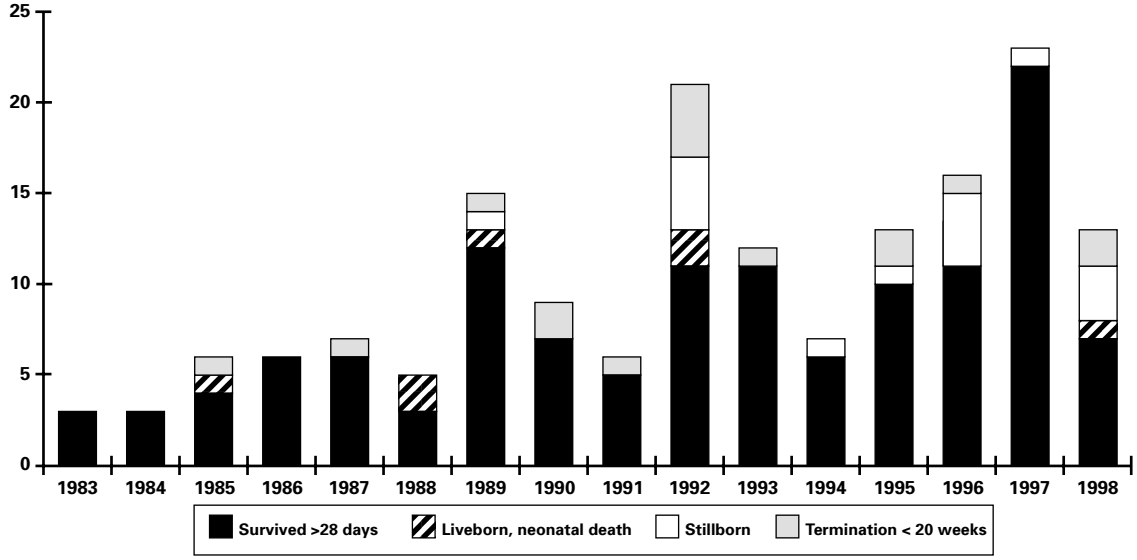
Table 3.23.3 Exomphalos, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**315**		**218**		**97**		**3.1**	***2.81***	***3.52***
Maternal Age											
<20	40,400	*4.0*	15	4.8	12	*5.5*	3	*3.1*	3.7	*2.08*	*6.13*
20–24	185,901	*18.6*	50	15.9	40	*18.3*	10	*10.3*	2.7	*2.00*	*3.55*
25–29	367,374	*36.7*	89	28.3	61	*28.0*	28	*28.9*	2.4	*1.96*	*3.00*
30–34	289,879	*29.0*	84	26.7	61	*28.0*	23	*23.7*	2.9	*2.32*	*3.61*
35–39	101,575	*10.1*	53	16.8	29	*13.3*	24	*24.7*	5.2	*3.94*	*6.88*
40+	15,718	*1.6*	15	4.8	9	*4.1*	6	*6.2*	9.5	*5.34*	*15.75*
Unknown#	273	*0.0*	9	2.9	6	*2.8*	3	*3.1*	N/A		
Country of birth											
Australia	753,424	*75.3*	210	66.7	140	*64.2*	70	*72.2*	2.8	*2.43*	*3.20*
Oceania inc NZ	20,620	*2.1*	6	1.9	2	*0.9*	4	*4.1*	2.9	*1.07*	*6.34*
UK inc Eire	50,596	*5.1*	10	3.2	8	*3.7*	2	*2.1*	2.0	*0.95*	*3.64*
Europe	58,931	*5.9*	17	5.4	11	*5.0*	6	*6.2*	2.9	*1.68*	*4.62*
Middle East	22,818	*2.3*	7	2.2	7	*3.2*	0	*0.0*	3.1	*1.23*	*6.32*
Asia	71,685	*7.2*	26	8.3	16	*7.3*	10	*10.3*	3.6	*2.37*	*5.33*
Nth America	5,003	*0.5*	0	0.0	0	*0.0*	0	*0.0*	0.0		
Sth America	4,689	*0.5*	1	0.3	1	*0.5*	0	*0.0*	2.1	*0.05*	*11.88*
Africa	10,944	*1.1*	3	1.0	1	*0.5*	2	*2.1*	2.7	*0.56*	*8.00*
Unknown#	2,950	*0.3*	35	11.1	32	*14.7*	3	*3.1*	N/A		
Region											
Barwon S W	70,984	*7.1*	19	6.0	10	*4.6*	9	*9.3*	2.7	*1.61*	*4.18*
Grampians	44,084	*4.4*	10	3.2	9	*4.1*	1	*1.0*	2.3	*1.09*	*4.17*
Loddon Mallee	63,104	*6.3*	25	7.9	22	*10.1*	3	*3.1*	4.0	*2.56*	*5.86*
Hume	51,348	*5.1*	17	5.4	14	*6.4*	3	*3.1*	3.3	*1.93*	*5.30*
Gippsland	56,679	*5.7*	19	6.0	17	*7.8*	2	*2.1*	3.4	*2.02*	*5.23*
Western Metro	126,711	*12.7*	36	11.4	23	*10.6*	13	*13.4*	2.8	*1.99*	*3.93*
Northern Metro	171,545	*17.1*	58	18.4	42	*19.3*	16	*16.5*	3.4	*2.59*	*4.40*
Eastern Metro	192,893	*19.3*	61	19.4	43	*19.7*	18	*18.6*	3.2	*2.44*	*4.09*
Southern Metro	214,947	*21.5*	65	20.6	35	*16.1*	30	*30.9*	3.0	*2.35*	*3.88*
Other	8,777	*0.9*	5	1.6	3	*1.4*	2	*2.1*	5.7	*1.85*	*13.27*
Unknown#	48	*0.0*	0	0.0	0	*0.0*	0	*0.0*	N/A		

* 95% of unknowns were in terminations before 20 weeks.

3.24 Gastroschisis

Figure 3.24 Gastroschisis, Number of Cases by Year



Number	3	3	6	6	7	5	15	9	6	21	12	7	13	16	23	13
N/10,000	0.5	0.5	1.0	1.0	1.1	0.8	2.3	1.3	0.9	3.2	1.9	1.1	2.0	2.5	3.7	2.1

Gastroschisis

British Paediatric Association code 756.71

Visceral herniation through an abdominal wall defect, lateral to an intact umbilical cord.

- The increase in reported prevalence of this rare condition in 1992 may have been a chance fluctuation or may have represented a true change. The finding that the 1994 numbers were again low reduced the urgency to investigate this increase. However, from 1995–1997, there was another increase, also reported in other Australian States and overseas.
- There have been small numbers of pregnancy terminations for this condition.
- Small proportions are stillborn (9%) or die in the neonatal period (4%). This is not often associated with a major malformation.
- Many (56%) of these babies are born with birth weight less than 2500 gms.
- There is an inverse association with maternal age, with a significant seven-fold increased prevalence in teenage mothers. A number of international studies are underway to examine this unusually high risk. There have been no cases born to the 15,718 women more than 39 years of age.
- There are no maternal country of birth nor regional associations.

Table 3.24.1 Gastroschisis, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**165**		**100**		**65**		**1.6**	***1.39***	***1.90***
Survived > 28 days	1,000,776	*98.6*	127	*77.0*	77	*77.0*	50	*76.9*	1.3		
Neonatal death	4,288	*0.4*	7	*4.2*	6	*6.0*	1	*1.5*			
Stillbirth	7,397	*0.7*	15	*9.1*	6	*6.0*	9	*13.8*			
Termination < 20 wks	2,402	*0.2*	16	*9.7*	11	*11.0*	5	*7.7*			
Sex											
Male	521,553	*51.4*	89	*53.9*	54	*54.0*	35	*53.8*	1.7	*1.4*	*2.11*
Female	492,558	*48.5*	67	*40.6*	39	*39.0*	28	*43.1*	1.4	*1.1*	*1.74*
Indeterminate	298	*0.0*	5	*3.0*	4	*4.0*	1	*1.5*			
Unknown#	454	*0.0*	4	*2.4*	3	*3.0*	1	*1.5*			
Plurality											
Singleton	987,675	*97.3*	162	*98.2*	98	*98.0*	64	*98.5*	1.6	*1.4*	*1.92*
Twin	26,192	*2.6*	3	*1.8*	2	*2.0*	1	*1.5*	1.1	*0.2*	*3.34*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**149**		**89**		**60**		**1.5**		
Birthweight											
< 1,000	6,784	*0.7*	12	*8.1*	4	*4.5*	8	*13.3*	17.7		
1,000–2,499	54,688	*5.4*	71	*47.7*	44	*49.4*	27	*45.0*	13.0		
2,500+	949,958	*93.8*	53	*35.6*	31	*34.8*	22	*36.7*	0.6		
Unknown	1,067	*0.1*	13	*8.7*	10	*11.2*	3	*5.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.24.2 Patterns of Birth Defects, Gastroschisis, 1983–1998

Type	Number	Per Cent
Isolated anomaly*	111	67.3
Other Associations:		
• Chromosomal	3	1.8
• Other Same System (Musculoskeletal)	1	0.6
• Other Different Systems	50	30.3
Total	**165**	**100.0**

* Isolated cases may include cases with one of the following minor condition: undescended testes. If a case has two or more of these conditions it is classified as a multiple system defect.

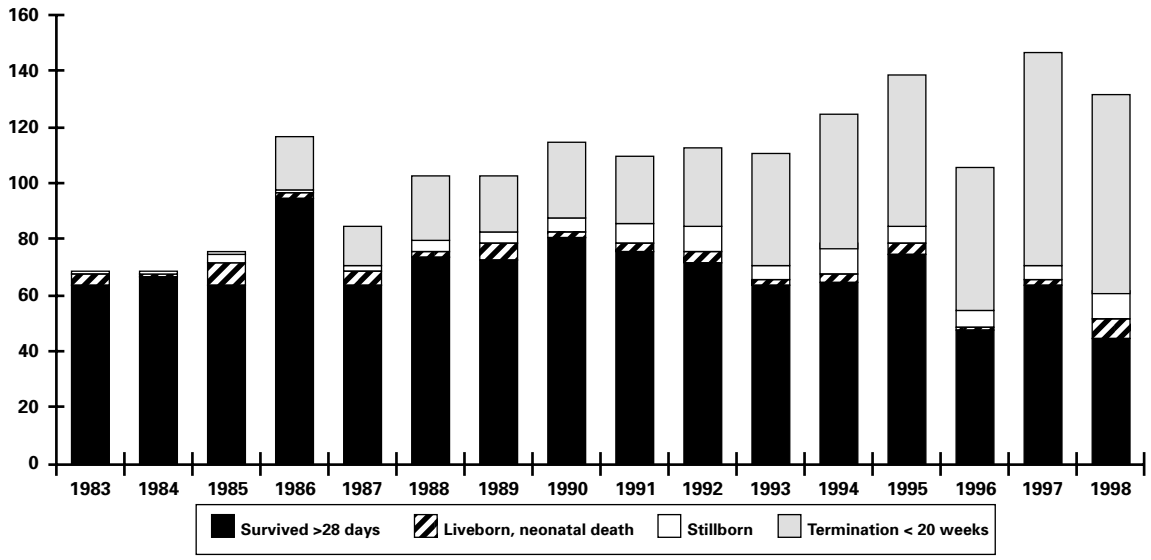
Table 3.24.3 Gastroschisis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**165**		**100**		**65**		**1.6**	***1.41***	***1.93***
Maternal Age											
<20	40,400	*4.0*	33	*20.0*	16	*16.0*	17	*26.2*	8.2	*5.62*	*11.48*
20–24	185,901	*18.6*	57	*34.5*	33	*33.0*	24	*36.9*	3.1	*2.34*	*4.00*
25–29	367,374	*36.7*	44	*26.7*	28	*28.0*	16	*24.6*	1.2	*0.87*	*1.61*
30–34	289,879	*29.0*	26	*15.8*	20	*20.0*	6	*9.2*	0.9	*0.59*	*1.32*
35–39	101,575	*10.1*	4	*2.4*	2	*2.0*	2	*3.1*	0.4	*0.11*	*1.01*
40+	15,718	*1.6*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Unknown#	273	*0.0*	1	*0.6*	1	*1.0*	0	*0.0*	N/A		
Country of birth											
Australia	753,424	*75.3*	127	*77.0*	75	*75.0*	52	*80.0*	1.7	*1.41*	*2.01*
Oceania inc NZ	20,620	*2.1*	3	*1.8*	2	*2.0*	1	*1.5*	1.5	*0.30*	*4.25*
UK inc Eire	50,596	*5.1*	9	*5.5*	4	*4.0*	5	*7.7*	1.8	*0.81*	*3.38*
Europe	58,931	*5.9*	3	*1.8*	3	*3.0*	0	*0.0*	0.5	*0.10*	*1.49*
Middle East	22,818	*2.3*	2	*1.2*	0	*0.0*	2	*3.1*	0.9	*0.11*	*3.16*
Asia	71,685	*7.2*	6	*3.6*	3	*3.0*	3	*4.6*	0.8	*0.31*	*1.82*
Nth America	5,003	*0.5*	1	*0.6*	1	*1.0*	0	*0.0*	2.0	*0.05*	*11.13*
Sth America	4,689	*0.5*	2	*1.2*	1	*1.0*	1	*1.5*	4.3	*0.52*	*15.40*
Africa	10,944	*1.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Unknown#	2,950	*0.3*	12	*7.3*	11	*11.0*	1	*1.5*	N/A		
Region											
Barwon S W	70,984	*7.1*	14	*8.5*	5	*5.0*	9	*13.8*	2.0	*1.08*	*3.31*
Grampians	44,084	*4.4*	9	*5.5*	4	*4.0*	5	*7.7*	2.0	*0.94*	*3.88*
Loddon Mallee	63,104	*6.3*	8	*4.8*	4	*4.0*	4	*6.2*	1.3	*0.55*	*2.50*
Hume	51,348	*5.1*	12	*7.3*	7	*7.0*	5	*7.7*	2.3	*1.21*	*4.09*
Gippsland	56,679	*5.7*	9	*5.5*	5	*5.0*	4	*6.2*	1.6	*0.73*	*3.02*
Western Metro	126,711	*12.7*	22	*13.3*	17	*17.0*	5	*7.7*	1.7	*1.09*	*2.62*
Northern Metro	171,545	*17.1*	24	*14.5*	16	*16.0*	8	*12.3*	1.4	*0.90*	*2.08*
Eastern Metro	192,893	*19.3*	27	*16.4*	15	*15.0*	12	*18.5*	1.4	*0.92*	*2.04*
Southern Metro	214,947	*21.5*	38	*23.0*	25	*25.0*	13	*20.0*	1.8	*1.25*	*2.43*
Other	8,777	*0.9*	2	*1.2*	2	*2.0*	0	*0.0*	2.3	*0.28*	*8.23*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.25 Trisomy 21

Figure 3.25 Trisomy 21, Number of Cases by Year



Number	69	69	76	117	85	103	103	115	110	113	111	127	139	106	147	133
N/10,000	11.4	11.4	12.4	19.1	13.8	16.2	16.0	17.2	16.9	17.0	17.1	19.6	21.8	16.8	23.6	21.4

Trisomy 21

British Paediatric Association code 758.00–758.09

Down syndrome—additional chromosome 21.

- This is the most common of the autosomal aneuploidies with an overall prevalence from 1983–1998 of 16.9 / 10,000 (1 in 592). The prevalence in the last four years was 20.8 / 1000.
- The number of reported trisomy 21 pregnancies (terminations plus births) has increased over the 16 years. This is because the proportion of older women (>35 years of age) giving birth has increased and more fetuses are being detected with this condition as a result of more prenatal diagnosis taking place. In addition there is more prenatal detection by maternal serum screening and ultrasound in women of all ages.
- There is a significant excess of males.
- There is no association with twinning.
- The expected association with maternal age is seen. From 1983–1994, 62% of the babies with trisomy 21 were born to women who were less than 35 years of age, but in the last four years (1995–1998), only 44% were identified in younger women. This shift in proportions is probably due to affected fetuses, that would once have spontaneously aborted and never been identified as affected, now being identified by prenatal testing in older women in the first and early second trimester.
- The overall prevalence for women 35–39 years is approximately 1 in 200 and for women 40 years and over it is 1 in 66.
- There is no significant association with mother's country of birth nor region of residence.

Table 3.25.1 Trisomy 21, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**1,720**		**1,195**		**525**		**16.9**	***16.15***	***17.75***
Survived > 28 days	1,000,776	*98.6*	1,088	*63.3*	856	*71.6*	232	*44.2*	10.9		
Neonatal death	4,288	*0.4*	56	*3.3*	42	*3.5*	14	*2.7*			
Stillbirth	7,397	*0.7*	79	*4.6*	52	*4.4*	27	*5.1*			
Termination < 20 wks	2,402	*0.2*	497	*28.9*	245	*20.5*	252	*48.0*			
Sex											
Male	521,553	*51.4*	926	*53.8*	662	*55.4*	264	*50.3*	17.8	*16.6*	*18.90*
Female	492,558	*48.5*	709	*41.2*	508	*42.5*	201	*38.3*	14.4	*13.3*	*15.45*
Indeterminate	298	*0.0*	11	*0.6*	7	*0.6*	4	*0.8*			
Unknown#	454	*0.0*	74	*4.3*	18	*1.5*	56	*10.7*			
Plurality											
Singleton	987,675	*97.3*	1,668	*97.0*	1156	*96.7*	512	*97.5*	16.9	*16.1*	*17.70*
Twin	26,192	*2.6*	48	*2.8*	36	*3.0*	12	*2.3*	18.3	*13.5*	*24.34*
Triplet	939	*0.1*	4	*0.2*	3	*0.3*	1	*0.2*	42.6	*11.6*	*109.05*
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**1,223**		**950**		**273**		**12.1**		
Birthweight											
< 1,000	6,784	*0.7*	61	*5.0*	30	*3.2*	31	*11.4*	89.9		
1,000–2,499	54,688	*5.4*	222	*18.2*	176	*18.5*	46	*16.8*	40.6		
2,500+	949,958	*93.8*	922	*75.4*	728	*76.6*	194	*71.1*	9.7		
Unknown	1,067	*0.1*	18	*1.5*	16	*1.7*	2	*0.7*			

95% of unknowns were in terminations before 20 weeks

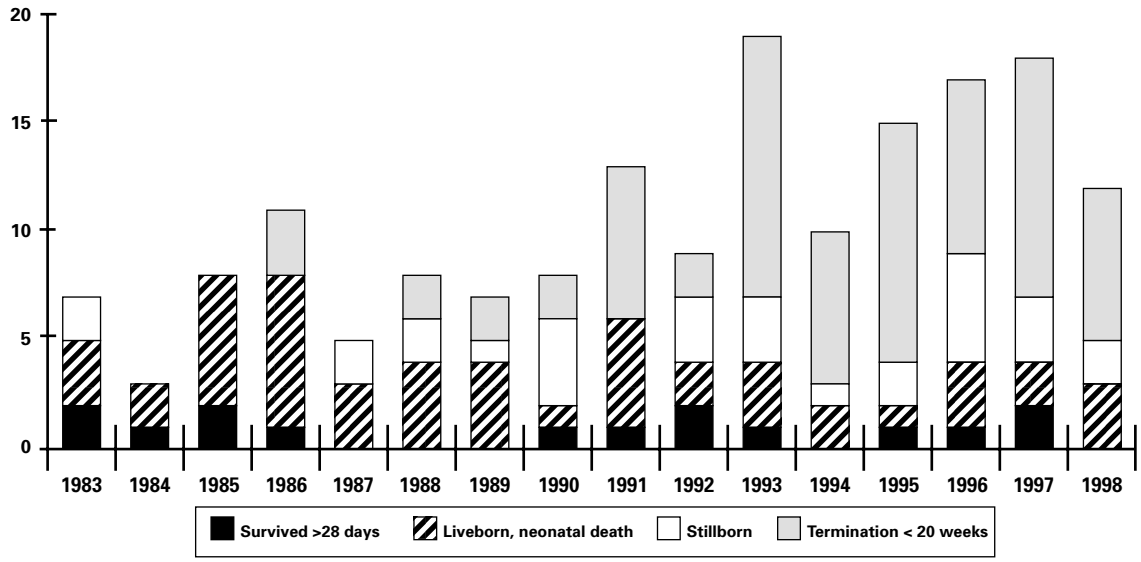
Table 3.25.2 Trisomy 21, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**1,713**		**1,188**		**525**		**17.1**	***16.38***	***17.84***
Maternal Age											
<20	40,400	*4.0*	29	*1.7*	24	*2.0*	5	*1.0*	7.2	*4.81*	*10.34*
20–24	185,901	*18.6*	146	*8.5*	117	*9.8*	29	*5.5*	7.9	*6.65*	*9.27*
25–29	367,374	*36.7*	310	*18.1*	249	*21.0*	61	*11.6*	8.4	*7.54*	*9.44*
30–34	289,879	*29.0*	453	*26.4*	324	*27.3*	129	*24.6*	15.6	*14.19*	*17.07*
35–39	101,575	*10.1*	485	*28.3*	306	*25.8*	179	*34.1*	47.7	*43.50*	*51.99*
40+	15,718	*1.6*	237	*13.8*	130	*10.9*	107	*20.4*	150.8	*132.54*	*171.59*
Unknown#	273	*0.0*	51	*3.0*	36	*3.0*	15	*2.9*	N/A		
Country of birth											
Australia	753,424	*75.3*	1,089	*63.6*	735	*61.9*	354	*67.4*	14.5	*13.60*	*15.31*
Oceania inc NZ	20,620	*2.1*	30	*1.8*	18	*1.5*	12	*2.3*	14.5	*9.82*	*20.81*
UK inc Eire	50,596	*5.1*	69	*4.0*	47	*4.0*	22	*4.2*	13.6	*10.69*	*17.37*
Europe	58,931	*5.9*	105	*6.1*	80	*6.7*	25	*4.8*	17.8	*14.65*	*21.67*
Middle East	22,818	*2.3*	33	*1.9*	20	*1.7*	13	*2.5*	14.5	*9.95*	*20.33*
Asia	71,685	*7.2*	114	*6.7*	77	*6.5*	37	*7.0*	15.9	*13.18*	*19.18*
Nth America	5,003	*0.5*	8	*0.5*	5	*0.4*	3	*0.6*	16.0	*6.89*	*31.50*
Sth America	4,689	*0.5*	8	*0.5*	7	*0.6*	1	*0.2*	17.1	*7.35*	*33.61*
Africa	10,944	*1.1*	18	*1.1*	14	*1.2*	4	*0.8*	16.4	*9.75*	*25.99*
Unknown#	2,950	*0.3*	239	*14.0*	185	*15.6*	54	*10.3*	N/A		
Region											
Barwon S W	70,984	*7.1*	100	*5.8*	73	*6.1*	27	*5.1*	14.1	*11.52*	*17.22*
Grampians	44,084	*4.4*	61	*3.6*	49	*4.1*	12	*2.3*	13.8	*10.67*	*17.91*
Loddon Mallee	63,104	*6.3*	95	*5.5*	77	*6.5*	18	*3.4*	15.1	*12.24*	*18.49*
Hume	51,348	*5.1*	87	*5.1*	58	*4.9*	29	*5.5*	16.9	*13.66*	*21.01*
Gippsland	56,679	*5.7*	80	*4.7*	63	*5.3*	17	*3.2*	14.1	*11.26*	*17.66*
Western Metro	126,711	*12.7*	207	*12.1*	146	*12.3*	61	*11.6*	16.3	*14.21*	*18.75*
Northern Metro	171,545	*17.1*	332	*19.4*	212	*17.8*	120	*22.9*	19.4	*17.36*	*21.58*
Eastern Metro	192,893	*19.3*	339	*19.8*	236	*19.9*	103	*19.6*	17.6	*15.78*	*19.58*
Southern Metro	214,947	*21.5*	367	*21.4*	244	*20.5*	123	*23.4*	17.1	*15.38*	*18.94*
Other	8,777	*0.9*	26	*1.5*	13	*1.1*	13	*2.5*	29.6	*19.34*	*43.55*
Unknown#	48	*0.0*	19	*1.1*	17	*1.4*	2	*0.4*	N/A		

95% of unknowns were in terminations before 20 weeks

3.26 Trisomy 13

Figure 3.26 Trisomy 13, Number of Cases by Year



Number	7	3	8	11	5	8	7	8	13	9	19	10	15	17	18	12
N/10,000	1.2	0.5	1.3	1.8	0.8	1.3	1.1	1.2	2.0	1.4	2.9	1.5	2.4	2.7	2.9	1.9

Trisomy 13

British Paediatric Association code 758.10–758.19

Patau syndrome—additional chromosome 13

- This is the least common of the autosomal trisomies compatible with life and has a total birth prevalence of 1.7/ 10,000. As with trisomy 21, there has been a marked increase in prevalence in the last four years, compared to the prevalence for 1983–1994, due to an increase in the number of fetuses detected in utero and pregnancies terminated.
- Only 8.8%, or 15 cases in the 16 year period, survived the neonatal period.
- There is no significant difference in male and female prevalence.
- There is the expected trend with advancing maternal age. Women 35 years and older have a significantly increased risk of having a baby with this condition compared with women less than 35 years. For women 40 years and older, the risk of having a baby with this condition is almost the same as that for a 30–34 year old woman having a baby with trisomy 21.
- There are no associations with mother's country of birth nor region of residence.

Table 3.26.1 Trisomy 13, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**170**		**108**		**62**		**1.7**	***1.44***	***1.95***
Survived > 28 days	1,000,776	*98.6*	15	*8.8*	11	*10.2*	4	*6.5*	0.1		
Neonataldeath	4,288	*0.4*	51	*30.0*	42	*38.9*	9	*14.5*			
Stillbirth	7,397	*0.7*	30	*17.6*	18	*16.7*	12	*19.4*			
Termination < 20 wks	2,402	*0.2*	74	*43.5*	37	*34.3*	37	*59.7*			
Sex											
Male	521,553	*51.4*	78	*45.9*	55	*50.9*	23	*37.1*	1.5	*1.2*	*1.88*
Female	492,558	*48.5*	78	*45.9*	47	*43.5*	31	*50.0*	1.6	*1.3*	*1.99*
Indeterminate	298	*0.0*	4	*2.4*	4	*3.7*	0	*0.0*			
Unknown#	454	*0.0*	10	*5.9*	2	*1.9*	8	*12.9*			
Plurality											
Singleton	987,675	*97.3*	166	*97.6*	106	*98.1*	60	*96.8*	1.7	*1.4*	*1.96*
Twin	26,192	*2.6*	4	*2.4*	2	*1.9*	2	*3.2*	1.5	*0.4*	*3.91*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**96**		**71**		**25**		**0.9**		
Birthweight											
<1,000	6,784	*0.7*	24	*25.0*	9	*12.7*	15	*60.0*	35.4		
1,000–2,499	54,688	*5.4*	34	*35.4*	30	*42.3*	4	*16.0*	6.2		
2,500+	949,958	*93.8*	36	*37.5*	30	*42.3*	6	*24.0*	0.4		
Unknown	1,067	*0.1*	2	*2.1*	2	*2.8*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

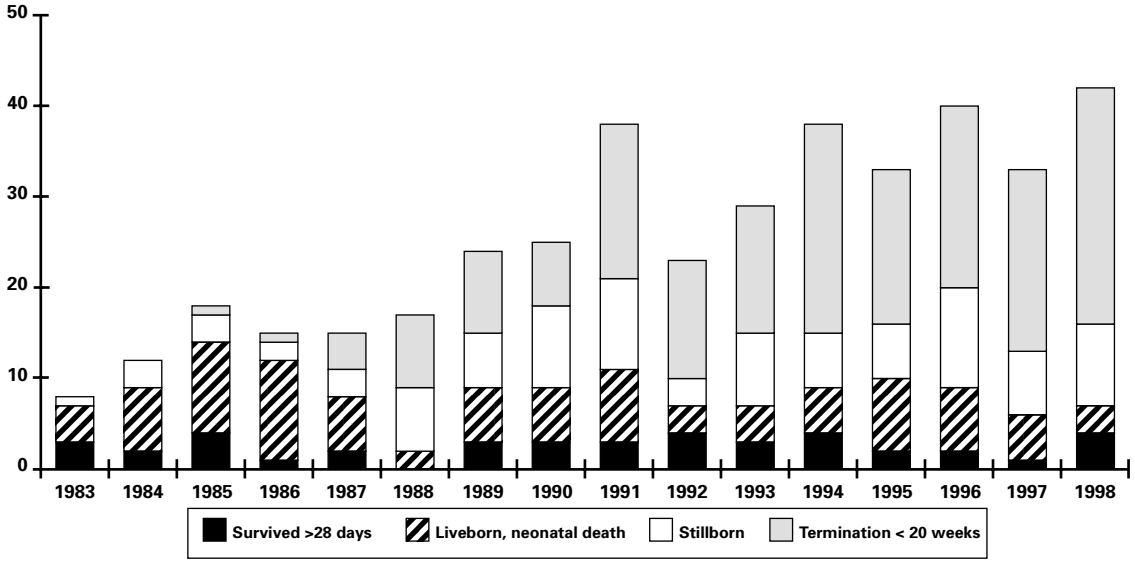
Table 3.26.2 Trisomy 13, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**170**		**108**		**62**		**1.7**	***1.46***	***1.98***
Maternal Age											
<20	40,400	*4.0*	2	*1.2*	2	*1.9*	0	*0.0*	0.5	*0.06*	*1.79*
20–24	185,901	*18.6*	21	*12.4*	17	*15.7*	4	*6.5*	1.1	*0.70*	*1.73*
25–29	367,374	*36.7*	38	*22.4*	27	*25.0*	11	*17.7*	1.0	*0.73*	*1.42*
30–34	289,879	*29.0*	37	*21.8*	28	*25.9*	9	*14.5*	1.3	*0.90*	*1.76*
35–39	101,575	*10.1*	41	*24.1*	20	*18.5*	21	*33.9*	4.0	*2.89*	*5.47*
40+	15,718	*1.6*	24	*14.1*	10	*9.3*	14	*22.6*	15.3	*9.79*	*22.75*
Unknown#	273	*0.0*	7	*4.1*	4	*3.7*	3	*4.8*	N/A		
Country of birth											
Australia	753,424	*75.3*	99	*58.2*	61	*56.5*	38	*61.3*	1.3	*1.07*	*1.61*
Oceania inc NZ	20,620	*2.1*	3	*1.8*	2	*1.9*	1	*1.6*	1.5	*0.30*	*4.25*
UK inc Eire	50,596	*5.1*	8	*4.7*	2	*1.9*	6	*9.7*	1.6	*0.68*	*3.11*
Europe	58,931	*5.9*	11	*6.5*	6	*5.6*	5	*8.1*	1.9	*0.93*	*3.34*
MiddleEast	22,818	*2.3*	3	*1.8*	3	*2.8*	0	*0.0*	1.3	*0.27*	*3.84*
Asia	71,685	*7.2*	10	*5.9*	8	*7.4*	2	*3.2*	1.4	*0.67*	*2.57*
Nth America	5,003	*0.5*	2	*1.2*	1	*0.9*	1	*1.6*	4.0	*0.48*	*14.43*
Sth America	4,689	*0.5*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Africa	10,944	*1.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Unknown#	2,950	*0.3*	34	*20.0*	25	*23.1*	9	*14.5*	N/A		
Region											
Barwon S W	70,984	*7.1*	13	*7.6*	7	*6.5*	6	*9.7*	1.8	*0.97*	*3.13*
Grampians	44,084	*4.4*	9	*5.3*	7	*6.5*	2	*3.2*	2.0	*0.94*	*3.88*
Loddon Mallee	63,104	*6.3*	7	*4.1*	6	*5.6*	1	*1.6*	1.1	*0.44*	*2.29*
Hume	51,348	*5.1*	7	*4.1*	2	*1.9*	5	*8.1*	1.4	*0.55*	*2.81*
Gippsland	56,679	*5.7*	5	*2.9*	4	*3.7*	1	*1.6*	0.9	*0.29*	*2.06*
Western Metro	126,711	*12.7*	16	*9.4*	8	*7.4*	8	*12.9*	1.3	*0.72*	*2.05*
Northern Metro	171,545	*17.1*	31	*18.2*	20	*18.5*	11	*17.7*	1.8	*1.23*	*2.57*
Eastern Metro	192,893	*19.3*	36	*21.2*	25	*23.1*	11	*17.7*	1.9	*1.31*	*2.58*
Southern Metro	214,947	*21.5*	45	*26.5*	29	*26.9*	16	*25.8*	2.1	*1.53*	*2.81*
Other	8,777	*0.9*	1	*0.6*	0	*0.0*	1	*1.6*	1.1	*0.03*	*6.35*
Unknown#	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

3.27 Trisomy 18

Figure 3.27 Trisomy 18, Number of Cases by Year



Number	8	12	18	19	15	17	24	25	38	23	29	38	33	48	33	42
N/10,000	1.3	2.0	2.9	3.1	2.4	2.7	3.7	3.7	5.8	3.5	4.5	5.9	5.2	7.6	5.3	6.8

Trisomy 18

British Paediatric Association code 758.20–758.29

Edward syndrome—additional chromosome 18.

- This trisomy is more than twice as common as trisomy 13 with a birth prevalence of 4.2 / 10,000. The prevalence in the last four years was 6.2 / 10,000, with a marked increase in the number of recognised affected pregnancies and subsequent termination.
- Only 41 cases (9.7%) in the 16 year period survived the neonatal period, or 6% of those born since 1995.
- There is a non-significant excess of females.
- Birth prevalence increased markedly with increasing maternal age. A significant difference is seen for women 30–34 years compared with women 25–29 years and these significant differences are even greater for older women. Women 40 years and older have a risk equivalent to 35–36 year old women having a baby with trisomy 21
- Asian-born women have a significantly increased risk of having a baby with this condition compared to Australian, European and Middle Eastern-born women.
- Women living in Eastern Metropolitan region have an increased risk compared with women living in Loddon Mallee region, almost certainly reflecting maternal age characteristics of the regions. (Refer to Table 2.5 in Births in Victoria, 1996–1998, p.15(6)).

Table 3.27.1 Trisomy 18, 1983–1998, by Selected Infant Characteristics

	All Births + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,014,863**		**422**		**266**		**156**		**4.2**	***3.76***	***4.55***
Survived > 28 days	1,000,776	*98.6*	41	*9.7*	32	*12.0*	9	*5.8*	0.4		
Neonatal death	4,288	*0.4*	95	*22.5*	72	*27.1*	23	*14.7*			
Stillbirth	7,397	*0.7*	94	*22.3*	61	*22.9*	33	*21.2*			
Termination < 20 wks	2,402	*0.2*	192	*45.5*	101	*38.0*	91	*58.3*			
Sex											
Male	521,553	*51.4*	179	*42.4*	119	*44.7*	60	*38.5*	3.4	*3.0*	*3.98*
Female	492,558	*48.5*	209	*49.5*	134	*50.4*	75	*48.1*	4.2	*3.7*	*4.87*
Indeterminate	298	*0.0*	6	*1.4*	6	*2.3*	0	*0.0*			
Unknown#	454	*0.0*	28	*6.6*	7	*2.6*	21	*13.5*			
Plurality											
Singleton	987,675	*97.3*	414	*98.1*	259	*97.4*	155	*99.4*	4.2	*3.8*	*4.60*
Twin	26,192	*2.6*	8	*1.9*	7	*2.6*	1	*0.6*	3.1	*1.3*	*6.02*
Triplet	939	*0.1*	0	*0.0*	0	*0.0*	0	*0.0*	0.0		
Other	48	*0.0*	0	*0.0*	0	*0.0*	0	*0.0*			
	All Births excluding TOPs (83–98)										
Total	**1,012,461**		**230**		**165**		**65**		**2.3**		
Birthweight											
< 1,000	6,784	*0.7*	64	*27.8*	33	*20.0*	31	*47.7*	94.3		
1,000–2,499	54,688	*5.4*	144	*62.6*	113	*68.5*	31	*47.7*	26.3		
2,500+	949,958	*93.8*	12	*5.2*	9	*5.5*	3	*4.6*	0.1		
Unknown	1,067	*0.1*	10	*4.3*	10	*6.1*	0	*0.0*			

95% of unknowns were in terminations before 20 weeks

Table 3.27.2 Trisomy 18, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% CI	
	No.	%	No.	%	No.	%	No.	%		LL	UL
Total	**1,001,120**		**422**		**266**		**156**		**4.2**	***3.82***	***4.61***
Maternal Age											
<20	40,400	*4.0*	6	*1.4*	4	*1.5*	2	*1.3*	1.5	*0.55*	*3.24*
20–24	185,901	*18.6*	30	*7.1*	24	*9.0*	6	*3.8*	1.6	*1.09*	*2.31*
25–29	367,374	*36.7*	78	*18.5*	58	*21.8*	20	*12.8*	2.1	*1.69*	*2.66*
30–34	289,879	*29.0*	96	*22.7*	61	*22.9*	35	*22.4*	3.3	*2.70*	*4.06*
35–39	101,575	*10.1*	111	*26.3*	57	*21.4*	54	*34.6*	10.9	*9.03*	*13.21*
40+	15,718	*1.6*	87	*20.6*	51	*19.2*	36	*23.1*	55.4	*44.61*	*68.63*
Unknown#	273	*0.0*	14	*3.3*	11	*4.1*	3	*1.9*	N/A		
Country of birth											
Australia	753,424	*75.3*	216	*51.2*	129	*48.5*	87	*55.8*	2.9	*2.50*	*3.28*
Oceania inc NZ	20,620	*2.1*	9	*2.1*	5	*1.9*	4	*2.6*	4.4	*2.00*	*8.29*
UK inc Eire	50,596	*5.1*	21	*5.0*	13	*4.9*	8	*5.1*	4.2	*2.57*	*6.35*
Europe	58,931	*5.9*	17	*4.0*	9	*3.4*	8	*5.1*	2.9	*1.68*	*4.62*
Middle East	22,818	*2.3*	5	*1.2*	4	*1.5*	1	*0.6*	2.2	*0.71*	*5.11*
Asia	71,685	*7.2*	53	*12.6*	23	*8.6*	30	*19.2*	7.4	*5.59*	*9.75*
Nth America	5,003	*0.5*	2	*0.5*	1	*0.4*	1	*0.6*	4.0	*0.48*	*14.43*
Sth America	4,689	*0.5*	3	*0.7*	3	*1.1*	0	*0.0*	6.4	*1.32*	*18.68*
Africa	10,944	*1.1*	4	*0.9*	4	*1.5*	0	*0.0*	3.7	*0.99*	*9.36*
Unknown#	2,950	*0.3*	92	*21.8*	75	*28.2*	17	*10.9*	N/A		
Region											
Barwon S W	70,984	*7.1*	23	*5.5*	12	*4.5*	11	*7.1*	3.2	*2.05*	*4.86*
Grampians	44,084	*4.4*	13	*3.1*	8	*3.0*	5	*3.2*	2.9	*1.57*	*5.04*
Loddon Mallee	63,104	*6.3*	16	*3.8*	10	*3.8*	6	*3.8*	2.5	*1.45*	*4.11*
Hume	51,348	*5.1*	15	*3.6*	11	*4.1*	4	*2.6*	2.9	*1.64*	*4.82*
Gippsland	56,679	*5.7*	19	*4.5*	11	*4.1*	8	*5.1*	3.4	*2.02*	*5.23*
Western Metro	126,711	*12.7*	53	*12.6*	34	*12.8*	19	*12.2*	4.2	*3.16*	*5.52*
Northern Metro	171,545	*17.1*	65	*15.4*	43	*16.2*	22	*14.1*	3.8	*2.95*	*4.86*
Eastern Metro	192,893	*19.3*	104	*24.6*	73	*27.4*	31	*19.9*	5.4	*4.43*	*6.56*
Southern Metro	214,947	*21.5*	107	*25.4*	60	*22.6*	47	*30.1*	5.0	*4.10*	*6.04*
Other	8,777	*0.9*	6	*1.4*	3	*1.1*	3	*1.9*	6.8	*2.51*	*14.90*
Unknown#	48	*0.0*	1	*0.2*	1	*0.4*	0	*0.0*	N/A		

95% of unknowns were in terminations before 20 weeks

4.0 Birth Defect Cases by Selected Maternal Characteristics

Table 4.1 Birth Defect Cases by Maternal Age Group, 1983–1998

Age Group	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
<20	1,405	*4.1*	40,400	*4.0*	347.77	329.9	365.6
20–24	6,026	*17.4*	185,901	*18.6*	324.15	316.1	332.2
25–29	11,971	*34.5*	367,374	*36.7*	325.85	320.1	331.6
30–34	9,857	*28.4*	289,879	*29.0*	340.04	333.4	346.6
35–39	4,227	*12.2*	101,575	*10.1*	416.15	403.9	428.4
40+	976	*2.8*	15,718	*1.6*	620.94	583.2	658.7
Unknown	216	*0.6*	273	*0.0*	N/A	N/A	N/A
Total	**34,678**	***100.0***	**1,001,120**	***100.0***			

- Except for women aged less than 20, the prevalence of birth defects increases with increasing age.
- Women aged 35+ comprise 15% of all mothers who have a pregnancy affected by a birth defect and have a relative risk of having a child with a malformation of 1.34 compared to younger women (95%CI 1.30–1.38,p<0.0001)
- If all mothers with pregnancies affected by chromosomal abnormalities (Trisomy 21 , Trisomy 18 and Trisomy 13) are excluded, then the proportion of women aged 35 or more is 13.1%, giving a relative risk of having a child with a malformation 1.13 compared to younger women (95% CI 1.10–1.17, p<0.0001)

Table 4.2 Birth Defect Cases by Country of Birth, 1983–1998

Country of Birth	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Australia	25,316	*73.0*	753,424	*75.3*	330.53	331.9	340.1
Oceania/NZ	731	*2.1*	20,620	*2.1*	347.72	329.3	379.8
UK inc Eire	1,689	*4.9*	50,596	*5.1*	328.48	318.2	349.5
Europe	1,866	*5.4*	58,391	*5.8*	313.75	305.3	333.8
Middle East	973	*2.8*	22,818	*2.3*	422.91	400.2	452.6
Asia	2,271	*6.5*	71,685	*7.2*	311.36	304.0	329.6
Nth America	162	*0.5*	5,003	*0.5*	317.81	274.8	372.9
Sth America	140	*0.4*	4,689	*0.5*	294.31	249.9	347.3
Africa	369	*1.1*	10,944	*1.1*	333.52	303.4	371.0
Unknown	1,161	*3.3*	2,950	*0.3*	N/A	N/A	N/A
Total	**34,678**	***100.0***	**1,001,120**	***100.0***			

- 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 4.3 Birth Defect Cases by Region, 1983–1998

Region	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Barwon	2,248	*6.5*	70,984	*7.1*	312.18	299.4	325.0
Grampians	1,488	*4.3*	44,084	*4.4*	331.87	315.1	348.6
Loddon Mallee	2,031	*5.9*	63,104	*6.3*	317.25	303.6	330.9
Hume	1,824	*5.3*	51,348	*5.1*	349.77	333.9	365.7
Gippsland	1,806	*5.2*	56,679	*5.7*	314.58	300.2	328.9
W Metro	4,697	*13.5*	126,711	*12.7*	364.37	354.1	374.7 N
Metro	6,478	*18.7*	171,545	*17.1*	372.38	363.4	381.3 E
Metro	6,230	*18.0*	192,893	*19.3*	317.27	309.5	325.1 S
Metro	7,415	*21.4*	214,947	*21.5*	339.06	331.4	346.7
Other	381	*1.1*	8,777	*0.9*	423.84	381.7	466.0
Missing	80	*0.2*	48	*0.0*	N/A	#NUM!	#NUM!
Total	**34,678**	***100.0***	**1,001,120**	***100.0***		**337.3**	**344.4**

* Refers to women who reside outside Victoria but who birth at a Victorian hospital.

- Women from Rural Regions (grouped) have a statistically significant decreased likelihood of having a baby born with a malformation when compared to Metropolitan Regions (grouped) (RR 0.9, 95% CI 0.88–0.92,p<0.0001). This is a small decreased risk.

Table 4.4 Birth Defect Cases by Aboriginality (excluding TOPs), 1983–1998

Aboriginality	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Yes	149	*0.5*	5,954	*0.6*	280.48	238.5	322.4
No	32,138	*99.5*	992,774	*99.4*	318.18	314.7	321.6
Total	**32,287**	***100.0***	**998,728**	***100.0***		**314.5**	**321.4**

- The prevalence of birth defects in pregnancies of women identified as Koori in the Perinatal Morbidity Statistics System is significantly less than those identified as non-Koori (RR 0.77, 95% CI 0.66–0.91, p=0.001).

Table 4.5 Birth Defect Cases by Marital Status (excluding TOPs), 1983–1998

Marital Status	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Single	3,284	*10.2*	90,481	*9.1*	356.98	344.9	369.1
Divorced	179	*0.6*	4,473	*0.4*	391.24	334.4	448.1
Widowed	20	*0.1*	677	*0.1*	295.42	167.9	423.0
Separated	233	*0.7*	6,911	*0.7*	331.36	289.2	373.6
Married	26,489	*82.0*	831,448	*83.3*	313.36	309.6	317.1
De facto	2,018	*6.3*	63,178	*6.3*	314.03	300.4	327.6
Unknown	64	*0.2*	1,560	*0.2*	403.85	306.2	501.5
Total	**32,287**	***100.0***	**998,728**	***100.0***		**314.5**	**321.4**

- If marital status is divided into unpartnered (single, separated, divorced, widowed) versus partnered (defacto, married), there is a statistically significant increased relative risk of 1.59 (95% CI 1.54–1.64, p<0.0001) for unpartnered women.

Table 4.6 Birth Defect Cases by Parity (excluding TOPs), 1983–1998

Parity	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
None	13,530	*41.9*	396,771	*39.7*	341.00	335.4	346.6
One	10,459	*32.4*	341,808	*34.2*	305.99	300.2	311.8
Two	5,238	*16.2*	170,619	*17.1*	307.00	298.8	315.2
Three	1,963	*6.1*	59,846	*6.0*	328.01	313.7	342.3
Four	631	*2.0*	18,115	*1.8*	348.33	321.6	375.0
Five or more	461	*1.4*	11,508	*1.2*	400.59	364.8	436.4
Unknown	5	*0.0*	61	*0.0*	819.67	131.3	1508.1
Total	**32,287**	***100.0***	**998,728**	**100.0**	**323.28**	**319.8**	**326.8**

- Primiparous women are more likely to have babies with birth defects than multiparous women (RR 1.09, 95% CI 1.07,1.12, p<0.0001).

5. Birth Defect Cases by Selected Infant Characteristics

Table 5.1 Birth Defect Cases by Number of Defects per Case 1983–1998

Number	No. Cases	%
1	13,949	40.0
2	8996	25.8
3	4575	13.1
4+	7327	21.0
Total	**34,847**	**100.0**

- 34,847 birth defect cases were notified to the BDR, comprising 55,743 individual birth defects
- 40% of cases had an isolated defect.
- The high proportion of cases with multiple defects (ie. 60%) is increased by our coding practice of classifying each manifestation of a syndrome separately.

Table 5.2 Birth Defect Cases by Infant Gender, 1983–1998

Sex	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Male	19,182	*55.0*	521,553	*51.4*	367.8	362.7	394.4
Female	14,910	*42.8*	492,558	*48.5*	302.7	297.9	330.2
Indeterminate	300	*0.9*	298	*0.0*			
Unknown	455	*1.3*	454	*0.0*			
Total	**34,847**	***100.0***	**1,014,863**	***100.0***			

- Birth defects were more commonly reported in males than in females (ie 3.7% of all male pregnancies versus 3.0% of all female pregnancies). This gives a relative risk of having a birth defect for males of 1.21 (95% CI 1.19–1.24, p<0.0001).
- There were 300 cases of indeterminate sex. All births of indeterminate sex are considered to be birth defects.

Table 5.3 Birth Defect Cases by Birthweight (excluding TOPs) , 1983–1998

Birthweight Group	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
<1000	1,560	*4.8*	6,748	*0.7*	2,311.8	2,211.2	2,521.0
1000–2499	4,747	*14.6*	54,688	*5.4*	868.0	844.4	948.1
2500 +	25,772	*79.4*	949,958	*93.8*	271.3	268.0	291.1
Unknown	366	*1.1*	1,067	*0.1*	3,430.2	3,145.3	3,916.5
Total	**32,445**	***100.0***	**1,012,461**	***100.0***	**320.5**	**317.0**	**339.6**

- Malformations were recorded among 10.3% of low birthweight infants (<2500 grams) compared with 2.7 % of infants with birthweights of 2500 grams or more.

Table 5.4 Birth Defect Cases by Gestational Age (excluding TOPs), 1983–1998

Gestation (weeks)	No. Cases	%	Total No Cofinements	%	N/10,000	95% Confidence Interval LL	UL
20–27	1,579	*4.9*	6,565	*0.6*	2,405.2	2,301.8	2,616.0
28–31	1,183	*3.6*	7,317	*0.7*	1,616.8	1,532.4	1,826.6
32–36	3,449	*10.6*	53,972	*5.3*	639.0	618.4	720.7
37–41	24,897	*76.7*	902,898	*89.2*	275.7	272.4	296.1
> 41	1046	*3.2*	35,900	*3.5*	291.4	274.0	393.3
Unknown	291	*0.9*	5809	*0.6*	500.9	444.8	751.6
Total	**32,445**	***100.0***	**1,012,461**	***100.0***	**320.5**	**317.0**	**339.6**

- 9.2% of premature infants (< 37 weeks) had a malformation compared to 2.8% among term and post-term infants (>= 37 weeks).

Table 5.5 Birth Defect Cases by Plurality, 1983–1998

Plurality	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Single	33,410	*95.9*	987,675	*97.3*	338.3	334.7	357.7
Twin	1,353	*3.9*	26,192	*2.6*	516.6	489.8	634.5
Triplet	69	*0.2*	939	*0.1*	734.8	567.9	1,350.5
Quad	6	*0.0*	48	*0.0*	1,250.0	314.4	3,896.3
Unknown	9	*0.0*	9	*0.0*	10,000.0	10,000.0	10,000.0
Total	**34,847**	***100.0***	**1,014,863**	*100.0*	**343.4**	**339.8**	**362.5**

- 3.4% of singleton births had a birth defect.
- 5.3% of multiple births (twins, triplets, quads) had a birth defect.
- Relative risk for multiple versus singleton birth is 1.55 (95% CI 1.48–1.64,p <0.0001).

Table 5.6 Birth Defect Cases by Rank (multiple pregnancies only), 1983–1998

Rank	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
1	715	*50.1*	13,415	*49.4*	533.0	495.0	697.6
2	677	*47.4*	13,414	*49.4*	504.7	467.7	669.6
3	21	*1.5*	325	*1.2*	646.2	378.9	1,697.7
4	2	*0.1*	12	*0.0*	1,666.7		
Unknown	13	*0.9*	13	*0.0*			
Total	**1,428**	***100.0***	**27,179**	***100.0***		**498.9**	**641.1**

- Within the multiple births, 715 (50.1%) were the first born and 677 (47.4%) were second born infants.

Table 5.7 Perinatal Mortality Associated with Birth Defect Cases (excluding TOPs), 1983–1998

Status	No. Cases	%	Total No. Cofinements	%	N/10,000	95% Confidence Interval LL	UL
Liveborn, surviving >28 days	29,032	*89.5*	1,000,776	*98.8*	290.1	286.8	309.4
Stillborn	1,553	*4.8*	7,397	*0.7*	2,099.5	2,006.7	2,302.1
NND	1,860	*5.7*	4,288	*0.4*	4,337.7	4,189.3	4,562.9
Total	**32,445**	***100.0***	**1,012,461**	***100.0***	**320.5**	**317.0**	**339.6**

1. Perinatal mortality is high among infants with birth defects.
- 1,553 (4.8%) infants were stillborn and 1,860 (5.7%) were neonatal deaths. This corresponds to a perinatal mortality rate of 105.2 / 1,000 births with a malformation.
- These mortality figures comprise all birth defect cases, including those where the cause of death may not be directly related to the birth defect.
- Relative risk of a perinatal death for a baby with a malformation compared to a baby without a malformation is 13.81 (95% CI 13.25,14.40,$p < 0.0001$).

2. Birth defects are a major contributor to perinatal mortality.
- Between 1983–1998 there were the 7,397 stillbirths, 1,553 (21%) had a birth defect.
- Between 1983–1998 there were 4,288 neonatal deaths, 1,860 (43.4%) had a birth defect.
- 29.2% of all perinatal deaths were associated with one or more birth defects.

References

1. Riley M. & Halliday J (1996), *Congenital Malformations in Victoria 1983–1994*, Perinatal Data Collection Unit, Victorian Government Department of Human Services, Melbourne.
2. Hurst T, Shafir E, Day P & Lancaster P. 1999, *Congenital Malformations Australia 1995 and 1996*, AIHW Cat. No. PER 8, Sydney: Australian Institute of Health and Welfare National Perinatal Statistics Unit (Birth Defects Series No.3)
3. The Consultative Council on Obstetric and Paediatric Mortality and Morbidity (2000), *Annual Report for the Year 1998, incorporating the 37th Survey of Perinatal Deaths*, Melbourne.
4. Lumley J., Palma S., Fischer M., Robertson H.(1988), 'The tip of the iceberg: A validation study of the Victorian Congenital Malformations / Birth Defects Register', *Australian Paediatric Journal*, no. 24: p.244–6
5. Kilkenny M., Riley M., Lumley J. (1995), 'Follow-up validation study study of the Victorian Congenital Malformations Register', *Journal of Paediatric & Child Health*, no. 31 p.323–5
6. Riley M. & Halliday J. (1999), *Births in Victoria 1996–1998*, Perinatal Data Collection Unit, Victorian Government Department of Human Services, Melbourne.

Appendix A: List of Exclusions

NB: There has been some variation in this list of exclusions between 1983–1998. Some excluded conditions may be included in this report if they were previously not excluded AND occur with other birth defects

Abnormal palmar creases

Accessory nipples

Anal fissure

Balanced autosomal translocation (unless ocurring 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

Hydrocoele

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

Pyloris stenosis

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

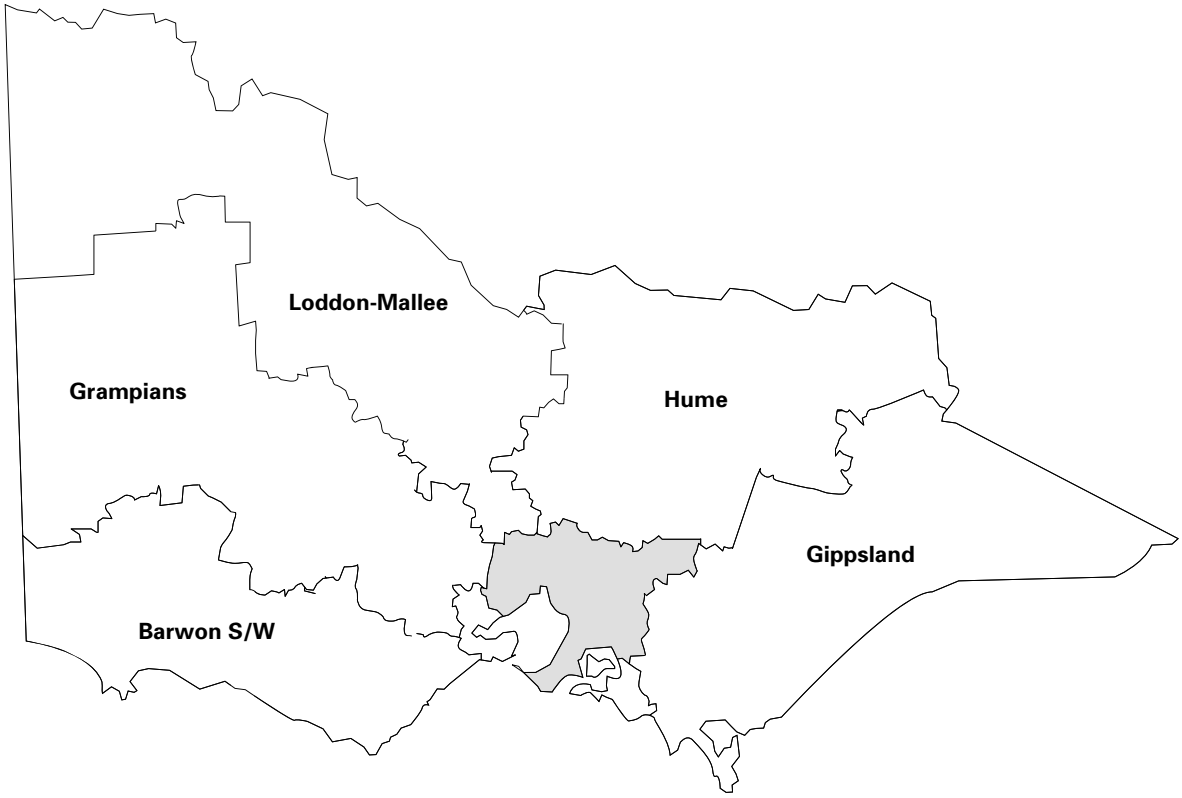
Appendix B: Routine Data Items Contained in the Birth Defects/Congenital Malformations Register

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 malformations, position code, source of notification

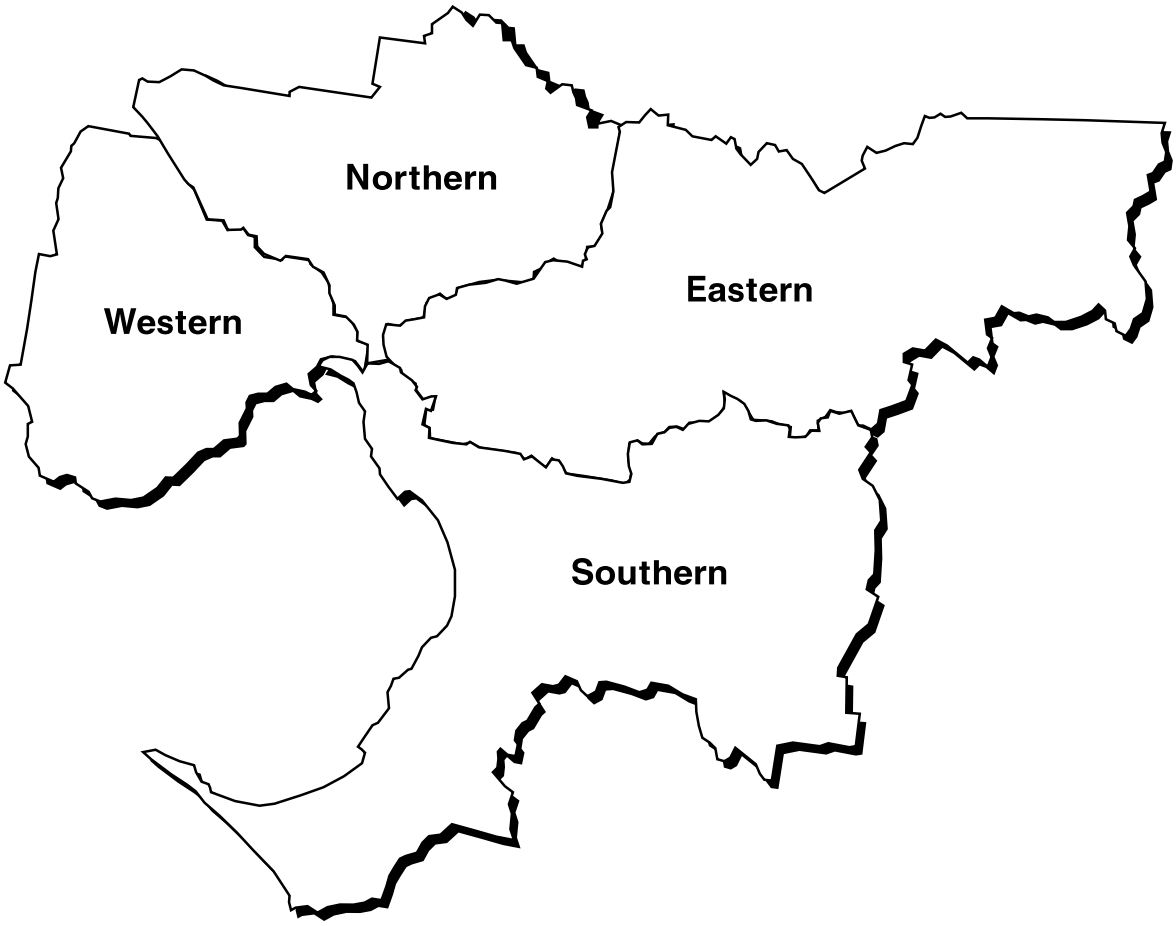
Other data items available from linkage to the Perinatal Morbidity Statistics Form:

(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 C: Department of Human Services—Rural Regions



Appendix C: Department of Human Services—Metropolitan Regions



Appendix D: Allocation of Postcodes to Current (1998) LGA Boundaries

Barwon South West

3211, 3212, 3214 to 3228, 3230, 3232,3233,3235 to 3243, 3249 to 3251, 3253, 3254, 3256, 3257, 3260, 3264 to 3282, 3284 to 3287, 3289, 3290, 3292 to 3294, 3300 to 3306, 3309 to 3312, 3314, 3315, 3323 to 3325, 3361, 3407, 3408, 3410

Grampians

3231, 3291, 3317 to 3319, 3321, 3322, 3328 to 3334, 3340 to 3343, 3345, 3350 to 3353, 3355 to 3357, 3360, 3363, 3364, 3370, 3371, 3373, 3375, 3377 to 3381, 3384 to 3401, 3405, 3409, 3412 to 3415, 3417 to 3420, 3422 to 3424, 3458 to 3461, 3467 to 3469, 3478, 3485, 3487 to 3489, 3491

Loddon Mallee

3430 to 3451, 3453, 3462 to 3465, 3472, 3475, 3480, 3482, 3483, 3490, 3494, 3496, 3498, 3500 to 3502, 3505 to 3507, 3509, 3512, 3515 to 3518, 3520, 3523, 3525, 3527, 3529 to 3537, 3539, 3540, 3542, 3544, 3546, 3549 to 3551, 3555 to 3576, 3578 to 3591, 3594 to 3601, 3612, 3620 to 3625, 3654

Hume

3521, 3522, 3603, 3604, 3606 to 3611, 3613 to 3618, 3627 to 3641, 3644 to 3649, 3651 to 3653, 3655, 3656, 3658 to 3678, 3682 to 3691, 3693 to 3701, 3704 to 3749, 3756, 3758, 3762 to 3764, 3778, 3779

Gippsland

3816 to 3825, 3828, 3831 to 3842, 3844, 3847, 3850 to 3904, 3909, 3921 to 3925, 3945, 3946, 3950 to 3968, 3971, 3979, 3984, 3987 to 3996

Western Metro

3000 to 3003, 3006, 3011 to 3042, 3051 to 3053, 3335 to 3338, 3427

Northern Metro

3043 to 3049, 3054 to 3099, 3121, 3428 to 3430, 3750 to 3754, 3757, 3759 to 3761

Eastern Metro

3101 to 3140, 3146 to 3160, 3166, 3168, 3170, 3178 to 3180, 3755, 3765 to 3767, 3770, 3775, 3777, 3782, 3785 to 3799

Southern Metro

3004, 3141 to 3146, 3148, 3161 to 3165, 3167, 3169, 3171 to 3177, 3181 to 3207, 3781 to 3783, 3802 to 3815, 3910 to 3920, 3926 to 3944, 3975 to 3978, 3980, 3981

Other

Postcodes outside Victoria