

Birth Defects in Victoria 1983–1998

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Contents

Acknowledgments	v
Abbreviations	vi
Definitions	vii
List of Figures	viii
List of Tables	ix
Highlights	xi
1. Introduction	1
1.1 Background	1
1.2 Purposes	1
1.3 Inclusion Criteria	1
1.4 Sources of Notification	2
1.5 Data Items	3
1.6 Data Quality	3
1.7 Explanatory Notes	3
1.8 Terminations of Pregnancy	3
1.9 Statistics	4
1.10 Regional Variations	4
2. Summary Tables	7
2.1 Birth Defects by Year, 1983–1998	7
2.2 Comparison of Prevalence of Birth Defects, 1983–1994 and 1995–1998	8
2.3 Order of Prevalence of Twenty Seven Selected Defects, 1995–1998	9
2.4 Summary of Patterns for Twenty-Four Selected Defects, 1983–1998	10
2.5 Birth Defects by Major Anatomical System	11
2.6 Birth Defects by Four-Digit Code	12
2.7 Indications for Terminations of Pregnancy for a Birth Defect Pre 20 weeks Gestation, 1989–1998	18
3. Major Birth Defects	21
3.1 Anencephaly	21
3.2 Spina Bifida	24
3.3 Encephalocele	27
3.4 Microcephalus	30
3.5 Hydrocephalus	33
3.6 Transposition of Great Vessels	36
3.7 Tetralogy of Fallot	39
3.8 Ventricular Septal Defect	42
3.9 Hypoplastic Left Heart Syndrome	45
3.10 Coarctation of Aorta	48
3.11 Cleft Palate	51
3.12 Cleft Lip	54
3.13 Cleft Lip and Palate	57
3.14 Oesophageal Atresia and/or Stenosis	60
3.15 Anorectal Atresia and/or Stenosis	63
3.16 Hypospadias	66
3.17 Renal Agenesis and Dysgenesis	69
3.18 Cystic Kidney Disease	72
3.19 Obstructive Defects of Renal Pelvis	75
3.20 Congenital Dislocation of Hip	79

3.21	Limb Reduction Defects	82
3.22	Diaphragmatic Hernia	85
3.23	Exomphalos	88
3.24	Gastroschisis	91
3.25	Trisomy 21	94
3.26	Trisomy 13	97
3.27	Trisomy 18	100
4.	Birth Defect Cases by Selected Maternal Factors.....	103
4.1	Birth Defect Cases by Maternal Age	103
4.2	Birth Defect Cases by Maternal Country of Birth	103
4.3	Birth Defect Cases by Humans Services Regions	104
4.4	Birth Defect Cases by Aboriginality	104
4.5	Birth Defect Cases by Marital Status	105
4.6	Birth Defect Cases by Parity	105
5.	Birth Defect Cases by Selected Infant Factors	107
5.1	Birth Defect Cases by Number of Defects per Case	107
5.2	Birth Defect Cases by Infant Gender	107
5.3	Birth Defect Cases by Birthweight	107
5.4	Birth Defect Cases by Gestational Age	108
5.5	Birth Defect Cases by Plurality	108
5.6	Birth Defect Cases by Rank	108
5.7	Perinatal Mortality Associated with Birth Defect Cases	109
	References	111
	Appendix A Exclusions	113
	Appendix B Routine Data Items in the Birth Defects/Congenital Malformations Register ..	114
	Appendix C Department of Human Services—Rural Regions.....	115
	Department of Human Services—Metropolitan Region	116
	Appendix D Allocation of Postcodes to Current LGA boundaries	117

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Abbreviations

a/s	atresia/stenosis	MMC	Monash Medical Centre
AIHW	Australian Institute of Health and Welfare	nec	not elsewhere classified
anom	anomalies	NND	neonatal death
ASD	atrial septal defect	NPSU	National Perinatal Statistics Unit
BDR	Birth Defects/Congenital Malformations Register	nutri	nutritional
CDH	congenital dislocation of hip	p	probability
chrom	chromosomal	PDA	patent ductus arteriosus
CI	confidence interval	PDCU	Perinatal Data Collection Unit
CMR	Birth Defects/Congenital Malformations Register	pns	peripheral nervous system
cns	central nervous system	RCH	Royal Children's Hospital
COA	coarctation of aorta	red	reduction
desc	described	RR	relative risk
dev	developmental	RWH	Royal Women's Hospital
df	degrees of freedom	s/insuff	stenosis/insufficiency
dysgen	dysgenesis	SB	stillbirth
electr	electrolyte	spec	specified
end	endocrine	sternocleido	sternocleidomastoid
g	grams	TGV	transposition of great vessels
HLHS	hypoplastic left heart syndrome	TOP	termination of pregnancy
inc	includes	trans	translocation
ICD	International Classification of Diseases	tris	trisomy
LGA	Local Government Area	udt	undescended testicle
MCH Nurse	Maternal & Child Health Nurse	unspe	unspecified
metab	metabolic	VCGS	Victorian Clinical Genetics Service
MHW	Mercy Hospital for Women	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.

List of Figures

Figure 2.1	Prevalence of Birth Defects as a Proportion of Births, 1983–1994	7
Figure 3.1	Anencephaly, Number of Cases by Year	21
Figure 3.2	Spina Bifida, Number of Cases by Year	24
Figure 3.3	Encephalocele, Number of Cases by Year	27
Figure 3.4	Microcephalus, Number of Cases by Year	30
Figure 3.5	Hydrocephalus, Number of Cases by Year	33
Figure 3.6	Transposition of Great Vessels, Number of Cases by Year	36
Figure 3.7	Tetralogy of Fallot, Number of Cases by Year	39
Figure 3.8	Ventricular Septal Defect, Number of Cases by Year	42
Figure 3.9	Hypoplastic Left Heart Syndrome, Number of Cases by Year	45
Figure 3.10	Coarctation of Aorta, Number of Cases by Year	48
Figure 3.11	Cleft Palate, Number of Cases by Year	51
Figure 3.12	Cleft Lip, Number of Cases by Year	54
Figure 3.13	Cleft Lip and Palate, Number of Cases by Year	57
Figure 3.14	Oesophageal Atresia and/or Stenosis, Number of Cases by Year	60
Figure 3.15	Anorectal Atresia and/or Stenosis, Number of Cases by Year	63
Figure 3.16	Hypospadias, Number of Cases by Year	66
Figure 3.17	Renal Agenesis and Dysgenesis, Number of Cases by Year	69
Figure 3.18	Cystic Kidney Disease, Number of Cases by Year	72
Figure 3.19	Obstructive Defects of Renal Pelvis, Number of Cases by Year	75
Figure 3.20	Congenital Dislocation of Hip, Number of Cases by Year	79
Figure 3.21	Limb Reduction Defects, Number of Cases by Year	82
Figure 3.22	Diaphragmatic Hernia, Number of Cases by Year	85
Figure 3.23	Exomphalos, Number of Cases by Year	88
Figure 3.24	Gastroschisis, Number of Cases by Year	91
Figure 3.25	Trisomy 21, Number of Cases by Year	94
Figure 3.26	Trisomy 13, Number of Cases by Year	97
Figure 3.27	Trisomy 18, Number of Cases by Year	100

List of Tables

Table 1.1	Sources of Notification, 1989–1998	2
Table 1.2	An Example: a Proportion of Spina Bifida Cases by Human Services Regions 1992–1995 and 1998	4
Table 2.1	Birth Defects by Year, 1983–1998	7
Table 2.2	Comparison of Prevalence of Birth Defects, 1983–1994 and 1995–1998	8
Table 2.3	Order of Prevalence of Twenty Seven Selected Defects	9
Table 2.4	Summary of Patterns of Birth Defects 1983–1998	10
Table 2.5	Birth Defects by Major Anatomical Site.....	11
Table 2.6	Birth Defects by Four Digit Code, 1983–1998	12
Table 2.7	Indications for Terminations of Pregnancy for Birth Defects Pre 20 weeks Gestation, 1989–1998	18
Table 3.1.1	Anencephaly, 1983–1998, by Selected Infant Characteristics	22
Table 3.1.2	Patterns of Birth Defects, Anencephaly, 1983–1998	22
Table 3.1.3	Anencephaly, 1983–1998, by Selected Maternal Characteristics	23
Table 3.2.1	Spina Bifida, 1983–1998, by Selected Infant Characteristics	25
Table 3.2.2	Patterns of Birth Defects, Spina Bifida, 1983–1998.....	25
Table 3.2.3	Spina Bifida, 1983–1998, by Selected Maternal Characteristics	26
Table 3.3.1	Encephalocele, 1983–1998, by Selected Infant Characteristics.....	28
Table 3.3.2	Patterns of Birth Defects, Encephalocele, 1983–1998	28
Table 3.3.3	Encephalocele, 1983–1998, by Selected Maternal Characteristics	29
Table 3.4.1	Microcephalus, 1983–1998, by Selected Infant Characteristics	31
Table 3.4.2	Patterns of Birth Defects, Microcephalus, 1983–1998	31
Table 3.4.3	Microcephalus, 1983–1998, by Selected Maternal Characteristics	32
Table 3.5.1	Hydrocephalus, 1983–1998, by Selected Infant Characteristics	34
Table 3.5.2	Patterns of Birth Defects, Hydrocephalus, 1983–1998	34
Table 3.5.3	Hydrocephalus, 1983–1998, by Selected Maternal Characteristics	35
Table 3.6.1	Transposition of Great Vessels, 1983–1998, by Selected Infant Characteristics	37
Table 3.6.2	Patterns of Birth Defects, Transposition of Great Vessels, 1983–1998	37
Table 3.6.3	Transposition of Great Vessels, 1983–1998, by Selected Maternal Characteristics	38
Table 3.7.1	Tetralogy of Fallot, 1983–1998, by Selected Infant Characteristics	40
Table 3.7.2	Patterns of Birth Defects, Tetralogy of Fallot, 1983–1998	40
Table 3.7.3	Tetralogy of Fallot, 1983–1998, by Selected Maternal Characteristics.....	41
Table 3.8.1	Ventricular Septal Defect, 1983–1998, by Selected Infant Characteristics	43
Table 3.8.2	Patterns of Birth Defects, Ventricular Septal Defect, 1983–1998	43
Table 3.8.3	Ventricular Septal Defect, 1983–1998, by Selected Maternal Characteristics	44
Table 3.9.1	Hypoplastic Left Heart Syndrome, 1983–1998, by Selected Infant Characteristics	46
Table 3.9.2	Patterns of Birth Defects, Hypoplastic Left Heart Syndrome, 1983–1998	46
Table 3.9.3	Hypoplastic Left Heart Syndrome, 1983–1998, by Selected Maternal Characteristics	47
Table 3.10.1	Coarctation of Aorta, 1983–1998, by Selected Infant Characteristics	49
Table 3.10.2	Patterns of Birth Defects, Coarctation of Aorta, 1983–1998	49
Table 3.10.3	Coarctation of Aorta, 1983–1998, by Selected Maternal Characteristics.....	50
Table 3.11.1	Cleft Palate, 1983–1998, by Selected Infant Characteristics.....	52
Table 3.11.2	Patterns of Birth Defects, Cleft Palate, 1983–1998	52
Table 3.11.3	Cleft Palate, 1983–1998, by Selected Maternal Characteristics	53
Table 3.12.1	Cleft Lip, 1983–1998, by Selected Infant Characteristics.....	55
Table 3.12.2	Patterns of Birth Defects, Cleft Lip, 1983–1998	55
Table 3.12.3	Cleft Lip, 1983–1998, by Selected Maternal Characteristics.....	56
Table 3.13.1	Cleft Lip and Palate, 1983–1998, by Selected Infant Characteristics	58
Table 3.13.2	Patterns of Birth Defects, Cleft Lip and Palate, 1983–1998	58
Table 3.13.3	Cleft Lip and Palate, 1983–1998, by Selected Maternal Characteristics	59
Table 3.14.1	Oesophageal Atresia and/or Stenosis, 1983–1998, by Selected Infant Characteristics	61
Table 3.14.2	Patterns of Birth Defects, Oesophageal Atresia and/or Stenosis, 1983–1998	61
Table 3.14.3	Oesophageal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics	62

Table 3.15.1	Anorectal Atresia and/or Stenosis, 1983–1998, by Selected Infant Characteristics	64
Table 3.15.2	Patterns of Birth Defects, Anorectal Atresia and/or Stenosis, 1983–1998	64
Table 3.15.3	Anorectal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics	65
Table 3.16.1	Hypospadias, 1983–1998, by Selected Infant Characteristics	67
Table 3.16.2	Patterns of Birth Defects, Hypospadias, 1983–1998	67
Table 3.16.3	Hypospadias, 1983–1998, by Selected Maternal Characteristics	68
Table 3.17.1	Renal Agenesis and Dysgenesis, 1983–1998, by Selected Infant Characteristics	70
Table 3.17.2	Patterns of Birth Defects, Renal Agenesis and Dysgenesis, 1983–1998.....	70
Table 3.17.3	Renal Agenesis and Dysgenesis, 1983–1998, by Selected Maternal Characteristics	71
Table 3.18.1	Cystic Kidney Disease, 1983–1998, by Selected Infant Characteristics	73
Table 3.18.2	Patterns of Birth Defects, Cystic Kidney Disease, 1983–1998	73
Table 3.18.3	Cystic Kidney Disease, 1983–1998, by Selected Maternal Characteristics	74
Table 3.19.1	Obstructive Defects of Renal Pelvis, 1983–1998, by Selected Infant Characteristics	77
Table 3.19.2	Patterns of Birth Defects, Obstructive Defects of Renal Pelvis, 1983–1998	77
Table 3.19.3	Obstructive Defects of Renal Pelvis, 1983–1998, by Selected Maternal Characteristics	78
Table 3.20.1	Congenital Dislocation of Hip, 1983–1998, by Selected Infant Characteristics	80
Table 3.20.2	Patterns of Birth Defects, Congenital Dislocation of Hip, 1983–1998	80
Table 3.20.3	Congenital Dislocation of Hip, 1983–1998, by Selected Maternal Characteristics	81
Table 3.21.1	Limb Reduction Defects, 1983–1998, by Selected Infant Characteristics	83
Table 3.21.2	Patterns of Birth Defects, Limb Reduction Defects, 1983–1998	83
Table 3.21.3	Limb Reduction Defects, 1983–1998, by Selected Maternal Characteristics	84
Table 3.22.1	Diaphragmatic Hernia, 1983–1998, by Selected Infant Characteristics	86
Table 3.22.2	Patterns of Birth Defects, Diaphragmatic Hernia, 1983–1998.....	86
Table 3.22.3	Diaphragmatic Hernia, 1983–1998, by Selected Maternal Characteristics	87
Table 3.23.1	Exomphalos, 1983–1998, by Selected Infant Characteristics	89
Table 3.23.2	Patterns of Birth Defects, Exomphalos, 1983–1998.....	89
Table 3.23.3	Exomphalos, 1983–1998, by Selected Maternal Characteristics	90
Table 3.24.1	Gastroschisis, 1983–1998, by Selected Infant Characteristics	92
Table 3.24.2	Patterns of Birth Defects, Gastroschisis, 1983–1998	92
Table 3.24.3	Gastroschisis, 1983–1998, by Selected Maternal Characteristics.....	93
Table 3.25.1	Trisomy 21, 1983–1998, by Selected Infant Characteristics.....	95
Table 3.25.2	Trisomy 21, 1983–1998, by Selected Maternal Characteristics.	96
Table 3.26.1	Trisomy 13, 1983–1998, by Selected Infant Characteristics.....	98
Table 3.26.2	Trisomy 13, 1983–1998, by Selected Maternal Characteristics	99
Table 3.27.1	Trisomy 18, 1983–1998, by Selected Infant Characteristics.....	101
Table 3.27.2	Trisomy 18, 1983–1998, by Selected Maternal Characteristics	102
Table 4.1	Birth Defect Cases by Maternal Age Group, 1983–1998	103
Table 4.2	Birth Defect Cases by Country of Birth, 1983–1998	103
Table 4.3	Birth Defect Cases by Region, 1983–1998	104
Table 4.4	Birth Defect Cases by Aboriginality, 1983–1998	104
Table 4.5	Birth Defect Cases by Marital Status, 1983–1998	105
Table 4.6	Birth Defect Cases by Parity, 1983–1998	105
Table 5.1	Birth Defect Cases by Number of Defects per Case, 1983–1998.	107
Table 5.2	Birth Defect Cases by Infant Gender, 1983–1998	107
Table 5.3	Birth Defect Cases by Birthweight, 1983–1998	107
Table 5.4	Birth Defect Cases by Gestational Age, 1983–1998	108
Table 5.5	Birth Defect Cases by Plurality, 1983–1998	108
Table 5.6	Birth Defect Cases by Rank, 1983–1998	108
Table 5.7	Perinatal Mortality Associated with Birth Defect Cases, 1983–1998	109

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 Syndrome, 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 versus 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 \geq 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

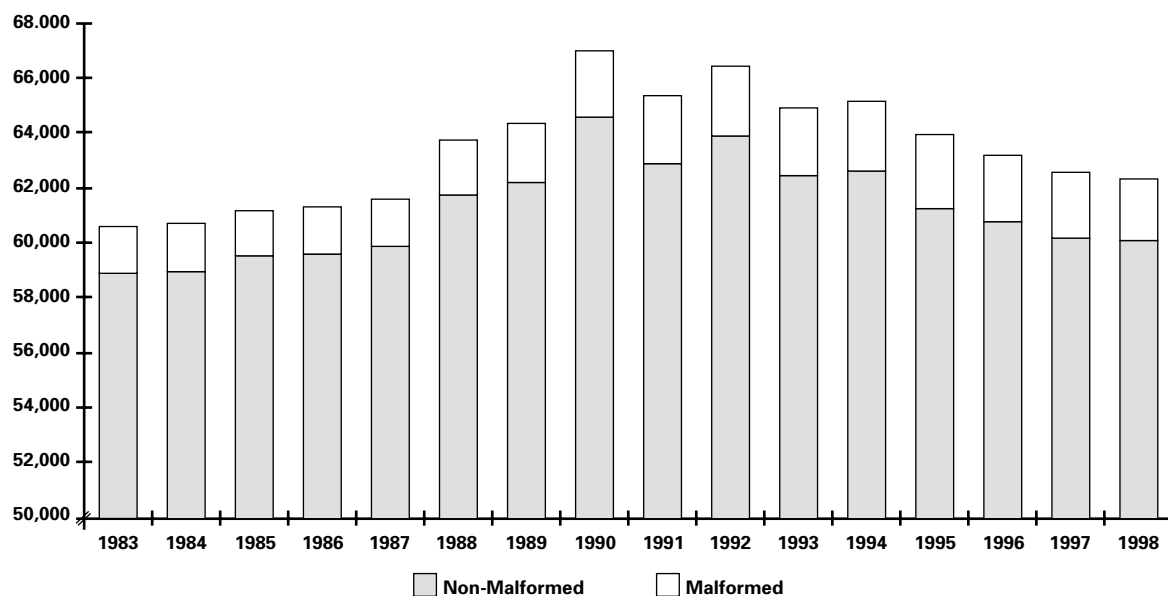
2.1 Birth Defects by Year, 1983–1998

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
<i>Total pregnancies (births + TOPs)</i>	<i>762,705</i>		<i>252,158</i>	
Anencephaly	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 chromosomal 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	Developmental 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 hydrocele 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. continued next page

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	<i>Bilateral</i>	293	2.9
75301	<i>Unilateral</i>	244	2.4
7531#	Cystic Kidney Disease	476	4.7
75311-3	<i>Polycystic</i>	154	1.5
75316	<i>Multicystic</i>	242	2.4
75310/4/8	<i>Other</i>	80	0.8
7532#	Obstructive Defects Renal Pelvis/Ureter	2,093	20.6
75320	Hydronephrosis	1,194	11.8
75321-9	<i>Other</i>	507	5.0
75330/1/3-9	<i>Other Specified Kidney Disorders</i>	243	2.4
75332	<i>Horseshoe Kidney</i>	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. *continued next page*

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	<i>Craniosynostosis</i>	221	2.2
75601-2/4-9	<i>Other</i>	345	3.4
75603	<i>Pierre Robin Syndrome</i>	115	1.1
7561	Spine	316	3.1
7562-3	Ribs & Sternum	138	1.4
7564	Chondrodystrophy	143	1.4
75643	<i>Achondroplasia</i>	37	0.4
75644	<i>Other Dwarfing</i>	33	0.3
75640-2/5-9	<i>Other</i>	73	0.7
7565	Osteodystrophies	122	1.2
75650	<i>Osteogenesis</i>	75	0.7
75651-9	<i>Other</i>	47	0.5
7566	Diaphragm	444	4.4
75660/2-9	<i>Other</i>	87	0.9
75661	<i>Diaphragmatic Hernia</i>	357	3.5
7567	Abdominal Wall	549	5.4
75670	<i>Exomphalos</i>	315	3.1
75671	<i>Gastroschisis</i>	165	1.6
75672-9	<i>Other</i>	69	0.7
7568	Other Specified of Muscle/ Tendon/ Fascia/ Connective Tissue	116	1.1
7569	Unspecified	18	0.2
757	Integument	467	4.6
7570	Hereditary Oedema of Legs	5	0.0
7571	Ichthyosis 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. *continued next page*

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 Sclerosis	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	<i>Haemangioma</i>	82	0.8
22819	<i>Cystic Hygroma</i>	192	1.9
2100-227/229	<i>Other</i>	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

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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	Developmental Delay/Problems*	222	2.2
(also 783-786)			

* 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
Multiple 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

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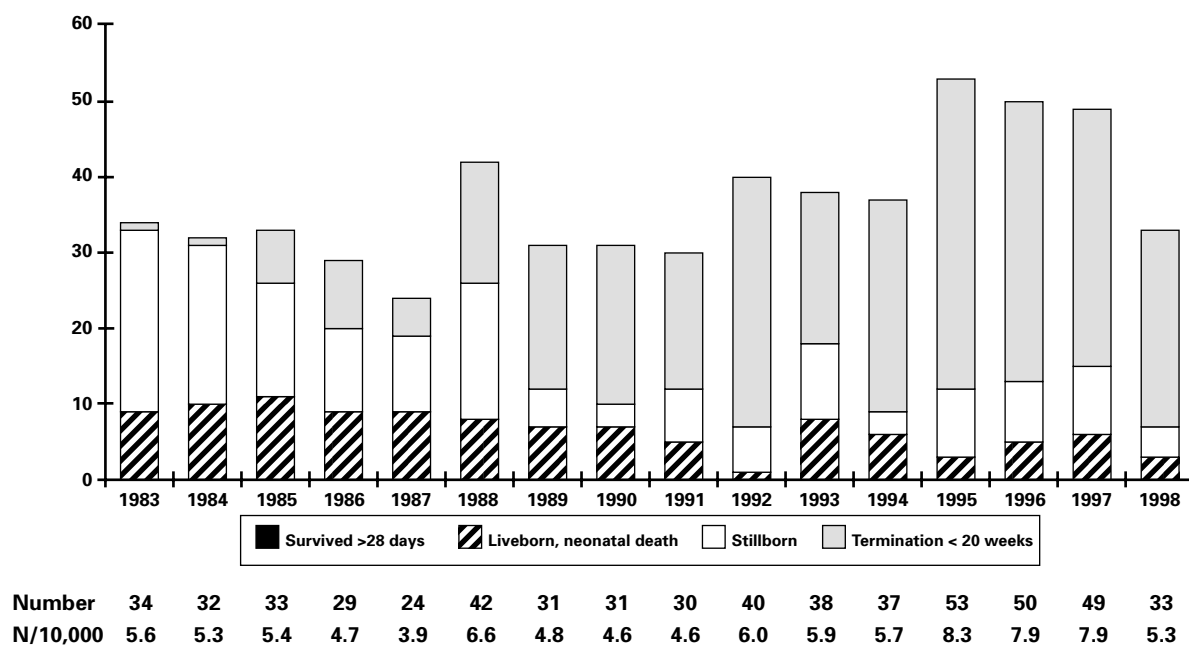
Table 2.7 Indications for Terminations of Pregnancy for a Birth Defect Pre 20 weeks Gestation, 1989–1998 (cont.)

Anomaly	1989	1990	1991	1992	1993	1994	1995	1996	1997	1998	Total
<i>Digestive System</i>	0	1	0	0	0	0	0	1	1	1	4
<i>Urinary System</i>											
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
<i>Musculoskeletal System</i>											
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



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 periconceptual 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.

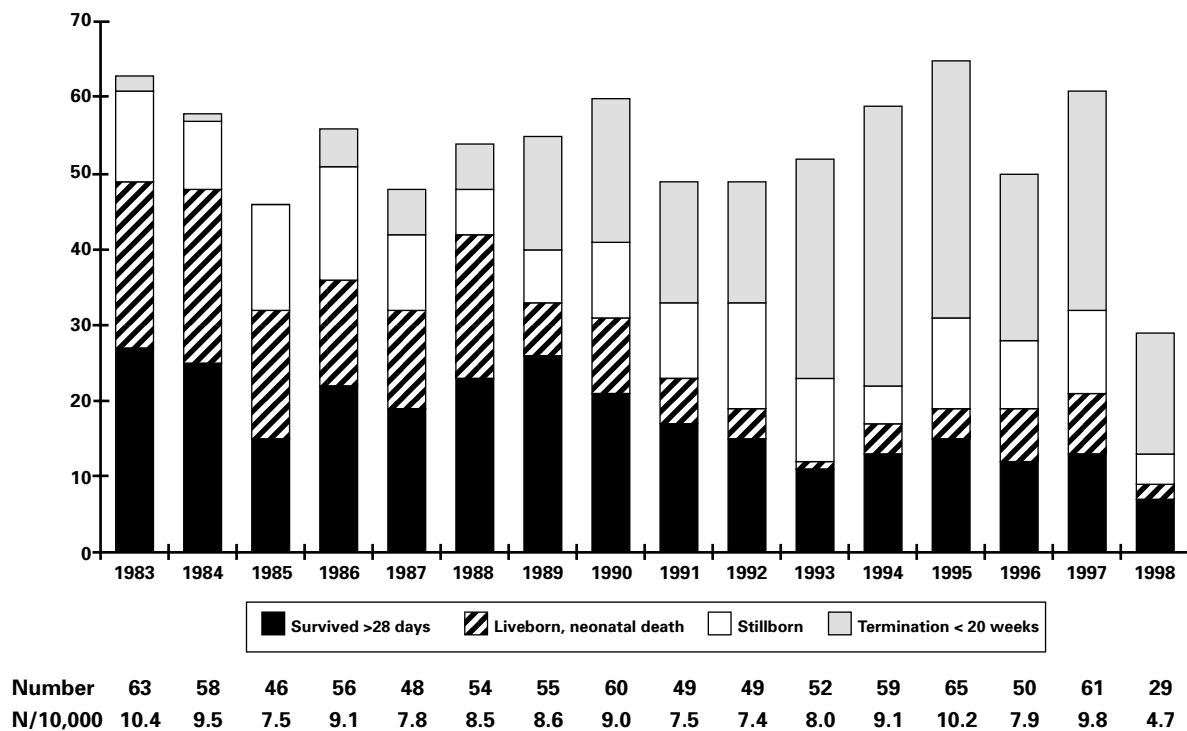
Table 3.1.3 Anencephaly, 1983–1998, by Selected Maternal Characteristics

	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



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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

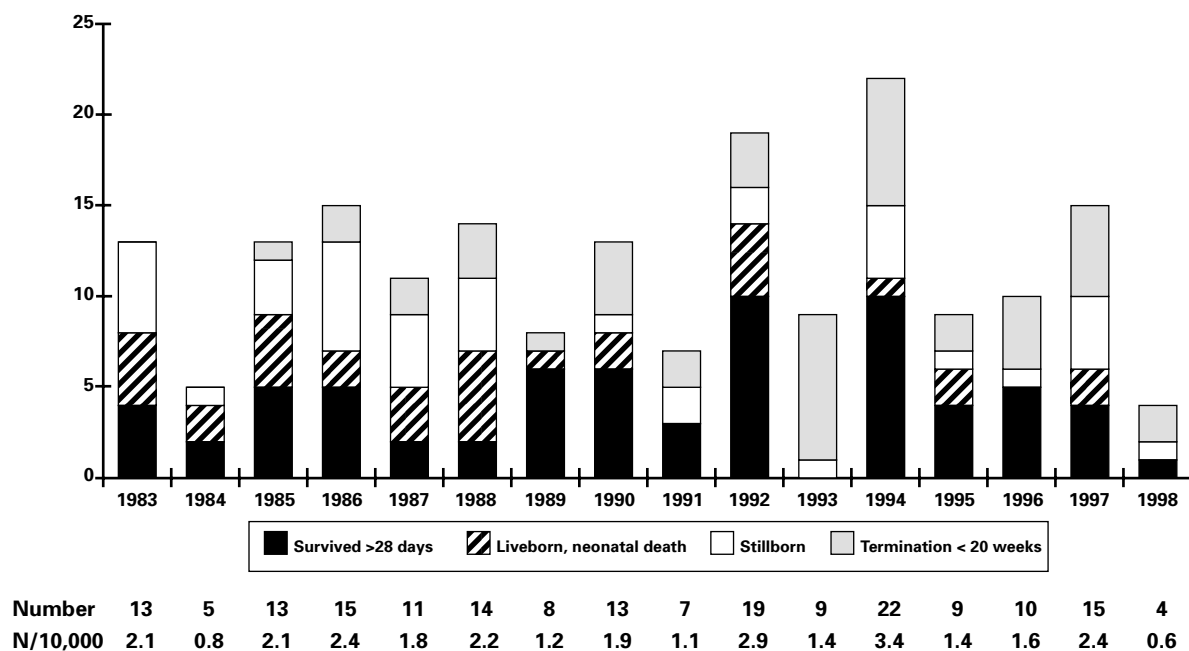
Table 3.2.3 Spina Bifida, 1983–1998, by Selected Maternal Characteristics

	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



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.	%		
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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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

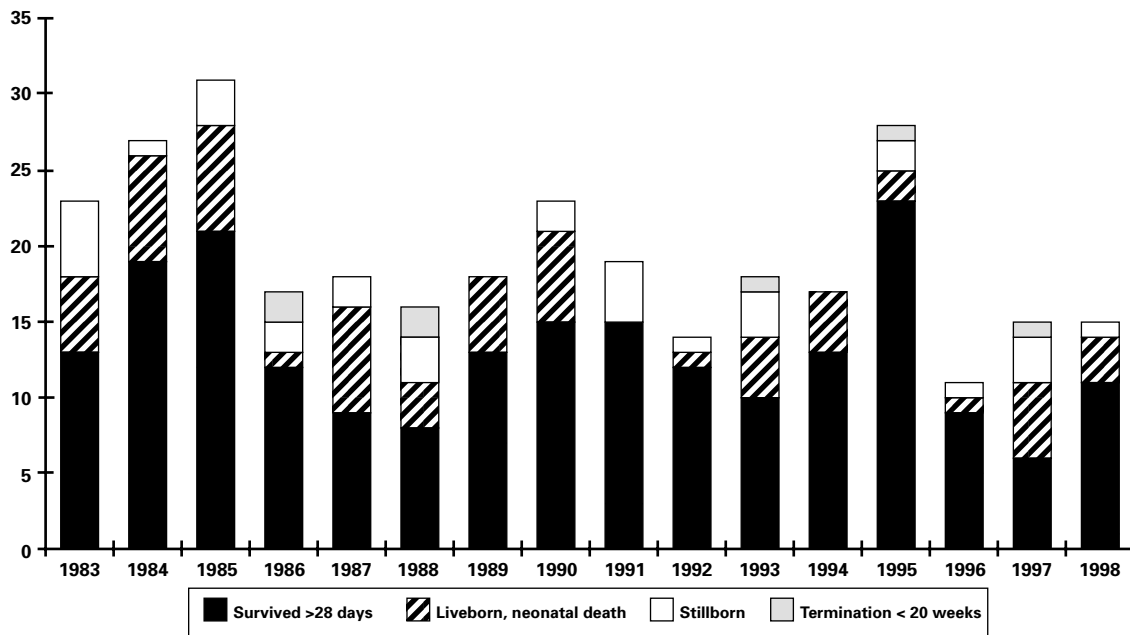


Figure 3.4 Microcephaly, Number of Cases by Year

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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

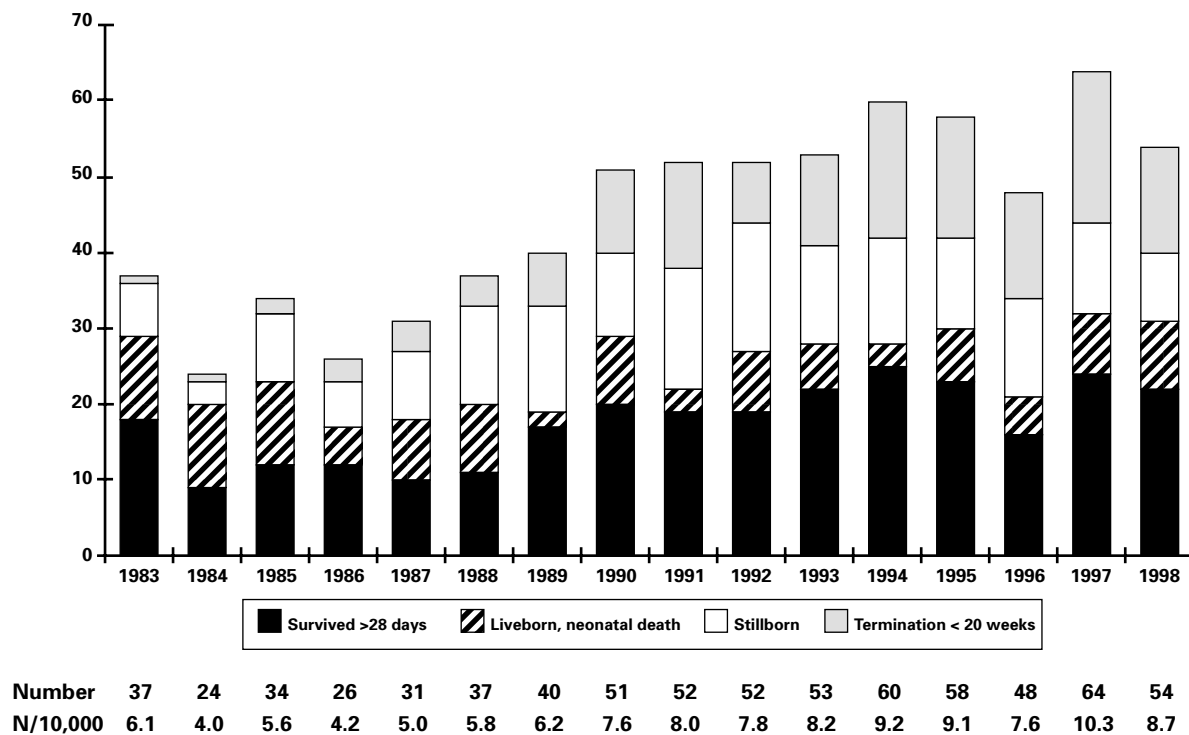
Table 3.4.3 Microcephaly, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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.

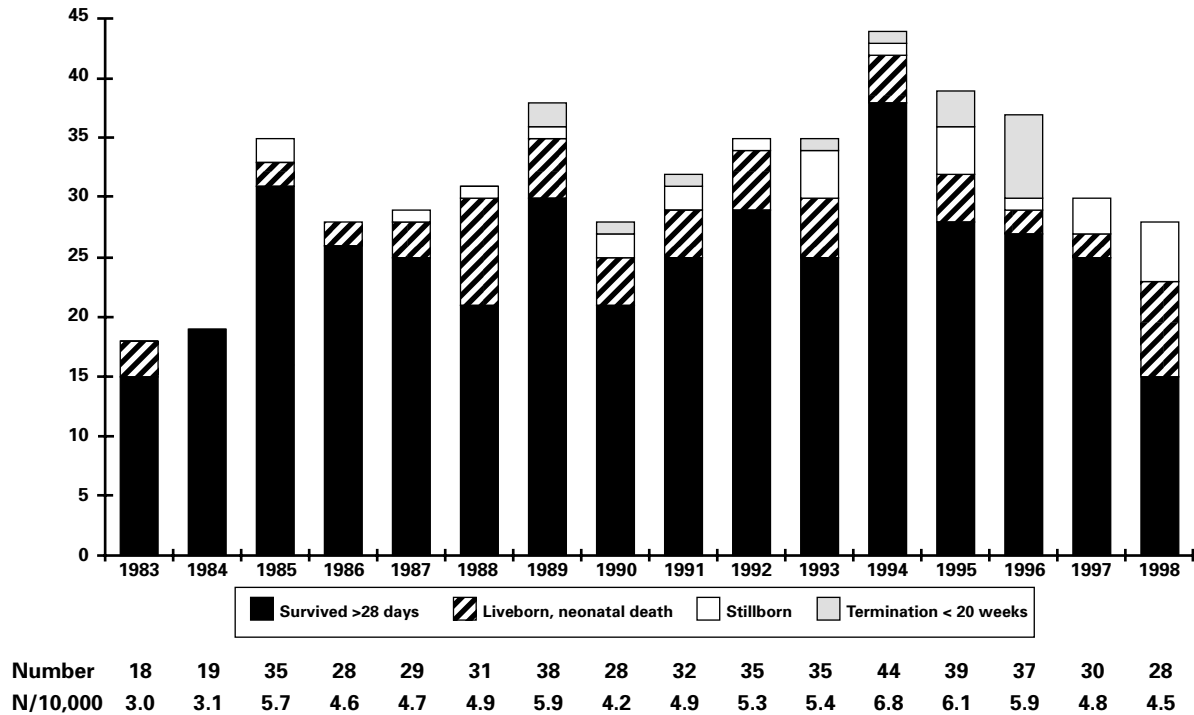
Table 3.5.3 Hydrocephalus, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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

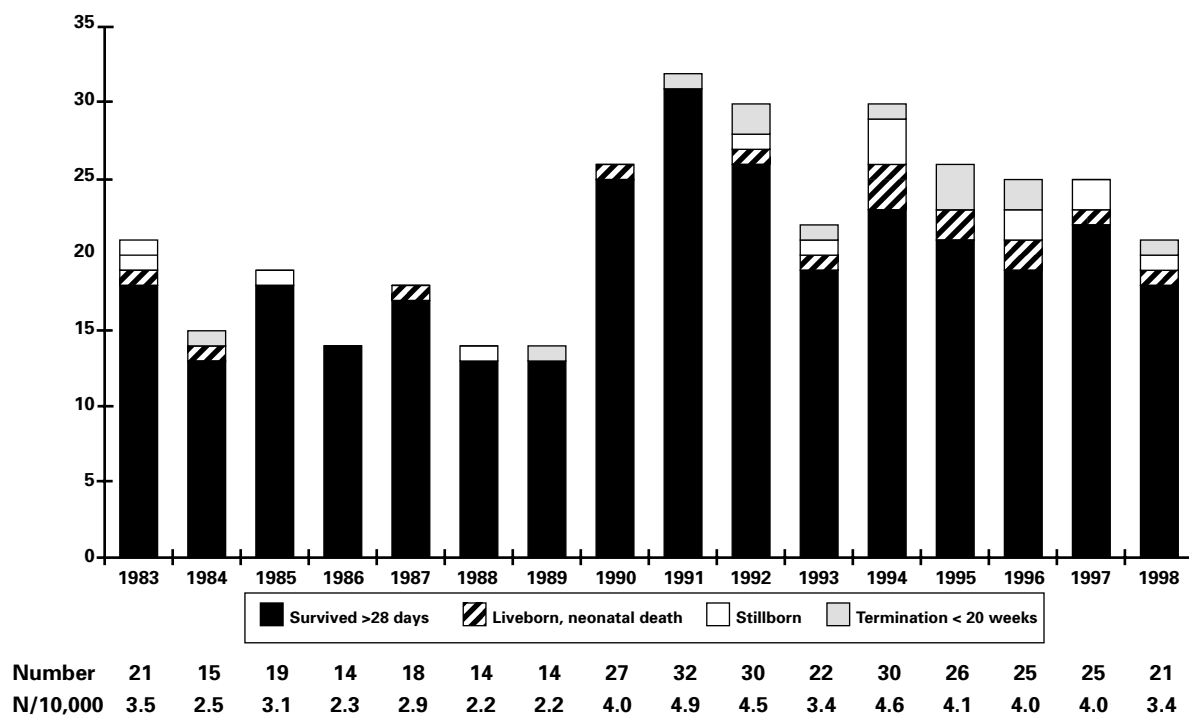
Table 3.6.3 Transposition of Great Vessels, 1983–1988, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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

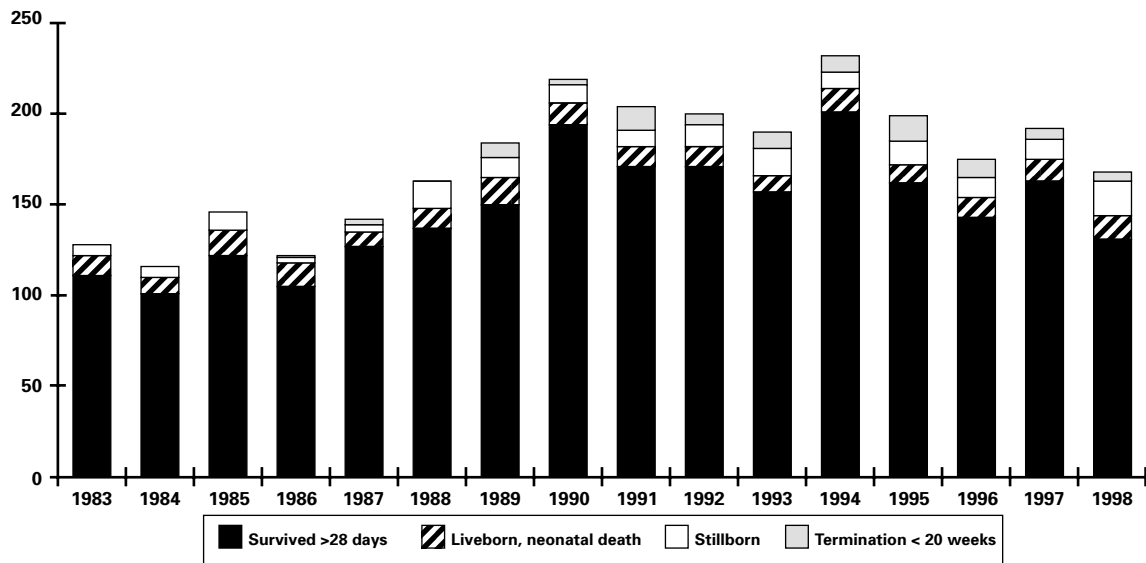
Table 3.7.3 Tetralogy of Fallot, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95%	CI
	No.	%	No.	%	No.	%	No.	%			
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 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.

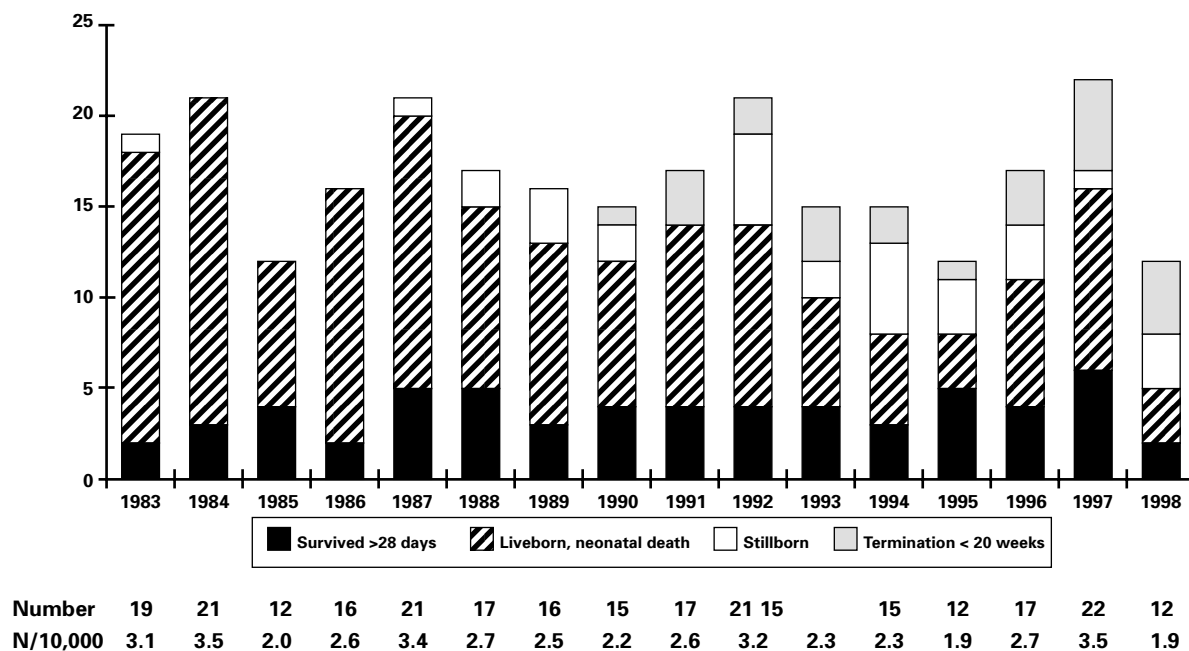
Table 3.8.3 Ventricular Septal Defect, 1983–1998, by Selected Maternal Characteristics

	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



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.

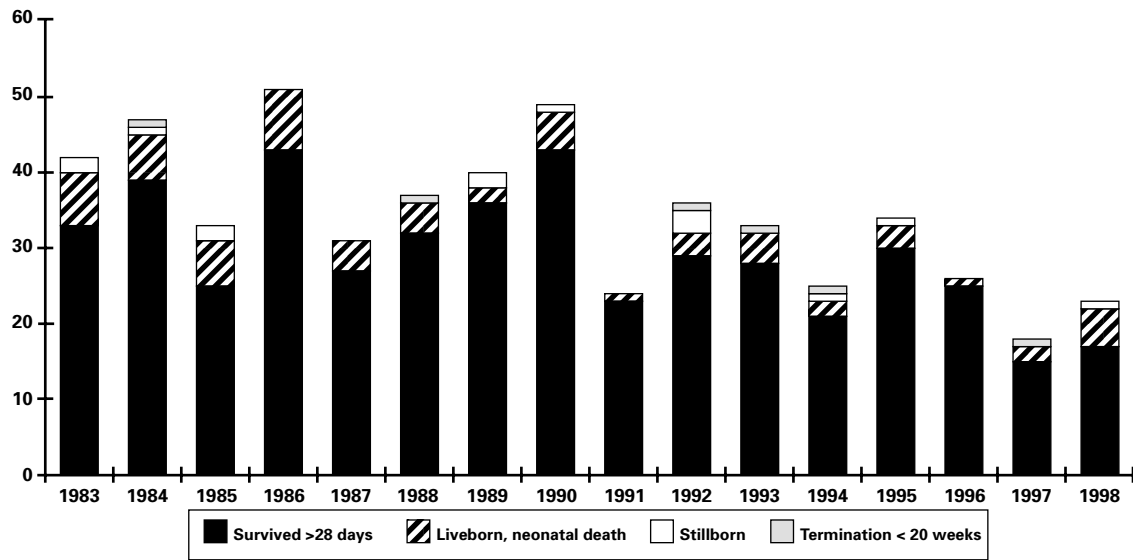
Table 3.9.3 Hypoplastic Left Heart Syndrome, 1983–1998, by Selected Maternal Characteristics

	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.

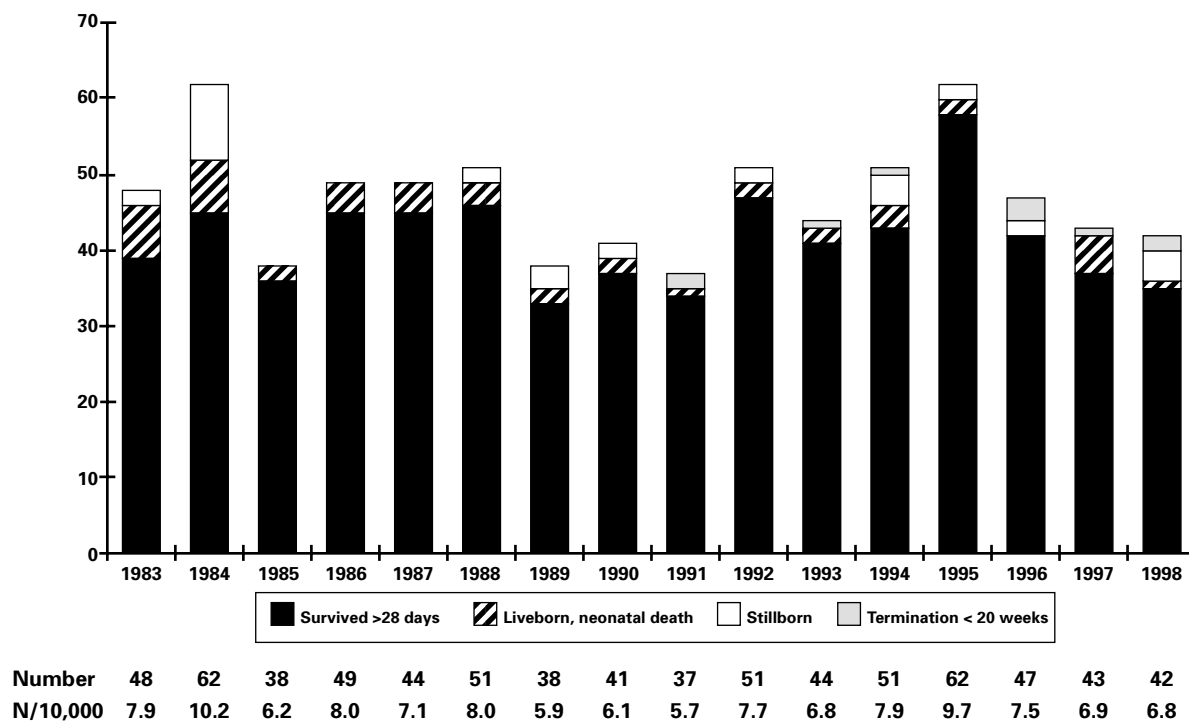
Table 3.10.3 Coarctation of Aorta, 1983–1998, by Selected Maternal Characteristics

	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



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.

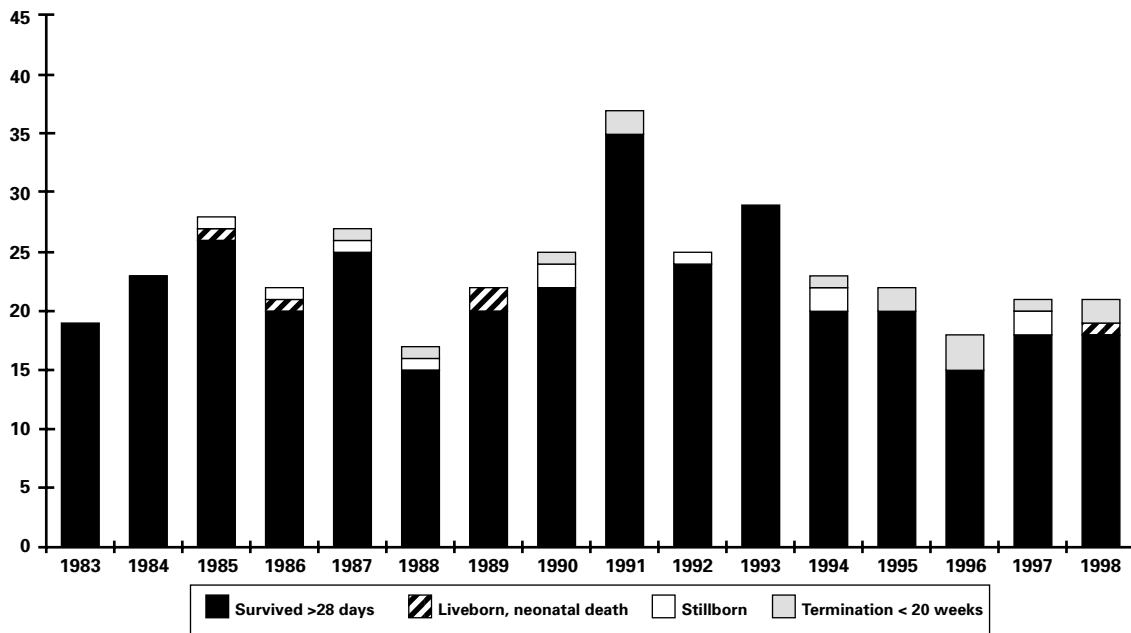
Table 3.11.3 Cleft Palate, 1983–1998, by Selected Maternal Characteristics

	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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

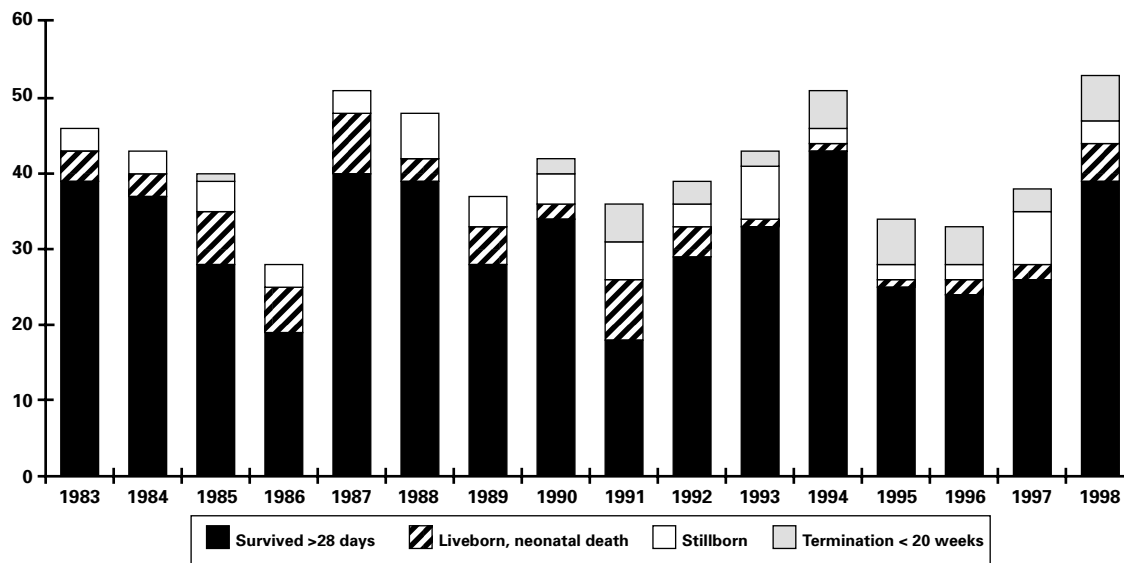
Table 3.12.3 Cleft Lip, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

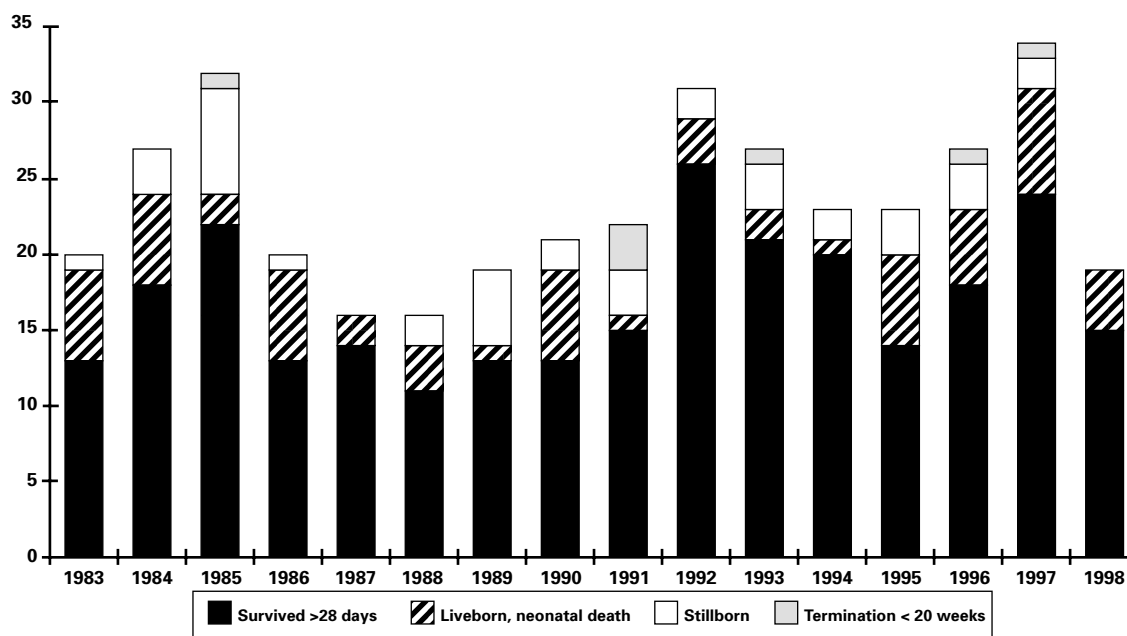
Table 3.13.3 Cleft Lip and Palate, 1983–1998, by Selected Maternal Characteristics

	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-oesophageal 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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

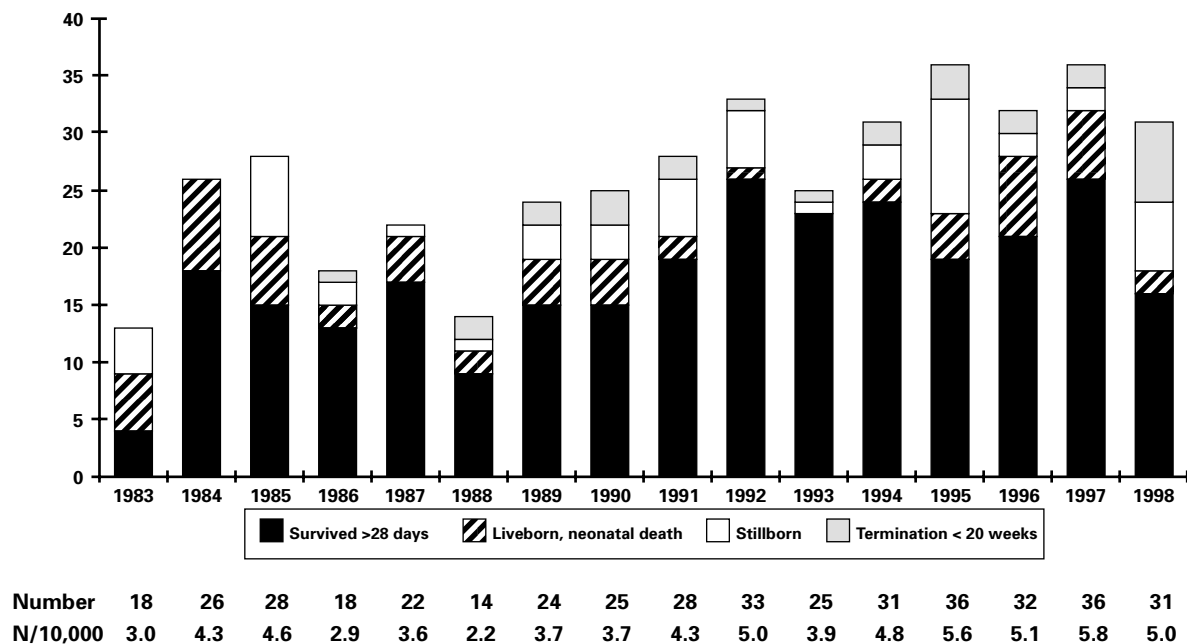
Table 3.14.3 Oesophageal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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.	%		
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.

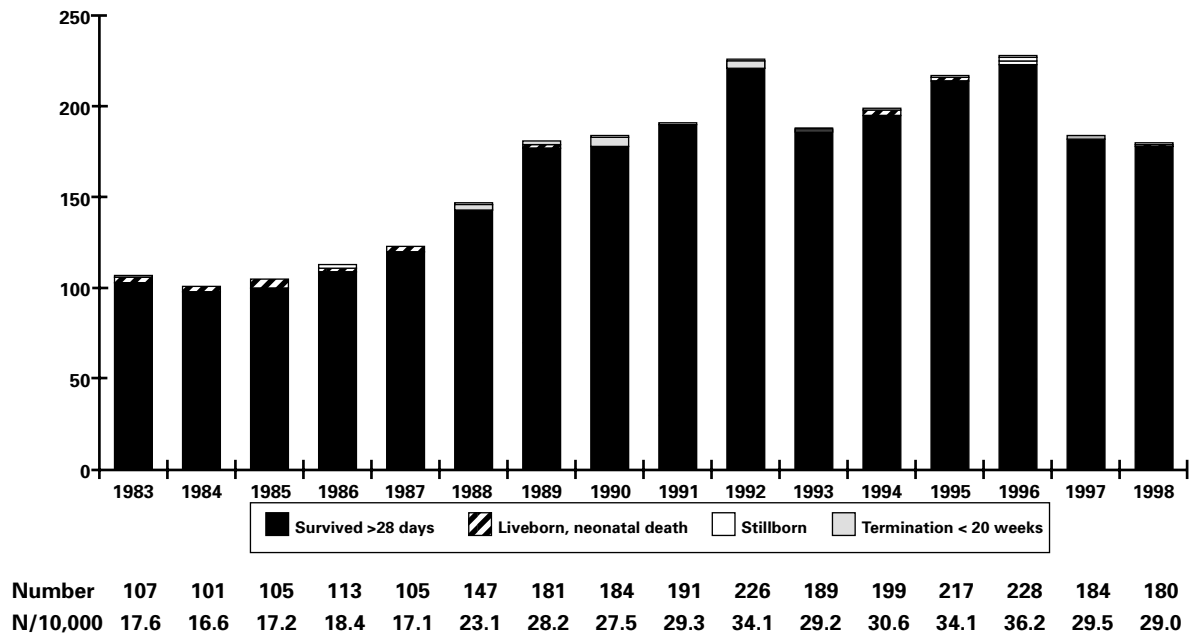
Table 3.15.3 Anorectal Atresia and/or Stenosis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

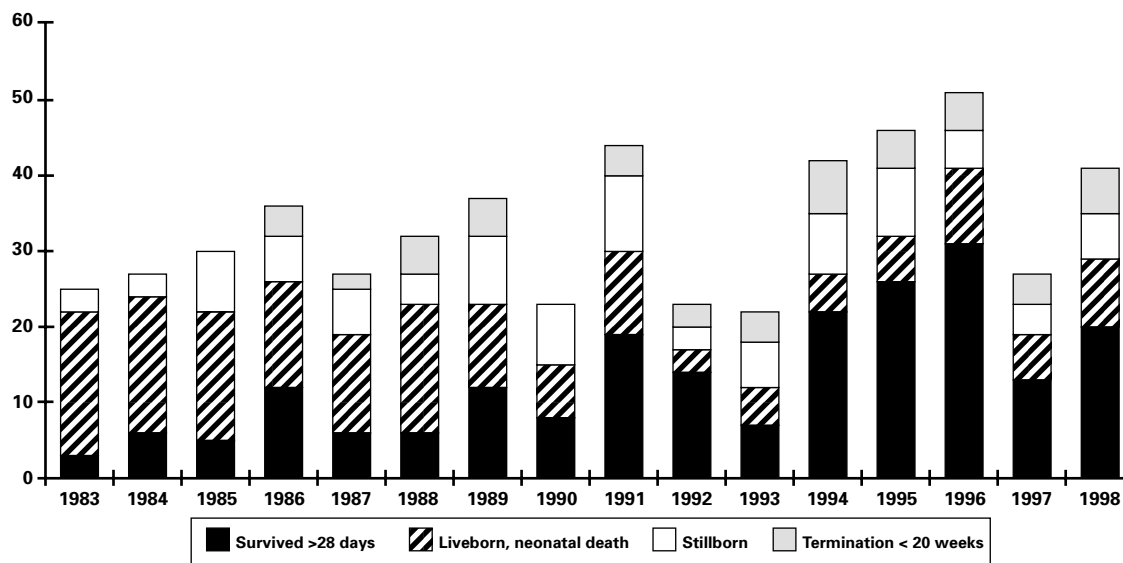
Table 3.16.3 Hypospadias, 1983–1998, by Selected Maternal Characteristics

	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



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 (Urinary)	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.

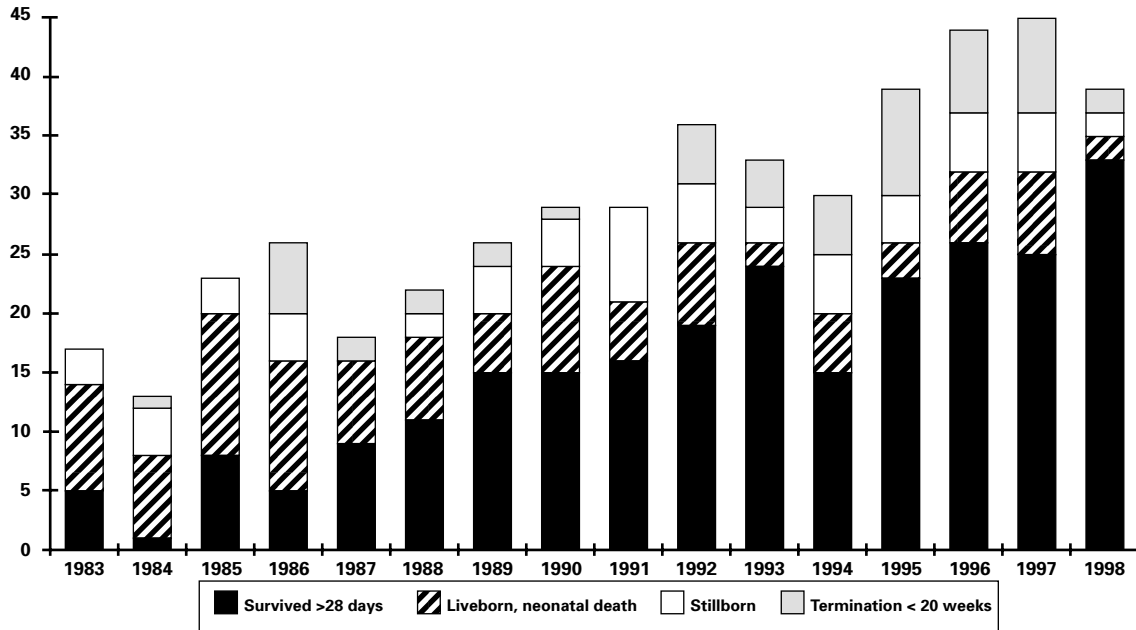
Table 3.17.3 Renal Agenesis and Dysgenesis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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 (Urinary)	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.

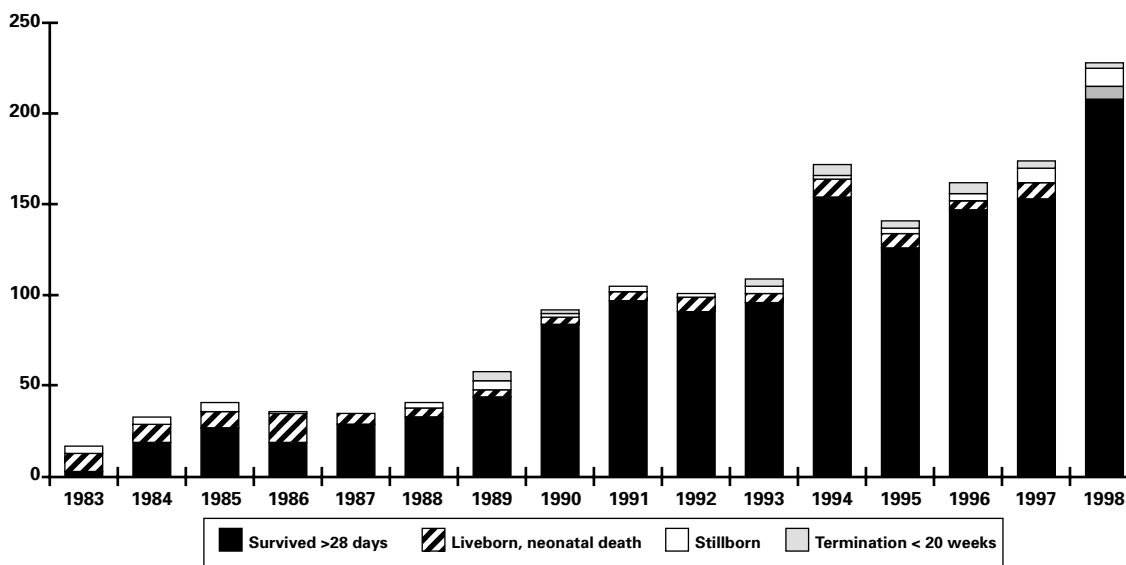
Table 3.18.3 Cystic Kidney Disease, 1983–1998, by Selected Maternal Characteristics

	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.

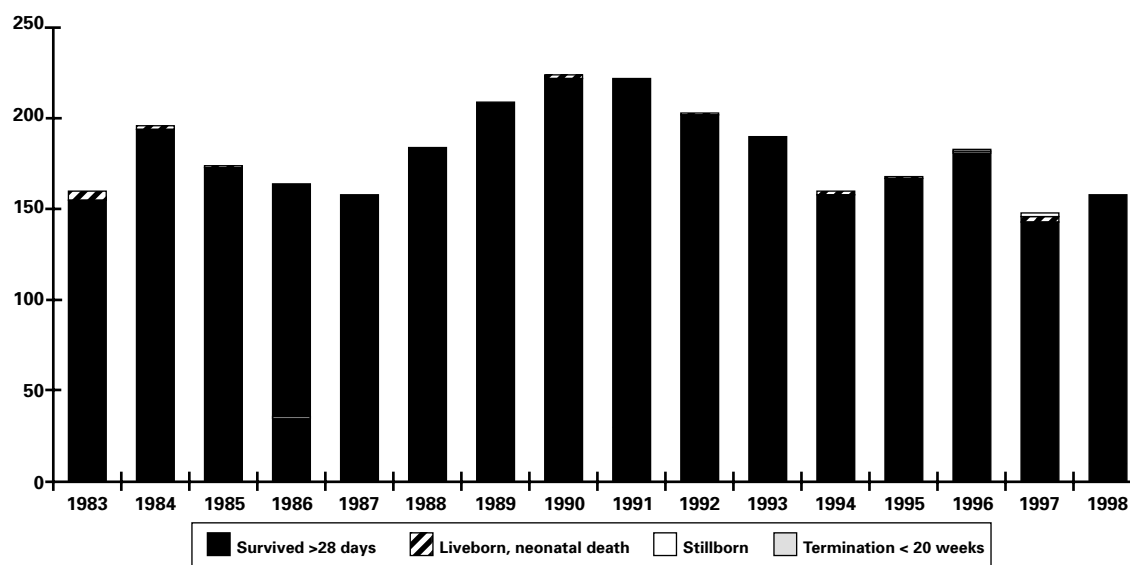
Table 3.19.3 Obstructive Defects of Renal Pelvis, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

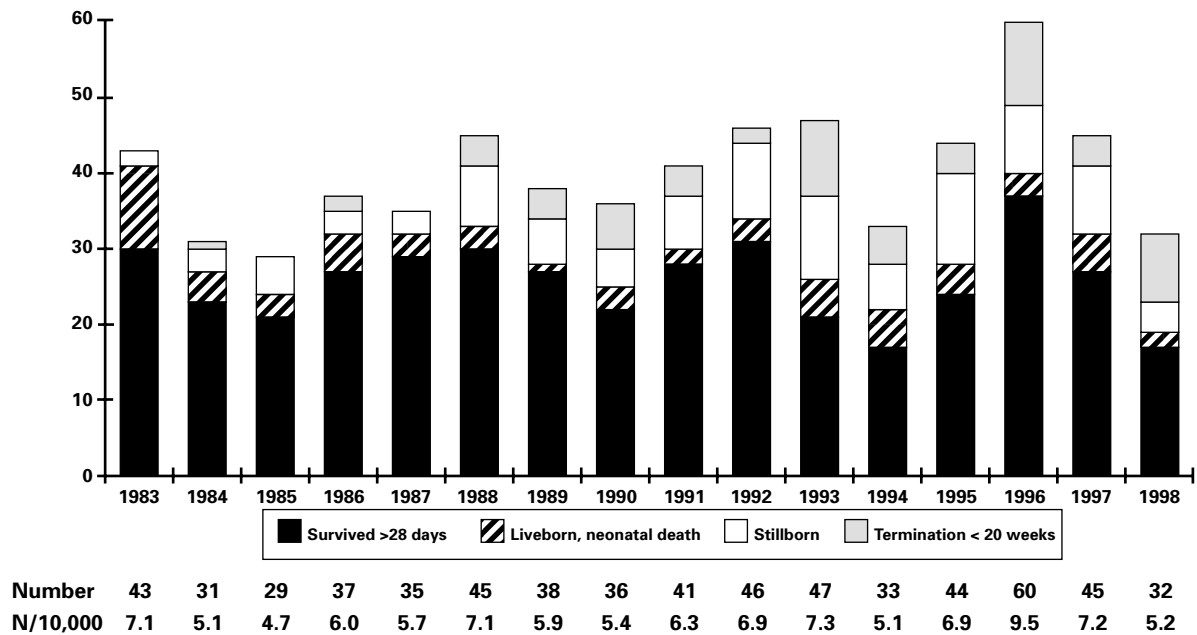
Table 3.20.3 Congenital Dislocation of Hip, 1983–1998, by Selected Maternal Characteristics

	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



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.

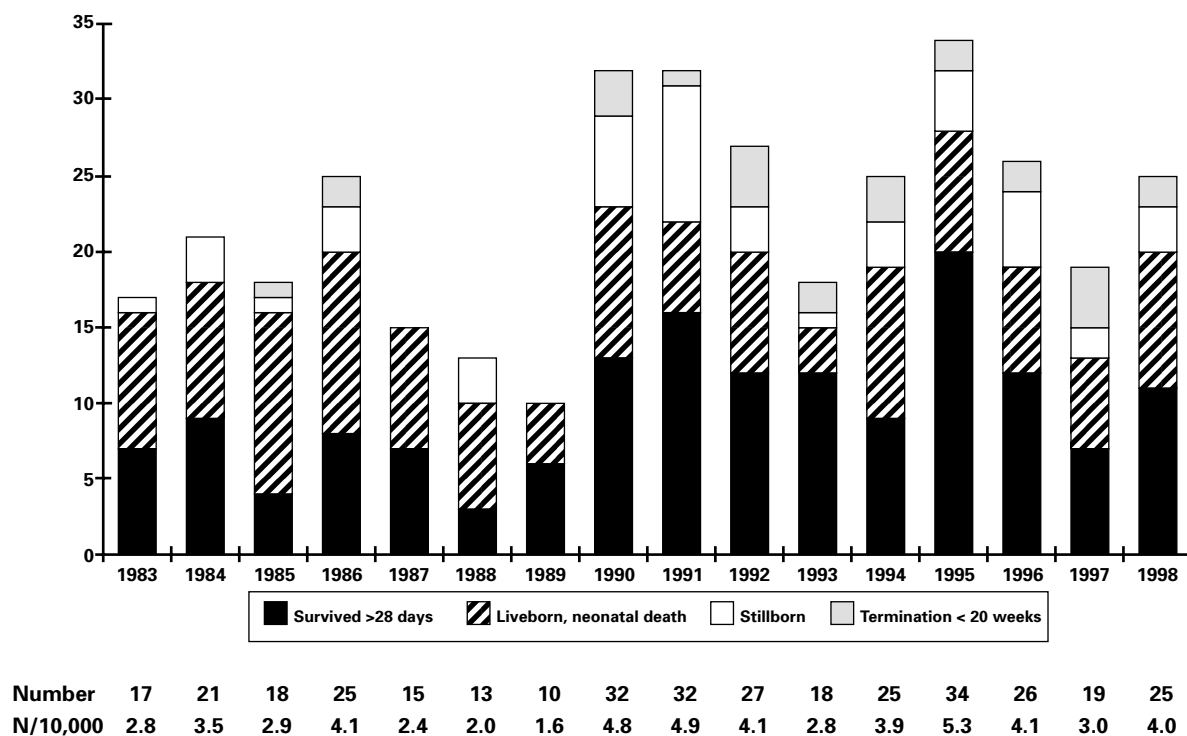
Table 3.21.3 Limb Reduction Defects, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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.

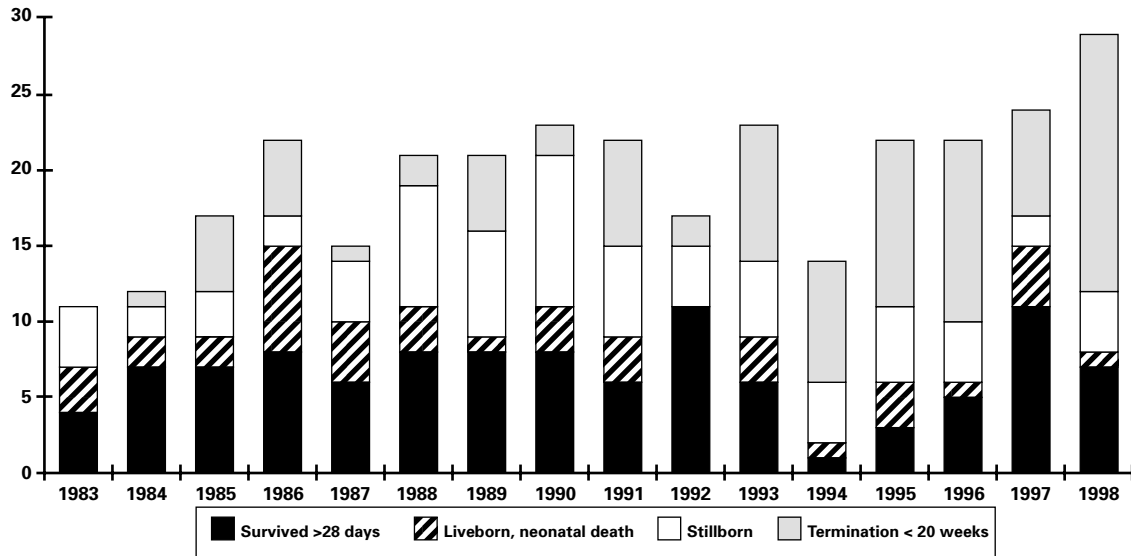
Table 3.22.3 Congenital Diaphragmatic Hernia, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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.

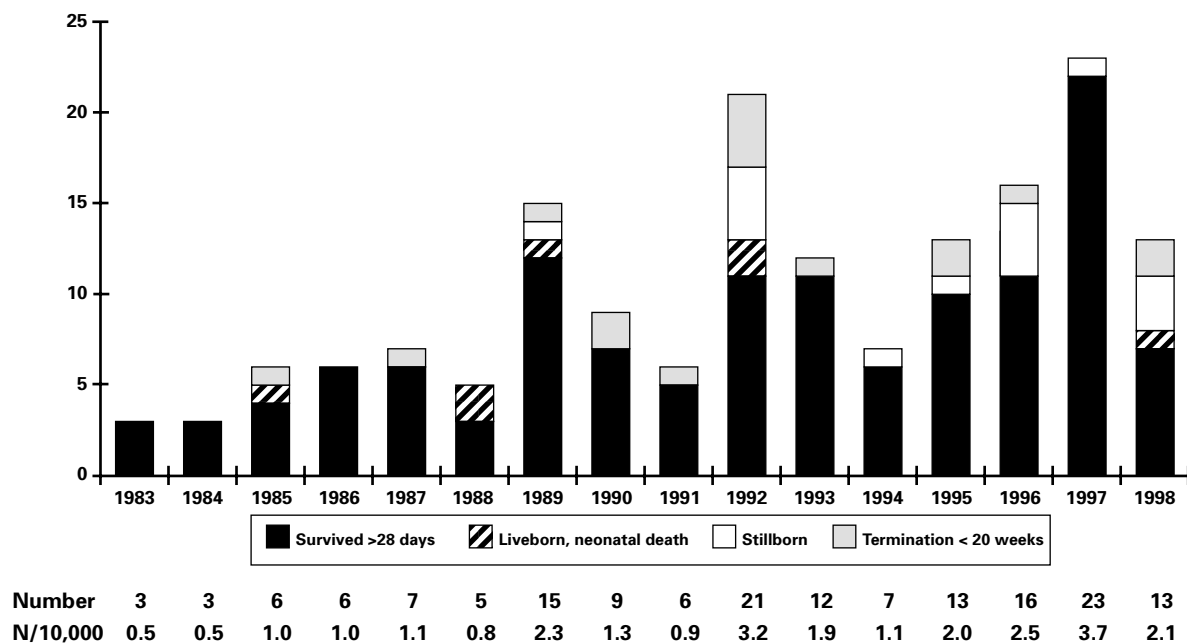
Table 3.23.3 Exomphalos, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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



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.

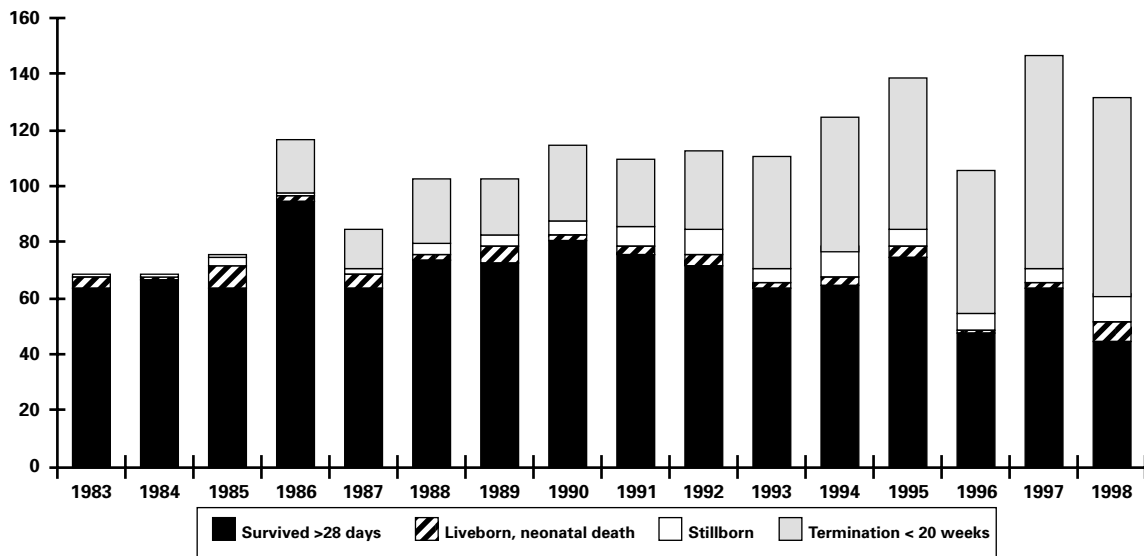
Table 3.24.3 Gastroschisis, 1983–1998, by Selected Maternal Characteristics

	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

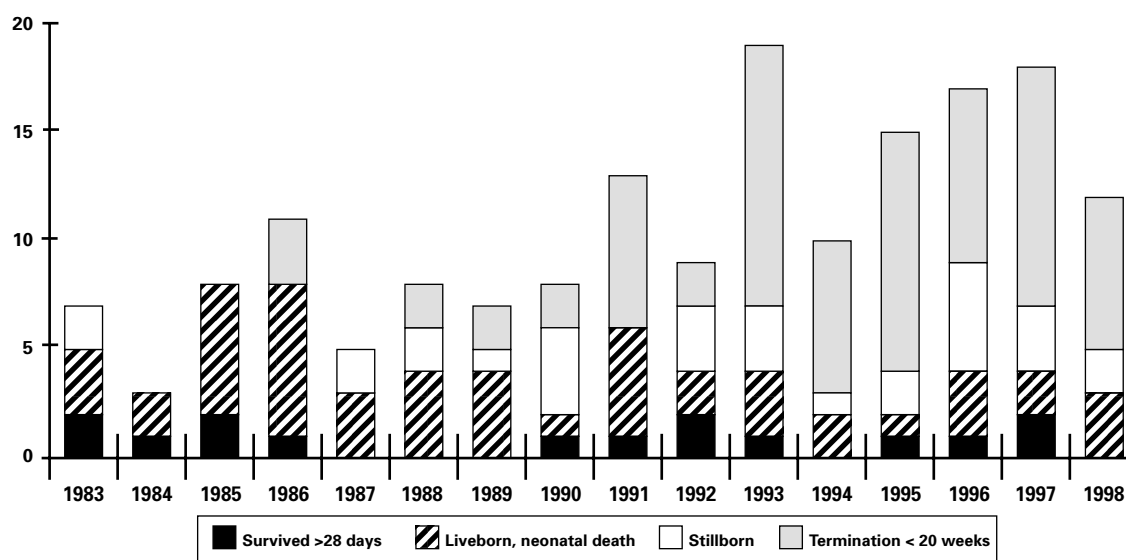
Table 3.25.2 Trisomy 21, 1983–1998, by Selected Maternal Characteristics

	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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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		
Neonatal death	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

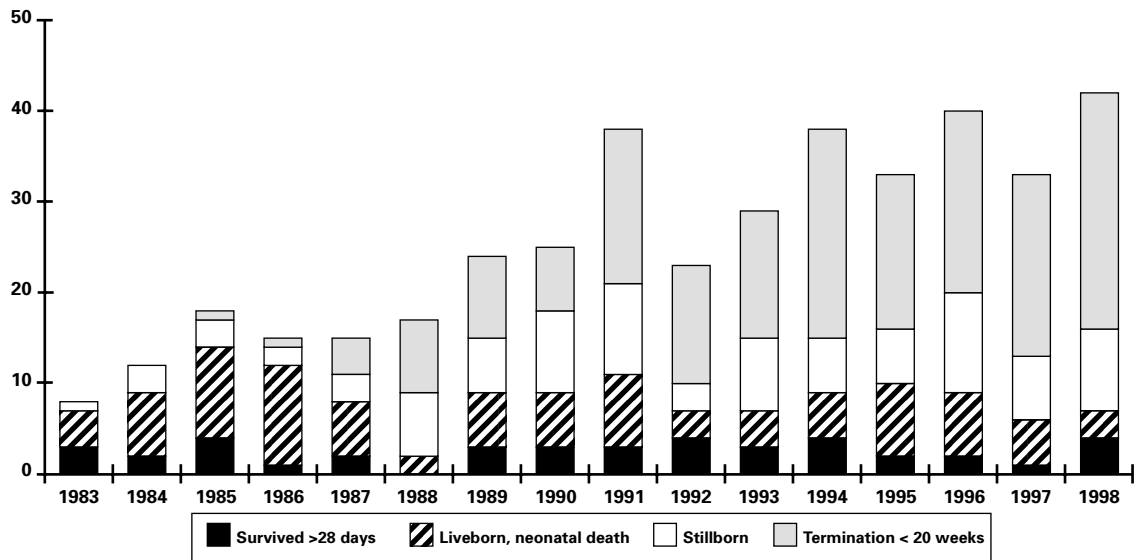
Table 3.26.2 Trisomy 13, 1983–1998, by Selected Maternal Characteristics

	All Confinements + TOPs (83–98)		Total Cases including TOPs (83–98)		1983–1994		1995–1998		Rate/ 10,000	95% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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% LL	CI UL
	No.	%	No.	%	No.	%	No.	%			
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% LL	Confidence Interval 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% LL	Confidence Interval 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% LL	Confidence Interval 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% LL	Confidence Interval 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
						LL	Interval 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
						LL	Interval 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% LL	Confidence Interval 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% LL	Confidence Interval 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
						LL	Interval 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
						LL	Interval 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
						LL	Interval 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% LL	Confidence Interval 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 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	Metatarsus varus
Accessory nipples	Micrognathia (unless severe)
Anal fissure	Mongolian spots
Balanced autosomal translocation (unless occurring with structural defects)	Occiput, flat/prominent
Birth injuries	Patent ductus arteriosus (< 37 weeks)
Birth marks (smaller than 4cm, not including giant naevus)	Philtrum, long/short
Bowing of legs (unless severe)	Plagiocephaly
Blocked tear ducts (dacryostenosis)	Pre-auricular sinus
Brushfield spots	Prominent forehead
Cephalhaematoma	Protruding tongue
Cleft gum	Ptosis
Clicky hips	Pylorus stenosis
Clinodactyly	Retrognathia (unless severe)
Craniotabes (unless severe)	Rocker-bottom feet (prominent heels)
Dermatoglyphic abnormalities	Sacral pits, dimples, sinuses
Ear abnormalities (minor)	Short sternum
Epicanthic folds	Simian creases
Gastro-oesophageal reflux	Single umbilical artery/2 vessels in cord
Haemangioma (<4 cm wide)	Skin folds/tags
Hernia—inguinal, umbilical	Slanting eyes
High-arched palate	Small mouth
Hydrocoele	Spina bifida occulta
Hypertelorism	Sternomastoid tumour
Imperforate hymen	Subluxating knee joint
Laryngeal stridor	Talipes (positional)
Laryngomalacia	Toe anomalies—minor
Low slung/set ears	Tongue tie
Macroglossia (large tongue)	Torticollis
Meckel's diverticulum	Ureteric reflux (ultrasound diagnosed)
Meconium ileus	Webbing of 2nd & 3rd toes/fingers
Mental retardations (unless occurring with a syndrome/structural defect)	Wide suture lines

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

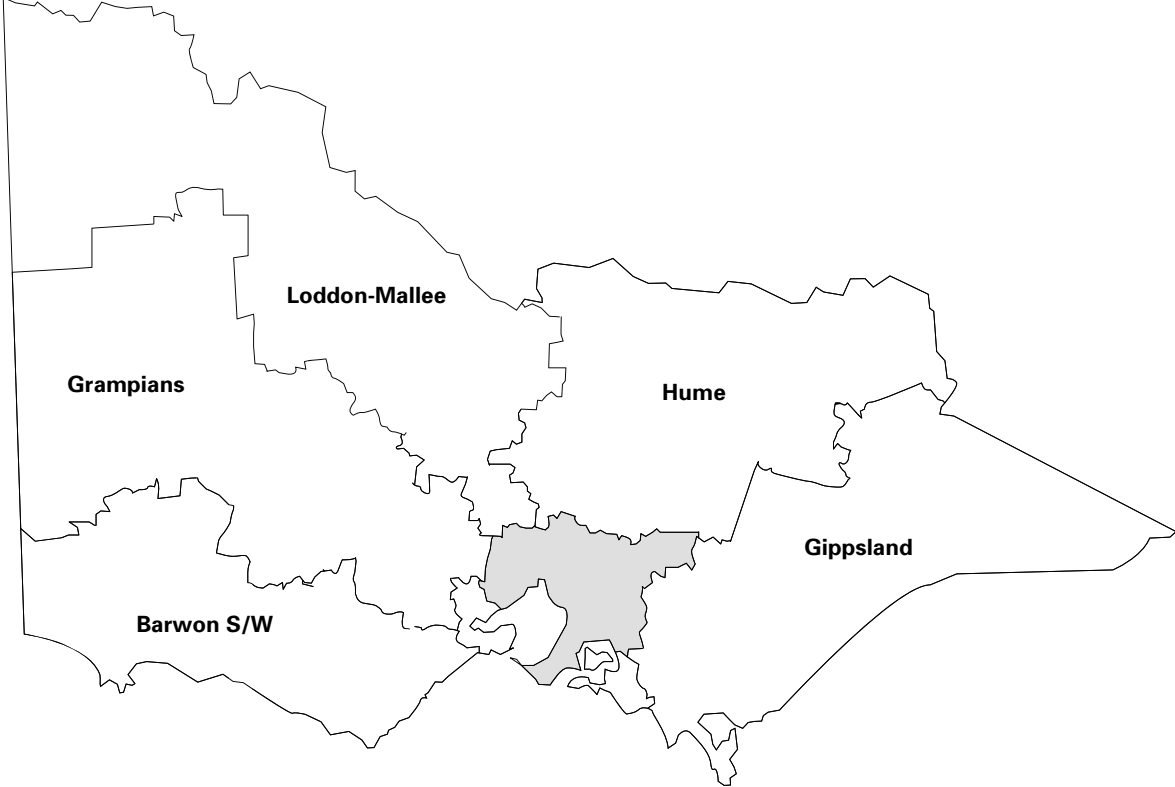
Maternal data: postcode, date of birth, method of prenatal diagnosis, name

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

