

PERINATAL DATA COLLECTION UNIT

Congenital Malformations in Victoria 1983-1994

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HIGHLIGHTS

- In the 12 year study period, 1983 to 1994, there have been 22,710 babies born with a congenital malformation at or after 20 weeks gestation. Another 1,311 were identified as terminations of pregnancy before 20 weeks gestation. This gives an overall prevalence of 315/10,000 or 3.1%.
- There has been no decline in the total number of anencephalic nor spina bifida fetuses, although the number of livebirths has declined due to prenatal diagnosis of affected fetuses. The overall prevalence of these malformations in pregnancy is 1/1900 for anencephaly, 1/1200 for spina bifida or 1/735 for either neural tube defect. However, there are now less than 20 babies per year liveborn with spina bifida.
- Advanced maternal age is significantly associated with microcephalus, hydrocephalus, transposition of the great arteries, ventricular septal defect (VSD), Down syndrome and other chromosome abnormalities.
- Teenage mothers have a significantly increased risk of having babies with spina bifida, hydrocephalus, renal agenesis and gastroschisis.
- There are some significant gender differences in birth prevalence with males at higher risk overall compared with females. Congenital malformations significantly associated with males are often cardiac (transposition of the great arteries, Tetralogy of Fallot, coarctation of aorta) or renal (anorectal atresia, renal agenesis, cystic kidney disease, obstructive defects of renal pelvis). Males are also at increased risk of cleft lip and palate, oesophageal atresia and Down syndrome, while females only have an increased risk for anencephaly, VSD and congenital dislocation of the hip.

- Congenital dislocation of the hip is the most common (sentinel) congenital malformation, reported in 29.3/10,000 births or 1/340 births, with female babies having a fourfold increased risk for this condition over male babies. VSD is the second most common with an overall prevalence of 26.3/10,000 or 1 / 380 births.
- There has been a significant decline in the number of reported microcephalic babies.
- Gastroschisis numbers have declined again, after a large increase in 1992.
- The number of babies living beyond 28 days with hypoplastic left heart, diaphragmatic hernia or exomphalos has not increased.
- Obstructive defects of the renal pelvis have become more prevalent, probably because of ultrasound detection in pregnancy.
- 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. We now have approximately 80 liveborn babies with Down syndrome each year in Victoria.

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. 2-3% of Victorian babies are born each year with a malformation and they are a major contributor to both perinatal mortality and morbidity.

It was the Yarram Inquiry of 1978 which highlighted the lack of routinely collected data on not only congenital malformations, but also on births in the

State. This led to the establishment of the Victorian Perinatal Data Collection Unit (VPDCU). Under the Health Act of 1982 one of the fundamental purposes for the establishment of the VPDCU was the formation of a Congenital Malformations/Birth Defects Register (CMR).

1.2 Purposes

The Victorian Congenital Malformations/Birth Defects Register is a state-wide population based surveillance system established to:

1. provide statistical information to organisations responsible for planning health care facilities for those with congenital malformations,
2. provide background information useful in assessing the factors which cause (or do not cause) birth defects,
3. to provide information for epidemiological research.

It 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 obtain information on internal errors in metabolism, haematological disorders, congenital infections, neoplasms, and developmental delay. There are certain isolated minor malformations that are not notifiable and these are listed in Appendix. A.

The CMR 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 CMR by December 31, 1995.

1.4 Sources of Notification

The CMR is a voluntary notification system. Data are obtained from multiple sources . Table 1 details the number of notifications of congenital malformation cases for 1989-1994 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 only been maintained since 1989. For the period 1989-1994 there were 22,837 notifications for a total of 12,457 cases. This approximates to 1.8 notifications per case.

Table 1.1 Sources of Notification, 1989-1994

Notification Source.	Number.	Percent(%)
1. Perinatal Unit.	9409	41.2
2. Perinatal Death Certificate.	1150	5.0
3. Autopsy Report.	742	3.2
4. Cytogenetics Report.	1361	6.0
5. M.C.H. Nurse.	2555	11.2
6. Hospitals.	7483	32.8
7. Other Professionals	86	0.4
8. Other (e.g. parent)	18	0.1
9. Unknown.	33	0.1
Total	22,837	100

1.5 Data Items

The CMR collects maternal, infant and fetal demographic data on all cases. All notifications (excluding terminations 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 CMR are listed in

Appendix. B. Further data items are available for each case from the Perinatal Data Collection Unit where all the information from the morbidity statistics forms are recorded.

1.6 Data Quality

Because of the voluntary nature of the CMR, it has been necessary to assess data quality by validation studies (1,2). The first, conducted in 1986, discovered that the proportion of congenital malformations notified to the CMR 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 CMR. The register has been updated from inpatient listings from the RCH detailing all children born since 1982 who have been admitted to the Royal Children's Hospital (RCH) with a congenital malformation since 1982. We have also obtained listings of all children born since 1982 who have visited the RCH Cardiology Unit, Orthopaedic Unit and the Victorian Clinical Genetics Service (VCGS) Metabolic Clinic, either as an inpatient or outpatient.

This procedure has also been adopted for the Mercy Hospital for Women, where the inpatient listings date back to 1989, and Monash Medical Centre where we have followed-up inpatients admitted since 1992.

1.7 Explanatory Notes

Cases versus Malformations: Section 2 describes *each individual malformation*. Sections 4 & 5 describe *congenital malformation cases* - the number of liveborn or stillborn infants affected by at least one congenital malformation. Thus, the number of congenital malformations 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 VPDCU to code the syndrome **and** all of its manifestations.

Differences with AIHW NPSU Report (3): 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 congenital malformations in the State of Victoria.

Graphs: In Section 3, Major congenital malformations, where the combined stillbirth (SB) and neonatal death (NND) figure for a specific malformation is greater than 25%, the graphs include a breakdown of the various perinatal outcomes (ie. termination < 20 weeks (TOP), SB, NND, survived >28 days.) Where there are more than ten terminations, the graphs depict the number of infants born with that particular condition and the number terminated between 1983-1994.

1.8 Terminations

Termination of pregnancy refers to an induced abortion by medical or mechanical means before 20 weeks gestation. If a termination occurs 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 VPDCU and the CMR.

Notification of terminations for malformations only began in 1986. While the capture of this 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 VPDCU staff member has been responsible for reviewing the medical records of all patients with terminations for malformations at both MMC and Royal Women's Hospital (RWH). A letter is also sent to all hospitals with maternity beds to obtain information on patients who meet this criteria.

For the purposes of this report, inclusion of data on terminations has been restricted to the overall prevalence of each specific condition. No termination data has been included in the breakdown of selected maternal and infant characteristics, nor in tables of birth defects by major anatomical site .

1.9 Statistics

Chi square tests for heterogeneity, linear trend and relative risk were done using Epi Info Version 6.1. Due to the large numbers in the study population, a p value of 0.01 was chosen as representing a significant difference.

1.10 Regional variations

Victoria is divided into eight regions for health data analysis, five rural and three metropolitan (see Appendix B for map). There was a change in region boundaries in 1992, but we have used the `old' regions throughout the report. Table 4.3 summarises the data for the 12 study years. In the text there are references to significant regional differences in birth prevalence. However, without further detailed analysis, it is not possible to provide any explanations for these findings. It must be remembered that there are very large numbers involved and heterogeneous populations within and between regions.



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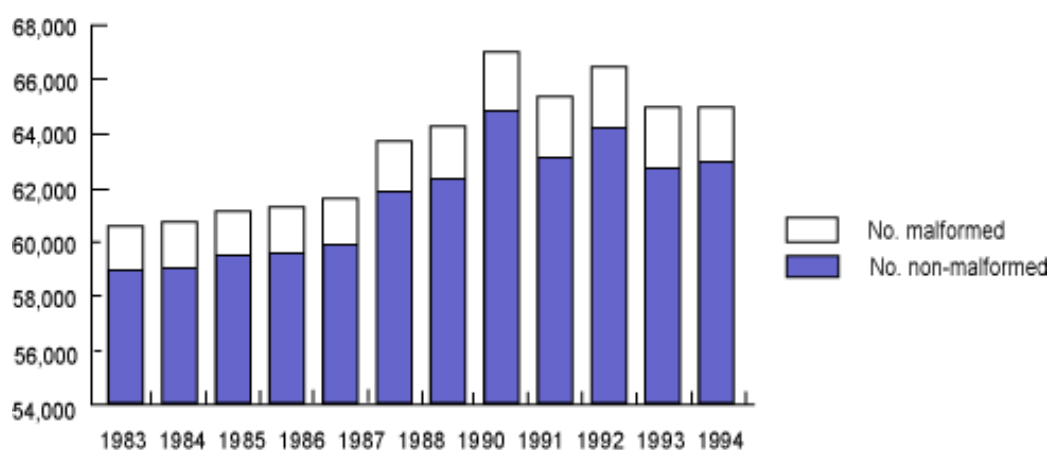
2. SUMMARY TABLES

2.1 Congenital malformations by year, 1983-1994

The combined birth prevalence of congenital malformations over 12 years is 298.3/10,000 or 3.0%. There have only been slight variations over the 12 years.

Table 2.1 Congenital malformations by year, 1983-1994

Year	Total Births 20 weeks and later	Malformations 20 weeks and later	Malformations, before 20 weeks (terminations)	N/10,000	%
1983	60628	1699	4	280.2	2.8
1984	60738	1752	11	288.5	2.9
1985	61186	1623	17	265.3	2.7
1986	61253	1643	83	270.6	2.7
1987	61567	1666	65	240.6	2.7
1988	63658	1870	109	293.8	2.9
1989	64245	1971	125	306.8	3.1
1990	66876	2171	147	324.6	3.2
1991	65248	2132	139	326.8	3.3
1992	66305	2128	156	320.9	3.2
1993	64737	2045	207	315.9	3.2
1994	64895	2010	248	309.7	3.1
Total	761,336	22,710	1,311	298.3	3.0



Figure

2.1 Number of congenital malformations as a proportion of total births, 1983-1994

2.2 Congenital malformations by major anatomical system, Victoria, 1984-1993

The following figures refer to individual congenital malformations, **not cases**.

Table 2.2 Congenital malformations by major anatomical site, 1983-1994

Codes	Malformations	Number	%	N/10,000
Total	36,175			
740-2	Nervous system	2441	6.75	32.06
743	Eye	625	1.73	8.21
744	Ear, face & neck	695	1.92	9.13
745-6	Heart	6624	18.31	87.00
747	Circulatory system	3248	8.98	42.66
748	Respiratory system	1057	2.92	13.88
749	Cleft palate/lip	1288	3.56	16.92
750-1	Digestive system	2727	7.54	35.82
752	Genital organs	2677	7.40	35.16
753	Urinary system	2114	5.84	27.77
755	Limbs	2399	6.63	31.51
754/6	Other musculo-skeletal	5702	15.76	74.89
757	Integument	286	0.79	3.76
758	Chromosomal	1821	5.03	23.92
759	Other & unspecified	809	2.24	10.63
760	Maternal conditions	4	0.01	0.05
7710-2	Congenital infections	48	0.13	0.63
140-239	Neoplasms	236	0.65	3.10
240-279	Endo/Nutri/Meta-bolic	960	2.65	12.61
280-289	Disease of blood	125	0.35	1.64
315/8	Dev Delay	57	0.16	0.75
7780	Hydrops	161	0.45	2.11
7786	Hydrocoele	71	0.20	0.93

Congenital Malformations in Victoria 1983-1994

3. MAJOR CONGENITAL MALFORMATIONS

Anencephaly

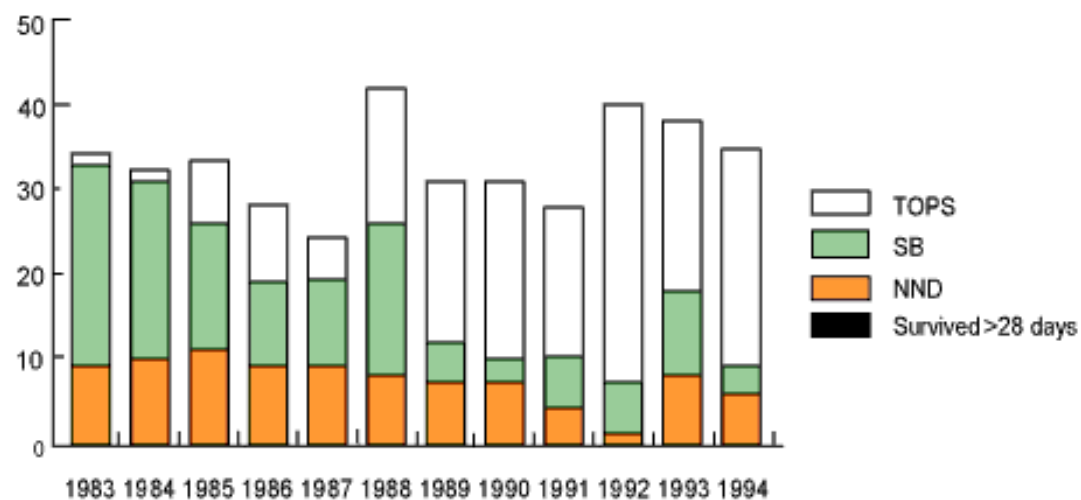


Figure 3.1.1 Anencephaly, number of cases by year

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
- The number of pregnancies reported to the CMR (sum of terminations and

births) has shown annual fluctuations with no evidence of a significant decline in the actual number of pregnancies. In 1983 the overall prevalence was 1/1784 of all pregnancies, compared with 1/1909 in 1994.

- When comparing absolute numbers of anencephalic babies with those of spina bifida (next section), we find 30-35 per year for anencephaly and 50-55 for spina bifida. On the basis of a study⁵ done in 1980 -1981, before widespread usage of ultrasound and amniocentesis in Victoria, we would expect equal proportions of these malformations. Therefore, this discrepancy is difficult to explain, unless incomplete ascertainment of early terminations of anencephalic fetuses, occurring after first trimester ultrasound, explains the 20 'missing cases' each year.

- There is an excess of females (two-fold increased relative risk for female fetuses RR = 1.93; 95% CI 1.46-2.56, p < 0.0001)

- Nine percent of anencephalic babies are one of a twin. This is more than twice the rate expected for babies with a malformation (RR = 2.30; 95%CI 1.51-3.51, p = 0.0001)

- There is a non-significant association of anencephaly with maternal age 20-30 years versus all other ages (RR = 1.39; 95%CI 1.05 - 1.84, p = 0.02).

- Women from Asia and the Middle East are at a non-significantly lower risk of having an anencephalic baby.

- The regional differences in birth prevalence are not significant

Table 3.1.1 Anencephaly, 1983-1994, by selected infant characteristics

	No. Cases	%	Total No. Births	%	N/10,000
Total live & stillbirths	221		761,336		2.9
Sex					
Male	76	34.4	391,073	51.4	1.9
Female	139	62.9	370,088	48.6	3.8
Indeterminate	6	2.7	175	0	342.9

Birthweight(g)

<1000	74	33.5	4709	0.6	157.1
1000-2499	104	47.1	40,317	5.3	25.8
2500+	27	12.2	715,328	94.0	0.4
Unknown	16	7.2	982	0.0	162.9

Plurality

Singleton	200	90.5	741,939	97.5	2.7
Twin	20	9.0	18,746	2.5	10.7
Triplet	1	0.5	615	0.1	16.3
Other	0	0	36	0	-

Outcome

Survived >28 days	0	0	752,298	98.8	-
Stillbirth	131	59.3	5611	0.7	233.5
NND	90	40.7	3427	0.5	262.6

Total TOPs (<20weeks)

1,311

**Total pregnancies
(inc.all TOPs)**

762,647

	No.cases	1983-88	1989-94	N/10,000
TOPs	176	39	137	
Livebirths& Stillbirths	221	154	67	
Total	397	193	204	5.2

