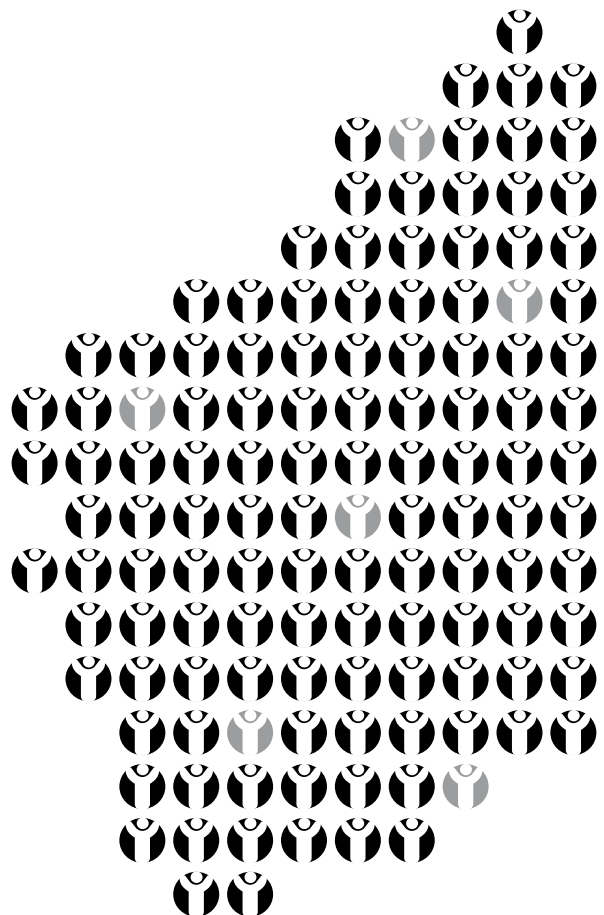


Report of the Birth Defects Registry of Western Australia 1980-2007

JANUARY 2009



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**REPORT OF THE
BIRTH DEFECTS REGISTRY OF WESTERN AUSTRALIA**

1980 - 2007

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We gratefully acknowledge the support and collaboration of our colleagues at Princess Margaret Hospital, the Telethon Institute for Child Health Research and, especially, at King Edward Memorial Hospital.

Our thanks also to the specialist advisers to the Registry, and to the members of our Advisory Committee and Consumer Reference group, for their guidance and support.

FOREWORD

The Western Australian Birth Defect Registry is an important state-wide resource that provides valuable data to health professionals, researchers, government agencies and community organisations.

During the past two years the Registry staff, lead by Professor Carol Bower, have embraced the opportunity to enter a new phase in the Registry's operations and engage in an active partnership with health consumers and community organisations.

This partnership, which was precipitated through the process of seeking a mandatory notification process for the Registry, has enabled the establishment of a community reference group. The reference group, consisting of representatives from parent support groups, the Health Consumers Council and the Telethon Institute for Child Health Research was formed following community workshops held in 2007.

Whilst the main work of the reference group is yet to be done due to delays with mandating the notification process, it is certainly an exciting new phase in the history of the Registry.

The reference group is looking forward to having the opportunity to work with the Registry staff and Advisory Committee to discuss issues such as raising awareness within the wider community of the existence, purpose and outputs of the Registry, the sensitivity and timing of informing parents of the Registry, access and security of data held on the Registry and ethical standards relating to research using data from the Registry.

I am privileged to have the opportunity to work with both the Registry staff and the dedicated members of the reference group who have shown such patience in the long process but remain committed to ensuring that the Western Australia Birth Defects Registry continues to provide world-class data to fulfil its aims.



Anne McKenzie
Consumer Research Liaison Officer
Telethon Institute for Child Health Research

SUMMARY

- The Western Australian Birth Defects Registry has a commitment to obtain high quality, complete and population-based information on birth defects for Western Australia (WA).
- For the purposes of the Registry, a birth defect is defined as a structural or functional abnormality that is present at conception or occurs before the end of pregnancy, and is diagnosed by six years of age.
- The numerator data in this report comprise malformations occurring in livebirths and stillbirths in WA and in pregnancies terminated because of fetal abnormalities. Birth defects diagnosed prenatally and in children up to six years of age are included. The denominator data are all births in WA.
- This report presents information on birth defects notified to the Registry for births and terminations of pregnancy occurring between 1 January 1980 and 31 December 2007. A summary of the Registry's activities over the past year is also provided.
- A total of 1072 cases of birth defects were notified to the Registry relating to births and terminations of pregnancy in 2007, a proportion of 3.6%. This is expected to increase as birth defects continue to be diagnosed up to the age of six years in children born in 2007 (Table 1).
- Birth defects were generally more common in male infants and multiple pregnancies and were reported slightly less frequently in Aboriginal infants compared with non-Aboriginal infants (Table 2).
- Birth defects were also generally reported less frequently for rural regions compared with metropolitan regions (Table 3).
- In 2007, musculo-skeletal defects (10.4 per 1000 births), cardiovascular defects (8.9 per 1000 births) and urogenital defects (9.0 per 1000 births) were the most common categories of birth defects (Table 4).
- From 1980 to 1995, rates of neural tube defects (births plus terminations of pregnancy) were around 2 per 1000 births but have fallen since 1995, thought to be due to a primary preventive effect of periconceptional folate (Figure 1). Very few infants (0.2 per 1000 in 2007) are now liveborn with a neural tube defect (Figure 7).
- Chromosomal anomalies generally have been increasing since 1980. The total rate for Down syndrome (livebirths, stillbirths and terminations of pregnancy) and the rate for terminations alone have steadily increased over time, reaching 2.4 per 1000 for total Down syndrome in 2007 and 1.5 per 1000 for terminations (Figure 8). Although the rate of Down syndrome in liveborn infants has remained around 1 per 1000 for many years, it was under 1 per 1000 in the most recent two years (2006-2007).
- Birth defects are a major cause of deaths. For 2007 births, a birth defect was present in 7.4% of stillbirths, 23.0% of neonatal deaths and 25% of post-neonatal deaths (Table 5).
- The major sources of notification to the Registry were hospitals and private practitioners, Department of Health databases (midwives', mortality and hospital morbidity systems) and investigative and treatment centres (Table 6).
- Research using Registry data is reported: age at diagnosis of birth defects; trends in anencephaly, spina bifida and encephalocele; holoprosencephaly; and association of birth defects with increased maternal pre-pregnancy weight.

INTRODUCTION

The Western Australian Birth Defects Registry has a commitment to obtain high quality, complete, and population-based information on birth defects for Western Australia, and to use this information to:

- establish how often birth defects occur in WA;
- carry out research into the causes and prevention of birth defects;
- investigate changes in the frequency of birth defects, which may point to a possible cause or provide reassurance that an increase has not occurred;
- give doctors and other health professionals information about birth defects in WA;
- provide local statistics to assist health workers who counsel families about their chance of having a child with a birth defect;
- evaluate screening for and treatment and prevention of birth defects and assist with planning health care facilities; and
- increase knowledge generally about birth defects.

This report provides routine statistics on notifications received by 31 August 2008 for births occurring between 1 January 1980 and 31 December 2007. The cases of birth defects comprise malformations occurring in livebirths and stillbirths in WA, and in pregnancies terminated because of fetal malformation.

Malformations diagnosed in children up to six years of age are included. Children not born in WA but resident in the State are not included in this report. They are, however, recorded on the Registry for such purposes as evaluation of treatment and planning of facilities for children with malformations in WA.

A summary of the Registry's activities over the past year is also provided.

ROUTINE STATISTICS

The numerator data in this report comprise malformations occurring in livebirths and stillbirths in WA and in pregnancies terminated because of fetal malformation. Malformations diagnosed in children up to six years of age are included. The denominator data in this Report are derived from information provided by the Department of Health and include only livebirths and stillbirths 20 weeks' gestation or more.

The proportion of births with birth defects has increased from under 5% in early birth cohorts (1980-1982) to a peak of 7.0% in 1996-1997 (Table 1). Children born from 2003 onwards are not yet six years of age, and hence the percentage with birth defects in these later years of birth can be expected to increase as birth defects continue to be diagnosed in children up to the age of six years.

Table 1
Birth Defects in Western Australia, 1980 - 2007

Year	Total Births in WA	Cases of birth defects notified	WA Births with defects %
1980	20825	984	4.7
1981	22240	1046	4.7
1982	22400	1068	4.8
1983	23082	1174	5.1
1984	22989	1192	5.2
1985	23402	1165	5.0
1986	23961	1224	5.1
1987	24242	1256	5.2
1988	25191	1310	5.2
1989	25582	1408	5.5
1990	26039	1482	5.7
1991	25058	1475	5.9
1992	25358	1595	6.3
1993	25370	1590	6.3
1994	25450	1630	6.4
1995	25448	1647	6.5
1996	25586	1753	6.9
1997	25257	1785	7.1
1998	25668	1680	6.5
1999	25743	1736	6.7
2000	25229	1701	6.7
2001	24932	1613	6.5
2002	24782	1494	6.0
2003	24681	1389	5.6
2004	25530	1392	5.5
2005	26984	1462	5.4
2006	28639	1362	4.8
2007	30066	1072	3.6

DEMOGRAPHIC INFORMATION

(i) Race, sex and plurality (Table 2)

Birth defects are generally more common in multiple births and male infants. There is a lower prevalence of birth defects reported in Aboriginal children. This is thought to be due in part to under-ascertainment of cases of birth defects in Aboriginal children.

Table 2
Birth Defects in Western Australian births
by Race, Sex and Plurality, 1980 - 2007
 (Percentages are for total WA births in each year)

Year of Birth	Race		Sex			Plurality		
	Non-aboriginal	Aboriginal	Male	Female	Indeterminate	Unknown	Single	Multiple
1980-84	5212	(4.9)	3204	(5.6)			5352	(4.9)
	252	(4.5)	2235	(4.1)			112	(4.7)
1985-89			3					
			22					
	6043	(5.2)	3767	(6.0)			6159	(5.2)
	320	(4.8)	2576	(4.3)			204	(6.4)
1990-94			4					
			16					
	7369	(6.1)	4480	(6.8)			7499	(6.1)
	403	(5.5)	3253	(5.3)			273	(8.1)
1995-99			2					
			37					
	8186	(6.8)	4867	(7.4)			8315	(6.8)
	416	(5.5)	3699	(6.0)			287	(7.6)
2000-04			3					
			33					
	7151	(6.2)	4361	(6.8)			7300	(6.0)
	440	(5.5)	3164	(5.1)			291	(7.1)
2005			4					
			62					
	1374	(5.4)	834	(6.1)			1401	(5.4)
	88	(5.4)	615	(4.7)			61	(6.8)
2006			0					
			13					
	1280	(4.8)	745	(5.1)			1308	(4.7)
	82	(4.5)	592	(4.2)			54	(6.6)
2007			1					
			24					
	1013	(3.6)	575	(3.7)			1037	(3.6)
	59	(3.2)	483	(3.3)			35	(4.0)
		0						
		14						

(ii) Area of residence

Table 3 shows that the proportion of births with a birth defect has increased gradually over time in all regions. Proportions tend to be higher in the two metropolitan regions than in the rural regions. This may be due to under-ascertainment from rural regions rather than a real difference.

Table 3
Numbers and Proportions of Cases of Birth Defects by Year of Birth and Health Region, 1980 - 2007
 (Proportions are per 100 births) No=Number, Prop=Proportion

Health Region (WA Dept of Health)	1980-84	1985-89	1990-94	1995-99	2000-04	2005	2006	2007
	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop	No Prop
North Metro	2061 5.0	2575 5.6	3212 6.7	3558 7.3	3154 6.4	592 5.5	556 4.9	490 4.1
South Metro	1868 5.5	2059 5.4	2630 6.3	3006 7.0	2749 6.4	524 5.6	491 4.8	362 3.4
Kimberley	103 4.1	111 3.8	164 5.5	177 5.6	190 5.6	33 4.8	28 4.1	19 2.6
Pilbara Gascoyne	207 3.4	264 4.1	246 4.4	231 4.9	214 5.1	36 4.0	37 4.4	27 3.4
Midwest Murchison	180 4.1	237 4.7	247 4.6	258 5.8	205 5.5	41 5.3	39 4.2	21 2.2
Wheatbelt	322 4.4	344 5.2	337 5.4	335 6.1	252 5.2	44 4.5	37 4.0	33 3.4
Goldfields SE Coastal	188 4.1	223 4.2	270 4.8	305 5.4	259 5.3	42 4.5	33 3.4	31 3.0
Great Southern	196 4.9	173 4.3	190 5.0	221 5.8	148 4.3	22 3.0	36 4.7	18 2.5
Southwest	335 4.5	364 4.5	463 5.8	488 5.9	394 4.7	117 6.5	81 4.1	66 3.2

DIAGNOSTIC INFORMATION

The definition of a birth defect, used by the Registry is: *a structural or functional abnormality that is present at conception or occurs before the end of pregnancy, and is diagnosed by six years of age*. This includes structural (eg spina bifida), chromosomal (eg Down syndrome) and metabolic (eg phenylketonuria) defects. Most minor malformations are excluded unless they are disfiguring or require treatment. Of all cases registered, about 90% have at least one major malformation (with or without a minor malformation); the remainder have only minor malformations. A list of exclusions can be found on page 34. Each individual defect (up to a maximum of 10 defects per case) is coded according to the 5-digit British Paediatric Association ICD-9 system. Syndrome diagnoses are coded along with the major individual defects seen in that infant (eg Down syndrome, VSD and duodenal atresia occurring in one child are all coded).

Table 4 shows the number and proportion per 1000 total births of the major categories of defects, as well as the more common or important defects individually, by year of birth. Since about a quarter of the cases registered have more than one defect, the total number of defects exceeds the total number of cases. Not all individual birth defects are reported in Table 4, but information on any birth defect is available on request.

Figures 1 - 6 show the prevalence per 1000 total births for selected malformations by year of birth, from 1980-2007 and Figures 7 and 8 show livebirths, terminations and total cases with neural tube defects and Down syndrome.

Some trends of note are:

- There has been a fall in **neural tube defects** in total since 1995, and in **anencephaly** and **spina bifida** when considered separately. This is believed to be due to increased maternal intake of periconceptional folate, as folic acid supplements, and in food (including foods voluntarily fortified with folic acid). The increase in spina bifida in 2005 (0.9 per 1000), has not continued.
- The apparent fall in prevalence of some defects in 2006-2007 is due to late diagnosis or registration of birth defects. For example, most cases of **undescended testes** are registered at the time surgery is undertaken, usually around 1-2 years of age.
- The gradual rise in **chromosomal defects** since 1980 is a result of the increasing numbers of pregnancies in women over 35 years of age, and may also be related to the increased use of first trimester screening. There has been little change, however, in the rate of **Down syndrome** in liveborn infants (Figure 8).
- There has been an increase in **Fetal Alcohol Syndrome (FAS)**, from a low of 0.1 per 1000 in 1980-1984 to 0.5 per 1000 in 2000-2004. There is evidence of under-diagnosis and under-reporting of FAS in WA and Registry efforts to enhance notification are likely to account for the increases seen.

Table 4**Numbers and Proportions of Cases of Birth Defects by Year of Birth and Diagnosis, 1980 - 2007**

(Proportions are per 1000 births and only calculated if number of cases is greater than 13.
No=Number, Prop=Proportion)

Diagnostic Category (British Paediatric Association Code)	80-84 No Prop	85-89 No Prop	90-94 No Prop	95-99 No Prop	00-04 No Prop	05 No Prop	06 No Prop	07 No Prop
NERVOUS SYSTEM DEFECTS (74000 - 74299)	471 4.2	523 4.3	598 4.7	596 4.7	586 4.7	127 4.7	105 3.7	87 2.9
Neural Tube Defects (74000 - 74209)	208 1.9	238 1.9	251 2.0	203 1.6	182 1.5	44 1.6	36 1.3	33 1.1
Anencephalus (74000 - 74029)	92 0.8	112 0.9	110 0.9	91 0.7	79 0.6	16 0.6	12	16 0.5
Spina Bifida (74100 - 74199)	96 0.9	105 0.9	114 0.9	95 0.7	88 0.7	23 0.9	21 0.7	13
Encephalocoele (74200 - 74209)	20 0.2	21 0.2	27 0.2	17 0.1	15 0.1	5	3	4
Microcephaly (74210)	65 0.6	58 0.5	74 0.6	71 0.6	72 0.6	15 0.6	11	8
Congenital Hydrocephalus (excludes those with NTD) (74230 - 74239)	77 0.7	78 0.6	112 0.9	125 1.0	91 0.7	26 1.0	25 0.9	20 0.7
Congenital Deafness (74287)	74 0.7	90 0.7	102 0.8	109 0.9	111 0.9	14 0.5	9	6
CONGENITAL ANOMALIES OF EYE (74300 - 74399)	126 1.1	138 1.1	181 1.4	179 1.4	142 1.1	27 1.0	14 0.5	13
Anophthalmia (74300 - 74309)	6	7	4	10	6	2	0	1
Microphthalmia (74310 - 74319)	17 0.2	24 0.2	24 0.2	31 0.2	21 0.2	2	0	2
Congenital Cataract and Lens Anomalies (74330 - 74339)	26 0.2	31 0.3	53 0.4	49 0.4	33 0.3	6	3	1
CONGENITAL ANOMALIES OF EAR, FACE AND NECK (74400 - 74499)	249 2.2	346 2.8	460 3.6	574 4.5	556 4.4	113 4.2	99 3.5	66 2.2
Anotia, Microtia (74400 - 74401, 74421)	18 0.2	27 0.2	34 0.3	40 0.3	27 0.2	6	6	3

Table 4 (continued)

Diagnostic Category (British Paediatric Association Code)	80-84 No Prop	85-89 No Prop	90-94 No Prop	95-99 No Prop	00-04 No Prop	05 No Prop	06 No Prop	07 No Prop
Branchial Remnants (74440 - 74448)	50 0.4	70 0.6	74 0.6	77 0.6	58 0.5	7	12	5
CARDIOVASCULAR DEFECTS (74500 - 74799)	868 7.8	1067 8.7	1512 11.9	1617 12.7	1565 12.5	286 10.6	307 10.7	267 8.9
Transposition of Great Vessels (74510 - 74519)	37 0.3	60 0.5	50 0.4	63 0.5	64 0.5	13	16 0.6	14 0.5
Tetralogy of Fallot (74520)	36 0.3	37 0.3	58 0.5	41 0.3	38 0.3	10	7	6
Ventricular Septal Defect (74540 - 74549)	438 3.9	532 4.3	835 6.6	908 7.1	863 6.9	169 6.3	172 6.0	153 5.1
Atrial Septal Defect (74551 - 74559)	157 1.4	155 1.3	234 1.8	261 2.0	220 1.8	36 1.3	42 1.5	43 1.4
Hypoplastic Left Heart Syndrome (74670)	24 0.2	22 0.2	32 0.3	22 0.2	19 0.2	5	6	4
Patent Ductus Arteriosus (74700)	149 1.3	162 1.3	221 1.7	255 2.0	210 1.7	48 1.8	54 1.9	29 1.0
Coarctation of Aorta (74710 - 74719)	51 0.5	68 0.6	76 0.6	63 0.5	81 0.6	15 0.6	16 0.6	10
RESPIRATORY SYSTEM DEFECTS (74800 - 74899)	92 0.8	108 0.9	101 0.8	151 1.2	142 1.1	24 0.9	31 1.1	31 1.0
Choanal Atresia (74800 - 74809)	16 0.1	20 0.2	17 0.1	17 0.1	16 0.1	1	2	1
GASTRO-INTESTINAL DEFECTS (74900 - 75199)	688 6.2	791 6.5	811 6.4	868 6.8	783 6.3	177 6.6	151 5.3	122 4.1
Cleft Palate only (74900 - 74909)	93 0.8	104 0.8	133 1.0	150 1.2	147 1.2	32 1.2	20 0.7	20 0.7
Cleft Lip only (74910 - 74919)	47 0.4	62 0.5	59 0.5	57 0.4	73 0.6	16 0.6	16 0.6	16 0.5
Cleft Lip and Palate (74920 - 74929)	86 0.8	111 0.9	68 0.5	102 0.8	97 0.8	12	16 0.6	23 0.8
Tracheo-Oesophageal Fistula, Oesophageal Atresia/Stenosis (75030 - 75038)	34 0.3	43 0.4	35 0.3	42 0.3	50 0.4	16 0.6	10	14 0.5

Table 4 (continued)

Diagnostic Category (British Paediatric Association Code)	80-84 No Prop	85-89 No Prop	90-94 No Prop	95-99 No Prop	00-04 No Prop	05 No Prop	06 No Prop	07 No Prop
Pyloric Stenosis (75051 - 75058)	215 1.9	255 2.1	237 1.9	224 1.8	145 1.2	53 2.0	33 1.2	4
Stenosis/Atresia Small Intestine (75110 - 75119)	34 0.3	30 0.2	33 0.3	38 0.3	39 0.3	8	3	9
Stenosis/Atresia Anus (75123 - 75125)	61 0.5	60 0.5	85 0.7	71 0.6	86 0.7	18 0.7	16 0.6	12
Hirschprung's Disease (75130 - 75133)	19 0.2	18 0.1	32 0.3	21 0.2	17 0.1	2	6	8
URO-GENITAL DEFECTS (75200 - 75399)	1493 13.4	1786 14.6	2340 18.4	2508 19.6	2194 17.5	394 14.6	330 11.5	271 9.0
Undescended Testis (treated) (75250 - 75254, 75257)	725 6.5	822 6.7	854 6.7	741 5.8	621 5.0	82 3.0	66 2.3	42 1.4
Hypospadias (75260, 75263 - 75269)	300 2.7	364 3.0	444 3.5	458 3.6	461 3.7	82 3.0	71 2.5	68 2.3
Renal Agenesis or Dysgenesis (75300 - 75306)	55 0.5	56 0.5	76 0.6	100 0.8	115 0.9	12	31 1.1	16 0.5
Cystic Kidney Disease (75310 - 75319)	28 0.3	52 0.4	92 0.7	95 0.7	124 1.0	19 0.7	23 0.8	13
Obstructive Defects Renal Pelvis (75320 - 75329)	50 0.4	102 0.8	199 1.6	291 2.3	280 2.2	96 3.6	83 2.9	81 2.7
Vesico-Ureteric Reflux (75344)	197 1.8	289 2.4	502 3.9	650 5.1	407 3.3	44 1.6	40 1.4	34 1.1
Other Anomalies of Ureter (75340 - 75343, 75345 - 75349)	75 0.7	96 0.8	190 1.5	226 1.8	162 1.3	35 1.3	32 1.1	27 0.9
MUSCULO-SKELETAL DEFECTS (75400 - 75699)	1684 15.1	1727 14.1	1989 15.6	2321 18.2	1954 15.6	394 14.6	400 14.0	313 10.4
Developmental Dysplasia of Hip (75430 - 75434, 75439)	757 6.8	745 6.1	777 6.1	938 7.3	716 5.7	150 5.6	170 5.9	144 4.8
Talipes (75450, 75454-75456,75473)	268 2.4	240 2.0	267 2.1	251 2.0	300 2.4	60 2.2	70 2.4	62 2.1
Polydactyly (75500 - 75509)	103 0.9	134 1.1	135 1.1	159 1.2	141 1.1	40 1.5	27 0.9	10
Syndactyly (75510 - 75519)	77 0.7	81 0.7	68 0.5	80 0.6	66 0.5	21 0.8	11	8

Table 4 (continued)

Diagnostic Category (British Paediatric Association Code)	80-84 No Prop	85-89 No Prop	90-94 No Prop	95-99 No Prop	00-04 No Prop	05 No Prop	06 No Prop	07 No Prop
Reduction Deformities Upper and/or Lower Limbs (75520 - 75549)	73 0.7	78 0.6	109 0.9	151 1.2	121 1.0	28 1.0	32 1.1	11
Craniosynostosis (75600, 75601)	46 0.4	59 0.5	83 0.7	75 0.6	58 0.5	12	13	9
Diaphragmatic Hernia (75661)	36 0.3	33 0.3	36 0.3	55 0.4	36 0.3	9	12	6
Exomphalos (75670)	21 0.2	38 0.3	47 0.4	38 0.3	59 0.5	10	13	7
Gastroschisis (75671)	15 0.1	22 0.2	32 0.3	52 0.4	38 0.3	7	17 0.6	11
CONGENITAL ANOMALIES OF INTEGUMENT (75700 - 75799)	339 3.0	380 3.1	517 4.1	709 5.6	542 4.3	64 2.4	58 2.0	28 0.9
Birth Marks, Naevus (75738)	196 1.8	220 1.8	288 2.3	436 3.4	328 2.6	28 1.0	29 1.0	13
CHROMOSOME DEFECTS (75800 - 75899)	222 2.0	322 2.6	457 3.6	549 4.3	670 5.4	157 5.8	159 5.6	121 4.0
Down Syndrome (75800 - 75809)	129 1.2	186 1.5	219 1.7	251 2.0	331 2.6	79 2.9	75 2.6	72 2.4
Trisomy 13 (75810 - 75819)	8	14 0.1	18 0.1	22 0.2	40 0.3	11	7	7
Trisomy 18 (75820 - 75829)	18 0.2	22 0.2	49 0.4	65 0.5	91 0.7	26 1.0	26 0.9	17 0.6
Turner Syndrome (75860 - 75861, 75869)	13	15 0.1	34 0.3	43 0.3	61 0.5	9	13	9
OTHER								
Congenital Hypothyroidism (24390 - 24399)	16 0.1	33 0.3	54 0.4	40 0.3	64 0.5	7	19 0.7	5
Adrenogenital Syndrome (25520 - 25529)	6	9	9	15 0.1	14 0.1	4	1	3
Disorders of Amino Acid Transport and Metabolism (27000 - 27099)	15 0.1	23 0.2	26 0.2	34 0.3	27 0.2	8	6	5

Table 4 (continued)

Diagnostic Category (British Paediatric Association Code)	80-84 No Prop	85-89 No Prop	90-94 No Prop	95-99 No Prop	00-04 No Prop	05 No Prop	06 No Prop	07 No Prop
Phenylketonuria (27010)	6	7	7	7	11	3	3	1
Disorders of Carbohydrate Transport and Metabolism (27100 - 27199)	13	8	14 0.1	8	8	0	0	0
Cystic Fibrosis (27700)	33 0.3	43 0.4	43 0.3	28 0.2	53 0.4	17 0.6	11	11
G6PD Deficiency (28220)	17 0.2	27 0.2	22 0.2	44 0.3	30 0.2	5	0	2
Thalasseмии (28240 - 28249)	3	2	6	6	3	0	1	1
Haemophilia (28600 - 28620)	11	8	4	18 0.1	22 0.2	1	1	2
Muscular Dystrophies and Myopathies (35900 - 35999)	35 0.3	35 0.3	23 0.2	29 0.2	13	2	4	3
Fetal Alcohol Syndrome (75992)	15 0.1	21 0.2	23 0.2	24 0.2	68 0.5	12	6	6
Congenital Rubella Syndrome (77100)	16 0.1	8	7	2	2	0	0	0
Non-Immune Fetal Hydrops (77800)	22 0.2	42 0.3	86 0.7	89 0.7	91 0.7	22 0.8	24 0.8	23 0.8

Figure 1: Neural tube defects

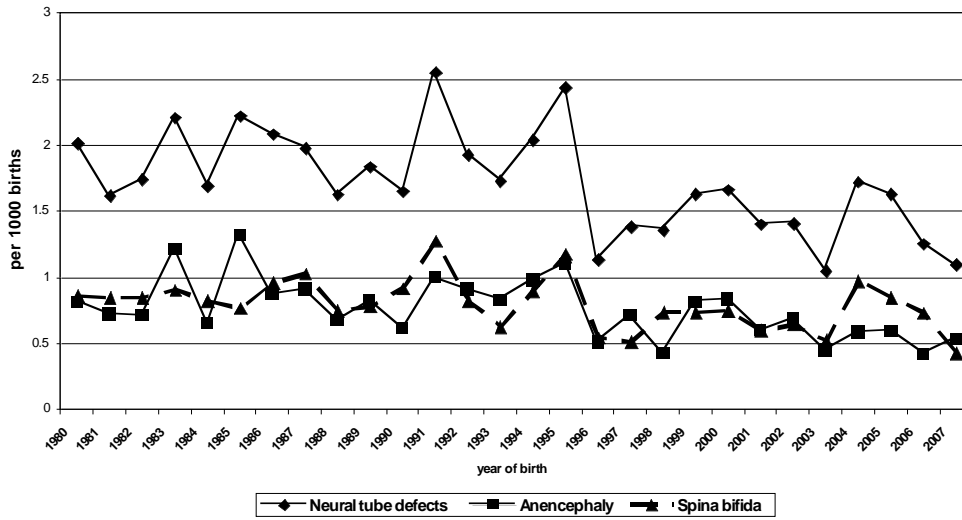


Figure 2: Cardiovascular defects

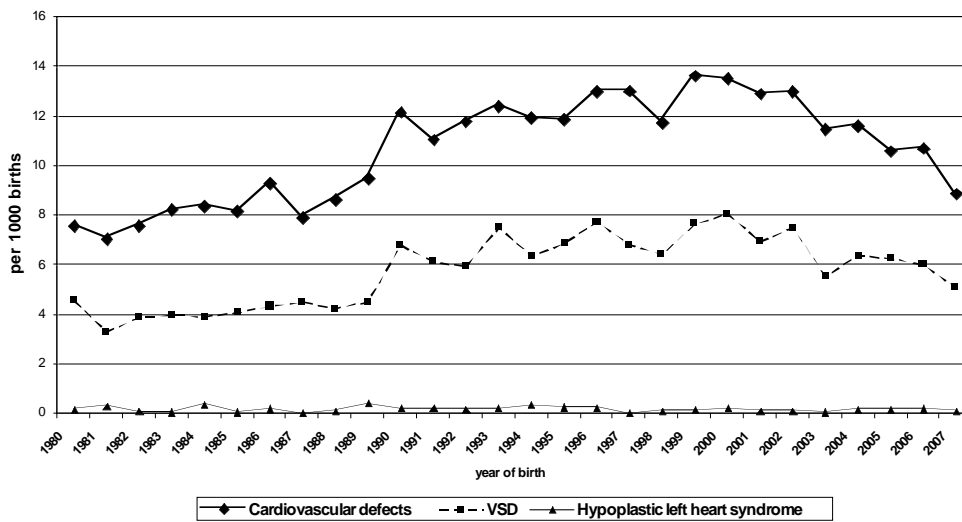


Figure 3: Cleft lip and palate

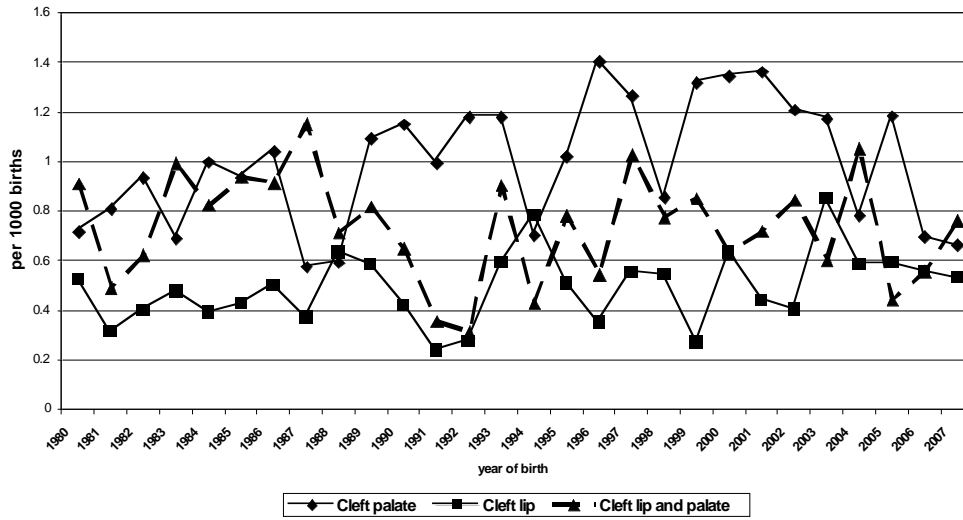


Figure 4: Hypospadias and renal agenesis/dysgenesis

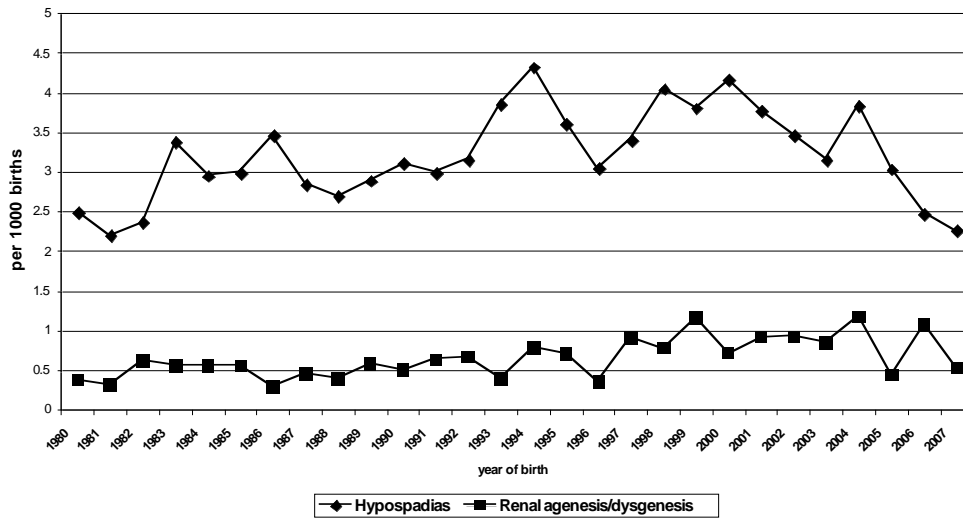


Figure 5: Exomphalos and gastroschisis

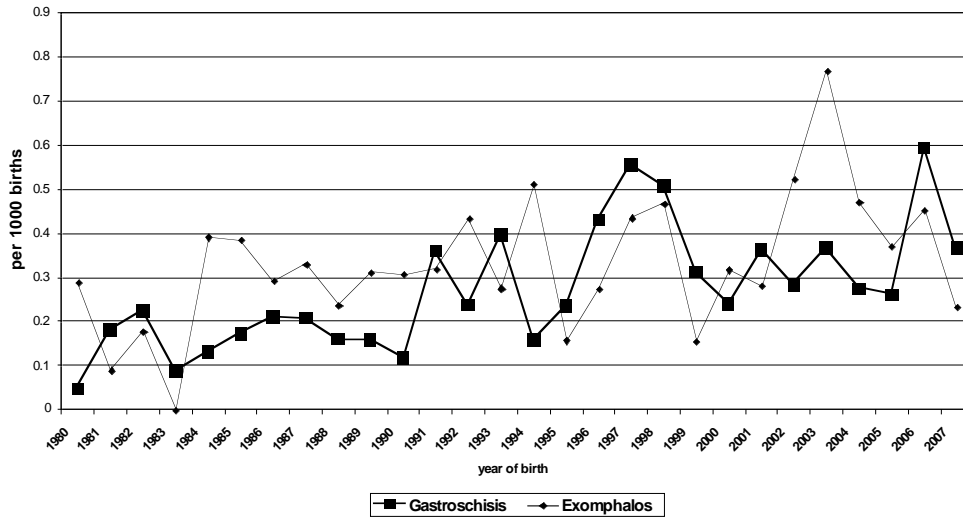


Figure 6: Chromosomal defects

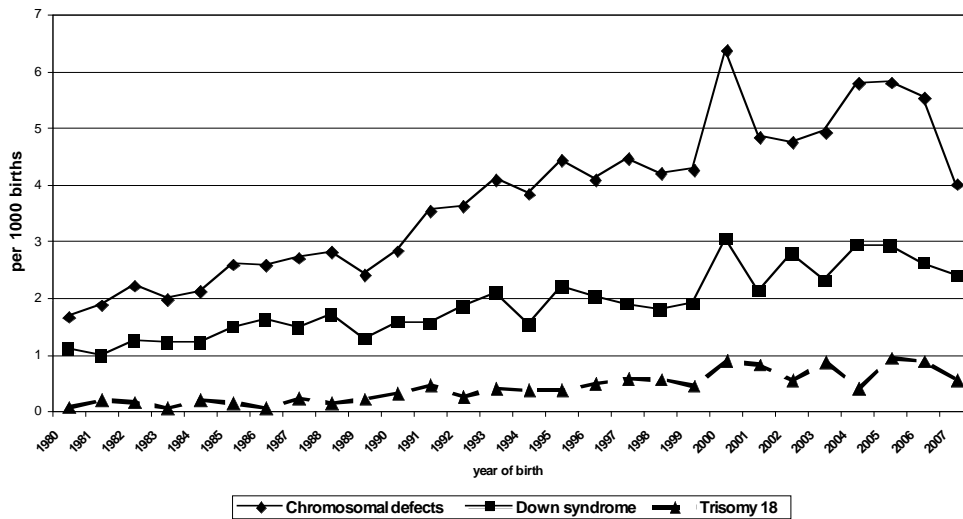


Figure 7: Neural tube defects: total, livebirths and terminations of pregnancy

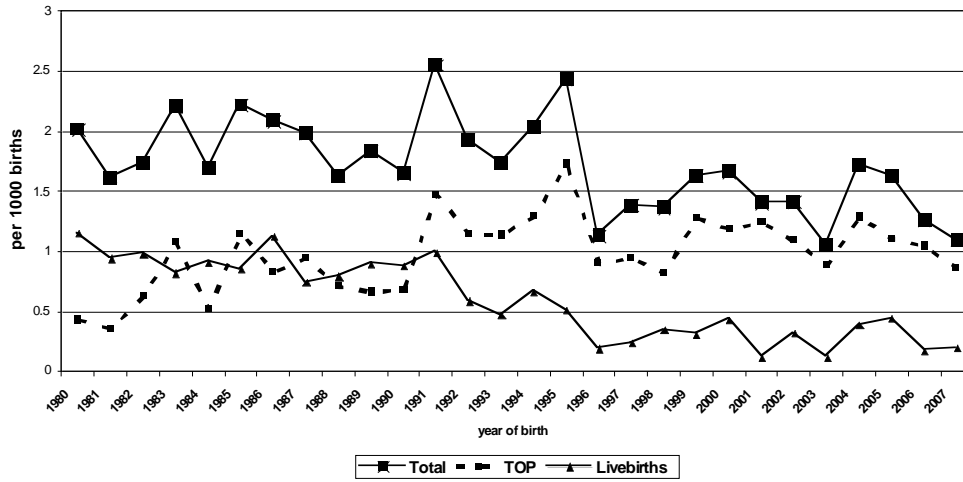
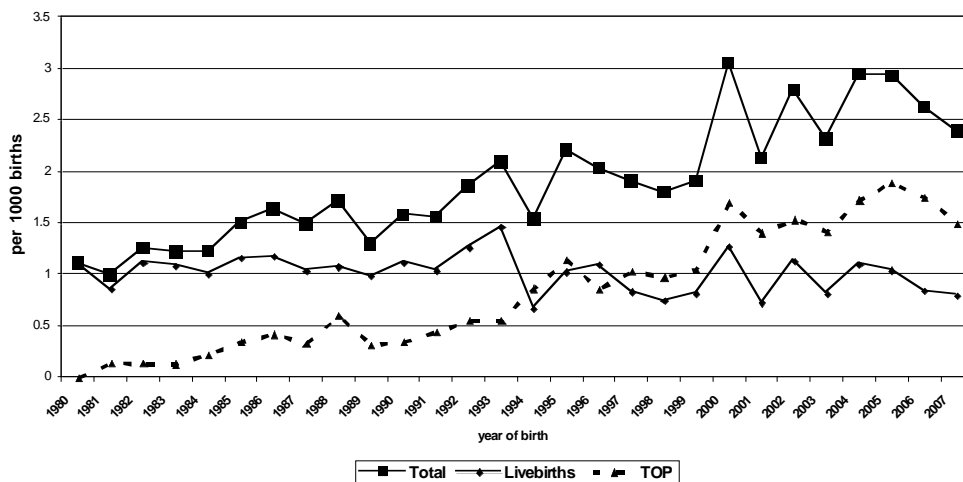


Figure 8: Down syndrome: total, livebirths and terminations of pregnancy



DEATHS

Table 5 shows the number (and percentage) of stillbirths, neonatal and post-neonatal deaths known to have a birth defect. Terminations of pregnancy are those which occurred following prenatal diagnosis of a fetal abnormality. Between 7% and 15% of stillbirths have a reported birth defect, as do 23% - 41% of neonatal deaths and 25% - 44% of post-neonatal deaths.

Rates of termination of pregnancy for fetal abnormality have increased from 1.2 per 1000 births in 1980-1984 to 7.4 per 1000 in 2005.

Table 5
Deaths with Birth Defects 1980 - 2007

Year of Birth	Stillbirths (% is of all stillbirths)		Neonatal (% is of all neonatal)		Postneonatal (% is of all postneonatal)		Terminations of Pregnancy
	No	%	No	%	No	%	No
1980-84	122	13.2	247	38.5	118	31.0	134
1985-89	124	13.2	267	41.0	97	24.7	272
1990-94	138	15.4	189	40.3	93	28.5	473
1995-99	135	14.9	146	39.4	81	38.4	635
2000-04	98	10.9	99	33.0	52	31.9	814
2005	21	10.4	21	28.8	21	43.8	185
2006	24	11.5	21	33.9	16	42.1	204
2007	14	7.4	14	23.0	7	25.0	175

NOTIFICATIONS

Information is collected from statutory data sources (Midwives' Notification System, death certifications, Hospital Morbidity System) and voluntary notification is sought from many others. Table 6 documents the numbers of notifications received from different sources by year of birth of the child. Most sources provide very consistent levels of notification. Of concern is the reduction in number of notifications from private practitioners in 2005-2007. Private practitioners are an important source of notifications and we are working to ensure that privacy and confidentiality issues are not a barrier to notification.

Table 6
Sources of Notifications by Year of Birth of Cases Notified, 1980 - 2007

Notifiers	1980-84	1985-89	1990-94	1995-99	2000-04	2005	2006	2007
MIDWIVES' FORMS	1650	1692	1613	1425	1355	238	219	136
DEATH CERTIFICATES	446	449	378	375	338	90	62#	24#
HOSPITAL MORBIDITY	507	975	1320	1987	1918	352	219	*
PAEDIATRIC HOSPITALS EXCL SPECIAL DEPTS	2822	2711	2971	2051	1383	328	301	199
PAEDIATRIC HOSPITALS SPECIAL DEPARTMENTS	640	1520	1897	1910	1527	333	273	253
OBSTETRIC HOSPITALS EXCL SPECIAL DEPTS	830	945	1055	1002	856	147	144	133
OBSTETRIC HOSPITALS SPECIAL DEPARTMENTS	141	188	408	595	673	145	164	174
OTHER HOSPITALS	319	102	51	30	28	6	4	1
CYTOGENETIC SERVICES	179	220	402	529	639	158	171	197
PATHOLOGY SERVICES	405	400	565	647	741	169	169	162
GENETICS SERVICES	808	1122	1501	1617	1452	251	195	155
PRIVATE PRACTITIONERS	2379	2614	3162	3616	3192	499	527	431
CHILD & COMMUNITY HEALTH NURSES & DOCTORS	766	435	422	252	84	17	3	6
RURAL PAEDIATRIC SERVICE	75	211	353	269	172	30	19	15
OTHER	464	161	58	60	3	0	0	0
REGISTER CHECK TOTAL	500	251	252	179	102	18	20	29
	12931	13996	16408	16544	14463	2781	2490	1916

Incomplete death data for 2006 and 2007; * Hospital morbidity data for 2007 births not yet available

REGISTRY ACTIVITIES

1. Provision of data

The Registry is a comprehensive source of information on birth defects in WA for use in all relevant areas of health service provision, policy development, research and evaluation. Provision of data from the Registry may take two forms: (1) unnamed tabulated information similar to that contained in this report; and (2) named or unnamed unit data for specific research projects. Requests for the latter must be submitted in writing to the Registry in the first instance, and then forwarded to the Department of Health WA Human Research Ethics Committee for approval (see Confidentiality Guidelines, page 34).

2. Information on malformations and teratogens

The Registry maintains an extensive library of reprints and acts as a resource for information not directly available from the Registry data. Staff also seek information from elsewhere or directs enquiries to other sources, as appropriate. Over the past year, 20 requests for information have been received. Two of these requests were from the Department of Health, 14 were from health professionals and institutions in WA, Australia or overseas and four were from the general public, the media or students. About a third of the requests required a considerable amount of computing, analysis and discussion, and responses to most of the remainder involved provision and/or interpretation of published data.

3. Presentations

In the past year, the following presentations were made:

- A comparison of encephalocoeles with other NTD: trends before and after folate promotion and voluntary food fortification in Western Australia. International Neural Tube Defects Conference, Monterey 2007.
- Consumer views on statutory notification to the Western Australian Birth Defects Registry International Clearinghouse for Birth Defects Surveillance and Research Annual Scientific Meeting, Chianciano, Italy, 2007
- Folate and neural tube defects: the politics of prevention. International Congress on Developmental Origins of Health and Disease, Perth 2007.
- A sorry state of affairs: Aboriginal child health in Australia. Slone Memorial Lecture, Slone Epidemiology Center, Boston University, USA, 2008.
- Epidemiology of hypospadias in Western Australia. 35th Annual Meeting of the International Clearinghouse for Birth Defects Surveillance and Research, Italy, 2008.
- Age at diagnosis of birth defects in children up to six years of age. 35th Annual Meeting of the International Clearinghouse for Birth Defects Surveillance and Research, Italy, 2008.
- Maternal and paternal occupational exposure to endocrine disrupting chemicals and risk of hypospadias. Population Health Congress, Brisbane, 2008.
- Fetal Alcohol Spectrum Disorder in Australia. National Fetal Alcohol Spectrum Disorder Workshop, Adelaide, 2008.
- Strategies for preventing Fetal Alcohol Spectrum Disorder. National Fetal Alcohol Spectrum Disorder Workshop, Adelaide, 2008.

4. National Perinatal Statistics Unit (NPSU)

WA data for 2002-2003 births with birth defects were provided to NPSU for inclusion in their report published in May 2008 (*Aberwardana S, Sullivan EA 2008. Congenital anomalies in Australia 2002-2003. Birth Anomalies Series no.3 Cat no. PER 41. Sydney: AIHW National Perinatal Statistics Unit*). Registry data for WA were also included in a NPSU report on neural tube defects in Australia, published in November, 2008 (*Aberwardana S, Sullivan EA 2008. Neural tube defects in Australia. An epidemiological report. Cat. no. PER 45. AIHW National Perinatal Statistics Unit*). Registry staff have contributed to the review of coding of birth defects being conducted by the NPSU.

5. International Clearinghouse for Birth Defect Surveillance and Research

Data based on 2006 births were provided to the International Clearinghouse, for inclusion in the 2008 Annual Report of the Clearinghouse. WA data contributed to a paper from the Clearinghouse on holoprosencephaly, published in 2008.

6. Research Activities

6.1. Age at diagnosis of birth defects

In order to investigate how many birth defects (and cases) are ascertained over the age of one year, we examined notifications to the Registry for children born in 2000, who had had a full six years for ascertainment. Of the 1700 cases in 2000, 18% had at least one birth defect diagnosed prenatally, 70% were diagnosed by one year of age and 12% were diagnosed between one and six years of age. Urogenital, musculoskeletal and cardiovascular defects were the commonest conditions diagnosed beyond one year of age. We are now going to estimate the marginal costs (in terms of staff time) of ascertaining cases over one year of age to assist in determining the value of this extended period of ascertainment.

6.2. Neural tube defects

Using Registry data on anencephaly, spina bifida and encephalocoele, we showed that, for 1996-2006, there was a 32% reduction in anencephaly, 23% in spina bifida and 34% in encephalocoele compared with 1980-1992, corresponding to the introduction of promotion of periconceptional folic acid supplements and voluntary fortification of food. The rates for Aboriginal infants were higher than for non-Aboriginal infants, for each type of neural tube defect and there was no decrease in rates seen for Aboriginal infants. These data will provide a useful baseline against which to monitor the effects of mandatory fortification on NTD, when it is introduced in Australia in September 2009. A paper based on this work has been accepted for publication.

6.3. Holoprosencephaly

WA Registry data contributed to an international study of holoprosencephaly, which has now been published. There were 963 cases of holoprosencephaly in over 7 million births 2000-2004 in data from 24 member registries of the International Clearinghouse for Birth Defects Surveillance and Research giving an overall prevalence of 1.31 per 10,000 births. There was a range of rates for individual registers (from 0.63 per 10,000 in Atlanta USA to 2.50 per 10,000 in Strasbourg France. The two Australian register rates were similar -1.73 per 10,000 births in Victoria and 1.92 per 10,000 in WA.

6.4. Pre-pregnancy maternal weight and risk of birth defects.

We examined the effect of maternal pre-pregnancy body mass index (BMI) on the odds of birth defects, using data from our population-based case-control study of 418 controls, 111 cases with heart defects (and of these, 38 had

conotruncal heart defects), 27 with neural tube defects, 86 cases with urinary tract defects, 48 cases with orofacial clefts, and 20 with limb reduction defects. We found that women with pre-pregnancy obesity (BMI 30+) had a two-fold increased odds of having an infant with neural tube defects, conotruncal heart defects, orofacial clefts and limb reduction defects and 30-40% increase in heart defects generally and urinary tract defects. None of the estimates was statistically significant, but these results are consistent with similar, statistically significant studies in the literature, raising the possibility of another means of primary prevention of birth defects. A paper reporting these findings has been accepted for publication.

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**BIRTH DEFECTS REGISTRY OF WESTERN AUSTRALIA
GUIDELINES FOR CONFIDENTIALITY AND RELEASE OF DATA**

1. Responsibility for the confidentiality of the birth defects data lies ultimately with the Director General of Health.
2. All staff are instructed regarding the need for and maintenance of confidentiality. The staff are responsible to the Director General for ensuring that confidentiality is maintained.
3. All routine reports from the collection are in statistical form without identification of individual patients, doctors or hospitals.
4. Non-routine release of tabulated, non-personally identified data from the Birth Defects Registry is the responsibility of and at the discretion of the Medical Officer of the Registry.
5. Access to patient identifiers by other than Registry staff or Registry research personnel will be given only by the express permission of the Director General, on the advice of the Department of Health WA Human Research Ethics Committee.

Such permission will be granted only if:

- (i) It is considered that such use of the data would promote the prevention or alleviation of handicap associated with congenital malformations (see Part IXA of the Health Act);
- (ii) It is considered unlikely to harm the patients or parents concerned in any way.

Should permission be granted for the release of personal identifiers, the following requirements will be made:

- (a) That only minimum identification necessary to the proposed use be given (eg for a case record review project in teaching hospitals, only unit medical record number would be supplied);
 - (b) That the use of the data be under the supervision of a registered medical practitioner;
 - (c) That the data be handled according to the code of confidentiality set down by the Confidentiality of Health Information Committee, particularly no confidential data to be released to any third party;
 - (d) That no approach be made directly to patients or their parents without:
 - (i) The knowledge and consent of all the notifiers of that patient to the Registry;
 - (ii) The knowledge and consent of the patient's present primary care physician (insofar as this person can be identified).
6. All persons who have access to name identified data for routine maintenance of the Registry or for research purposes shall complete a signed declaration binding them to respect the confidentiality of the information obtained therein, and to follow this code of practice.
 7. Any costs incurred in fulfilling these guidelines are to be borne by the requesting body.
 8. Final reports or papers for publication are to be vetted by the Medical Officer of the Registry before publication.
 9. Where a member of the Registry staff has made a significant intellectual contribution to the work using the Registry data, then the staff member(s) concerned should be author(s) on any relevant publication arising from that work.

REGISTRY EXCLUSION LIST

(if in doubt please notify or contact us on 9340 2735)

Accessory nipples	Meconium ileus
Balanced translocation (in normal individual without birth defect(s))	Mental retardation
Blocked tear duct	Metatarsus adductus (unless splinted)
Birth mark, Naevus, Haemangioma (unless multiple or >4cm - give size)	Mongolian blue spot
Broncho-pulmonary dysplasia	Motor impairment
Clicky hips	Oesophageal reflux
Congenital Infection (if no birth defects)	Paroxysmal atrial tachycardia
Congenital pneumonia	Patent foramen ovale
Cerebral palsy	Persistent fetal circulation
Delayed milestones	Perthe's disease
Deviated nasal septum	Pilonidal sinus
Ear anomalies - minor	Positional/Postural foot deformity
Epigastric hernia	Sacral dimple
Epilepsy	Sacral sinus (unrelated to occult spinal dysraphism)
Failure to thrive	Single palmar crease
Galactosaemia - Duarte Variant	Single umbilical artery
Hiatus hernia	Skin tag
Hydatid Of Morgagni	Strabismus
Hydrocele testis	Thalassemia Minor
Hydrops fetalis - immune (include if non-immune hydrops)	Toe anomalies - minor
Hypoglycaemia	Tongue tie
Imperforate hymen	Tricuspid incompetence
Inguinal hernia	Trigger finger/thumb (unless treated)
Intrauterine growth retardation	Umbilical hernia
Intussusception	Undescended testis (unless treated)
Labial adhesion or fusion	Webbing of 2nd & 3rd toes
Large fontanelles	Wide suture lines
Laryngeal stridor	Wolf Parkinson White syndrome
Laryngomalacia	
Low birth weight	

