

## 10. BIRTH DEFECTS

### Birth defects among stillborn and liveborn infants

A birth defect is any structural defect detected during pregnancy or at birth, excluding birth injuries and minor anomalies such as skin tags, positional talipes, birthmarks, or clicky hips. Descriptions of some common birth defects are shown in Appendix 1. A list of common exclusions is shown in Appendix 2.

From 1 January 1998, doctors, hospitals and laboratories are required to notify birth defects detected during pregnancy, at birth, or up to one year of life under the *NSW Public Health Act 1991*. Information reported is included in the NSW Birth Defects Register (BDR). The quality of information received by the BDR has improved since 1998, particularly in relation to pregnancy outcome.

This chapter reports birth defects detected during pregnancy or in the first year of life for 1998–2003 and birth defects detected during pregnancy or at birth for 2004.

### Trends in reported birth defects

Between 1998 and 2004, the reported number of infants with birth defects has remained stable at just over 2 per cent (Table 114). In 2004, 997 cases of birth defects detected during pregnancy or at birth were reported.

### Birth defects by diagnostic category

The most common categories of birth defects for births of more than 20 weeks gestation or with a birth weight greater

TABLE 114

BIRTH DEFECT CASES, NSW 1998–2004#

Year	Birth defect cases	Births	Rate/1,000 births
1998	1941	85627	22.7
1999	1828	86468	21.1
2000	1858	87279	21.3
2001	1775	85285	20.8
2002	1739	85398	20.4
2003	1761	85853	20.5
2004	997	85016	11.7

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

# For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

than 400 grams are presented in Table 115. Birth defects are classified using the British Paediatric Association (BPA) Classification of Diseases, which is primarily organised by body system. For infants with more than one defect, each defect is counted separately. The number of birth defects reported therefore exceeds the number of affected infants.

In 1998–2004, defects of the cardiovascular system were most commonly reported, followed by defects of the musculoskeletal system and defects of the genito-urinary system (Table 115). This is a similar pattern to previous years. In 2003, the overall rate of defects was lower than the previous 5 years (35.4 versus 38.1 per 1,000), due to a lower overall birth defect rate among infants.

TABLE 115

BIRTH DEFECTS AMONG STILLBIRTHS AND LIVE BIRTHS BY DIAGNOSTIC CATEGORY, NSW 1998–2004

Diagnostic category	No. defects			1998–2004	Rate/1,000 births			
	1998–2002	2003	2004		1998–2002	2003	2004	1998–2004
<b>Defects of nervous system</b>								
Anencephaly	49	11	6	66	0.1	0.1	0.1	0.1
Spina Bifida	140	25	21	186	0.3	0.3	0.2	0.3
Encephalocele	27	6	6	39	0.1	0.1	0.1	0.1
Microcephaly	116	23	11	150	0.3	0.3	0.1	0.2
Congenital hydrocephalus	174	38	33	245	0.4	0.4	0.4	0.4
Other nervous system defects	366	58	41	465	0.9	0.7	0.5	0.8
TOTAL	872	161	118	1151	2.0	1.9	1.4	1.9
<b>Defects of eye</b>								
Anophthalmos–microphthalmos	55	11	15	81	0.1	0.1	0.2	0.1
Buphthalmos–congenital glaucoma	30	2	1	33	0.1	0.0	0.0	0.1
Congenital cataract	83	15	5	103	0.2	0.2	0.1	0.2
Other eye defects	176	27	23	226	0.4	0.3	0.3	0.4
TOTAL	344	55	44	443	0.8	0.6	0.5	0.7
<b>Defects of ear, face and neck</b>								
Absence–stricture auditory canal	56	9	7	72	0.1	0.1	0.1	0.1
Absent auricle	9	0	1	10	0.0	0.0	0.0	0.0
Defects of face and neck	43	9	8	60	0.1	0.1	0.1	0.1
Other ear defects	89	12	11	112	0.2	0.1	0.1	0.2
TOTAL	197	30	27	254	0.5	0.3	0.3	0.4
<b>Defects of cardiovascular system</b>								
Transposition of great vessels	210	52	26	288	0.5	0.6	0.3	0.5
Tetralogy of Fallot	148	35	16	199	0.3	0.4	0.2	0.3
Ventricular septal defect	902	160	102	1164	2.1	1.9	1.2	1.9
Atrial septal defect	834	157	79	1070	1.9	1.8	0.9	1.8

TABLE 115 (continued)

## BIRTH DEFECTS AMONG STILLBIRTHS AND LIVE BIRTHS BY DIAGNOSTIC CATEGORY, NSW 1998–2004#

Diagnostic category	No. defects			Rate/1,000 births				
	1998–2002	2003	2004	1998–2004	1998–2002	2003	2004	1998–2004
<b>Defects of cardiovascular system (cont.)</b>								
Heart valve defects	640	106	60	806	1.5	1.2	0.7	1.3
Patent ductus arteriosus > 37 weeks	470	81	69	620	1.1	0.9	0.8	1.0
Coarctation of aorta	198	38	22	258	0.5	0.4	0.3	0.4
Other defects of aorta	111	27	16	154	0.3	0.3	0.2	0.3
Defects of pulmonary artery	144	27	15	186	0.3	0.3	0.2	0.3
Other cardiovascular defects	774	148	114	1036	1.8	1.7	1.3	1.7
TOTAL	4431	831	519	5781	10.3	9.7	6.1	9.6
<b>Defects of respiratory system</b>								
Defects of nose	77	9	8	94	0.2	0.1	0.1	0.2
Defects of larynx, trachea and bronchus	50	9	4	63	0.1	0.1	0.0	0.1
Defects of lung	93	7	10	110	0.2	0.1	0.1	0.2
TOTAL	220	25	22	267	0.5	0.3	0.3	0.4
<b>Defects of gastrointestinal system</b>								
Cleft palate only	365	103	67	535	0.8	1.2	0.8	0.9
Cleft lip only	145	34	31	210	0.3	0.4	0.4	0.3
Cleft palate and cleft lip	235	46	40	321	0.5	0.5	0.5	0.5
Oesophageal atresia only	8	3	2	13	0.0	0.0	0.0	0.0
Oesophageal atresia with TOF	87	14	9	110	0.2	0.2	0.1	0.2
Tracheo-oesophageal fistula (TOF) only	28	1	2	31	0.1	0.0	0.0	0.1
Atresia/stenosis of small intestine	142	22	8	172	0.3	0.3	0.1	0.3
Atresia/stenosis of anus	133	27	15	175	0.3	0.3	0.2	0.3
Other gastrointestinal defects	487	95	47	629	1.1	1.1	0.6	1.0
TOTAL	1630	345	221	2196	3.8	4.0	2.6	3.7
<b>Defects of genitourinary system</b>								
Defects of female genitals	46	11	15	72	0.1	0.1	0.2	0.1
Undescended testis	349	48	26	423	0.8	0.6	0.3	0.7
Hypospadias	886	170	121	1177	2.1	2.0	1.4	2.0
Epispadias	18	2	0	20	0.0	0.0	.	0.0
Chordee	133	32	12	177	0.3	0.4	0.1	0.3
Indeterminate sex–ambiguous genitalia	53	8	10	71	0.1	0.1	0.1	0.1
Renal agenesis–dysgenesis	215	31	30	276	0.5	0.4	0.4	0.5
Obstructive defects of renal pelvis and ureter	747	126	66	939	1.7	1.5	0.8	1.6
Other genito-urinary system defects	635	117	67	819	1.5	1.4	0.8	1.4
TOTAL	3082	545	347	3974	7.2	6.3	4.1	6.6
<b>Defects of musculoskeletal system</b>								
Congenital dislocation of the hips	668	146	55	869	1.6	1.7	0.6	1.4
Talipes equinovarus	294	78	42	414	0.7	0.9	0.5	0.7
Polydactyly	479	103	85	667	1.1	1.2	1.0	1.1
Syndactyly	114	17	18	149	0.3	0.2	0.2	0.2
Reduction deformities of limbs	256	34	36	326	0.6	0.4	0.4	0.5
Craniosynostosis	326	53	5	384	0.8	0.6	0.1	0.6
Diaphragmatic hernia	127	21	25	173	0.3	0.2	0.3	0.3
Exomphalos	72	14	14	100	0.2	0.2	0.2	0.2
Gastroschisis	94	16	14	124	0.2	0.2	0.2	0.2
Other musculoskeletal defects	919	132	108	1159	2.1	1.5	1.3	1.9
TOTAL	3349	614	402	4365	7.8	7.2	4.7	7.3
<b>Defects of integumentary system</b>								
Cystic hygroma	339	62	29	430	0.8	0.7	0.3	0.7
Cystic hygroma	53	8	6	67	0.1	0.1	0.1	0.1
<b>Chromosomal defects</b>								
Trisomy 21	513	103	79	695	1.2	1.2	0.9	1.2
Trisomy 13	35	7	7	49	0.1	0.1	0.1	0.1
Trisomy 18	91	19	13	123	0.2	0.2	0.2	0.2
Turner syndrome	68	13	6	87	0.2	0.2	0.1	0.1
Other chromosomal defects	278	66	34	378	0.6	0.8	0.4	0.6
TOTAL	985	208	139	1332	2.3	2.4	1.6	2.2
Situs inversus	20	12	3	35	0.0	0.1	0.0	0.1
<b>Congenital malformation syndromes</b>								
Congenital cytomegalovirus infection	195	38	31	264	0.5	0.4	0.4	0.4
Congenital cytomegalovirus infection	4	0	0.0	4	0.0	0	0.0	0.0
Congenital toxoplasmosis	1	0	0.0	1	0.0	0	0.0	0.0
Non-immune hydrops foetalis	126	19	20	165	0.3	0.2	0.2	0.3
Other and unspecified birth defects	550	87	15	652	1.3	1.0	0.2	1.1
TOTAL	16398	3040	1943	21381	38.1	35.4	22.9	35.6

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

# For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

## Infant characteristics

In the period 1998–2004, a single defect was reported in 63.8 per cent of infants, 2 defects in 18.7 per cent, 3 defects in 8.0 per cent, and 4 or more defects in 10.0 per cent of cases.

The sex was male in 58.1 per cent of infants, female in 41.4 per cent, indeterminate in 0.3 per cent of infants, and was not stated for 0.2 per cent.

Birth defects were more common in preterm and post-term infants than infants born at term (Table 116). Birth defects were also more common in infants born of a multiple

pregnancy than a singleton pregnancy: in 1998–2004, 2.0 per cent of singleton babies, 2.5 per cent of twins, and 4.2 per cent of triplets were born with a birth defect.

About 11 per cent of infants born with birth defects died in the perinatal period, over half of which were stillbirths (Table 117). These figures comprise all birth defect cases, including those where the cause of death may not be directly related to the birth defect(s). By comparison, the perinatal mortality rate among all births reported to the NSW Midwives Data Collection was less than one per cent in 2004 (see Chapter 4).

**TABLE 116**

### BIRTH DEFECT CASES BY GESTATIONAL AGE, NSW 1998–2004<sup>#</sup>

Gestational age (weeks)	1998–2002		2003		Year 2004		1998–2004		Rate/1,000 births
	No.	%	No.	%	No.	%	No.	%	
20–27	552	6.0	131	7.4	94	9.4	777	6.5	186.8
28–31	275	3.0	50	2.8	37	3.7	362	3.0	81.3
32–36	1080	11.8	197	11.2	134	13.4	1411	11.9	41.3
37–41	6723	73.5	1229	69.8	713	71.5	8665	72.8	15.9
42 +	204	2.2	29	1.6	18	1.8	251	2.1	18.1
Not stated	307	3.4	125	7.1	1	0.1	433	3.6	–
TOTAL	9141	100.0	1761	100.0	997	100.0	11899	100.0	19.8

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

<sup>#</sup> For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

**TABLE 117**

### BIRTH DEFECT CASES BY PREGNANCY OUTCOME, NSW 1998–2004<sup>#</sup>

Pregnancy outcome	1998–2002		2003		Year 2004		1998–2004	
	No.	%	No.	%	No.	%	No.	%
Stillbirth	541	5.9	121	6.9	104	10.4	766	6.4
Liveborn–neonatal death	397	4.3	69	3.9	56	5.6	522	4.4
Liveborn–postneonatal death	84	0.9	10	0.6	4	0.4	98	0.8
Liveborn surviving	8119	88.8	1561	88.6	833	83.6	10513	88.4
TOTAL	9141	100.0	1761	100.0	997	100.0	11899	100.0

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

<sup>#</sup> For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported. Postneonatal deaths are likely to be under-reported.

## Maternal characteristics

After 30 years of age, the incidence of birth defects increased with increasing maternal age (Table 118). While the rate of birth defects is higher in older women, the majority of births occur in younger women. In 1998–2004, 74.7 per cent of babies with birth defects were born to women aged less than 35 years.

In 1998–2004, 286 babies of Aboriginal or Torres Strait Islander mothers were reported to have birth defects. The rate of birth defects among these babies was 19.1 per 1,000 compared with 18.9 per 1,000 for non-Aboriginal mothers.

**TABLE 118**

### BIRTH DEFECT CASES BY MATERNAL AGE, NSW 1998–2004<sup>#</sup>

Maternal age (years)	1998–2002		2003		Year 2004		1998–2004		Rate/1,000 births
	No.	%	No.	%	No.	%	No.	%	
Under 20	416	4.6	62	3.5	41	4.1	519	4.4	19.8
20–24	1360	14.9	228	12.9	151	15.1	1739	14.6	19.0
25–29	2643	28.9	404	22.9	261	26.2	3308	27.8	18.3
30–34	2514	27.5	487	27.7	323	32.4	3324	27.9	17.3
35–39	1328	14.5	255	14.5	172	17.3	1755	14.7	19.2
40–44	329	3.6	67	3.8	44	4.4	440	3.7	25.1
45+	20	0.2	3	0.2	4	0.4	27	0.2	34.1
Not stated	531	5.8	255	14.5	1	0.1	787	6.6	–
TOTAL	9141	100.0	1761	100.0	997	100.0	11899	100.0	19.8

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

<sup>#</sup> For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

## Birth defects among terminations of pregnancy, spontaneous abortions and unknown outcomes of pregnancy

In the period 1998–2003, on average about 280 terminations of pregnancy per year were reported to the NSW Birth Defects Register (Table 119). To date, 172 terminations of pregnancy have been reported to the Register for 2004. This number is expected to increase as outcomes for mothers with defects detected during pregnancy in 2004 continue to be reported. Of the 1,852 terminations of pregnancy reported in 1998–2004, 1,372 (74.1 per cent)

were associated with a chromosomal abnormality, the most common of which was Trisomy 21 (Down syndrome), and 231 (12.5 per cent) were associated with a neural tube defect (Tables 119 and 120).

For spontaneous abortions, cytogenetic analysis is only carried out in cases of habitual abortion; the numbers presented, therefore, underestimate the number of spontaneous abortions that occur due to birth defects. Descriptions of some diagnostic terms used here are included in Appendix 1.

**TABLE 119**

### PREGNANCIES WITH FETUSES AFFECTED BY BIRTH DEFECTS AND RESULTING IN SPONTANEOUS ABORTION, TERMINATION OF PREGNANCY OR UNKNOWN OUTCOME, NSW 1998–2004

Pregnancy outcome	1998	1999	2000	2001	Year	2003	2004	1998–2004
	No.	No.	No.	No.	2002	No.	No.	No.
Spontaneous abortion	84	119	124	171	202	232	280	1212
Termination of pregnancy								
less than 20 weeks gestation	254	310	262	257	285	312	172	1852
Unknown outcome	13	16	22	19	7	18	–	95
TOTAL	351	445	408	447	494	562	452	3159

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

TABLE 120

## BIRTH DEFECTS AMONG SPONTANEOUS ABORTIONS, TERMINATIONS OF PREGNANCY AND UNKNOWN OUTCOME OF PREGNANCY BY DIAGNOSTIC CATEGORY, NSW 1998–2004

Diagnostic category	1998–2002			Year 2003			2004			1998–2004		
	Spont. abortion No.	Termination of pregnancy less than 20 weeks gestation No.	Unknown outcome No.	Spont. abortion No.	Termination of pregnancy less than 20 weeks gestation No.	Unknown outcome No.	Spont. abortion No.	Termination of pregnancy less than 20 weeks gestation No.	Unknown outcome No.	Spont. abortion No.	Termination of pregnancy less than 20 weeks gestation No.	Unknown outcome No.
<b>Defects of nervous system</b>												
Neural tube defects	8	183	3	0	29	0	1	19	9	231	3	
Other nervous system defects	4	142	6	1	12	0	1	19	6	173	6	
TOTAL	12	325	9	1	41	0	2	38	15	404	9	
<b>Defects of eye</b>	0	2	0	0	1	0	0	0	0	3	0	
<b>Defects of ear, face and neck</b>	0	11	1	1	1	0	0	2	14	1		
<b>Defects of cardiovascular system</b>	9	172	7	4	37	0	1	24	14	233	7	
<b>Defects of respiratory system</b>	0	25	3	2	1	0	0	3	2	29	3	
<b>Defects of gastrointestinal system</b>	3	110	4	5	25	0	2	23	10	158	4	
<b>Defects of musculoskeletal system</b>	30	317	9	4	47	0	7	49	41	413	9	
<b>Defects of genitourinary system</b>	8	153	6	3	28	0	2	27	13	208	6	
<b>Defects of integumentary system</b>	1	2	0	0	0	0	1	0	2	0		
<b>Cystic hygroma</b>	7	84	0	2	30	3	6	20	15	134	3	
<b>Chromosomal defects</b>												
Trisomy 21	53	486	16	19	126	11	25	60	97	672	27	
Trisomy 13	30	73	10	7	16	1	9	9	46	98	11	
Trisomy 18	38	181	8	9	47	0	15	20	62	248	8	
Turner syndrome	65	77	2	26	23	0	33	13	124	113	2	
Other chromosomal defects	494	195	25	169	33	6	192	13	855	241	31	
TOTAL	680	1012	61	230	245	18	274	115	1184	1372	79	
<b>Situs inversus</b>	0	5	0	0	0	0	0	0	0	5	0	
<b>Congenital malformation syndromes</b>	2	11	0	0	2	0	0	0	2	13	0	
<b>Non-immune hydrops foetalis</b>	7	45	2	2	8	1	2	6	11	59	3	
<b>Other and unspecified birth defects</b>	3	36	3	2	3	0	0	6	5	45	3	
TOTAL	762	2310	105	256	469	22	297	313	1315	3092	127	

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

## Trends in selected birth defects

Trends in a selection of common birth defects are shown in Table 121 and Figures 14 to 17. For 1998–2003, malformations reported up to one year of age are included; for 2004, malformations reported during pregnancy or at birth are included.

The reported number of liveborn and stillborn infants with neural tube defects was 42 in 1998 and 39 in 2003, and 33 have been reported for 2004 to date. The number of reported terminations of pregnancy was 38 in 1998, 28 in 2003, and 19 in 2004 (Figure 14).

Over the period 1998–2004, the number of cases of isolated cleft palate ranged from 60 to 88 per year, and for total cleft lip (including cases of cleft lip and cleft palate) from 71 to 89 per year. Termination of pregnancy was usually associated with other defects such as neural tube defects, chromosomal abnormalities, or multiple abnormalities in addition to the cleft lip and/or cleft palate.

The reported number of liveborn and stillborn infants with chromosomal defects was 201 in 1998 and 207 in 2003, and the number of reported terminations of pregnancy associated with chromosomal defects rose from 156 in 1998 to 244 in 2003. The number of infants born with Down syndrome was 111 in 1998 and 102 in 2003, while the number of reported terminations of pregnancy associated with Down syndrome rose from 74 in 1998 to 125 in 2003.

In 1998, 19 liveborn infants and 2 stillborn infants had a diaphragmatic hernia, and there were 3 terminations of pregnancy for this condition. In 2003, there were 17 liveborn infants and 4 stillborn infants who had a diaphragmatic hernia, and there were no terminations of pregnancy (Figure 17).

**TABLE 121**

**SELECTED BIRTH DEFECT CASES BY YEAR, NSW 1998–2004<sup>#</sup>**

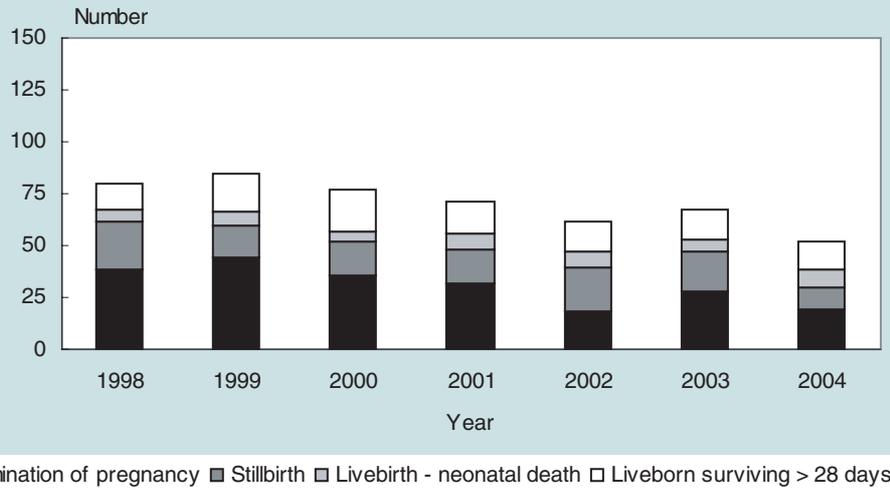
Birth defect	Year													
	1998		1999		2000		2001		2002		2003		2004	
	No.	Rate/ 1,000												
Neural tube defects	80	0.9	85	1.0	77	0.9	71	0.8	62	0.7	67	0.8	52	0.6
Anencephalus	34	0.4	24	0.3	29	0.3	25	0.3	20	0.2	18	0.2	15	0.2
Spina bifida	42	0.5	57	0.7	42	0.5	39	0.5	33	0.4	46	0.5	31	0.4
Encephalocele	10	0.1	8	0.1	13	0.1	8	0.1	10	0.1	7	0.1	6	0.1
Cleft palate	68	0.8	67	0.8	79	0.9	67	0.8	60	0.7	88	1.0	61	0.7
Total cleft lip	89	1.0	84	1.0	71	0.8	88	1.0	76	0.9	85	1.0	74	0.9
Hypospadias	191	2.2	199	2.3	191	2.2	173	2.0	133	1.6	171	2.0	121	1.4
Limb reduction defects	54	0.6	56	0.6	61	0.7	42	0.5	21	0.2	31	0.4	27	0.3
Chromosomal abnormalities	357	4.2	412	4.8	412	4.7	370	4.3	441	5.2	451	5.3	253	3.0
Down syndrome	185	2.2	199	2.3	214	2.5	180	2.1	221	2.6	227	2.6	139	1.6
Renal agenesis and dysgenesis	100	1.2	80	0.9	82	0.9	75	0.9	63	0.7	68	0.8	60	0.7
Exomphalos	25	0.3	17	0.2	28	0.3	22	0.3	22	0.3	20	0.2	24	0.3
Gastroschisis	18	0.2	18	0.2	20	0.2	23	0.3	18	0.2	19	0.2	15	0.2
Diaphragmatic hernia	24	0.3	41	0.5	22	0.3	28	0.3	23	0.3	21	0.2	25	0.3

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

<sup>#</sup> Includes terminations of pregnancy, stillbirths and livebirths. From 1 January 1998 birth defects became notifiable under the *NSW Public Health Act 1991*. This resulted in increased reporting of birth defects, particularly those associated with termination of pregnancy. For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

**FIGURE 14**

**NEURAL TUBE DEFECTS: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004\***

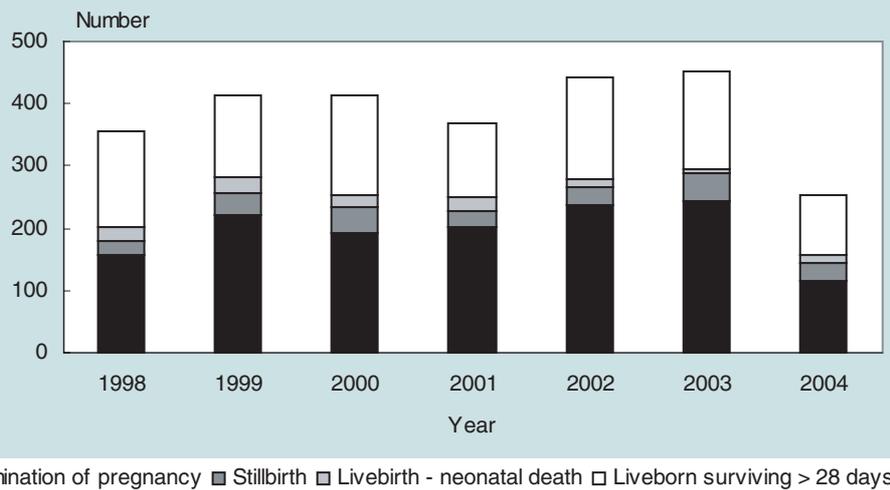


Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

# For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

**FIGURE 15**

**CHROMOSOMAL ABNORMALITIES: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004\***

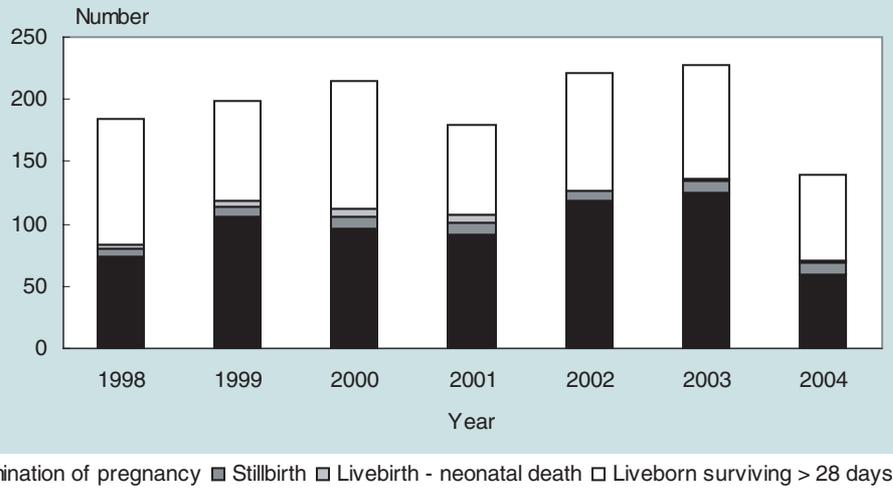


Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

# For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

**FIGURE 16**

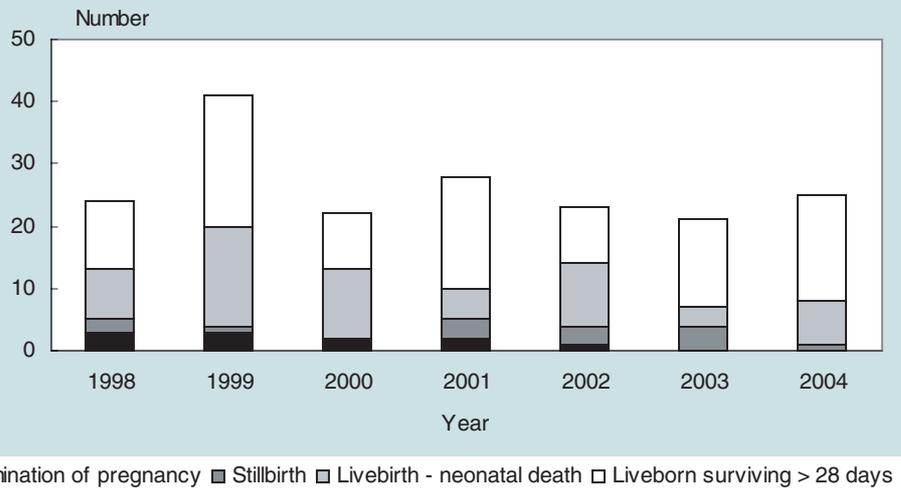
**DOWN SYNDROME: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004\***



Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.  
 # For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

**FIGURE 17**

**DIAPHRAGMATIC HERNIA: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004\***



Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.  
 # For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.

## Birth defects by NSW health areas

Crude rates of reported birth defects for NSW health areas and rates standardised for maternal age are shown in Table 122. The denominator population includes livebirths and stillbirths among NSW residents as reported to the MDC. The rate of birth defects increases with increasing maternal age (Table 118). In order to allow direct comparison of geographic areas, rates have been standardised to the maternal age distribution of births in NSW in 1991.

Information shown in this table reflects the reporting practices of the various areas. From 1 January 1998 doctors, hospitals and laboratories are required to notify birth defects detected during pregnancy, at birth or up to one year of life under the *NSW Public Health Act 1991*. Thus, higher rates of reported birth defects may be expected from 1998 onwards compared to previous years.

In interpreting birth defect rates among NSW areas, it should also be noted that infants with birth defects who are born to mothers resident in areas close to interstate borders may be transferred interstate for care and therefore may not be reported to the BDR.

Over the period 1998–2004, standardised rates of reported birth defects were lowest in the Greater Southern Area and highest in the Hunter & New England Area. Review of cases showed slightly increased reported rates of a range of birth defects in the Hunter & New England Area compared to NSW overall including: unstable hips (but not dislocated hips), isolated atrial septal defect and ventricular septal defect, and first degree hypospadias. The range and pattern of these defects suggests that enumeration of less severe conditions is better in the Hunter & New England Health Area compared with NSW as a whole.

**TABLE 122**

### BIRTH DEFECTS IN NSW HEALTH AREAS, 1998–2004\*

Health Area	1998–2002			2003			2004			1998–2004			99% confidence intervals
	No.	Crude rate per 1,000 births	Standardised rate per 1,000 births	No.	Crude rate per 1,000 births	Standardised rate per 1,000 births	No.	Crude rate per 1,000 births	Standardised rate per 1,000 births	No.	Crude rate per 1,000 births	Standardised rate per 1,000 births	
Sydney South West	2196	22.8	21.4	455	23.0	18.9	250	13.2	12.5	2901	21.5	19.8	18.8–20.9
South Eastern Sydney & Illawarra	1698	24.3	21.7	354	25.0	20.2	164	11.4	10.3	2216	22.5	19.9	18.6–21.1
Sydney West	1898	23.6	22.4	359	22.2	18.4	220	13.7	13.5	2477	22.0	20.6	19.5–21.7
Northern Sydney & Central Coast	1536	23.4	20.9	285	21.3	14.6	172	13.0	11.7	1993	21.6	18.8	17.4–20.2
Hunter & New England	1371	26.9	25.8	282	28.6	24.2	177	18.0	16.0	1830	25.9	24.2	22.7–25.8
North Coast	516	21.2	20.8	83	17.9	16.4	64	13.5	12.0	663	19.6	19.0	17.1–21.1
Greater Southern	388	17.7	16.4	72	18.5	15.3	46	11.8	11.5	506	17.0	15.7	13.8–17.6
Greater Western	428	20.6	19.6	78	19.7	18.9	61	15.9	15.6	567	19.8	19.0	16.9–21.2
TOTAL NSW	10031	23.3	21.8	1968	22.9	18.9	1154	13.6	12.8	13153	21.9	20.1	19.7–20.6

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

\* Cases exclude terminations of pregnancy, stillbirths and livebirths where the place of residence is unknown. For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.