# 9. 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 1996–2001 and birth defects detected during pregnancy or at birth for 2002.

# Trends in reported birth defects

Between 1996 and 2002, the reported number of infants with birth defects has remained stable at just over two per cent (Table 106). In 2002, 926 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 birthweight

# TABLE 106

BIRTH DEFECT CASES, NSW 1996-2002#

Year	Birth defect cases	Births	Rate/1,000 births
1996	1875	85706	21.9
1997	1991	87416	22.8
1998	1941	85627	22.7
1999	1828	86468	21.1
2000	1858	87279	21.3
2001	1774	85285	20.8
2002	926	85398	10.8

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

# For 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, cases reported during pregnancy or at birth are reported.

greater than 400 grams are presented in Table 107. Birth defects are classified using the British Paediatric Association (BPA) Classification of Diseases, which is primarily organised by body system.<sup>1</sup> 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 1996–2002, defects of the cardiovascular system were most commonly reported, followed by defects of the musculoskeletal system and defects of the genito-urinary system (Table 107). This is a similar pattern to previous years. In 2001, the overall rate of defects was lower than the previous five years (34.8 versus 41.0 per 1,000), due to fewer babies being born with multiple defects.

#### TABLE 107

BIRTH DEFECTS AMONG STILLBIRTHS AND LIVE BIRTHS BY DIAGNOSTIC CATEGORY, NSW 1996-2002

Diagnostic category		No. defects				Rate/1,000 b	irths	
	1996–2000	2001	2002	1996-2002	1996–2000	2001	2002	1996–2002
Defects of nervous system								
Anencephaly	48	9	12	69	0.1	0.1	0.1	0.1
Spina Bifida	147	26	20	193	0.3	0.3	0.2	0.3
Encephalocele	32	5	6	43	0.1	0.1	0.1	0.1
Microcephaly	131	17	12	160	0.3	0.2	0.1	0.3
Congenital hydrocephalus	180	32	25	237	0.4	0.4	0.3	0.4
Other nervous system defects	438	64	28	530	1.0	0.8	0.3	0.9
TOTAL	976	153	103	1232	2.3	1.8	1.2	2.0
Defects of eye								
Anophthalmos-microphthalmos	65	9	1	75	0.2	0.1	0.0	0.1
Buphthalmos-congenital glaucoma	a 22	6	2	30	0.1	0.1	0.0	0.0
Congenital cataract	89	10	4	103	0.2	0.1	0.0	0.2
Other eye defects	179	38	8	225	0.4	0.4	0.1	0.4
TOTAL	355	63	15	433	0.8	0.7	0.2	0.7
Defects of ear, face and neck								
Absence/ stricture auditory canal	47	12	15	74	0.1	0.1	0.2	0.1
Absent auricle	8	2	1	11	0.0	0.0	0.0	0.0
Defects of face and neck	45	5	4	54	0.1	0.1	0.0	0.1
Other ear defects	92	12	23	127	0.2	0.1	0.3	0.2
TOTAL	192	31	43	266	0.4	0.4	0.5	0.4
Defects of cardiovascular system								
Transposition of great vessels	206	39	31	276	0.5	0.5	0.4	0.5
Tetralogy of Fallot	149	17	20	186	0.3	0.2	0.2	0.3
Ventricular septal defect	978	161	83	1222	2.3	1.9	1.0	2.0
Atrial septal defect	935	144	69	1148	2.2	1.7	0.8	1.9

# TABLE 107 (continued)

BIRTH DEFECTS AMONG STILLBIRTHS AND LIVE BIRTHS BY DIAGNOSTIC CATEGORY, NSW 1996-2002#

Diagnostic category	1996–2000	No. defects 2001	2002	1996–2002	1996–2000	Rate/1 2001	,000 births 2002	1996–2002
Defects of cardiovascular system (co	nt)							
Heart valve defects	744	114	51	909	1.7	1.3	0.6	1.5
Patent ductus arteriosus > 37 weeks	534	67	50	651	1.2	0.8	0.6	1.1
Coarctation of aorta	186	49	16	251	0.4	0.6	0.2	0.4
Other defects of aorta	110	16	10	136	0.3	0.2	0.2	0.4
Defects of pulmonary artery	155	25	4	184	0.4	0.3	0.0	0.3
Other cardiovascular defects	867	112	68	1047	2.0	1.3	0.8	1.7
TOTAL	4864	744	402	6010	11.2	8.7	4.7	10.0
Defects of respiratory system								
Defects of nose	77	12	9	98	0.2	0.1	0.1	0.2
Defects of larynx, trachea and bronchi	us 54	6	3	63	0.1	0.1	0.0	0.1
Defects of lung	97	15	13	125	0.2	0.2	0.2	0.2
Other respiratory defects	1	0	0.0	1	0.0	0	0.0	0.0
TOTAL	229	33	25	287	0.5	0.4	0.3	0.5
	225	55	20	207	0.5	0.4	0.5	0.5
Defects of gastrointestinal system	004	07		500	0.0	0.0	0.0	0.0
Cleft palate only	381	67	55	503	0.9	0.8	0.6	0.8
Cleft lip only	164	23	28	215	0.4	0.3	0.3	0.4
Cleft palate and cleft lip	233	59	37	329	0.5	0.7	0.4	0.5
Oesophageal atresia only	5	4	1	10	0.0	0.0	0.0	0.0
Oesophageal atresia with TOF	100	11	5	116	0.2	0.1	0.1	0.2
Tracheo-oesophageal fistula (TOF) or		4	3	35	0.1	0.0	0.0	0.1
Atresia–stenosis of small intestine	136	31	19	186	0.3	0.4	0.2	0.3
	146		19					
Atresia-stenosis of anus		21		179	0.3	0.2	0.1	0.3
Other gastrointestinal defects	509	91	22	622	1.2	1.1	0.3	1.0
TOTAL	1702	311	182	2195	3.9	3.6	2.1	3.6
Defects of genitourinary system								
Defects of female genitals	44	16	1	61	0.1	0.2	0.0	0.1
Undescended testis	386	73	16	475	0.9	0.9	0.2	0.8
Hypospadias	929	172	94	1195	2.1	2.0	1.1	2.0
	26	3	2	31	0.1	0.0	0.0	0.1
Epispadias								
Chordee	147	25	17	189	0.3	0.3	0.2	0.3
Indeterminate sex-ambiguous genitali		12	6	80	0.1	0.1	0.1	0.1
Renal agenesis–dysgenesis	212	41	21	274	0.5	0.5	0.2	0.5
Obstructive defects of renal pelvis								
and ureter	804	148	35	987	1.9	1.7	0.4	1.6
Other genitourinary system defects	680	139	40	859	1.6	1.6	0.5	1.4
TOTAL	3290	629	232	4151	7.6	7.4	2.7	6.9
	5250	023	202	4151	7.0	7.7	2.1	0.5
Defects of musculoskeletal system	700	110	00	0.07	4 7		0.7	4.5
Congenital dislocation of the hips	726	119	62	907	1.7	1.4	0.7	1.5
Talipes equinovarus	283	58	33	374	0.7	0.7	0.4	0.6
Polydactyly	478	83	93	654	1.1	1.0	1.1	1.1
Syndactyly	121	11	21	153	0.3	0.1	0.2	0.3
Reduction deformities of limbs	311	51	25	387	0.7	0.6	0.3	0.6
Craniosynostosis	398	54	11	463	0.9	0.6	0.1	0.8
	136	27	19	182	0.3	0.0	0.1	0.3
Diaphragmatic hernia								
Exomphalos	75	16	11	102	0.2	0.2	0.1	0.2
Gastroschisis	87	21	18	126	0.2	0.2	0.2	0.2
Other musculoskeletal defects	1154	150	73	1377	2.7	1.8	0.9	2.3
TOTAL	3769	590	366	4725	8.7	6.9	4.3	7.8
Defects of integumentary system	384	61	26	471	0.9	0.7	0.3	0.8
Cystic hygroma	54	16	6	76	0.3	0.2	0.0	0.0
	54	10	0	10	0.1	0.2	0.1	0.1
Chromosomal defects	E 40	0.0	75	700	4.0	10	0.0	10
Trisomy 21	546	88	75	709	1.3	1.0	0.9	1.2
Trisomy 13	35	6	3	44	0.1	0.1	0.0	0.1
Trisomy 18	89	11	17	117	0.2	0.1	0.2	0.2
Turner syndrome	56	11	9	76	0.1	0.1	0.1	0.1
Other chromosomal defects	249	51	26	326	0.6	0.6	0.3	0.5
TOTAL	975	167	130	1272	2.3	2.0	1.5	2.1
situs inversus	18	5	3	26	0.0	0.1	0.0	0.0
Congenital malformation syndromes	197	29	17	243	0.5	0.3	0.2	0.4
Congenital rubella syndrome	2	0	0.0	2	0.0	0	0.0	0.0
Congenital cytomegalovirus infection		0	0.0	9	0.0	0	0.0	0.0
Congenital toxoplasmosis	. 1	0	0.0	1	0.0	Õ	0.0	0.0
Non-immune hydrops foetalis	131	28	13	172	0.3	0.3	0.2	0.3
Other and unspecified birth defects	571	111	12	694	1.3	1.3	0.1	1.2
OTAL	17719	2971	1575	22265	41.0	34.8	18.4	36.9

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health. # For 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, cases reported during pregnancy or at birth are reported.

#### Infant characteristics

In the period 1996–2002, a single defect was reported in 63.5 per cent of infants, two defects in 18.0 per cent, three defects in 8.2 per cent, and four or more defects in 10.4 per cent of cases.

The sex was male in 58.3 per cent of infants, female in 41.1 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 postterm infants than infants born at term (Table 108). Birth defects were also more common in infants born of a multiple pregnancy than a singleton pregnancy: in 1996–2002, 2.0 per cent of singleton babies, 2.6 per cent of twins, and 3.2 per cent of triplets were born with a birth defect.

About 10 per cent of infants born with birth defects died in the perinatal period, with stillbirths contributing over half the perinatal deaths (Table 109). 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 8.7 per 1,000 in 2001 (see Chapter 4).

#### TABLE 108

Gestational age									
(weeks)	1990	6–2000	:	2001	20	002	199	96–2002	
	No.	%	No.	%	No.	%	No.	%	Rate/1,000 birth
20–27	519	5.5	110	6.2	82	8.9	711	5.8	175.0
28–31	287	3.0	49	2.8	21	2.3	357	2.9	82.7
32–36	1095	11.5	215	12.1	99	10.7	1409	11.6	41.5
37–41	7088	74.7	1284	72.4	698	75.4	9070	74.4	16.6
42 +	224	2.4	40	2.3	25	2.7	289	2.4	20.3
Not stated	280	2.9	76	4.3	1	0.1	357	2.9	-
TOTAL	9493	100.0	1774	100.0	926	100.0	12193	100.0	20.2

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

For 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, cases reported during pregnancy or at birth are reported.

#### TABLE 109

#

BIRTH DEFECT CASES BY PREGNANCY OUTCOME, NSW 1996-2002#

Pregnancy outcome					Year					
	199	6–2000	:	2001	20	002	199	1996-2002		
	No.	%	No.	%	No.	%	No.	%		
Stillbirth	554	5.8	98	5.5	96	10.4	748	6.1		
Liveborn–neonatal death	418	4.4	88	5.0	43	4.6	549	4.5		
Liveborn-postneonatal death	87	0.9	10	0.6	6	0.6	103	0.8		
Liveborn surviving	8434	88.8	1578	89.0	781	84.3	10793	88.5		
TOTAL	9493	100.0	1774	100.0	926	100.0	12193	100.0		

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

# For 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, cases reported during pregnancy or at birth are reported.

#### **Maternal characteristics**

After 30 years of age, the incidence of birth defects increased with increasing maternal age (Table 110). While the rate of birth defects is higher in older women, the majority of births occur in younger women: in 1996–2002, 77.2 per cent of babies with birth defects were born to women aged less than 35 years.

In 1996–2002, 205 babies of Aboriginal or Torres Strait Islander mothers were reported to have birth defects. The rate of birth defects among these babies was 14.6 per 1,000 compared with 20.4 per 1,000 for non-Aboriginal mothers.

#### TABLE 110

BIRTH DEFECT CASES BY MATERNAL AGE, NSW 1996-2002#

Maternal age					Year				
(years)	1990	6–2000	:	2001	20	002	199	96–2002	
	No.	%	No.	%	No.	%	No.	%	Rate/1,000 births
Under 20	468	4.9	80	4.5	38	4.1	586	4.8	20.9
20–24	1480	15.6	275	15.5	144	15.6	1899	15.6	19.5
25–29	2790	29.4	514	29.0	259	28.0	3563	29.2	18.8
30–34	2600	27.4	470	26.5	292	31.5	3362	27.6	18.2
35–39	1353	14.3	262	14.8	151	16.3	1766	14.5	20.4
40–44	320	3.4	54	3.0	35	3.8	409	3.4	26.4
45+	17	0.2	3	0.2	5	0.5	25	0.2	38.8
Not stated	465	4.9	116	6.5	2	0.2	583	4.8	_
TOTAL	9493	100.0	1774	100.0	926	100.0	12193	100.0	20.2

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health. # For 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, 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 1996–2001, about 220 terminations of pregnancy per year were reported to the NSW Birth Defects Register (Table 111). Following the introduction of a requirement to notify birth defects under the *NSW Public Health Act 1991* from 1 January 1998, the number of terminations reported rose to about 250 per year.

Of the total 1,420 terminations of pregnancy reported in 1996–2002, 935 (65.8 per cent) were associated with a chromosomal abnormality, the most common of which was Trisomy 21(Down syndrome), and 245 (17.3 per cent) were associated with a neural tube defect (Table 112). In 1996–2002, 51.6 per cent of terminations were carried out in women aged less than 35 years (Table 113).

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 111

PREGNANCIES WITH FETUSES AFFECTED BY BIRTH DEFECTS AND RESULTING IN SPONTANEOUS ABORTION, TERMINATION OF PREGNANCY OR UNKNOWN OUTCOME, NSW 1996–2002

Pregnancy outcome	1996–2000 No.	2001 No.	Year 2002 <sup>#</sup> No.	1996–2002 No.
Spontaneous abortion Termination of pregnancy less than	461	170	163	794
20 weeks gestation	1054	257	109	1420
Unknown outcome	378	21	0	399
TOTAL	1893	448	272	2613

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health. # For 2002, numbers refer only to outcomes for pregnancies

For 2002, numbers refer only to outcomes for pregnancies with fetuses affected by birth defects where outcomes have been reported to date.

# TABLE 112

BIRTH DEFECTS AMONG SPONTANEOUS ABORTIONS, TERMINATIONS OF PREGNANCY AND UNKNOWN OUTCOME OF PREGNANCY BY DIAGNOSTIC CATEGORY, NSW 1996–2002

Diagnostic category					Year						
		1996-2000			2001			2002		1996–200	
		Termination of pregnancy less than 20 weeks gestation		Spont. abortion	Termination of pregnancy less than 20 weeks gestation		Spont. abortion	Termination of pregnancy less than 20 weeks gestation		Termination of pregnancy less than 20 weeks gestation	
	No.	No.	No.	No.	No.	No.	No.	No.	No.		. No.
Defects of nervous system											
Neural tube defects	5	195	5	3	33	1	1	17	9	245	6
Other nervous system											
defects	4	111	7	1	28	2	0	12	5	151	9
TOTAL	9	306	12	4	61	3	1	29	14	396	15
Defects of eye	0	3	0	0	1	0	0	0	0	4	0
Defects of ear, face and											
neck	0	11	0	0	2	1	0	1	0	14	1
Defects of cardiovascular											
system	7	166	22	1	26	4	0	17	8	209	26
Defects of respiratory											
system	0	27	4	0	3	0	0	3	0	33	4
Defects of gastrointestinal								_		100	
system	4	96	4	0	20	2	0	7	4	123	6
Defects of genitourinary system	8	167	10	2	19	2	0	12	10	198	12
Defects of musculoskeletal		107	10	2	19	2	0	12	10	190	12
system	22	326	16	9	45	4	2	22	33	393	20
Defects of the											
integumentary system	1	1	0	0	0	0	0	1	1	2	0
Cystic hygroma	7	77	12	1	13	0	0	8	8	98	12
Chromosomal defects											
Trisomy 21	39	322	148	8	92	3	9	33	56		151
Trisomy 13	23	41	20	6	17	5	6	9	35	67	25
Trisomy 18	22	128	54	12	34	2	7	10	41	172	56
Turner syndrome	46	53	23	10	20	0	14	5	70	78	23
Other chromosomal											
defects	314	131	99	129	41	9	126	15	569		108
TOTAL	444	675	344	165	204	19	162	72	771		363
Situs inversus	0	3	0	0	1	0	0	2	0	6	0
Congenital malformation											
syndromes	0	17	1	2	3	0	0	0	2	20	1
Non-immune hydrops											
foetalis	6	48	7	0	4	0	1	7	7	59	7
Other and unspecified	0		10	0	-	•			-		10
birth defects	3	30	10	0	5	0	0	1	3	36	10
TOTAL	511	1953	442	184	407	35	166	182	861	2542	477

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

TABLE 113 TRENDS IN REP	ORTED 1	ERMI	NATION	IS OF PI	REGNA	NCYAS	SOCI	TED W	/ITH BI	RTH DE	FECTS	BYMA	TERNA	L AGE	i, 199	6–200	2	
Year	Maternal age (years)																	
	1	15–19 20–24 25–29 30–34 35–39 40–44 45 + Not stated TOTAL														TOTAL		
	No	%	No	%	No	%	No	%	No	%	No	%	No	%	No	%	No	%
1996	3	2.9	16	15.2	22	21.0	23	21.9	24	22.9	11	10.5	0	0.0	6	5.7	105	100.0
1997	3	2.4	13	10.4	33	26.4	32	25.6	25	20.0	13	10.4	1	0.8	5	4.0	125	100.0
1998	3	1.2	19	7.5	56	22.0	46	18.1	64	25.2	52	20.5	4	1.6	10	3.9	254	100.0
1999	6	1.9	20	6.5	58	18.7	71	22.9	92	29.7	42	13.5	4	1.3	17	5.5	310	100.0
2000	2	0.8	14	5.4	41	15.8	65	25.0	80	30.8	45	17.3	3	1.2	10	3.8	260	100.0
2001	11	4.3	13	5.1	36	14.0	70	27.2	64	24.9	44	17.1	4	1.6	15	5.8	257	100.0
2002	1	0.9	7	6.4	18	16.5	31	28.4	23	21.1	14	12.8	0	0.0	15	13.8	109	100.0
1996–2002	29	2.0	102	7.2	264	18.6	338	23.8	372	26.2	221	15.6	16	1.1	78	5.5	1420	100.0

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 114 and Figures 14 to 21. For 1996–2001, malformations reported up to one year of age are included; for 2002, malformations reported during pregnancy or at birth are included.

The reported number of liveborn and stillborn infants with neural tube defects was 44 in 1996 and 39 in 2001, and 37 have been reported for 2002 to date. The number of reported terminations of pregnancy was 37 in 1996, 32 in 2001, and 16 in 2002.

Over the period 1996–2002, the number of cases of isolated cleft palate ranged from 48 in 2002 to 79 in 2000, and for total cleft lip (including cases of cleft lip and cleft palate) from 68 in 2002 to 89 in 1999 (Figures 15 and 16). 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 number of reported cases of hypospadias varied from 94 in 2002 to 199 in 1999 (Figure 17), and cases of limb

reduction defects varied from 18 in 2002 to 61 in 1997 and 2000 (Figure 18).

The number of reported terminations of pregnancy for chromosomal abnormalities, including Down syndrome, increased following the introduction of a requirement to notify birth defects under the *NSW Public Health Act 1991* from 1 January 1998 (Figures 19 and 20). The reported number of liveborn and stillborn infants with chromosomal defects was 184 in 1996 and 167 in 2001, and the number of reported terminations of pregnancy associated with chromosomal defects rose from 48 in 1995 to 221 in 1999. The number of infants born with Down syndrome was 112 in 1996 and 88 in 2001, while the number of reported terminations of pregnancy associated with Down syndrome rose from 20 in 1995 to 106 in 1999.

There was a trend towards improved notification of cases of renal agenesis and dysgenesis, which peaked in 1998. The increased reporting is due partly to the introduction of notification requirements in 1998, but also to improved diagnosis of less severe forms of renal dysgenesis in infants (Figure 21).

#### TABLE 114

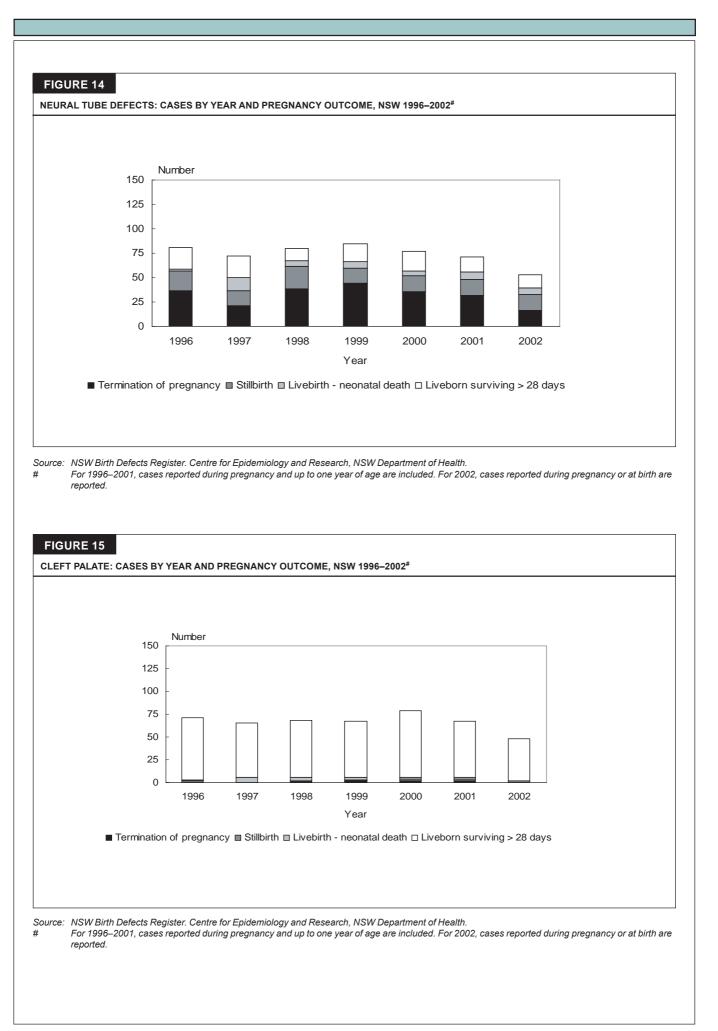
#

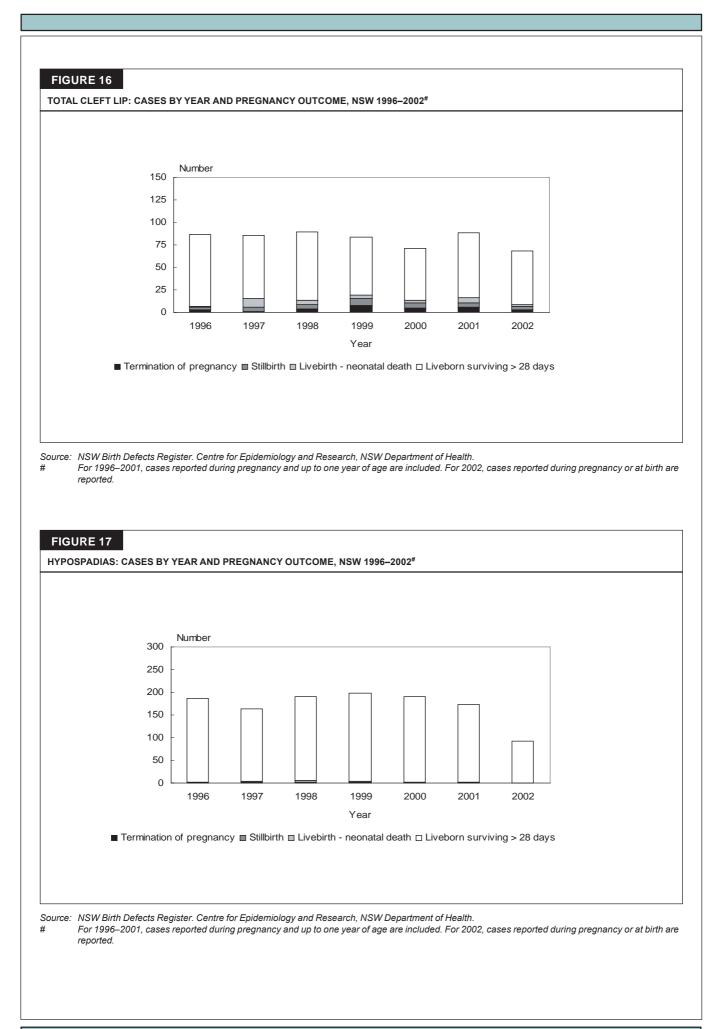
#### SELECTED BIRTH DEFECT CASES BY YEAR, NSW 1996-2002#

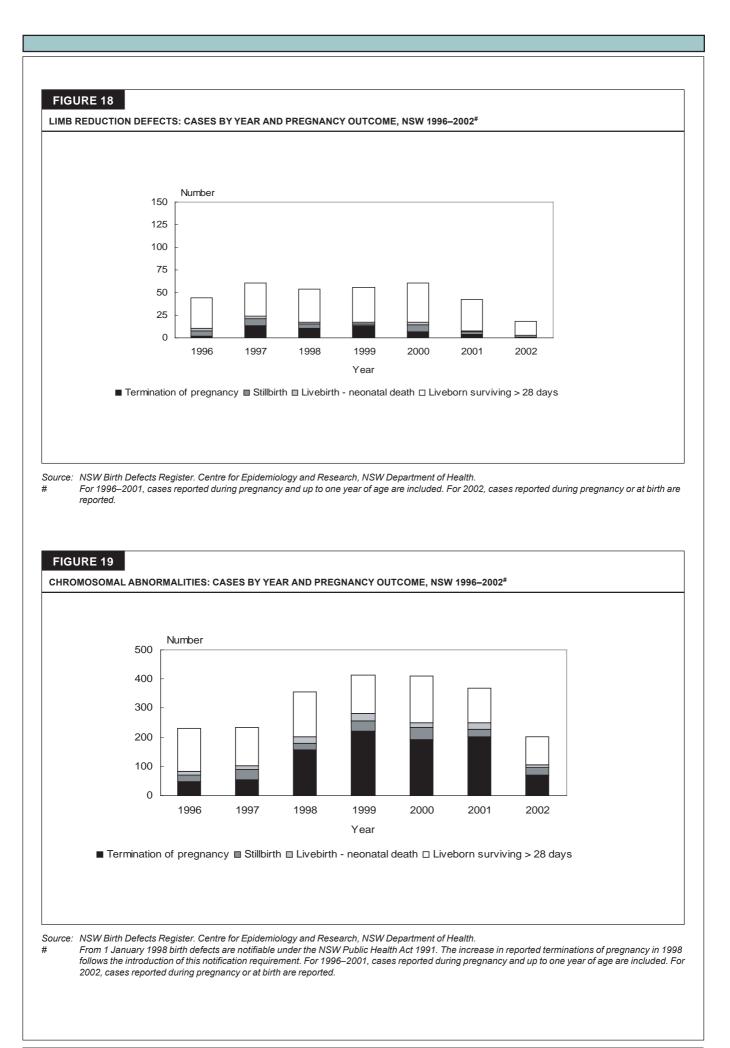
Birth defect						Ye	ar							
	1	996	19	997		1998	1	999	2	000	2	2001	2002	
	No.	Rate/ 1,000	No.	Rate/ 1,000										
Neural tube defects	81	0.9	72	0.8	80	0.9	85	1.0	77	0.9	71	0.8	53	0.6
Cleft palate	71	0.8	65	0.7	68	0.8	67	0.8	79	0.9	67	0.8	48	0.6
Total cleft lip	87	1.0	86	1.0	89	1.0	84	1.0	71	0.8	88	1.0	68	0.8
Hypospadias	186	2.2	163	1.9	191	2.2	199	2.3	191	2.2	173	2.0	94	1.1
Limb reduction defects	44	0.5	61	0.7	54	0.6	56	0.6	61	0.7	42	0.5	18	0.2
Chromosomal abnormalities	232	2.7	235	2.7	357	4.2	412	4.8	411	4.7	369	4.3	202	2.4
Down syndrome	132	1.5	139	1.6	185	2.2	199	2.3	213	2.4	180	2.1	108	1.3
Renal agenesis and dysgenesis	66	0.8	85	1.0	100	1.2	80	0.9	82	0.9	75	0.9	40	0.5

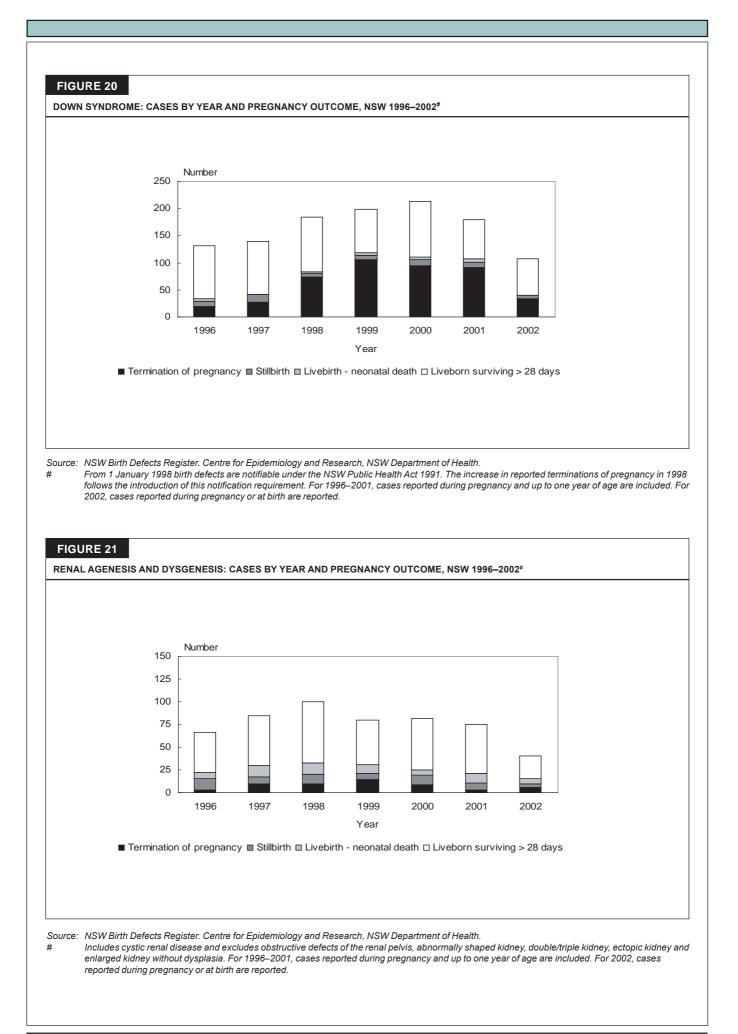
Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.

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 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, 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 115. 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 110 on page 85). 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 these tables 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 1996–2002, standardised rates of reported birth defects were lowest in the Southern Health Area and highest in the Hunter Health Area. Review of cases showed slightly increased reported rates of a range of birth defects in the Hunter Area compared to NSW overall including: unstable hips (but not dislocated hips), congenital hydronephrosis, isolated atrial septal defect and ventricular septal defect, microcephaly and congenital hydrocephalus. The range and pattern of these defects suggests that enumeration of birth defects, including less severe conditions, is better in the Hunter Health Area compared with NSW as a whole.

Birth defect rates may vary markedly from year to year for some areas, where the numbers of reported birth defects are small. For these areas, small variations in numbers of birth defect cases may result in a marked variation in the birth defect rate. The wide confidence intervals for some areas reflect this variability.

#### TABLE 115

BIRTH DEFECTS IN NSW HEALTH AREAS, 1996-2002#

Health Area		1996-200	00		2001	Year		2002			1	996-2002		
	No.		Standar- dised rate per 1,000 births	No.		Standar- dised rate per 1,000 births	No.		Standar- dised rate per	No.	Crude rate per 1,000 births	Standar- dised rate per 1,000 births		99% idence tervals
Central Sydney	780	22.9	21.3	163	24.3	15.8	75	11.2	12.5	1018	21.5	19.2	17.4	21.1
Northern Sydney	1121	24.5	23.1	207	22.0	16.4	125	13.2	11.6	1453	22.5	20.7	18.6	22.8
Western Sydney	1286	24.0	23.4	258	23.4	15.2	132	11.7	11.7	1676	22.1	20.5	19.1	21.9
Wentworth South Western	636	25.8	25.4	93	19.5	14.7	60	12.8	12.6	789	23.2	22.1	20.1	24.3
Sydney	1447	23.2	22.6	266	21.5	14.3	133	10.5	10.4	1846	21.1	19.6	18.4	20.9
Central Coast	465	24.6	24.3	80	21.7	14.5	30	8.3	8.1	575	21.9	20.7	18.4	23.1
Hunter	977	27.6	27.5	193	28.2	19.0	104	14.8	15.0	1274	25.9	24.5	22.7	26.4
Illawarra South Eastern	489	22.0	21.4	81	18.7	14.9	57	13.0	13.2	627	20.2	19.3	17.3	21.4
Sydney	1224	26.1	24.1	232	24.3	13.5	119	12.5	12.4	1575	23.9	20.9	19.3	22.7
Northern Rivers	318	21.6	22.4	50	17.9	16.0	28	10.1	10.0	396	19.5	19.6	17.1	22.3
Mid North Coast	359	24.0	23.4	78	27.4	19.9	47	16.8	16.9	484	23.5	22.0	19.4	24.8
New England	278	23.0	23.2	65	28.8	22.5	29	12.6		372	22.3	21.8	18.9	25.0
Macquarie	191	23.2	22.8	31	19.7	16.3	20	13.3		242	21.4	20.7	17.3	24.5
Mid Western	255	21.5	20.9	48	21.1	15.2	20	9.3	9.3	323	19.8	18.6	15.9	21.6
Far West	67	23.8	23.1	9	15.7	16.3	4	8.0		80	20.5	20.3	14.6	27.4
Greater Murray	295	19.6	19.0	42	16.3	11.6	19	7.5	7.4	356	17.7	16.6	14.3	19.1
Southern	198	21.9	19.7	26	15.4	10.0	13	8.7	9.2	237	19.4	17.1	14.1	20.4
TOTAL NSW	10386	24.0	23.2	1922	22.5	15.5	1015	11.9	11.8	13323	22.1	20.5	20.0	21.0

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 1996–2001, cases reported during pregnancy and up to one year of age are included. For 2002, cases reported during pregnancy or at birth are reported.

#### Reference

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1. British Paediatric Association. British Paediatric Association Classification of Diseases. London: British Paediatric Association, 1979.