

9. Congenital Conditions

Congenital conditions among stillborn and liveborn infants

From 1 January 1998, doctors, hospitals and laboratories are required under the *NSW Public Health Act 1991* to notify certain congenital conditions detected during pregnancy or in a baby up to one year of age. Information reported is included in the NSW Register of Congenital Conditions, formerly known as the NSW Birth Defects Register.

There are three types of conditions that are reported to the Register:

- Conditions that affect the growth, development and health of the baby that are present before birth, such as cleft lip, dislocated hip and problems with the development of the heart, lungs or other organs
- Conditions due to changes in the number of the baby's chromosomes, such as Down syndrome
- Four conditions due to changes in the baby's inherited genetic information: cystic fibrosis, phenylketonuria, congenital hypothyroidism and thalassaemia major.

Descriptions of some common congenital conditions are shown in Appendix 1. A list of common exclusions is shown in Appendix 2.

This chapter reports congenital conditions detected during pregnancy or in the first year of life for 2003-2008 and congenital conditions detected during pregnancy or at birth for 2009.

Trends in reported congenital conditions

Between 2003 and 2008, 1.8% to 2.1% of infants were reported to have congenital conditions, (Table 103). In 2009, 831 cases of congenital conditions detected during pregnancy or at birth have been reported to date.

Table 103. Cases of congenital conditions, NSW 2003–2009#

Year	Cases of congenital conditions	Births	Rate/1000 births
	No.	No.	
2003	1756	85405	20.6
2004	1751	84607	20.7
2005	1617	89337	18.1
2006	1768	91554	19.3
2007	1829	94640	19.3
2008	1881	94874	19.8
2009	831	94733	8.8

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.
For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Congenital conditions by diagnostic category

The most common categories of congenital conditions for stillborn and live born babies are presented in Table 104. Congenital conditions are classified using the British Paediatric Association (BPA) Classification of Diseases, which is primarily organised by body system. For infants with more than one condition, each condition is counted separately. The number of congenital conditions reported therefore exceeds the number of affected infants.

In 2002–2008, conditions of the cardiovascular system were most commonly reported, followed by conditions of the musculoskeletal system and conditions of the genitourinary system (Table 104). This is a similar pattern to previous years. In 2008, the overall rate of conditions was lower than the previous 5 years (36.0 versus 44.0 per 1,000 respectively).

Table 104. Congenital conditions among stillbirths and liveborn infants by diagnostic category, NSW 2003–2009#

Diagnostic category	No. conditions				Rate/1,000 births			
	2003-2007	2008	2009	2003-2009	2003-2007	2008	2009	2003-2009
Nervous system								
Anencephaly	51	7	5	63	0.1	0.1	0.1	0.1
Spina Bifida	138	38	27	203	0.4	0.4	0.3	0.3
Encephalocele	30	8	5	43	0.1	0.1	0.1	0.1
Microcephaly	101	19	10	130	0.3	0.2	0.1	0.2
Congenital hydrocephalus	175	33	23	231	0.5	0.3	0.2	0.4
Other nervous system anomalies	291	76	42	409	0.8	0.8	0.4	0.6
TOTAL	786	181	112	1079	2.2	1.9	1.2	1.7
Eye								
Anophthalmos-microphthalmos	58	8	11	77	0.2	0.1	0.1	0.1
Buphthalmos-congenital glaucoma	27	7	2	36	0.1	0.1	0.0	0.1
Congenital cataract	86	12	4	102	0.2	0.1	0.0	0.2
Other eye anomalies	180	45	11	236	0.5	0.5	0.1	0.4
TOTAL	351	72	28	451	1.0	0.8	0.3	0.7
Ear, face and neck								
Absence-stricture auditory canal	50	8	6	64	0.1	0.1	0.1	0.1
Absent auricle	4	1	0	5	0.0	0.0	0	0.0
Anomalies of face and neck	38	5	2	45	0.1	0.1	0.0	0.1
Other ear anomalies	65	12	7	84	0.2	0.1	0.1	0.1
TOTAL	157	26	15	198	0.4	0.3	0.2	0.3
Cardiovascular system								
Transposition of great vessels	242	51	46	339	0.7	0.5	0.5	0.5
Tetralogy of Fallot	160	33	20	213	0.5	0.3	0.2	0.3
Ventricular septal defect	804	203	86	1093	2.3	2.1	0.9	1.7
Atrial septal defect	821	194	102	1117	2.3	2.0	1.1	1.8
Heart valve anomalies	502	119	60	681	1.4	1.3	0.6	1.1
Patent ductus arteriosus > 37 weeks	444	94	79	617	1.3	1.0	0.8	1.0
Coarctation of aorta	211	48	28	287	0.6	0.5	0.3	0.5
Other anomalies of aorta	127	27	8	162	0.4	0.3	0.1	0.3
Anomalies of pulmonary artery	155	27	15	197	0.4	0.3	0.2	0.3
Other cardiovascular anomalies	744	153	131	1028	2.1	1.6	1.4	1.6
TOTAL	4210	949	575	5734	12.0	10.0	6.1	9.0
Respiratory system								
Nose	60	8	6	74	0.2	0.1	0.1	0.1
Larynx, trachea and bronchus	37	7	3	47	0.1	0.1	0.0	0.1
Lung	95	31	20	146	0.3	0.3	0.2	0.2
TOTAL	192	46	29	267	0.5	0.5	0.3	0.4
Gastrointestinal system								
Cleft palate only	430	87	47	564	1.2	0.9	0.5	0.9
Cleft lip only	144	38	24	206	0.4	0.4	0.3	0.3
Cleft palate and cleft lip	216	49	47	312	0.6	0.5	0.5	0.5
Oesophageal atresia only	17	2	2	21	0.0	0.0	0.0	0.0
Tracheo-oesophageal fistula (TOF) only	20	4	4	28	0.1	0.0	0.0	0.0
Oesophageal atresia with TOF	84	26	11	121	0.2	0.3	0.1	0.2
Atresia-stenosis of small intestine	123	26	11	160	0.4	0.3	0.1	0.3
Atresia-stenosis of anus	134	39	30	203	0.4	0.4	0.3	0.3
Other gastrointestinal anomalies	483	106	24	613	1.4	1.1	0.3	1.0
TOTAL	1651	377	200	2228	4.7	4.0	2.1	3.5
Genitourinary system								
Anomalies of female genitals	63	15	5	83	0.2	0.2	0.1	0.1
Undescended testis	312	89	20	421	0.9	0.9	0.2	0.7
Hypospadias	776	127	76	979	2.2	1.3	0.8	1.5
Epispadias	5	3	1	9	0.0	0.0	0.0	0.0
Chordee	149	43	8	200	0.4	0.5	0.1	0.3
Indeterminate sex-ambiguous genitalia	55	10	8	73	0.2	0.1	0.1	0.1
Renal agenesis-dysgenesis	174	30	20	224	0.5	0.3	0.2	0.4
Obstructive anomalies of renal pelvis and ureter	700	158	37	895	2.0	1.7	0.4	1.4
Other genitourinary system anomalies	677	142	61	880	1.9	1.5	0.6	1.4
TOTAL	2911	617	236	3764	8.3	6.5	2.5	5.9

Table 104. (Continued)

Diagnostic category	No. conditions				Rate/1,000 births			
	2003-2007	2008	2009	2003-2009	2003-2007	2008	2009	2003-2009
Musculoskeletal system								
Congenital dislocation of the hips	640	123	31	794	1.8	1.3	0.3	1.3
Talipes equinovarus	370	82	31	483	1.1	0.9	0.3	0.8
Polydactyly	485	95	60	640	1.4	1.0	0.6	1.0
Syndactyly	123	24	13	160	0.4	0.3	0.1	0.3
Reduction deformities of limbs	251	64	63	378	0.7	0.7	0.7	0.6
Craniosynostosis	197	47	2	246	0.6	0.5	0.0	0.4
Diaphragmatic hernia	129	30	25	184	0.4	0.3	0.3	0.3
Exomphalos	66	15	26	107	0.2	0.2	0.3	0.2
Gastroschisis	97	19	31	147	0.3	0.2	0.3	0.2
Other musculoskeletal anomalies	733	165	118	1016	2.1	1.7	1.2	1.6
TOTAL	3091	664	400	4155	8.8	7.0	4.2	6.5
Integumentary system								
Cystic hygroma	242	58	12	312	0.7	0.6	0.1	0.5
Cystic hygroma	48	14	8	70	0.1	0.1	0.1	0.1
Chromosomal anomalies								
Trisomy 21	450	88	61	599	1.3	0.9	0.6	0.9
Trisomy 13	36	4	8	48	0.1	0.0	0.1	0.1
Trisomy 18	92	16	8	116	0.3	0.2	0.1	0.2
Turner syndrome	72	15	5	92	0.2	0.2	0.1	0.1
Other chromosomal anomalies	331	62	37	430	0.9	0.7	0.4	0.7
TOTAL	981	185	119	1285	2.8	2.0	1.3	2.0
Situs inversus								
Situs inversus	28	6	2	36	0.1	0.1	0.0	0.1
Congenital malformation syndromes								
Congenital malformation syndromes	191	34	27	252	0.5	0.4	0.3	0.4
Congenital cytomegalovirus infection								
Congenital cytomegalovirus infection	1	2	1	4	0.0	0.0	0.0	0.0
Non-immune hydrops foetalis								
Non-immune hydrops foetalis	99	24	14	137	0.3	0.3	0.1	0.2
Other and unspecified anomalies								
Other and unspecified anomalies	486	149	20	655	1.4	1.6	0.2	1.0
TOTAL	15425	3404	1798	20627	44.0	36.0	19.0	32.5

Source: NSW Register of Congenital Conditions (HOIST), Centre for Epidemiology and Research, NSW Ministry of Health.
For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Infant characteristics

In the period 2003–2009, a single condition was reported in 63.4% of infants, two conditions in 18.2%, three conditions in 8.3%, and four or more conditions in 10.1% of cases. The sex was male in 57.7% of infants, female in 41.8%, indeterminate in 0.3% of infants, and was not stated for 0.2%.

Congenital conditions were more common in preterm and post-term infants than infants born at term (Table 105). Congenital conditions were also more common in infants born of a multiple pregnancy than a singleton pregnancy: in

2003–2009, 1.8% of singleton babies, 2.7% of twins, and 3.3% of triplets were born with a congenital condition.

About 11% of infants born with congenital conditions died in the perinatal period, over half of which were stillbirths (Table 106). These figures comprise all cases of congenital conditions, including those where the cause of death may not be directly related to the congenital condition(s). By comparison, the perinatal mortality rate among all births reported to the NSW Perinatal Data Collection was less than 1% in 2009 (see Chapter 4).

Table 105. Cases of congenital conditions by gestational age, NSW 2003–2009#

Gestational age (weeks)	Year									
	2003-2007		2008		2009		2003-2009		No. of cases/1,000 births	
	No.	%	No.	%	No.	%	No.	%		
20–27	599	6.9	136	7.2	87	10.5	822	7.2	182.2	
28–31	258	3.0	61	3.2	22	2.6	341	3.0	74.6	
32–36	1155	13.2	228	12.1	120	14.4	1503	13.1	40.4	
37–41	6313	72.4	1376	73.2	593	71.4	8282	72.4	14.3	
42 +	135	1.5	16	0.9	8	1.0	159	1.4	16.9	
Not stated	261	3.0	64	3.4	1	0.1	326	2.9	0	
TOTAL	8721	100.0	1881	100.0	831	100.0	11433	100.0	18.0	

Source: NSW Register of Congenital Conditions (HOIST), Centre for Epidemiology and Research, NSW Ministry of Health.
For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Table 106. Cases of congenital conditions by pregnancy outcome, NSW 2003–2009#

Pregnancy outcome	Year							
	2003-2007		2008		2009		2003-2009	
	No.	%	No.	%	No.	%	No.	%
Stillbirth	593	6.8	124	6.6	89	10.7	806	7.0
Liveborn/ neonatal death	333	3.8	68	3.6	45	5.4	446	3.9
Liveborn/ postneonatal death	42	0.5	15	0.8	17	2.0	74	0.6
Liveborn surviving	7753	88.9	1674	89.0	680	81.8	10107	88.4
TOTAL	8721	100.0	1881	100.0	831	100.0	11433	100.0

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.

For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Maternal characteristics

After 35 years of age, the incidence of congenital conditions increased with increasing maternal age (Table 107). While the rate of congenital conditions is higher in older women, the majority of births occur in younger women. In 2003–2009, 71.9% of babies with congenital conditions were born to women aged less than 35 years.

In 2003–2009, 414 babies of Aboriginal or Torres Strait Islander mothers were reported to have congenital conditions. The rate of congenital conditions among these babies was 22.6 per 1,000 compared with 17.2 per 1,000 for babies born to non-Aboriginal mothers.

Table 107. Cases of congenital conditions by maternal age, NSW 2003–2009#

Maternal age (years)	Year								No. of cases/ 1,000 births
	2003-2007		2008		2009		2003-2009		
	No.	%	No.	%	No.	%	No.	%	
Under 20	351	4.0	75	4.0	46	5.5	472	4.1	20.1
20–24	1198	13.7	252	13.4	112	13.5	1562	13.7	17.7
25–29	2132	24.4	409	21.7	205	24.7	2746	24.0	16.0
30–34	2649	30.4	553	29.4	239	28.8	3441	30.1	16.2
35–39	1477	16.9	379	20.1	180	21.7	2036	17.8	17.6
40–44	377	4.3	105	5.6	44	5.3	526	4.6	23.3
45+	32	0.4	6	0.3	5	0.6	43	0.4	38.5
Not stated	505	5.8	102	5.4	0	0.0	607	5.3	0.0
TOTAL	8721	100.0	1881	100.0	831	100.0	11433	100.0	18.0

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.

For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Congenital conditions among terminations of pregnancy, spontaneous abortions and unknown outcomes of pregnancy

The number of terminations of pregnancy reported to the Register increased from 313 in 2003 to 354 in 2009 (Table 108). The number of terminations of pregnancy for 2009 is expected to increase as pregnancy outcomes for babies with conditions detected during pregnancy in 2009 continue to be followed-up.

Of the 2,304 terminations of pregnancy reported in 2003–2009, 1,830 (79.4%) were associated with a chromosomal anomaly, the most common of which was Trisomy 21 (Down

syndrome), and 171 (7.4%) were associated with a neural tube defect (Table 109).

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 congenital conditions. Descriptions of some diagnostic terms used here are included in Appendix 1.

Table 108. Pregnancies with fetuses affected by congenital conditions resulting in spontaneous termination, spontaneous abortion or unknown outcome, NSW 2003–2009

Pregnancy outcome	Year							2003–2009
	2003	2004	2005	2006	2007	2008	2009	
	No.	No.	No.	No.	No.	No.	No.	No.
Spontaneous abortion	233	298	322	363	334	330	237	2117
Termination of pregnancy less than 20 weeks gestation	313	306	303	361	328	339	354	2304
Unknown outcome	17	38	29	23	23	35	0	165
TOTAL	563	642	654	747	685	704	591	4586

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.

Table 109. Congenital conditions among terminations, spontaneous abortions and unknown outcomes of pregnancy by diagnostic category, NSW 2003–2009

Diagnostic category	Year															
	2003–2007				2008				2009				2003–2009			
	Spont. abortion	Termination of pregnancy less than 20 weeks gestation	Unknown	No.	Spont. abortion	Termination of pregnancy less than 20 weeks gestation	Unknown	No.	Spont. abortion	Termination of pregnancy less than 20 weeks gestation	Unknown	No.	Spont. abortion	Termination of pregnancy less than 20 weeks gestation	Unknown	No.
Nervous system																
Neural tube defects	5	122	1	0	19	0	0	0	0	30	0	0	5	171	1	
Other nervous system defects	7	104	0	0	13	1	0	0	0	25	1	0	7	142	1	
TOTAL	12	226	1	0	32	1	0	0	0	55	1	0	12	313	2	
Eye	0	3	0	0	0	0	0	0	0	2	0	0	0	5	0	
Ear, face and neck	1	9	0	0	4	0	0	0	0	3	0	0	1	16	0	
Cardiovascular system	19	239	4	5	54	0	5	54	7	48	0	31	341	4		
Respiratory system	7	27	1	1	5	0	1	5	1	8	0	9	40	1		
Gastrointestinal system	13	141	0	6	22	1	4	22	4	20	1	23	183	1		
Musculoskeletal system	24	324	4	15	50	1	4	50	4	95	1	43	469	5		
Genitourinary system	9	166	1	5	30	0	0	30	0	15	0	14	211	1		
Integumentary system	1	0	0	0	0	0	0	0	0	0	0	1	0	0	0	
Cystic hygroma	24	133	9	8	25	7	8	25	5	21	7	37	179	16		
Chromosomal anomalies																
Trisomy 21	121	692	64	35	168	21	29	168	29	172	21	185	1032	85		
Trisomy 13	64	84	8	16	24	0	10	24	10	21	0	90	129	8		
Trisomy 18	69	210	15	16	42	3	10	42	10	40	3	95	292	18		
Turner syndrome	174	108	7	30	18	6	19	18	19	13	6	223	139	13		
Other chromosomal anomalies	1100	175	28	225	31	2	162	31	162	32	2	1487	238	30		
TOTAL	1528	1269	122	322	283	32	230	283	230	278	32	2080	1830	154		
Situs inversus	0	2	0	0	0	0	0	0	0	0	0	0	2	0	0	
Congenital malformation syndromes	0	16	0	2	2	0	1	2	1	3	0	3	21	0	0	
Non-immune hydrops foetalis	17	48	2	3	7	0	3	7	3	11	0	23	66	2	2	
Other and unspecified conditions	4	31	1	0	9	0	0	9	0	9	0	4	49	1	1	
TOTAL	1659	2634	145	367	523	42	255	523	255	568	42	2281	3725	187	187	

Source: NSW Register of Congenital Conditions (HOIST), Centre for Epidemiology and Research, NSW Ministry of Health.

Trends in selected congenital conditions

Trends in a selection of common congenital conditions are shown in Table 110 and Figures 5 to 8. For 2003–2008, conditions reported up to one year of age are included. For 2009, conditions reported during pregnancy or at birth are included.

Over the period 2003–2009, the rate of neural tube defects has remained fairly stable at about 0.7 per 1,000 births. The reported number of live born and stillborn infants with neural tube defects was 39 in 2003 and 35 in 2009. The number of reported terminations of pregnancy was 28 in both 2003 and 2009 (Figure 5).

Over the period 2003–2008, the number of cases of isolated cleft palate ranged from 64 to 90 per year, and for total cleft lip (including cases of cleft lip and cleft palate) from 66 to 91 per year. Termination of pregnancy was usually associated with other conditions such as neural tube conditions, chromosomal anomalies, or multiple anomalies in addition to the cleft lip and/or cleft palate.

The reported number of live born and stillborn infants reported with chromosomal anomalies was 209 in 2003 and 185 in 2008. The number of reported terminations of pregnancy associated with chromosomal conditions rose from 245 in 2003 to 283 in 2008. The number of infants born with Down syndrome was 102 in 2003 and 85 in 2008, while the number of reported terminations of pregnancy associated with Down syndrome rose from 126 in 2003 to 168 in 2008.

In 2003, 17 live born infants were reported to have a diaphragmatic hernia, 4 babies were stillborn and there were no terminations of pregnancy associated with this condition. In 2008, there were 28 live born infants reported with a diaphragmatic hernia, 2 stillborn babies and 2 terminations of pregnancy (Figure 8).

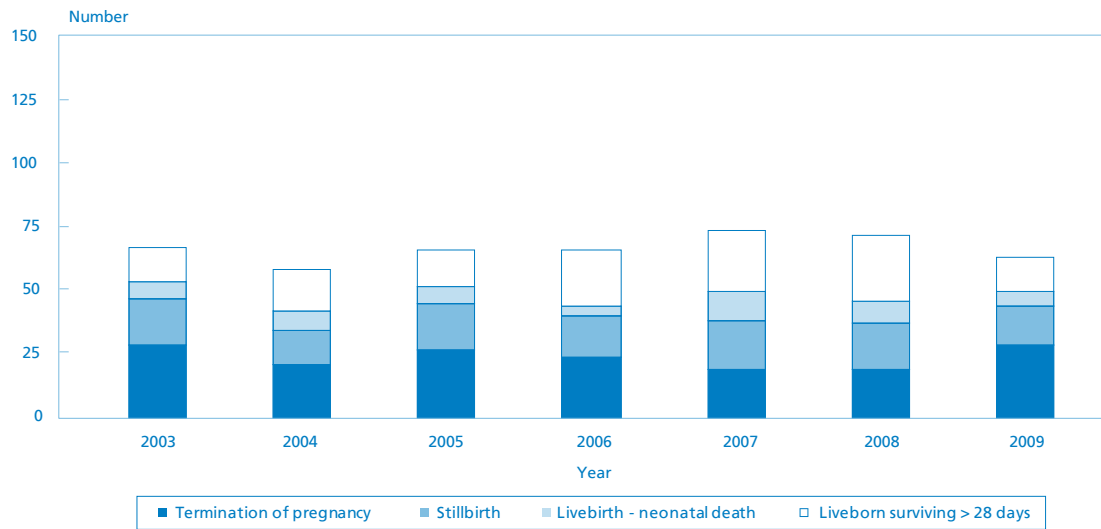
Table 110. Selected congenital conditions by year, NSW 2003–2009#

Condition	2003		2004		2005		2006		2007		2008		2009	
	No.	Rate/1000	No.	Rate/1000	No.	Rate/1000	No.	Rate/1000	No.	Rate/1000	No.	Rate/1000	No.	Rate/1000
Neural tube defects	67	0.8	58	0.7	66	0.7	66	0.7	74	0.8	72	0.8	63	0.7
Anencephalus	18	0.2	16	0.2	19	0.2	15	0.2	26	0.3	18	0.2	13	0.1
Spina bifida	46	0.5	35	0.4	40	0.4	45	0.5	42	0.4	45	0.5	45	0.5
Encephalocele	7	0.1	8	0.1	8	0.1	8	0.1	8	0.1	9	0.1	9	0.1
Cleft palate	88	1.0	74	0.9	90	1.0	64	0.7	74	0.8	82	0.9	43	0.5
Total cleft lip	83	1.0	79	0.9	66	0.7	85	0.9	73	0.8	91	1.0	73	0.8
Hypospadias	171	2.0	150	1.8	146	1.6	149	1.6	162	1.7	127	1.3	77	0.8
Limb reduction defects	31	0.4	36	0.4	38	0.4	43	0.5	49	0.5	53	0.6	45	0.5
Chromosomal abnormalities	454	5.3	447	5.3	409	4.6	480	5.2	454	4.8	468	4.9	397	4.2
Down syndrome	228	2.7	233	2.8	209	2.3	252	2.8	218	2.3	256	2.7	233	2.5
Renal agenesis and dysgenesis	67	0.8	82	1.0	73	0.8	86	0.9	63	0.7	70	0.7	44	0.5
Exomphalos	19	0.2	28	0.3	15	0.2	22	0.2	25	0.3	22	0.2	30	0.3
Gastroschisis	18	0.2	16	0.2	19	0.2	21	0.2	30	0.3	19	0.2	32	0.3
Diaphragmatic hernia	21	0.2	30	0.4	28	0.3	25	0.3	28	0.3	32	0.3	30	0.3

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.

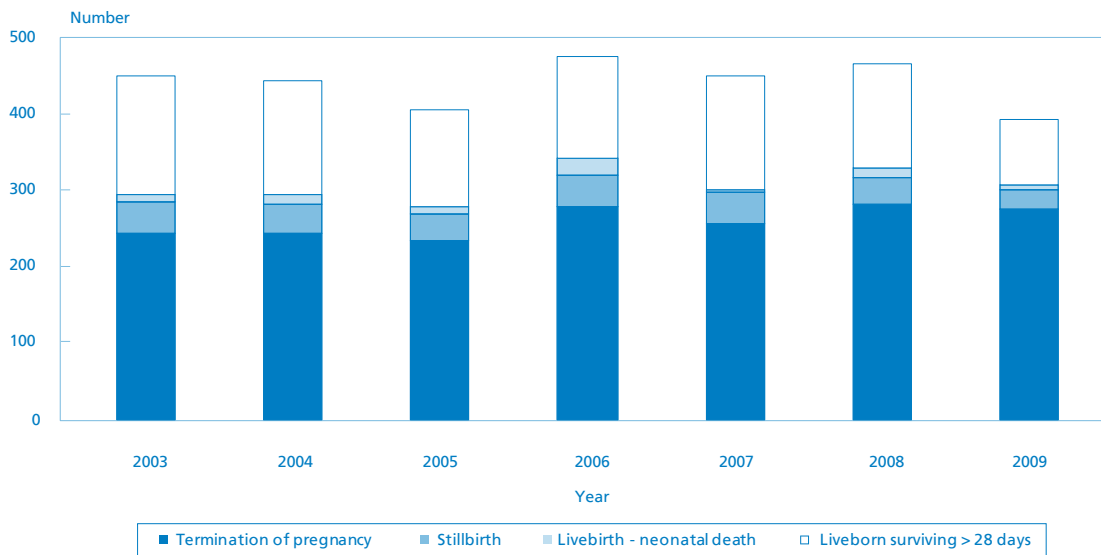
For 2003–2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are included.

Figure 5. Neural tube anomalies: cases by year and pregnancy outcome, NSW 2003–2009#



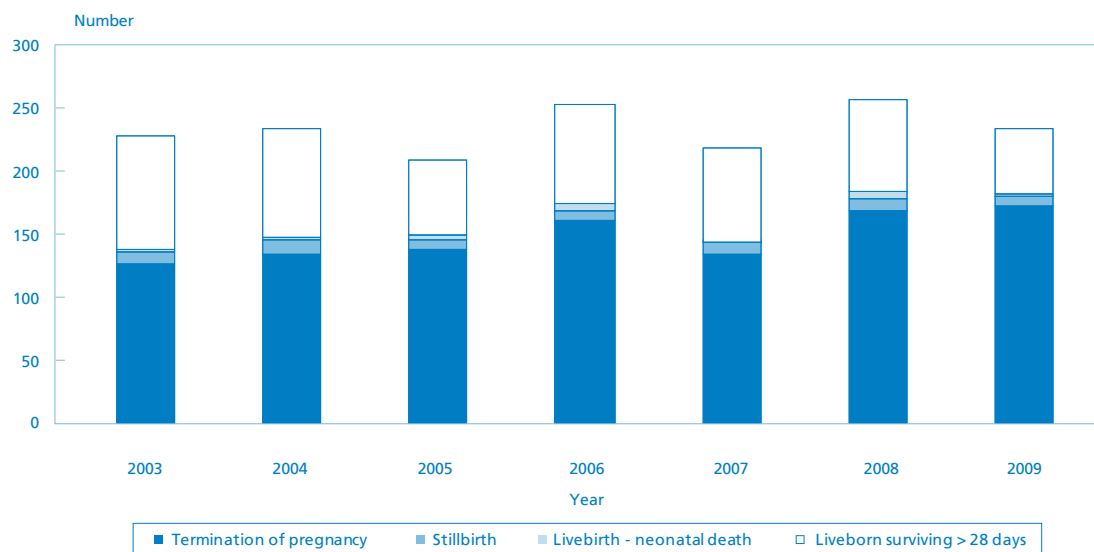
Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.
 # For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.

Figure 6. Chromosomal anomalies: cases by year and pregnancy outcome, NSW 2003-2009#



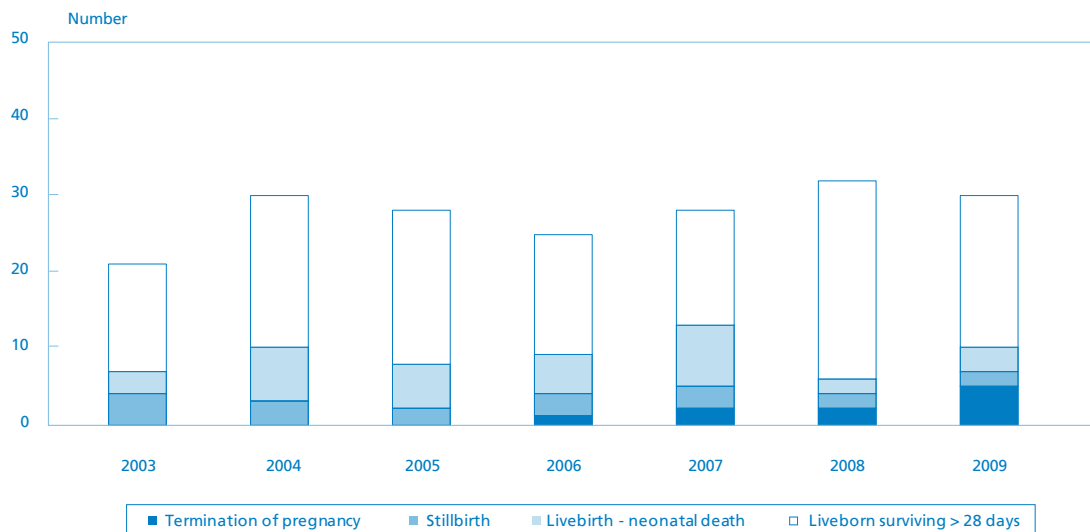
Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.
 # For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are reported.

Figure 7. Down syndrome: cases by year and pregnancy outcome, NSW 2003-2009#



Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.
 # For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are reported.

Figure 8. Diaphragmatic hernia: cases by year and pregnancy outcome, NSW 2003-2009#



Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Ministry of Health.
 # For 2003-2008, cases reported during pregnancy and up to one year of age are included. For 2009, cases reported during pregnancy or at birth are reported.