

# 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 2002-2007 and congenital conditions detected during pregnancy or at birth for 2008.

## Trends in reported congenital conditions

Between 2002 and 2004, about 2% of infants were reported to have congenital conditions, falling to 1.8% to 1.9% for the 3-year period 2005 to 2007 (Table 103). In 2008, 831 cases of congenital conditions detected during pregnancy or at birth have been reported to date.

Table 103. Cases of congenital conditions, NSW 2002–2008#

| Year | Cases of congenital conditions | Births | Rate/1000 births |
|------|--------------------------------|--------|------------------|
|      | No.                            | No.    |                  |
| 2002 | 1736                           | 85398  | 20.3             |
| 2003 | 1756                           | 85853  | 20.5             |
| 2004 | 1751                           | 85016  | 20.6             |
| 2005 | 1617                           | 89840  | 18.0             |
| 2006 | 1768                           | 92038  | 19.2             |
| 2007 | 1828                           | 94912  | 19.3             |
| 2008 | 831                            | 95171  | 8.7              |

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research. NSW Department of Health.  
## For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are included.

## Congenital conditions by diagnostic category

The most common categories of congenital conditions for stillborn and liveborn 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 2007, the overall rate of conditions was similar to the previous 5 years (33.8 versus 34.5 per 1,000 respectively).

**Table 104. Congenital conditions among stillbirths and liveborn infants by diagnostic category, NSW 2002–2008#**

| Diagnostic category                    | No. defects |      |      |           | Rate/1,000 births |      |      |           |
|--|-------------|------|------|-----------|-------------------|------|------|-----------|
|  | 2002–2006   | 2007 | 2008 | 2002–2008 | 2002–2006         | 2007 | 2008 | 2002–2008 |
| <b>Nervous system</b>                  |             |      |      |           |                   |      |      |           |
| Anencephaly                            | 51          | 14   | 7    | 72        | 0.1               | 0.1  | 0.1  | 0.1       |
| Spina Bifida                           | 126         | 36   | 21   | 183       | 0.3               | 0.4  | 0.2  | 0.3       |
| Encephalocele                          | 30          | 6    | 2    | 38        | 0.1               | 0.1  | 0.0  | 0.1       |
| Microcephaly                           | 109         | 19   | 8    | 136       | 0.2               | 0.2  | 0.1  | 0.2       |
| Congenital hydrocephalus               | 163         | 40   | 21   | 224       | 0.4               | 0.4  | 0.2  | 0.4       |
| Other nervous system anomalies         | 256         | 78   | 35   | 369       | 0.6               | 0.8  | 0.4  | 0.6       |
| TOTAL                                  | 735         | 193  | 94   | 1022      | 1.7               | 2.0  | 1.0  | 1.6       |
| <b>Eye</b>                             |             |      |      |           |                   |      |      |           |
| Anophthalmos-microphthalmos            | 51          | 10   | 3    | 64        | 0.1               | 0.1  | 0.0  | 0.1       |
| Buphthalmos-congenital glaucoma        | 29          | 6    | 2    | 37        | 0.1               | 0.1  | 0.0  | 0.1       |
| Congenital cataract                    | 83          | 17   | 1    | 101       | 0.2               | 0.2  | 0.0  | 0.2       |
| Other eye anomalies                    | 174         | 40   | 13   | 227       | 0.4               | 0.4  | 0.1  | 0.4       |
| TOTAL                                  | 337         | 73   | 19   | 429       | 0.8               | 0.8  | 0.2  | 0.7       |
| <b>Ear, face and neck</b>              |             |      |      |           |                   |      |      |           |
| Absence-stricture auditory canal       | 55          | 11   | 8    | 74        | 0.1               | 0.1  | 0.1  | 0.1       |
| Absent auricle                         | 5           | 0    | 1    | 6         | 0.0               | 0    | 0.0  | 0.0       |
| Anomalies of face and neck             | 35          | 13   | 3    | 51        | 0.1               | 0.1  | 0.0  | 0.1       |
| Other ear anomalies                    | 79          | 14   | 9    | 102       | 0.2               | 0.1  | 0.1  | 0.2       |
| TOTAL                                  | 174         | 38   | 21   | 233       | 0.4               | 0.4  | 0.2  | 0.4       |
| <b>Cardiovascular system</b>           |             |      |      |           |                   |      |      |           |
| Transposition of great vessels         | 235         | 51   | 30   | 316       | 0.5               | 0.5  | 0.3  | 0.5       |
| Tetralogy of Fallot                    | 168         | 30   | 14   | 212       | 0.4               | 0.3  | 0.1  | 0.3       |
| Ventricular septal defect              | 810         | 171  | 84   | 1065      | 1.8               | 1.8  | 0.9  | 1.7       |
| Atrial septal defect                   | 818         | 159  | 59   | 1036      | 1.9               | 1.7  | 0.6  | 1.6       |
| Heart valve anomalies                  | 507         | 87   | 72   | 666       | 1.2               | 0.9  | 0.8  | 1.1       |
| Patent ductus arteriosus > 37 weeks    | 444         | 91   | 45   | 580       | 1.0               | 1.0  | 0.5  | 0.9       |
| Coarctation of aorta                   | 209         | 40   | 22   | 271       | 0.5               | 0.4  | 0.2  | 0.4       |
| Other anomalies of aorta               | 127         | 19   | 16   | 162       | 0.3               | 0.2  | 0.2  | 0.3       |
| Anomalies of pulmonary artery          | 149         | 28   | 18   | 195       | 0.3               | 0.3  | 0.2  | 0.3       |
| Other cardiovascular anomalies         | 716         | 147  | 91   | 954       | 1.6               | 1.5  | 1.0  | 1.5       |
| TOTAL                                  | 4183        | 823  | 451  | 5457      | 9.5               | 8.7  | 4.7  | 8.7       |
| <b>Respiratory system</b>              |             |      |      |           |                   |      |      |           |
| Nose                                   | 64          | 11   | 5    | 80        | 0.1               | 0.1  | 0.1  | 0.1       |
| Larynx, trachea and bronchus           | 42          | 4    | 1    | 47        | 0.1               | 0.0  | 0.0  | 0.1       |
| Lung                                   | 85          | 27   | 15   | 127       | 0.2               | 0.3  | 0.2  | 0.2       |
| TOTAL                                  | 191         | 42   | 21   | 254       | 0.4               | 0.4  | 0.2  | 0.4       |
| <b>Gastrointestinal system</b>         |             |      |      |           |                   |      |      |           |
| Cleft palate only                      | 418         | 80   | 65   | 563       | 1.0               | 0.8  | 0.7  | 0.9       |
| Cleft lip only                         | 160         | 17   | 32   | 209       | 0.4               | 0.2  | 0.3  | 0.3       |
| Cleft palate and cleft lip             | 204         | 50   | 46   | 300       | 0.5               | 0.5  | 0.5  | 0.5       |
| Oesophageal atresia only               | 14          | 4    | 2    | 20        | 0.0               | 0.0  | 0.0  | 0.0       |
| Tracheo-oesophageal fistula (TOF) only | 20          | 5    | 3    | 28        | 0.0               | 0.1  | 0.0  | 0.0       |
| Oesophageal atresia with TOF           | 78          | 21   | 14   | 113       | 0.2               | 0.2  | 0.1  | 0.2       |

Table 104. (Continued)

| Diagnostic category                                | No. defects  |             |             |              | Rate/1,000 births |             |             |             |
|--|--------------|-------------|-------------|--------------|-------------------|-------------|-------------|-------------|
|  | 2002–2006    | 2007        | 2008        | 2002–2008    | 2002–2006         | 2007        | 2008        | 2002–2008   |
| Atresia-stenosis of small intestine                | 134          | 22          | 12          | 168          | 0.3               | 0.2         | 0.1         | 0.3         |
| Atresia-stenosis of anus                           | 129          | 27          | 23          | 179          | 0.3               | 0.3         | 0.2         | 0.3         |
| Other gastrointestinal anomalies                   | 474          | 99          | 32          | 605          | 1.1               | 1.0         | 0.3         | 1.0         |
| <b>TOTAL</b>                                       | <b>1631</b>  | <b>325</b>  | <b>229</b>  | <b>2185</b>  | <b>3.7</b>        | <b>3.4</b>  | <b>2.4</b>  | <b>3.5</b>  |
| <b>Genitourinary system</b>                        |              |             |             |              |                   |             |             |             |
| Anomalies of female genitals                       | 55           | 10          | 7           | 72           | 0.1               | 0.1         | 0.1         | 0.1         |
| Undescended testis                                 | 274          | 96          | 16          | 386          | 0.6               | 1.0         | 0.2         | 0.6         |
| Hypospadias  | 748          | 161         | 68          | 977          | 1.7               | 1.7         | 0.7         | 1.6         |
| Epispadias   | 7            | 0           | 3           | 10           | 0.0               | 0.0         | 0.0         | 0.0         |
| Chordee  | 144          | 34          | 14          | 192          | 0.3               | 0.4         | 0.1         | 0.3         |
| Indeterminate sex-ambiguous genitalia              | 54           | 10          | 4           | 68           | 0.1               | 0.1         | 0.0         | 0.1         |
| Renal agenesis-dysgenesis                          | 175          | 33          | 25          | 233          | 0.4               | 0.3         | 0.3         | 0.4         |
| Obstructive anomalies of renal pelvis and ureter   | 661          | 155         | 39          | 855          | 1.5               | 1.6         | 0.4         | 1.4         |
| Other genitourinary system anomalies               | 641          | 144         | 60          | 845          | 1.5               | 1.5         | 0.6         | 1.3         |
| <b>TOTAL</b>                                       | <b>2759</b>  | <b>643</b>  | <b>236</b>  | <b>3638</b>  | <b>6.3</b>        | <b>6.8</b>  | <b>2.5</b>  | <b>5.8</b>  |
| <b>Musculoskeletal system</b>                      |              |             |             |              |                   |             |             |             |
| Congenital dislocation of the hips                 | 659          | 116         | 31          | 806          | 1.5               | 1.2         | 0.3         | 1.3         |
| Talipes equinovarus                                | 375          | 69          | 35          | 479          | 0.9               | 0.7         | 0.4         | 0.8         |
| Polydactyly  | 497          | 89          | 77          | 663          | 1.1               | 0.9         | 0.8         | 1.1         |
| Syndactyly   | 108          | 36          | 18          | 162          | 0.2               | 0.4         | 0.2         | 0.3         |
| Reduction deformities of limbs                     | 214          | 63          | 48          | 325          | 0.5               | 0.7         | 0.5         | 0.5         |
| Craniosynostosis                                   | 218          | 38          | 5           | 261          | 0.5               | 0.4         | 0.1         | 0.4         |
| Diaphragmatic hernia                               | 125          | 26          | 22          | 173          | 0.3               | 0.3         | 0.2         | 0.3         |
| Exomphalos   | 63           | 17          | 14          | 94           | 0.1               | 0.2         | 0.1         | 0.1         |
| Gastroschisis                                      | 85           | 30          | 17          | 132          | 0.2               | 0.3         | 0.2         | 0.2         |
| Other musculoskeletal anomalies                    | 702          | 154         | 104         | 960          | 1.6               | 1.6         | 1.1         | 1.5         |
| <b>TOTAL</b>                                       | <b>3046</b>  | <b>638</b>  | <b>371</b>  | <b>4055</b>  | <b>7.0</b>        | <b>6.7</b>  | <b>3.9</b>  | <b>6.5</b>  |
| <b>Integumentary system</b>                        |              |             |             |              |                   |             |             |             |
| Cystic hygroma                                     | 46           | 11          | 11          | 68           | 0.1               | 0.1         | 0.1         | 0.1         |
| <b>Chromosomal anomalies</b>                       |              |             |             |              |                   |             |             |             |
| Trisomy 21   | 468          | 85          | 47          | 600          | 1.1               | 0.9         | 0.5         | 1.0         |
| Trisomy 13   | 35           | 6           | 4           | 45           | 0.1               | 0.1         | 0.0         | 0.1         |
| Trisomy 18   | 94           | 18          | 11          | 123          | 0.2               | 0.2         | 0.1         | 0.2         |
| Turner syndrome                                    | 72           | 17          | 10          | 99           | 0.2               | 0.2         | 0.1         | 0.2         |
| Other chromosomal defects                          | 321          | 69          | 34          | 424          | 0.7               | 0.7         | 0.4         | 0.7         |
| <b>TOTAL</b>                                       | <b>990</b>   | <b>195</b>  | <b>106</b>  | <b>1291</b>  | <b>2.3</b>        | <b>2.1</b>  | <b>1.1</b>  | <b>2.1</b>  |
| <b>Situs inversus</b>                              | <b>26</b>    | <b>6</b>    | <b>4</b>    | <b>36</b>    | <b>0.1</b>        | <b>0.1</b>  | <b>0.0</b>  | <b>0.1</b>  |
| <b>Congenital malformation syndromes</b>           | <b>193</b>   | <b>40</b>   | <b>14</b>   | <b>247</b>   | <b>0.4</b>        | <b>0.4</b>  | <b>0.1</b>  | <b>0.4</b>  |
| <b>Congenital cytomegalovirus infection</b>        | <b>1</b>     | <b>0</b>    | <b>0</b>    | <b>1</b>     | <b>0.0</b>        | <b>0.0</b>  | <b>0.0</b>  | <b>0.0</b>  |
| <b>Non-immune hydrops foetalis</b>                 | <b>98</b>    | <b>20</b>   | <b>16</b>   | <b>134</b>   | <b>0.2</b>        | <b>0.2</b>  | <b>0.2</b>  | <b>0.2</b>  |
| <b>Other and unspecified congenital conditions</b> | <b>453</b>   | <b>112</b>  | <b>24</b>   | <b>589</b>   | <b>1.0</b>        | <b>1.2</b>  | <b>0.3</b>  | <b>0.9</b>  |
| <b>TOTAL</b>                                       | <b>15110</b> | <b>3211</b> | <b>1637</b> | <b>19958</b> | <b>34.5</b>       | <b>33.8</b> | <b>17.2</b> | <b>31.8</b> |

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

# For 2002–2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.

## Infant characteristics

In the period 2002-2008, a single condition was reported in 64.0% of infants, two conditions in 18.2%, three conditions in 8.2%, and four or more conditions in 9.5% 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 2002-2008, 1.8% of singleton babies, 2.6% of twins, and 3.2% 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 Midwives Data Collection was less than 1% in 2008 (see Chapter 4).

**Table 105. Cases of congenital conditions by gestational age, NSW 2002–2008#**

| Gestational age (weeks) | 2002–2006 |       | 2007 |       | Year 2008 |       | 2000–2008 |       | No. of cases/1,000 births |
|-------------------------|-----------|-------|------|-------|-----------|-------|-----------|-------|---------------------------|
|                         | No.       | %     | No.  | %     | No.       | %     | No.       | %     |                           |
| 20 – 27                 | 593       | 6.9   | 112  | 6.1   | 90        | 10.8  | 795       | 7.0   | 178.9                     |
| 28 – 31                 | 268       | 3.1   | 44   | 2.4   | 19        | 2.3   | 331       | 2.9   | 72.8                      |
| 32 – 36                 | 1081      | 12.5  | 268  | 14.7  | 120       | 14.4  | 1469      | 13.0  | 40.1                      |
| 37 – 41                 | 6292      | 72.9  | 1327 | 72.6  | 599       | 72.1  | 8218      | 72.8  | 14.4                      |
| 42 +                    | 163       | 1.9   | 15   | 0.8   | 3         | 0.4   | 181       | 1.6   | 16.9                      |
| Not stated              | 231       | 2.7   | 62   | 3.4   | 0         | 0     | 293       | 2.6   | -                         |
| TOTAL                   | 8628      | 100.0 | 1828 | 100.0 | 831       | 100.0 | 11287     | 100.0 | 18.0                      |

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

# For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are included.

**Table 106. Cases of congenital conditions by pregnancy outcome, NSW 2002–2008#**

| Pregnancy outcome            | 2002–2006 |       | 2007 |       | Year 2008 |       | 2002–2008 |       |
|------------------------------|-----------|-------|------|-------|-----------|-------|-----------|-------|
|                              | No.       | %     | No.  | %     | No.       | %     | No.       | %     |
| Stillbirth                   | 589       | 6.8   | 109  | 6.0   | 87        | 10.5  | 785       | 7.0   |
| Liveborn/ neonatal death     | 327       | 3.8   | 67   | 3.7   | 47        | 5.7   | 441       | 3.9   |
| Liveborn/ postneonatal death | 57        | 0.7   | 5    | 0.3   | 8         | 1.0   | 70        | 0.6   |
| Liveborn surviving           | 7655      | 88.7  | 1647 | 90.1  | 689       | 82.9  | 9991      | 88.5  |
| TOTAL                        | 8628      | 100.0 | 1828 | 100.0 | 831       | 100.0 | 11287     | 100.0 |

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

# For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.

## 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 2002–2008, 73.1% of babies with congenital conditions were born to women aged less than 35 years.

In 2002–2008, 377 babies of Aboriginal or Torres Strait Islander mothers were reported to have congenital conditions. The rate of congenital conditions among these babies was 21.4 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 2002–2008<sup>#</sup>**

| Maternal age (years) | 2002–2006 |       | 2007 |       | Year 2008 |       | 2002–2008 |       | No. of cases/1,000 births |
|----------------------|-----------|-------|------|-------|-----------|-------|-----------|-------|---------------------------|
|                      | No.       | %     | No.  | %     | No.       | %     | No.       | %     |                           |
| Under 20             | 347       | 4.0   | 71   | 3.9   | 37        | 4.5   | 455       | 4.0   | 18.9                      |
| 20 – 24              | 1225      | 14.2  | 220  | 12.0  | 121       | 14.6  | 1566      | 13.9  | 17.6                      |
| 25 – 29              | 2144      | 24.8  | 465  | 25.4  | 179       | 21.5  | 2788      | 24.7  | 16.2                      |
| 30 – 34              | 2669      | 30.9  | 522  | 28.6  | 246       | 29.6  | 3437      | 30.5  | 16.3                      |
| 35 – 39              | 1408      | 16.3  | 338  | 18.5  | 189       | 22.7  | 1935      | 17.1  | 17.5                      |
| 40 – 44              | 365       | 4.2   | 78   | 4.3   | 56        | 6.7   | 499       | 4.4   | 23.2                      |
| 45+                  | 32        | 0.4   | 6    | 0.3   | 3         | 0.4   | 41        | 0.4   | 38.9                      |
| Not stated           | 438       | 5.1   | 128  | 7.0   | 0         | 0.0   | 566       | 5.0   | -                         |
| TOTAL                | 8628      | 100.0 | 1828 | 100.0 | 831       | 100.0 | 11287     | 100.0 | 18.0                      |

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research. NSW Department of Health.  
<sup>#</sup> For 2002–2007, cases reported during pregnancy and up to one year of age are included. For 2008, 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 285 in 2002 to 324 in 2007 (Table 108). To date, 281 terminations of pregnancy have been reported to the Register for 2008. This number is expected to increase as pregnancy outcomes for babies with conditions detected during pregnancy in 2008 continue to be reported.

Of the 2,173 terminations of pregnancy reported in 2002–2008, 1,729 (79.6%) were associated with a chromosomal

anomaly, the most common of which was Trisomy 21 (Down syndrome), and 156 (7.2%) 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 abortion, termination of pregnancy or unknown outcome, NSW 2002–2008**

| Pregnancy outcome                                     | Year |      |      |      |      |      |      |           |
|---|------|------|------|------|------|------|------|-----------|
|   | 2002 | 2003 | 2004 | 2005 | 2006 | 2007 | 2008 | 2002–2008 |
|   | No.  | No.  | No.  | No.  | No.  | No.  | No.  | No.       |
| Spontaneous abortion                                  | 202  | 233  | 298  | 322  | 363  | 334  | 322  | 2074      |
| Termination of pregnancy less than 20 weeks gestation | 285  | 313  | 306  | 303  | 361  | 324  | 281  | 2173      |
| Unknown outcome                                       | 7    | 17   | 38   | 29   | 23   | 28   | 0    | 142       |
| TOTAL   | 494  | 563  | 642  | 654  | 747  | 686  | 603  | 4389      |

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

Table 109. Congenital conditions among spontaneous abortions, terminations of pregnancy and unknown outcomes of pregnancy by diagnostic category, NSW 2002–2008

| Diagnostic category                                | Year            |   |         |     |                 |   |         |     |                 |   |         |     |   |         |
|--|-----------------|---|---------|-----|-----------------|---|---------|-----|-----------------|---|---------|-----|---|---------|
|  | 2002–2006       |   |         |     | 2007            |   |         |     | 2008            |   |         |     | 2002–2008   |         |
|  | 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. | Termination of pregnancy less than 20 weeks gestation | Unknown |
| <b>Nervous system</b>                              |                 |   |         |     |                 |   |         |     |                 |   |         |     |   |         |
| Neural tube defects                                | 5               | 121   | 1       | 1   | 19              | 1   | 1       | 0   | 16              | 6   | 156     | 2   |   |         |
| Other nervous system defects                       | 5               | 113   | 0       | 2   | 14              | 0   | 0       | 0   | 13              | 7   | 140     | 0   |   |         |
| TOTAL  | 10              | 234   | 1       | 3   | 33              | 1   | 1       | 0   | 29              | 13  | 296     | 2   |   |         |
| Eye  | 0               | 3   | 0       | 0   | 0               | 0   | 0       | 0   | 0               | 0   | 3       | 0   |   |         |
| Ear, face and neck                                 | 1               | 10  | 0       | 0   | 0               | 0   | 0       | 0   | 4               | 1   | 14      | 0   |   |         |
| Cardiovascular system                              | 16              | 235   | 5       | 3   | 35              | 1   | 1       | 5   | 53              | 24  | 323     | 6   |   |         |
| Respiratory system                                 | 7               | 21  | 1       | 0   | 10              | 0   | 0       | 1   | 5               | 8   | 36      | 1   |   |         |
| Gastrointestinal system                            | 12              | 121   | 0       | 1   | 33              | 0   | 0       | 6   | 22              | 19  | 176     | 0   |   |         |
| Musculoskeletal system                             | 19              | 282   | 5       | 7   | 77              | 1   | 1       | 15  | 49              | 41  | 408     | 6   |   |         |
| Genitourinary system                               | 8               | 144   | 1       | 1   | 38              | 0   | 0       | 5   | 30              | 14  | 212     | 1   |   |         |
| Integumentary system                               | 1               | 1   | 0       | 0   | 0               | 0   | 0       | 0   | 0               | 1   | 1       | 0   |   |         |
| Cystic hygroma                                     | 20              | 125   | 7       | 5   | 28              | 2   | 2       | 7   | 21              | 32  | 174     | 9   |   |         |
| <b>Chromosomal anomalies</b>                       |                 |   |         |     |                 |   |         |     |                 |   |         |     |   |         |
| Trisomy 21   | 116             | 677   | 53      | 22  | 131             | 14  | 14      | 32  | 130             | 170   | 938     | 67  |   |         |
| Trisomy 13   | 54              | 86  | 6       | 16  | 16              | 2   | 2       | 16  | 19              | 86  | 121     | 8   |   |         |
| Trisomy 18   | 60              | 199   | 13      | 20  | 48              | 4   | 4       | 16  | 32              | 96  | 279     | 17  |   |         |
| Turner syndrome                                    | 166             | 99  | 7       | 30  | 22              | 0   | 0       | 29  | 17              | 225   | 138     | 7   |   |         |
| Other chromosomal anomalies                        | 1004            | 183   | 26      | 241 | 41              | 5   | 5       | 221 | 29              | 1466  | 253     | 31  |   |         |
| TOTAL  | 1400            | 1244  | 105     | 329 | 258             | 25  | 25      | 314 | 227             | 2043  | 1729    | 130 |   |         |
| <b>Situs inversus</b>                              | 0               | 3   | 0       | 0   | 1               | 0   | 0       | 0   | 0               | 0   | 4       | 0   |   |         |
| <b>Congenital malformation syndromes</b>           | 0               | 13  | 0       | 0   | 4               | 0   | 0       | 2   | 2               | 2   | 19      | 0   |   |         |
| <b>Non-immune hydrops foetalis</b>                 | 12              | 52  | 1       | 6   | 6               | 1   | 1       | 3   | 7               | 21  | 65      | 2   |   |         |
| <b>Other and unspecified congenital conditions</b> | 4               | 31  | 2       | 0   | 8               | 0   | 0       | 0   | 10              | 4   | 49      | 2   |   |         |
| TOTAL  | 1510            | 2519  | 128     | 355 | 531             | 31  | 31      | 358 | 459             | 2223  | 3509    | 159 |   |         |

NSW Register of Congenital Conditions (HOIST), Centre for Epidemiology and Research, NSW Department 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 2002–2007, malformations reported up to one year of age are included. For 2008, malformations reported during pregnancy or at birth are included.

Over the period 2002 to 2007, 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 43 in 2002 and 55 in 2007, and 30 cases have been reported for 2008 to date. The number of reported terminations of pregnancy was 18 in both 2002 and 2007, and 16 have been reported to date for 2008 (Figure 5).

Over the period 2002–2008, the number of cases of isolated cleft palate ranged from 60 to 90 per year, and for total cleft lip (including cases of cleft lip and cleft palate) from 66 to 85 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 204 in 2002 and 195 in 2007. The number of reported terminations of pregnancy associated with chromosomal conditions rose from 236 in 2002 to 281 in 2006 and then fell to 256 in 2007. The number of infants born with Down syndrome was 103 in 2002 and 85 in 2007, while the number of reported terminations of pregnancy associated with Down syndrome rose from 118 in 2002 to 160 in 2006 and then fell to 131 in 2007.

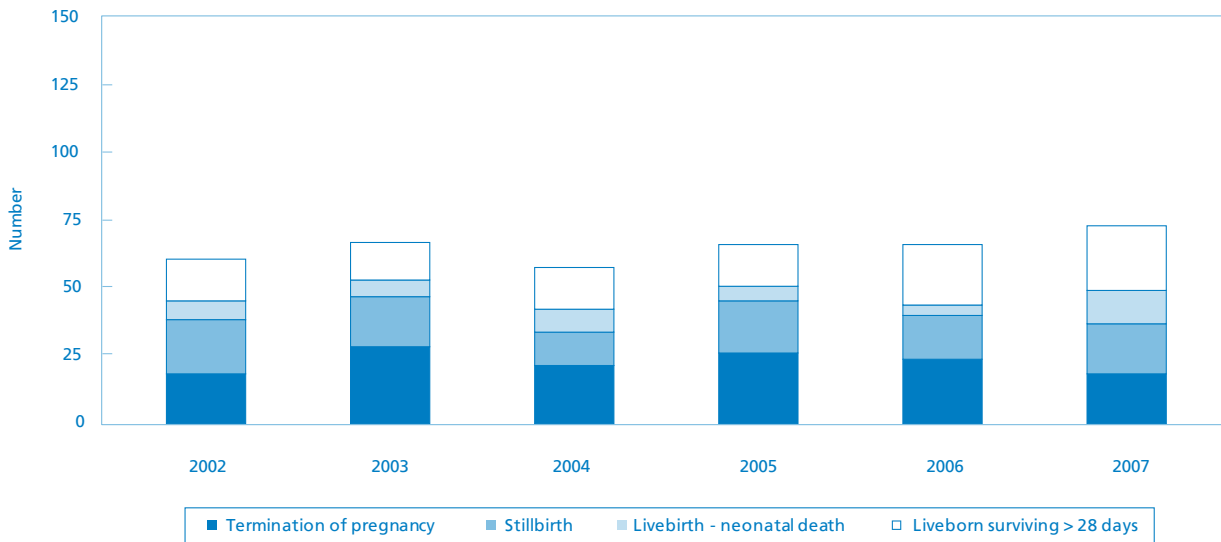
In 2002, 19 liveborn infants were reported to have a diaphragmatic hernia, 3 babies were stillborn and there was 1 termination of pregnancy associated with this condition. In 2008, there were 21 liveborn infants reported with a diaphragmatic hernia, 1 stillborn baby and terminations of pregnancy (Figure 8).

**Table 110. Selected congenital conditions by year, NSW 2002–2008#**

| Condition                     | 2002 |               | 2003 |               | 2004 |               | 2005 |               | 2006 |               | 2007 |               | 2008 |               |
|-------------------------------|------|---------------|------|---------------|------|---------------|------|---------------|------|---------------|------|---------------|------|---------------|
|                               | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 | No.  | Rate/<br>1000 |
| Neural tube defects           | 61   | 0.7           | 67   | 0.8           | 58   | 0.7           | 66   | 0.7           | 66   | 0.7           | 73   | 0.8           | 46   | 0.5           |
| Anencephalus                  | 20   | 0.2           | 18   | 0.2           | 16   | 0.2           | 19   | 0.2           | 15   | 0.2           | 25   | 0.3           | 16   | 0.2           |
| Spina bifida                  | 32   | 0.4           | 46   | 0.5           | 35   | 0.4           | 40   | 0.4           | 45   | 0.5           | 42   | 0.4           | 27   | 0.3           |
| Encephalocele                 | 10   | 0.1           | 7    | 0.1           | 8    | 0.1           | 8    | 0.1           | 8    | 0.1           | 8    | 0.1           | 3    | 0.0           |
| Cleft palate                  | 60   | 0.7           | 88   | 1.0           | 74   | 0.9           | 90   | 1.0           | 64   | 0.7           | 74   | 0.8           | 63   | 0.7           |
| Total cleft lip               | 76   | 0.9           | 83   | 1.0           | 79   | 0.9           | 66   | 0.7           | 85   | 0.9           | 73   | 0.8           | 82   | 0.9           |
| Hypospadias                   | 133  | 1.6           | 171  | 2.0           | 150  | 1.8           | 146  | 1.6           | 149  | 1.6           | 162  | 1.7           | 68   | 0.7           |
| Limb reduction defects        | 21   | 0.2           | 31   | 0.4           | 36   | 0.4           | 38   | 0.4           | 43   | 0.5           | 49   | 0.5           | 43   | 0.5           |
| Chromosomal abnormalities     | 440  | 5.2           | 454  | 5.3           | 447  | 5.3           | 409  | 4.6           | 480  | 5.2           | 451  | 4.8           | 333  | 3.5           |
| Down syndrome                 | 221  | 2.6           | 228  | 2.7           | 233  | 2.7           | 209  | 2.3           | 252  | 2.7           | 216  | 2.3           | 177  | 1.9           |
| Renal agenesis and dysgenesis | 63   | 0.7           | 67   | 0.8           | 82   | 1.0           | 73   | 0.8           | 86   | 0.9           | 63   | 0.7           | 45   | 0.5           |
| Exomphalos                    | 22   | 0.3           | 19   | 0.2           | 28   | 0.3           | 15   | 0.2           | 22   | 0.2           | 25   | 0.3           | 20   | 0.2           |
| Gastroschisis                 | 18   | 0.2           | 18   | 0.2           | 16   | 0.2           | 19   | 0.2           | 21   | 0.2           | 30   | 0.3           | 17   | 0.2           |
| Diaphragmatic hernia          | 23   | 0.3           | 21   | 0.2           | 30   | 0.4           | 28   | 0.3           | 25   | 0.3           | 28   | 0.3           | 24   | 0.3           |

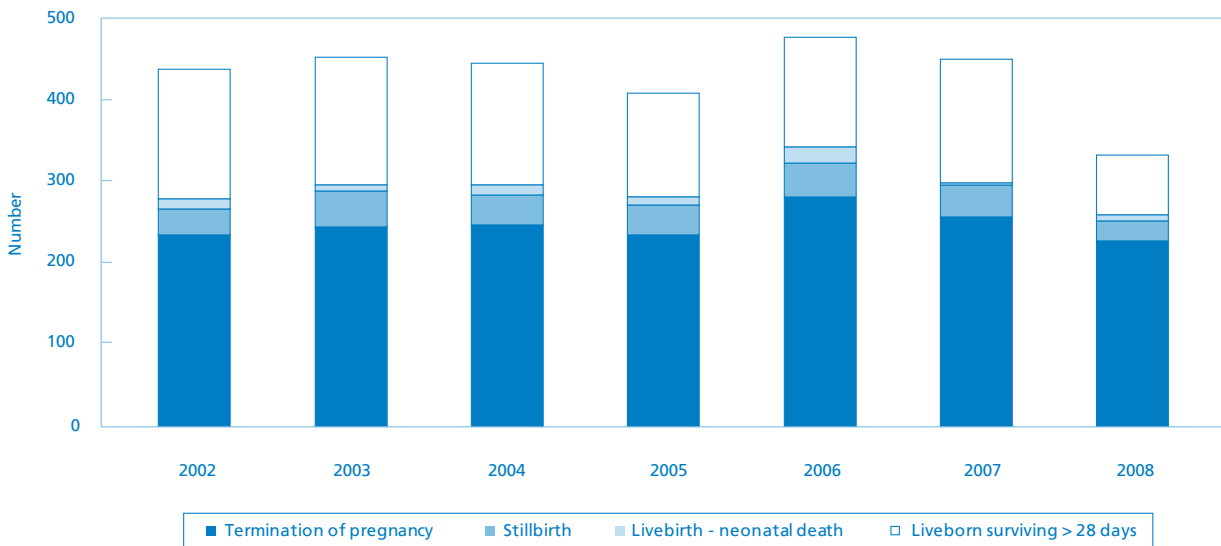
Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research. NSW Department of Health.

# For 2002–2007, 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 5. Neural tube anomalies: cases by year and pregnancy outcome, NSW 2002–2008#**

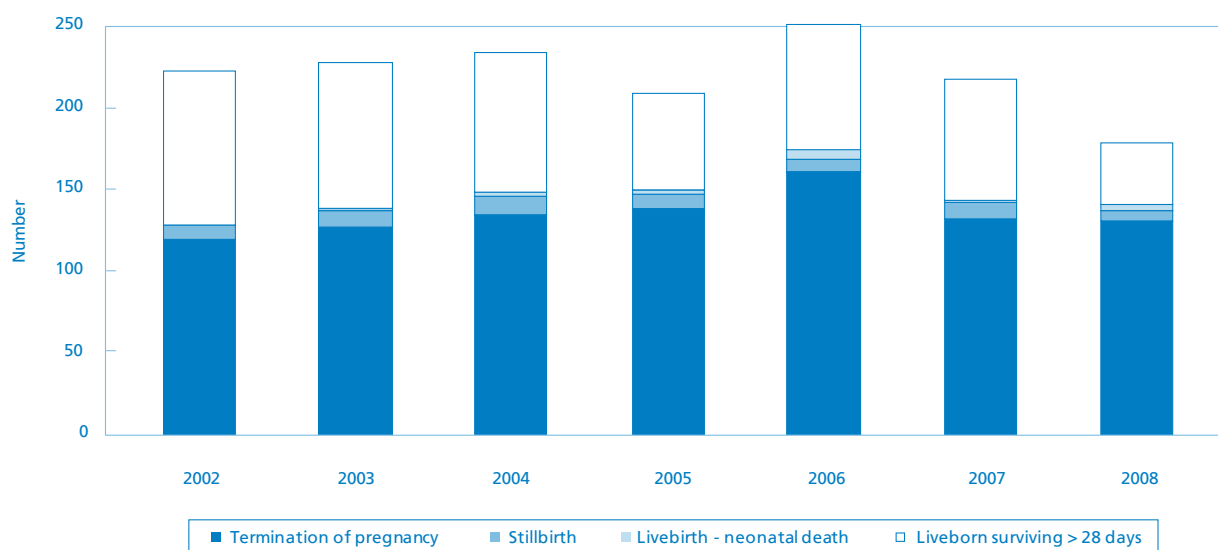
Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Department of Health.  
 # For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.



**Figure 6. Chromosomal anomalies: cases by year and pregnancy outcome, NSW 2002–2008#**

Source: NSW Register of Congenital Conditions (HOIST). Centre for Epidemiology and Research, NSW Department of Health.  
 # For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.

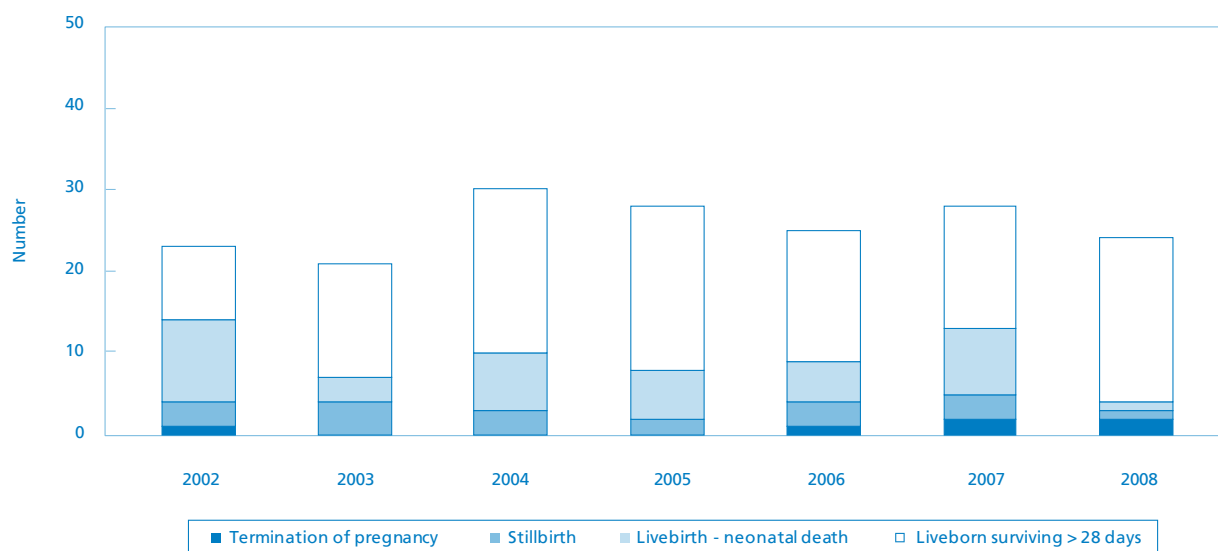




**Figure 7. Down Syndrome: cases by year and pregnancy outcome, NSW 2002–2008#**

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

# For 2002-2007, 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 8. Diaphragmatic Hernia: cases by year and pregnancy outcome, NSW 2002–2008#**

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

# For 2002-2007, cases reported during pregnancy and up to one year of age are included. For 2008, cases reported during pregnancy or at birth are reported.

## Congenital conditions in NSW health areas

Crude rates of reported congenital conditions for NSW health areas and rates standardised for maternal age are shown in Table 111. The denominator population includes live births and stillbirths among NSW residents as reported to the MDC. The rate of congenital conditions increases with increasing maternal age (Table 107). In order to allow direct comparison of geographic areas, rates have been standardised to the maternal age distribution of births in NSW in 1991.

In interpreting congenital condition rates among NSW areas, it should also be noted that infants with congenital conditions 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 Register.

Over the period 2002–2008, standardised rates of reported congenital conditions were lowest in the Greater Southern Area and highest in the Hunter and New England Area. Review of cases showed slightly increased reported rates of a wide range of congenital conditions in the Hunter and New England Area compared to NSW overall, including: unstable hips (but not dislocated hips), talipes equinovarus, first degree hypospadias, undescended testis, obstructive conditions of the renal pelvis and ureter, spina bifida, ventricular septal defect, atrial septal defect, patent ductus arteriosus, coarctation of the aorta and chromosomal anomalies. There was no clustering by small geographic area. The range and pattern of these conditions suggests that reporting of congenital abnormalities is better in the Hunter and New England Health Area compared with NSW as a whole.

**Table 111. Cases of congenital conditions by health area, 2002–2008\***

| Health area                      | Year         |                          |                                 |              |                          |                                 |              |                          |                                 |              |                          |                                 | 99% confidence intervals |
|----------------------------------|--------------|--------------------------|---------------------------------|--------------|--------------------------|---------------------------------|--------------|--------------------------|---------------------------------|--------------|--------------------------|---------------------------------|--------------------------|
|                                  | 2002–2006    |                          |                                 | 2007         |                          |                                 | 2008         |                          |                                 | 2002–2008    |                          |                                 |                          |
|                                  | No. of cases | Crude rate / 1000 births | Stand-ardised rate/ 1000 births | No. of cases | Crude rate / 1000 births | Stand-ardised rate/ 1000 births | No. of cases | Crude rate / 1000 births | Stand-ardised rate/ 1000 births | No. of cases | Crude rate / 1000 births | Stand-ardised rate/ 1000 births |                          |
| Sydney South West                | 2171         | 21.9                     | 20.2                            | 434          | 20.2                     | 18.2                            | 247          | 11.4                     | 9.3                             | 2852         | 20.1                     | 18.3                            | 17.3-19.3                |
| South Eastern Sydney & Illawarra | 1764         | 24.3                     | 21.5                            | 348          | 21.9                     | 19.3                            | 187          | 11.8                     | 9.6                             | 2299         | 22.0                     | 19.4                            | 18.1-20.7                |
| Sydney West                      | 1731         | 20.8                     | 19.6                            | 412          | 22.5                     | 21.3                            | 217          | 11.7                     | 10.5                            | 2360         | 19.7                     | 18.5                            | 17.5-19.6                |
| Northern Sydney & Central Coast  | 1565         | 23.0                     | 19.1                            | 335          | 22.8                     | 18.0                            | 165          | 11.3                     | 8.0                             | 2065         | 21.2                     | 17.3                            | 16.0-18.8                |
| Hunter & New England             | 1428         | 27.9                     | 26.1                            | 289          | 26.2                     | 24.6                            | 134          | 12.3                     | 10.2                            | 1851         | 25.3                     | 23.5                            | 22.0-25.0                |
| North Coast                      | 423          | 17.4                     | 16.2                            | 88           | 16.7                     | 14.6                            | 62           | 11.5                     | 10.8                            | 573          | 16.4                     | 15.2                            | 13.5-17.0                |
| Greater Southern                 | 352          | 17.7                     | 15.6                            | 59           | 14.4                     | 11.7                            | 39           | 9.5                      | 9.2                             | 450          | 16.0                     | 14.1                            | 12.3-16.1                |
| Greater Western                  | 428          | 21.5                     | 20.7                            | 100          | 24.2                     | 23.0                            | 37           | 8.9                      | 8.1                             | 565          | 20.1                     | 19.2                            | 17.1-21.5                |
| TOTAL NSW                        | 9862         | 22.5                     | 20.5                            | 2065         | 21.8                     | 19.5                            | 1088         | 11.4                     | 9.5                             | 13015        | 20.7                     | 18.7                            | 18.3-19.2                |

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