10. BIRTH DEFECTS

Birth defects among stillborn and liveborn infants

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

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

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

Trends in reported birth defects

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

Birth defects by diagnostic category

The most common categories of birth defects for births of more than 20 weeks gestation or with a birth weight greater than 400 grams are presented in Table 115. Birth defects are classified using the British Paediatric Association (BPA) Classification of Diseases, which is primarily organised by body system. For infants with more than one defect, each defect is counted separately. The number of birth defects reported therefore exceeds the number of affected infants.

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

---

### TABLE 114
BIRTH DEFECT CASES, NSW 1998–2004*

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

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

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

### TABLE 115
BIRTH DEFECT CASES AMONG STILLBIRTHS AND LIVE BIRTHS BY DIAGNOSTIC CATEGORY, NSW 1998–2004

<table>
<thead>
<tr>
<th></th>
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<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>No. defects</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Defects of nervous system</td>
<td>49</td>
<td>11</td>
<td>6</td>
<td>66</td>
<td>0.1</td>
</tr>
<tr>
<td>Anencephaly</td>
<td>49</td>
<td>11</td>
<td>6</td>
<td>66</td>
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<tr>
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<td>25</td>
<td>21</td>
<td>186</td>
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<tr>
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<td>6</td>
<td>6</td>
<td>39</td>
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</tr>
<tr>
<td>Microcephaly</td>
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<td>11</td>
<td>150</td>
<td>0.3</td>
</tr>
<tr>
<td>Congenital hydrocephalus</td>
<td>174</td>
<td>38</td>
<td>33</td>
<td>245</td>
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<tr>
<td>Other nervous system defects</td>
<td>366</td>
<td>58</td>
<td>41</td>
<td>465</td>
<td>0.9</td>
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<tr>
<td>TOTAL</td>
<td>872</td>
<td>161</td>
<td>118</td>
<td>1151</td>
<td>2.0</td>
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<td>Defects of eye</td>
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<td>11</td>
<td>15</td>
<td>81</td>
<td>0.1</td>
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<td>Anophthalmos–microphthalmos</td>
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<td>1</td>
<td>33</td>
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<td>Buphthalmos–congenital glaucoma</td>
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<td>7</td>
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<td>Absence–stricture auditory canal</td>
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<td>1</td>
<td>10</td>
<td>0.0</td>
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<td>11</td>
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<td>35</td>
<td>16</td>
<td>199</td>
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<td>2004</td>
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<td>806</td>
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<td>148</td>
<td>114</td>
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<td>15</td>
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<td>10</td>
<td>71</td>
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<td>Renal agenesis–dysgenesis</td>
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<td>31</td>
<td>30</td>
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<td>117</td>
<td>67</td>
<td>819</td>
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<tr>
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<td>545</td>
<td>347</td>
<td>3974</td>
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<td>85</td>
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<td>36</td>
<td>326</td>
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<td>53</td>
<td>5</td>
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<td>21</td>
<td>25</td>
<td>173</td>
<td>0.3</td>
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<td>Exomphalos</td>
<td>72</td>
<td>14</td>
<td>14</td>
<td>100</td>
<td>0.2</td>
</tr>
<tr>
<td>TOTAL</td>
<td>3349</td>
<td>614</td>
<td>402</td>
<td>4365</td>
<td>7.8</td>
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<td>Defects of integumentary system</td>
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<td>6</td>
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<td>Trisomy 21</td>
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<td>103</td>
<td>79</td>
<td>695</td>
<td>1.2</td>
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<td>7</td>
<td>7</td>
<td>49</td>
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<tr>
<td>Trisomy 18</td>
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<td>19</td>
<td>13</td>
<td>123</td>
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<td>13</td>
<td>6</td>
<td>87</td>
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<td>66</td>
<td>34</td>
<td>378</td>
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</tr>
<tr>
<td>TOTAL</td>
<td>985</td>
<td>208</td>
<td>139</td>
<td>1332</td>
<td>2.3</td>
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<tr>
<td>Situs inversus</td>
<td>20</td>
<td>12</td>
<td>3</td>
<td>35</td>
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<tr>
<td>TOTAL</td>
<td>16398</td>
<td>3040</td>
<td>1943</td>
<td>21381</td>
<td>38.1</td>
</tr>
</tbody>
</table>

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

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

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

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

Birth defects were more common in preterm and post-term infants than infants born at term (Table 116). Birth defects were also more common in infants born of a multiple pregnancy than a singleton pregnancy: in 1998–2004, 2.0 per cent of singleton babies, 2.5 per cent of twins, and 4.2 per cent of triplets were born with a birth defect.

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

### TABLE 116

<table>
<thead>
<tr>
<th></th>
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</thead>
<tbody>
<tr>
<td>20–27</td>
<td>552</td>
<td>131</td>
<td>94</td>
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<td>28–31</td>
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<td>32–36</td>
<td>1080</td>
<td>197</td>
<td>134</td>
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<td>6723</td>
<td>1229</td>
<td>713</td>
<td>8665</td>
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<td>42+</td>
<td>204</td>
<td>29</td>
<td>18</td>
<td>251</td>
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<td>433</td>
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<td>9141</td>
<td>1761</td>
<td>997</td>
<td>11899</td>
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</table>

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

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

### TABLE 117

<table>
<thead>
<tr>
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<tbody>
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<td>Stillbirth</td>
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<tr>
<td>Liveborn–postneonatal death</td>
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<td>4</td>
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<td>8119</td>
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<td>11899</td>
</tr>
</tbody>
</table>

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

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

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

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

<table>
<thead>
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<tr>
<td></td>
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<td>%</td>
<td>No.</td>
<td>%</td>
</tr>
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<td>404</td>
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</table>

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

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

In the period 1998–2003, on average about 280 terminations of pregnancy per year were reported to the NSW Birth Defects Register (Table 119). To date, 172 terminations of pregnancy have been reported to the Register for 2004. This number is expected to increase as outcomes for mothers with defects detected during pregnancy in 2004 continue to be reported. Of the 1,852 terminations of pregnancy reported in 1998–2004, 1,372 (74.1 per cent) were associated with a chromosomal abnormality, the most common of which was Trisomy 21 (Down syndrome), and 231 (12.5 per cent) were associated with a neural tube defect (Tables 119 and 120).

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

<table>
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<tr>
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<td>119</td>
<td>124</td>
<td>171</td>
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<td>Termination of pregnancy</td>
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<td>257</td>
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<td>312</td>
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<tr>
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<td>19</td>
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<td>18</td>
<td>–</td>
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<tr>
<td>TOTAL</td>
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<td>445</td>
<td>408</td>
<td>447</td>
<td>494</td>
<td>562</td>
<td>452</td>
<td>3159</td>
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</tbody>
</table>

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.
<table>
<thead>
<tr>
<th></th>
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<th></th>
<th></th>
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<tr>
<td></td>
<td>Spont. abortion</td>
<td>Unknown outcome</td>
<td>Spont. abortion</td>
<td>Unknown outcome</td>
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<td>1</td>
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<tr>
<td>Defects of respiratory system</td>
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<td>4</td>
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<td>Trisomy 18</td>
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<td>Turner syndrome</td>
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<td>Other and unspecified birth defects</td>
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<td>36</td>
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<td>TOTAL</td>
<td>762</td>
<td>2310</td>
<td>105</td>
<td>256</td>
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</tbody>
</table>

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.
Trends in selected birth defects

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

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

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

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

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

### TABLE 121

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

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 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.
FIGURE 14
NEURAL TUBE DEFECTS: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004*

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

FIGURE 15
CHROMOSOMAL ABNORMALITIES: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004*

Source: NSW Birth Defects Register. Centre for Epidemiology and Research, NSW Department of Health.
# For 1998–2003, cases reported during pregnancy and up to one year of age are included. For 2004, cases reported during pregnancy or at birth are reported.
FIGURE 16
DOWN SYNDROME: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004*

Number

Year

1998 1999 2000 2001 2002 2003 2004

Termination of pregnancy  Stillbirth  Livebirth - neonatal death  Liveborn surviving > 28 days

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

FIGURE 17
DIAPHRAGMATIC HERNIA: CASES BY YEAR AND PREGNANCY OUTCOME, NSW 1998–2004*

Number

Year

1998 1999 2000 2001 2002 2003 2004

Termination of pregnancy  Stillbirth  Livebirth - neonatal death  Liveborn surviving > 28 days

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

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

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

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

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

### TABLE 122

<table>
<thead>
<tr>
<th>Health Area</th>
<th>1998–2002 No.</th>
<th>1998–2002 Crude rate per 1,000 births</th>
<th>1998–2002 Standardised rate per 1,000 births</th>
<th>2003 No.</th>
<th>2003 Crude rate per 1,000 births</th>
<th>2003 Standardised rate per 1,000 births</th>
<th>2004 No.</th>
<th>2004 Crude rate per 1,000 births</th>
<th>2004 Standardised rate per 1,000 births</th>
<th>1998–2004 No.</th>
<th>1998–2004 Crude rate per 1,000 births</th>
<th>1998–2004 Standardised rate per 1,000 births</th>
<th>99% confidence intervals</th>
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</thead>
<tbody>
<tr>
<td>Sydney South West</td>
<td>2196</td>
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<td>21.4</td>
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<td>18.9</td>
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<td>12.5</td>
<td>2901</td>
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<td>18.8–20.9</td>
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<td>21.7</td>
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<td>1830</td>
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<td>22.7–25.8</td>
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<td>83</td>
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<td>16.4</td>
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<td>663</td>
<td>19.6</td>
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<td>17.1–21.1</td>
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<td>72</td>
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<td>15.3</td>
<td>46</td>
<td>11.8</td>
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<td>506</td>
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<td>13.8–17.6</td>
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<td>19.6</td>
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<td>1968</td>
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<td>20.1</td>
<td>19.7–20.6</td>
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</tbody>
</table>

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

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