Trisomy 21 (Down Syndrome) in Queensland: 1 July 2007 to 30 June 2010

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Trisomy 21 (or Down syndrome) is a congenital anomaly which is characterised by an excess of chromosome 21 material. It remains the most common chromosomal abnormality in Queensland, having occurred at an approximate rate of 9.4 cases per 10,000 births in 2004[1]. It is typically associated with a well known pattern of major and minor anomalies, although the long term outlook for individuals with this condition can be quite favourable. People with Down syndrome are now living longer and leading fuller, richer lives[2].

This report describes the epidemiology of Trisomy 21 in Queensland for the three year period 2007/2008 to 2009/2010. The data are sourced from the Queensland Hospital Admitted Data Collection (QHAPDC: terminations of pregnancy prior to 20 weeks duration) and the Queensland Perinatal Data Collection (QPDC: live births and fetal deaths of at least 20 weeks gestation or 400 grams in birth weight). In combination, these sources allow for the best available surveillance of diagnosed congenital anomalies from conception to the end of the perinatal period. However, it is recognised that this does not provide full surveillance, as events such as spontaneous abortions are not captured.

Table 1: Trisomy 21 in Queensland (2007/2008 to 2009/2010)

Measure	2007/ 2008	2008/ 2009	2009/ 2010
Incidence (per 10,000 fetuses) [n]	18.7 [140]	16.2 [124]	13.8 [105]
Live births (per 10,000 live births) [n]	10.8 [65]	7.4 [46]	7.1 [44]
Fetal deaths (per 10,000 fetal deaths) [n]	155.0 [6]	274.6 [12]	313.3 [12]
Total births (per 10,000 live births and fetal deaths) [n]	11.7 [71]	9.3 [58]	9.0 [56]
Early TOP (per 10,000 early TOP) [n]	48.4 [69]	45.8 [66]	35.5 [49]
Percent terminated (%)	49.3	53.2	46.7

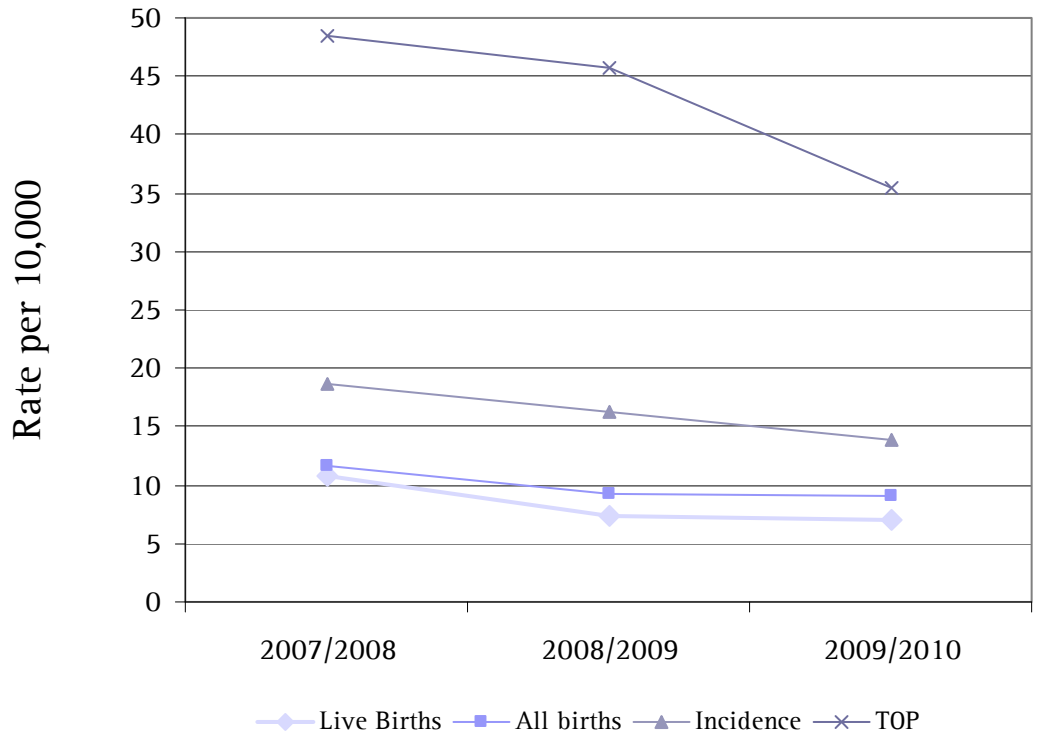
Sources: Queensland Hospital Admitted Patient Data Collection (extracted August 2011);
Queensland Perinatal Data Collection (extracted August 2011; 2010 data was preliminary at the time of extraction)

There were 369 fetuses affected by Trisomy 21 in Queensland between 2007/2008 and 2009/2010, yielding an incidence rate of 16.2 cases per 10,000 fetuses. Of these, 184 were terminated prior to 20 weeks gestation. Trisomy 21 was detected in 30 fetal deaths and in 155 live births. This corresponds to a fetal death rate of 248.6 affected fetuses per 10,000 fetal deaths and a live birth rate of of 8.4 affected fetuses per 10,000 live births. The total birth rate (live births plus fetal deaths) was 10.0 affected fetuses per 10,000 births which is marginally higher than that reported in 2004 (9.4 affected fetuses per 10,000 births)[1].

The incidence of Trisomy 21 declined in Queensland between 2007/2008 and 2009/2010 from 18.7 to 13.8 affected fetuses per 10,000 fetuses (Table 1, Figure 1). There was also a decrease in live birth rates and total birth rates (live births plus fetal deaths), as well as in early terminations of pregnancy (<20 weeks duration). Fetal death rates increased sharply over the study period (Table 1: from 155.0 to 313.3 affected fetuses per 10,000 fetal deaths).

[1]Roselli T 2006. Summary statistics on congenital anomalies in Queensland 1988-2008. Information circular 75. Health Statistics Centre. Queensland Health.
[2]National Institute of Child Development and Health: http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm .

Figure 1. Live birth, total birth and incidence rates for Trisomy 21 in Queensland 2007/2008 to 2009/2010



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