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Trisomy 21 (Down Syndrome) in Queensland 2007-2008

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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 2004¹. 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 lives².

The most recent published material¹ on congenital anomalies in Queensland used data that was drawn from the Queensland Perinatal Data Collection (QPDC). This includes all live births in Queensland, as well as deaths of fetuses which are at least of 20 weeks gestation or 400 grams in birth weight. However, information is not included in the QPDC on terminations of pregnancy (TOP) prior to 20 weeks gestation. Early detection of Trisomy 21 may result in TOP for this condition. Queensland Health has now added early TOP to its surveillance of congenital abnormalities in Queensland. In this publication, we include the TOP data to update earlier estimates of the incidence of Trisomy 21 in Queensland.

There were 127 fetuses affected by Trisomy 21 in Queensland during 2007-2008 (Table 1). This

corresponds to an incidence rate of 17.1 cases per 10,000 fetuses. Of these, 50.4% were terminated at prior to 20 weeks gestation. Trisomy 21 was detected in 8 fetal deaths and 55 live births. This corresponds to a fetal death rate of 1.4 cases per 10,000 births and a live birth rate of of 9.2 per 10,000 births. The total birth rate (live births plus fetal deaths) was 10.5 per 10,000, which is marginally higher than reported in 2004 (9.4 per 10,000)¹.

Birth rates for Trisomy 21 have risen slightly in Queensland since

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

Measure	Rate
Incidence (per 10,000 fetuses)	17.1
Live births (per 10,000 births*)	9.2
Fetal deaths (per 10,000 births*)	1.4
Total births* (per 10,000 births*)	10.5
Proportion of fetuses terminated at <20 weeks	50.4
gestation	

^{*}Includes live births and fetal deaths (>20 wk gestation or 400g birth wgt)

Sources: Queensland Admitted Patient Data Collection, Queensland Health – TOP prior to 20 weeks gestation (extracted June 2009)

Queensland Perinatal Data Collection, Queensland Health – Live births and

deaths in fetuses of at least 20 weeks gestation or 400 grams birth weight (extracted June 2009)

2004. The reasons behind this are not clear and it may simply reflect improved surveillance of congenital anaomalies in Queensland in recent years. Alternatively, it may reflect a decrease in the early TOP (i.e. prior to 20 weeks gestation) for Trisomy 21 over the intervening period, although this remains speculative. The data required to assess trends in TOP are not yet available.

Related publications:

STAT Bite #18: Maternal characteristics in pregnancies affected by Trisomy 21 (Down Syndrome) in Queensland 2007-2008.

¹Roselli T 2006. Summary statistics on congenital anomalies in Queensland 1988-2008. Information circular 75. Health Statistics Centre. Queensland Health.

²National Institute of Child Development and Health: http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm .