



Queensland  
Government

## Congenital Anomalies in Queensland: 1 July 2007 to 30 June 2010

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### Abstract

The purpose of this report is to describe the epidemiology of congenital anomalies in Queensland for the three year period 1 July 2007 to 30 June 2010.

*What is known about this topic?* Congenital anomalies affect roughly 1 in 20 births in Australia and are associated with high rates of mortality and morbidity<sup>1</sup>. The public health costs of major congenital anomalies are high and include screening programs for the early detection of congenital anomalies, early termination of pregnancy, and management of lifetime disability among those who survive.

*What does this report add?* This document presents the incidence of congenital anomalies in Queensland, with the inclusion of early terminations of pregnancy (i.e.: less than 20 weeks duration). Information concerning the early termination of pregnancies affected by congenital anomalies was not available prior to July 2007.

*What are the implications?* This report provides a mechanism for reporting and monitoring the incidence of congenital anomalies in Queensland.

### Introduction

The epidemiology of congenital anomalies in Australia has not been fully captured in recent years. The latest national report, which was prepared by the Australian Institute of Health and Welfare (AIHW)<sup>2</sup>, only includes data for 2002 – 2003, while local (Queensland) data has been limited by the exclusion of early terminations of pregnancy (less than 20 weeks duration)<sup>3 4</sup>.

The purpose of this report is to describe the epidemiology of congenital anomalies in Queensland for the period 1 July 2007 to 30 June 2010, with the inclusion of early terminations of pregnancy (i.e.: less than 20 weeks duration). The data are sourced from the Queensland Hospital Admitted Data Collection (QHAPDC: early terminations of pregnancy) 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. Data sources and methods used in the calculation of case rates in this report are summarised in

Appendix 1. The ICD-10-AM codes used to identify early terminations of pregnancy where a congenital anomaly was present are provided in Appendix 2.

The congenital anomalies reported here are those agreed upon by the National Congenital Anomalies Steering Committee (NCASC) (refer to Appendix 3). These are based on the 'sentinel' conditions reported by the International Clearing House for Birth Defects Surveillance and Research (ICHBDSR). As with the earlier national report<sup>2</sup>, undescended testis and prune belly were excluded on the grounds that the NCASC determined that they were difficult to define.

## Background

Congenital anomalies affect roughly 1 in 20 births in Australia and are associated with high rates of mortality and morbidity<sup>1</sup>. The public health costs of major congenital anomalies are high and include screening programs for the early detection of congenital anomalies, early termination of pregnancy, and management of lifetime disability among those who survive.

The most common congenital anomalies in Queensland prior to 2005 were Trisomy 21, neural tube defects, and facial clefts<sup>3</sup>. These have been described in the peer reviewed literature and a number of protective and risk factors have been identified. This report focuses on the epidemiology of these three defects in Queensland between 1 July 2007 and 30 June 2010. The remaining 'sentinel' defects, as defined by the ICHBDSR, are presented in Table 1 but are not described in detail in the body of this report.

**Trisomy 21 (Down syndrome):** Trisomy 21 is the most common chromosomal anomaly both in Queensland and nationally. There is evidence to suggest that birth rates for Trisomy 21 are on the decline, although the reasons for this are not entirely clear. While a declining birth rate may reflect a decrease in the incidence of Trisomy 21, it is equally plausible that this reflects an increase in the termination rates for pregnancies affected by this anomaly. Advancing maternal age (AMA) has been identified as the primary risk factor, and accordingly, some states have reported that they have explicit or implicit policies for offering diagnostic screening to women on the basis of advanced maternal age<sup>5</sup>.

**Neural tube defects (NTD):** NTD are also common locally and nationally. In Australia, spina bifida is the most prevalent of the neural tube defects, followed by anencephaly. NTD are associated with high rates of perinatal mortality and significant lifetime disability in those who survive. Most NTD are diagnosed early in the pregnancy and many affected women opt for a termination of pregnancy. NTD are viewed as potentially preventable through increased folic acid intake around the time of conception<sup>6</sup>. As a result, Australia moved to the mandatory (and legally enforceable) fortification of food with folic acid in 2009<sup>7</sup>. The impact of this on the incidence of NTD in Australia has yet to be established.

**Facial clefts:** Facial clefts are also common congenital anomalies, with cleft lip (with or without cleft palate) being more prevalent than cleft palate (without cleft lip). Mortality rates are generally low and few pregnancies are terminated prior to 20 weeks duration. Multiple defects are usually present in fetuses that were terminated as well as in those that died *in utero*<sup>2</sup>. There is evidence to suggest that folic acid intake around the time of conception reduces the risk of cleft lip (with or without cleft palate), although this remains controversial<sup>8</sup>. Epidemiological studies have produced mixed results, which may in part, be due to differences in study design, along with variations in the way that folic acid intake was assessed (e.g. through dietary intake versus vitamin supplements). Nonetheless, two studies<sup>8 9</sup> have reported that folic acid has a protective effect on cleft lip with or without cleft palate, although the authors concede that other dietary factors may be involved. Folic acid intake appears to have no effect on the incidence of cleft palate in the absence of cleft lip. Facial clefts have also been linked to periconceptional alcohol consumption<sup>10</sup>, although folic acid intake may modify alcohol-related risks.

## Results

This document represents the first time that the incidence of congenital anomalies has been reported in Queensland with the inclusion of early termination of pregnancies (at less than 20 weeks duration) affected by congenital anomalies, which was not available prior to July 2007. Previous reports were thus based on congenital anomalies birth rates as estimated from the Queensland Perinatal Data Collection.

**Trisomy 21:** The incidence of trisomy 21 declined in Queensland between 2007/2008 and 2009/2010 (Figure 1.1, Table 1) from 18.7 to 13.8 affected fetuses per 10,000 fetuses. There was a sharp drop in the number of early terminations of pregnancy in 2009/2010 when compared to the previous two years. The birth rates (as derived from live births and fetal deaths) decreased in the first two years, but remained stable in the 2009/2010 year. The overall drop in incidence rates was steady across the study period, with the reduction being 2.5 affected fetuses per 10,000 fetuses between 2007/2008 and 2008/2009 and 2.4 affected fetuses per 10,000 between 2008/2009 and 2009/2010.

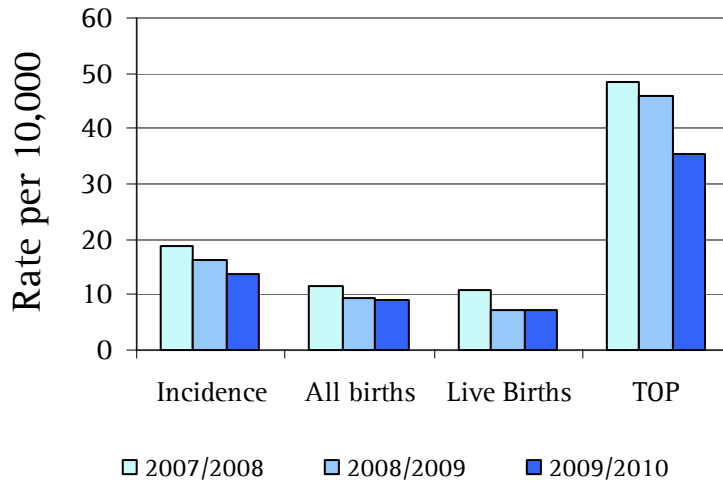
**Neural tube defects:** The incidence of neural tube defects in Queensland did not change between 2007/2008 and 2008/2009 (Figure 2.1, Table 1), with the respective rates being 9.7 and 9.9 affected fetuses per 10,000 fetuses. There was a drop in the incidence rate to 8.1 affected fetuses per 10,000 fetuses for 2009/2010, although the basis for this is uncertain. There was a drop in the termination rate in 2009/2010 when compared to previous years, which was offset by an increase in fetal deaths (Figure 2.2, Table 1). The trends reported for neural tube defects overall, were evident in the rates for anencephaly (Figures 3.1

and 3.2, Table 1), and spina bifida (Figures 4.1 and 4.2, Table 1), but not in those for encephalocele (Table 2). Encephalocele was rare in Queensland with only 14 cases reported across the study period.

Mortality rates for neural tube defects remain high. For anencephaly, only 9 of the affected fetuses were born alive (aggregated across the whole study period) and these died shortly after they were born. Survival rates for spina bifida were better, with nearly 40% of affected fetuses being born alive.

**Facial clefts:** Facial clefts were reported in 233 fetuses over the reporting period. Roughly 60% of these (n=138) were cleft lip (with or without cleft palate), while the remainder were cleft palate (without cleft lip). The incidence of cleft lip (with or without cleft palate) increased from 5.6 to 7.1 affected fetuses per 10,000 between 2007/2008 and 2009/2010 (Figure 6.1, Table 1), which is consistent with previously reported trends in birth rates<sup>3</sup>. In contrast, the incidence of cleft palate (without cleft lip) declined slightly between 2007/2008 and 2009/2010 from 4.5 to 3.8 affected fetuses per 10,000 (Figure 5.1, Table 1). This is partly consistent with the earlier study<sup>3</sup>, which reported an increase in birth rates to 2003 but a decline in birth rates thereafter. Terminations of pregnancy are not common for these anomalies.

**Figure 1.1: Trisomy21 in Queensland – July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)**



**Figure 1.2: Trisomy21 in Queensland – July 2007 to June 2010: fetal death rates**

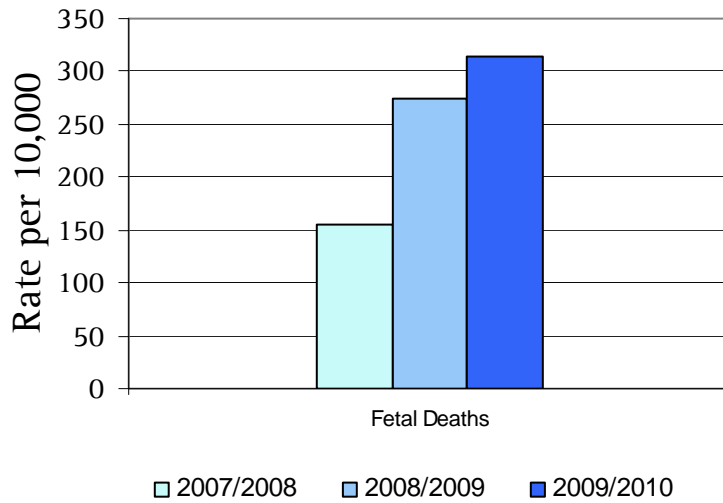


Figure 2.1: Neural tube defects in Queensland – July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)

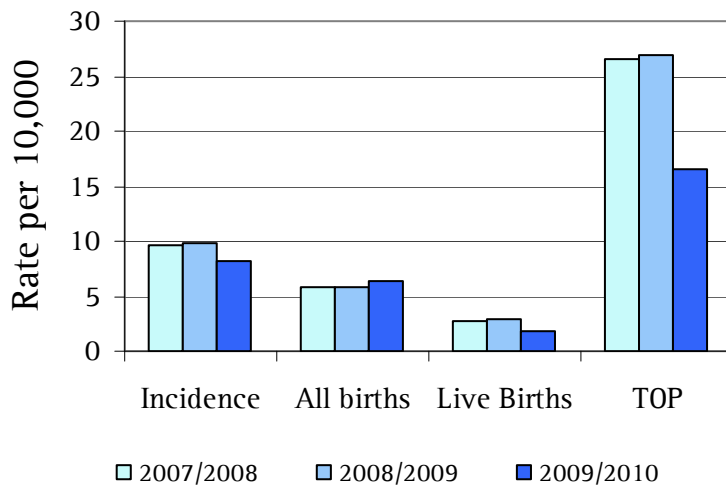


Figure 2.2: Neural tube defects in Queensland – July 2007 to June 2010: fetal death rates

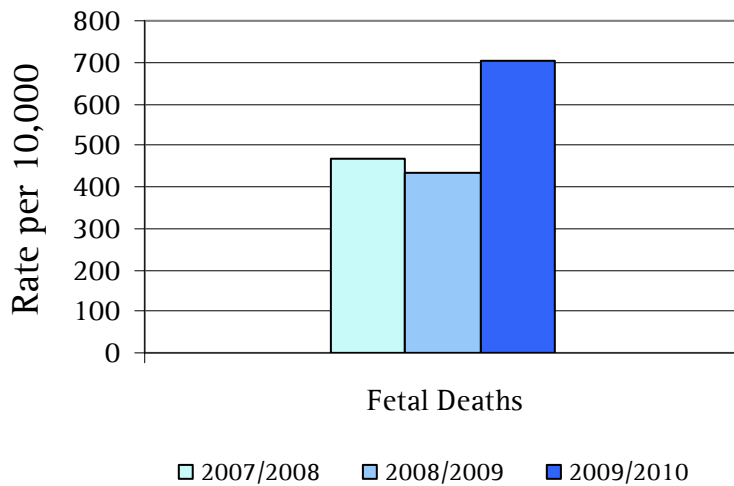


Figure 3.1: Anencephaly in Queensland – July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)

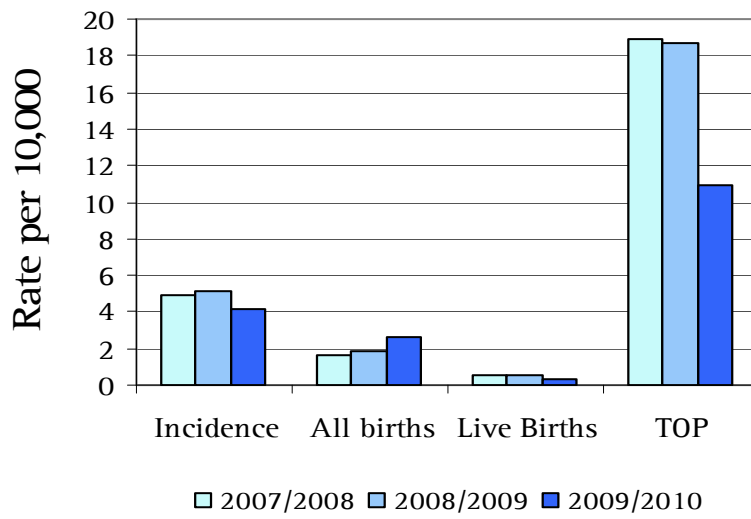


Figure 3.2: Anencephaly in Queensland July 2007 to June 2010: fetal death rates

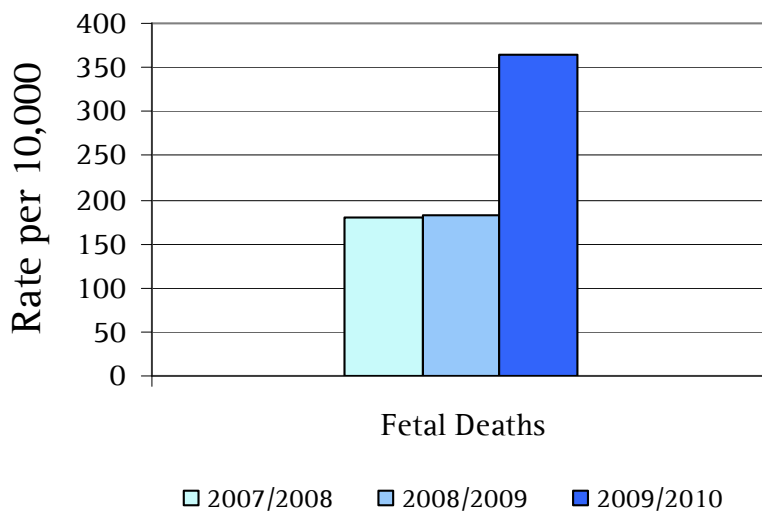


Figure 4.1: Spina bifida in Queensland – July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)

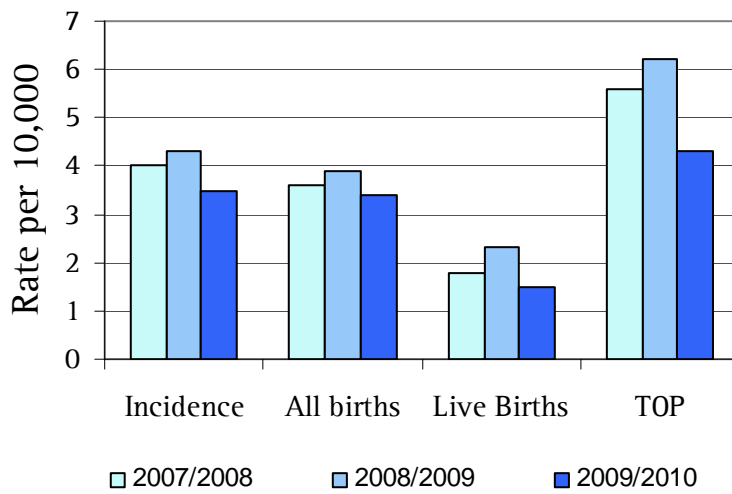
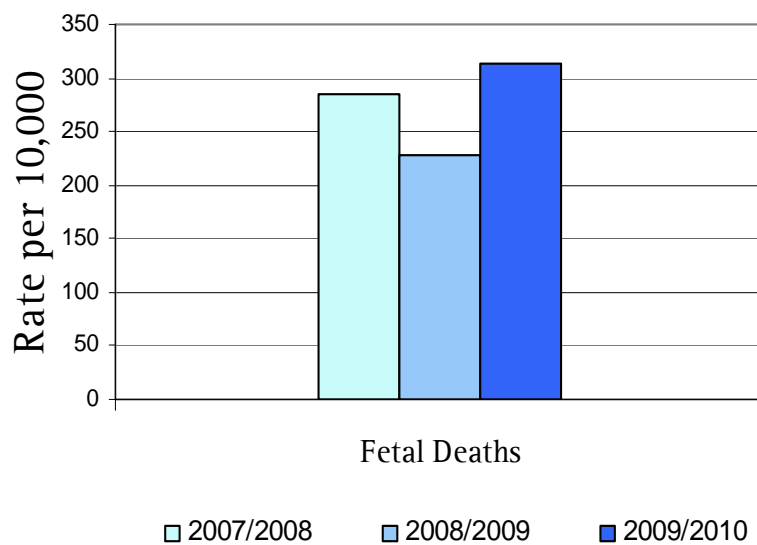
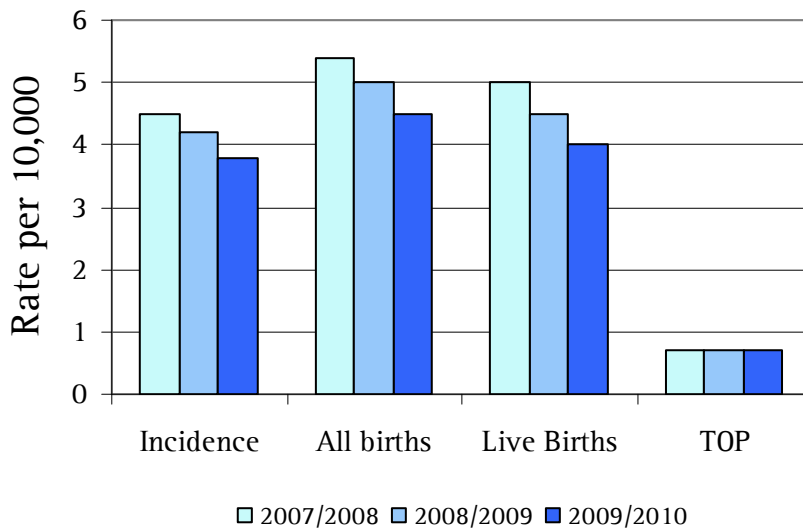


Figure 4.2: Spina bifida in Queensland July 2007 to June 2010: fetal death rates

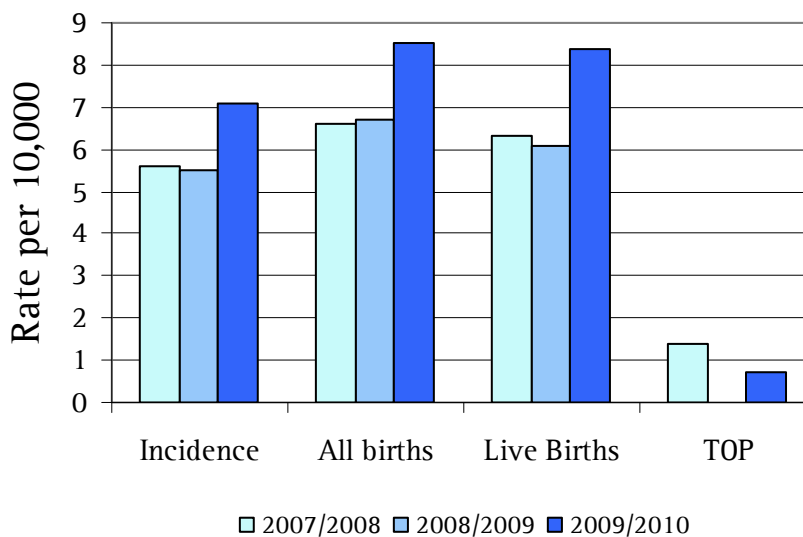




**Figure 5.1: Cleft palate without cleft lip in Queensland July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)**



**Figure 6.1: Cleft lip with or without cleft palate in Queensland July 2007 to June 2010: incidence, birth rates and early terminations of pregnancy (TOP)**



**Table 1. Reported rates for key congenital anomalies in Queensland 2007/2008 to 2009/2010**

Sources: Queensland Hospital Admitted Patient Data Collection, Queensland Health (Extracted June, 2011)

Terminations of pregnancy with less than 20 weeks duration

Perinatal Data Collection, Queensland Health (Extracted June, 2011; 2010 data preliminary at time of extraction) Live births and fetal deaths

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Neural Tube Defects	Live Births (1)	17	2.8		18	2.9		12	1.9	
	Fetal Deaths (2)	18	465.1		19	434.8		27	705	
	All births (3)	35	5.8		37	5.9		39	6.3	
	Births and TOP (4) (5)	73	12.0	9.7	76	12.2	9.9	62	10	8.1
	TOP (6)	38	26.6		39	27.0		23	16.6	
	Percent terminated (7)		52.1			51.3			37.1	
	Anencephaly	Live Births (1)	3	0.5		4	0.6		2	0.3
Fetal Deaths (2)		7	180.9		8	183.1		14	365.5	
All births (3)		10	1.6		12	1.9		16	2.6	
Births and TOP (4) (5)		37	6.1	4.9	39	6.3	5.1	31	5	4.1
TOP (6)		27	18.9		27	18.7		15	10.9	
Percent terminated (7)			73.0			69.2			48.4	
Spina bifida		Live Births (1)	11	1.8		14	2.3		9	1.5
	Fetal Deaths (2)	11	284.2		10	228.8		12	313.3	
	All births (3)	22	3.6		24	3.9		21	3.4	
	Births and TOP (4) (5)	30	4.9	4	33	5.3	4.3	27	4.3	3.5
	TOP (6)	8	5.6		9	6.2		6	4.3	
	Percent terminated (7)		26.7			27.3			22.2	
	Encephalocele	Live Births (1)	3	0.5		0	0		1	0.2
Fetal Deaths (2)		0	0		1	22.9		1	26.1	
All births (3)		3	0.5		1	0.2		2	0.3	
Births and TOP (4) (5)		6	1	0.8	4	0.6	0.5	4	0.6	0.5
TOP (6)		3	2.1		3	2.1		2	1.4	
Percent terminated (7)			50			75			50	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Trisomy21	Live Births (1)	65	10.8		46	7.4		44	7.1	
	Fetal Deaths (2)	6	155		12	274.6		12	313.3	
	All births (3)	71	11.7		58	9.3		56	9	
	Births and TOP (4) (5)	140	23.1	18.7	124	19.9	16.2	105	16.9	13.8
	TOP (6)	69	48.4		66	45.8		49	35.5	
	Percent terminated (7)		49.3			53.2			46.7	
Trisomy18	Live Births (1)	4	0.7		7	1.1		5	0.8	
	Fetal Deaths (2)	4	103.4		9	205.9		13	339.4	
	All births (3)	8	1.3		16	2.6		18	2.9	
	Births and TOP (4) (5)	28	4.6	3.7	36	5.8	4.7	36	5.8	4.7
	TOP (6)	20	14		20	13.9		18	13	
	Percent terminated (7)		71.4			55.6			50	
Trisomy 13	Live Births (1)	3	0.5		1	0.2		1	0.2	
	Fetal Deaths (2)	0	0		2	45.8		0	0	
	All births (3)	3	0.5		3	0.5		1	0.2	
	Births and TOP (4) (5)	14	2.3	1.9	22	3.5	2.9	5	0.8	0.7
	TOP (6)	11	7.7		19	13.2		4	2.9	
	Percent terminated (7)		78.6			86.4			80	
Microcephaly	Live Births (1)	7	1.2		7	1.1		2	0.3	
	Fetal Deaths (2)	3	77.5		2	45.8		3	78.3	
	All births (3)	10	1.6		9	1.4		5	0.8	
	Births and TOP (4) (5)	10	1.6	1.3	9	1.4	1.2	5	0.8	0.7
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Arhinencephaly	Live Births (1)	5	0.8		3	0.5		1	0.2	
	Fetal Deaths (2)	0	0		4	91.5		4	104.4	
	All births (3)	5	0.8		7	1.1		5	0.8	
	Births and TOP (4) (5)	9	1.5	1.2	11	1.8	1.4	10	1.6	1.3
	TOP (6)	4	2.8		4	2.8		5	3.6	
	Percent terminated (7)		44.4			36.4			50.0	
Hydrocephaly	Live Births (1)	19	3.2		17	2.8		7	1.1	
	Fetal Deaths (2)	4	103.4		10	228.8		6	156.7	
	All births (3)	23	3.8		27	4.3		13	2.1	
	Births and TOP (4) (5)	23	3.8	3.1	27	4.3	3.5	14	2.2	1.8
	TOP (6)	0	0		0	0		1	0.7	
	Percent terminated (7)		0			0			7.1	
Anophthalmos/Microphthalmos	Live Births (1)	2	0.3		4	0.6		6	1	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	2	0.3		4	0.6		6	1	
	Births and TOP (4) (5)	3	0.5	0.4	4	0.6	0.5	6	1	0.8
	TOP (6)	1	0.7		0	0		0	0	
	Percent terminated (7)		33.3			0			0	
Microtia	Live Births (1)	2	0.3		3	0.5		2	0.3	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	2	0.3		3	0.5		2	0.3	
	Births and TOP (4) (5)	2	0.3	0.3	3	0.5	0.4	2	0.3	0.3
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Transposition of the greater vessels	Live Births (1)	20	3.3		22	3.6		15	2.4	
	Fetal Deaths (2)	1	25.8		3	68.6		2	52.2	
	All births (3)	21	3.5		25	4		17	2.7	
	Births and TOP (4) (5)	21	3.5	2.8	25	4	3.3	17	2.7	2.2
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	
Tetralogy of Fallot	Live Births (1)	13	2.2		13	2.1		10	1.6	
	Fetal Deaths (2)	1	25.8		2	45.8		2	52.2	
	All births (3)	14	2.3		15	2.4		12	1.9	
	Births and TOP (4) (5)	14	2.3	1.9	15	2.4	2	12	1.9	1.6
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	
Hypoplastic left heart syndrome	Live Births (1)	9	1.5		6	1		9	1.5	
	Fetal Deaths (2)	10	258.4		4	91.5		2	52.2	
	All births (3)	19	3.1		10	1.6		11	1.8	
	Births and TOP (4) (5)	21	3.5	2.8	11	1.8	1.4	11	1.8	1.4
	TOP (6)	2	1.4		1	0.7		0	0	
	Percent terminated (7)		9.5			9.1			0	
Coarctation of the aorta	Live Births (1)	10	1.7		14	2.3		14	2.3	
	Fetal Deaths (2)	2	51.7		2	45.8		1	26.1	
	All births (3)	12	2		16	2.6		15	2.4	
	Births and TOP (4) (5)	12	2	1.6	16	2.6	2.1	15	2.4	2
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Choanal atresia	Live Births (1)	5	0.8		6	1		4	0.6	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	5	0.8		6	1		4	0.6	
	Births and TOP (4) (5)	5	0.8	0.7	6	1	0.8	5	0.8	0.7
	TOP (6)	0	0		0	0		1	0.7	
	Percent terminated (7)		0			0			20	
	Cleft palate without cleft lip	Live Births (1)	30	5		28	4.5		25	4.0
Fetal Deaths (2)		3	77.5		3	68.6		3	78.3	
All births (3)		33	5.4		31	5		28	4.5	
Births and TOP (4) (5)		34	5.6	4.5	32	5.1	4.2	29	4.7	3.8
TOP (6)		1	0.7		1	0.7		1	0.7	
Percent terminated (7)			2.9			3.1			3.4	
Cleft lip with or without cleft palate		Live Births (1)	38	6.3		38	6.1		52	8.4
	Fetal Deaths (2)	2	51.7		4	91.5		1	26.1	
	All births (3)	40	6.6		42	6.7		53	8.5	
	Births and TOP (4) (5)	42	6.9	5.6	42	6.7	5.5	54	8.7	7.1
	TOP (6)	2	1.4		0	0		1	0.7	
	Percent terminated (7)		4.8			0			1.9	
	Oesophageal atresia/stenosis	Live Births (1)	17	2.8		15	2.4		14	2.3
Fetal Deaths (2)		1	25.8		2	45.8		3	78.3	
All births (3)		18	3		17	2.7		17	2.7	
Births and TOP (4) (5)		18	3	2.4	17	2.7	2.2	17	2.7	2.2
TOP (6)		0	0		0	0		0	0	
Percent terminated (7)			0			0			0	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Small intestinal atresia/stenosis	Live Births (1)	13	2.2		4	0.6		9	1.5	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	13	2.1		4	0.6		9	1.4	
	Births and TOP (4) (5)	13	2.1	1.7	4	0.6	0.5	9	1.4	1.2
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	
Anorectal atresia/stenosis	Live Births (1)	16	2.7		20	3.2		9	1.5	
	Fetal Deaths (2)	0	0		4	91.5		2	52.2	
	All births (3)	16	2.6		24	3.9		11	1.8	
	Births and TOP (4) (5)	16	2.6	2.1	25	4	3.3	13	2.1	1.7
	TOP (6)	0	0		1	0.7		2	1.4	
	Percent terminated (7)		0			4			15.4	
Hirschsprung's disease	Live Births (1)	7	1.2		10	1.6		9	1.5	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	7	1.2		10	1.6		9	1.4	
	Births and TOP (4) (5)	7	1.2	0.9	10	1.6	1.3	9	1.4	1.2
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	
Epispadias	Live Births (1)	0	0		2	0.3		2	0.3	
	Fetal Deaths (2)	0	0		0	0		0	0	
	All births (3)	0	0		2	0.3		2	0.3	
	Births and TOP (4) (5)	0	0	0	2	0.3	0.3	2	0.3	0.3
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	

Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Renal agenesis/dysgenesis	Live Births (1)	18	3		28	4.5		31	5.0	
	Fetal Deaths (2)	4	103.4		16	366.1		5	130.5	
	All births (3)	22	3.6		44	7.1		36	5.8	
	Births and TOP (4) (5)	23	3.8	3.1	46	7.4	6	39	6.3	5.1
	TOP (6)	1	0.7		2	1.4		3	2.2	
	Percent terminated (7)		4.3			4.3			7.7	
Cystic kidney	Live Births (1)	20	3.3		31	5		28	4.5	
	Fetal Deaths (2)	3	77.5		7	160.2		1	26.1	
	All births (3)	23	3.8		38	6.1		29	4.7	
	Births and TOP (4) (5)	25	4.1	3.3	38	6.1	5	29	4.7	3.8
	TOP (6)	2	1.4		0	0		0	0	
	Percent terminated (7)		8			0			0	
Bladder exstrophy	Live Births (1)	0	0		1	0.2		1	0.2	
	Fetal Deaths (2)	0	0		1	22.9		0	0	
	All births (3)	0	0		2	0.3		1	0.2	
	Births and TOP (4) (5)	0	0	0	2	0.3	0.3	1	0.2	0.1
	TOP (6)	0	0		0	0		0	0	
	Percent terminated (7)		0			0			0	
Polydactyly	Live Births (1)	52	8.6		46	7.4		49	7.9	
	Fetal Deaths (2)	0	0		1	22.9		0	0	
	All births (3)	52	8.6		47	7.6		49	7.9	
	Births and TOP (4) (5)	52	8.6	6.9	48	7.7	6.3	49	7.9	6.4
	TOP (6)	0	0		1	0.7		0	0	
	Percent terminated (7)		0			2.1			0	



Congenital anomaly	Measure	Queensland 2007/2008			Queensland 2008/2009			Queensland 2009/2010		
		Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000	Count	Rate /10,000	HSC incidence rate /10,000
Limb reduction defects	Live Births (1)	20	3.3		15	2.4		23	3.7	
	Fetal Deaths (2)	6	155		6	137.3		4	104.4	
	All births (3)	26	4.3		21	3.4		27	4.3	
	Births and TOP (4) (5)	28	4.6	3.7	24	3.9	3.1	30	4.8	3.9
	TOP (6)	2	1.4		3	2.1		3	2.2	
	Percent terminated (7)		7.1			12.5			10	
Diaphragmatic hernia	Live Births (1)	16	2.7		12	1.9		19	3.1	
	Fetal Deaths (2)	2	51.7		3	68.6		2	52.2	
	All births (3)	18	3		15	2.4		21	3.4	
	Births and TOP (4) (5)	20	3.3	2.7	15	2.4	2	22	3.5	2.9
	TOP (6)	2	1.4		0	0		1	0.7	
	Percent terminated (7)		10			0			4.5	
Exomphalos	Live Births (1)	1	0.2		6	1		6	1	
	Fetal Deaths (2)	2	51.7		2	45.8		3	78.3	
	All births (3)	3	0.5		8	1.3		9	1.4	
	Births and TOP (4) (5)	5	0.8	0.7	8	1.3	1	13	2.1	1.7
	TOP (6)	2	1.4		0	0		4	2.9	
	Percent terminated (7)		40			0			30.8	
Gastroschisis	Live Births (1)	34	5.6		27	4.4		33	5.3	
	Fetal Deaths (2)	3	77.5		3	68.6		3	78.3	
	All births (3)	37	6.1		30	4.8		36	5.8	
	Births and TOP (4) (5)	38	6.3	5.1	31	5	4	37	5.9	4.9
	TOP (6)	1	0.7		1	0.7		1	0.7	
	Percent terminated (7)		2.6			3.2			2.7	

- (1) Includes all births with at least 20 weeks gestational age or at least 400 grams of birthweight. The rate is per 10,000 live births.
- (2) Includes fetal deaths with at least 20 weeks gestational age or at least 400 grams of birthweight. The rate is per 10,000 fetal deaths.
- (3) Includes all live births and all fetal deaths. The rate is per 10,000 live births plus fetal deaths
- (4) Includes all live births and fetal deaths and all terminations of pregnancy at less than 20 weeks duration. The rate is per 10,000 live births plus fetal deaths.
- (5) The adjusted rate is per 10,000 live births plus fetal deaths plus all terminations of pregnancy before 20 weeks duration
- (6) TOP: termination of pregnancy. The rate is per 10,000 TOP at less than 20 weeks duration
- (7) TOP with a congenital anomaly as a proportion of live births plus fetal deaths plus TOP with a congenital anomaly
- (8) Abeywardana S & Sullivan EA 2008. Congenital Anomalies in Australia 2002-2003. Birth anomalies series no. 3 Cat. No. PER 41. Sydney: AIHW National Perinatal Statistics Unit

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## Appendix 1: Data sources and methods used in the calculation of case rates used in this report

Measure	Data Source	Numerator	Denominator	Case rate
<b>1. Live births</b>	QPDC	Live births in Queensland during the study period where the selected CA was detected	All live births in Queensland during the study period.	The number of <u>live births</u> where the selected CA was detected as a rate per 10,000 <u>live births</u>
<b>2. Fetal deaths</b>	QPDC	Fetal deaths in Queensland during the study period where the selected CA was detected	All fetal deaths in Queensland during the study period	The number of <u>fetal deaths</u> where the selected CA was detected as a rate per 10,000 <u>fetal deaths</u>
<b>3. All births</b> (live births plus fetal deaths)	QPDC	All births in Queensland during the study period where the selected CA was detected	All births in Queensland during the study period	The number of <u>births</u> where the selected CA was detected as a rate per 10,000 <u>births</u>
<b>4. Early Termination of pregnancy (TOP) *</b> (less than 20 weeks duration)	QHAPDC	Early TOP (prior to 20 weeks) in Queensland during the study period where the selected CA was detected	All early TOP (prior to 20 weeks) in Queensland during the study period	The number of <u>early TOP</u> where the selected CA was detected as a rate per 10,000 <u>early TOP</u>  Note: The data is also reported as percent terminated
<b>5. All births and early TOP</b> (NPSU rate)	QPDC/QHAPDC	All births plus early TOP in Queensland during the study period where the selected CA was detected	All births in Queensland during the study period.  <b>Note:</b> NPSU does not include early TOP in the denominator when reporting this rate	The number of <u>births and early TOP</u> where the selected CA was detected as a rate per 10,000 <u>births</u>
<b>6. All births and early TOP</b> (HSC incidence rate)	QPDC/QHAPDC	All births plus early TOP in Queensland during the study period where the selected CA was detected	All births and all early TOP in Queensland during the study period.	The number of <u>births and early TOP</u> where the selected CA was detected as a rate per 10,000 <u>births and TOP</u> .  <b>Note: This rate represents the incidence of each selected CA in Queensland</b>

\*Caveat: Criminal proceedings in 2009 in relation to a terminated pregnancy may have impacted on early termination rates. Doctors in some facilities ceased performing terminations of pregnancy during the second half of 2009. This was due to legal concerns arising from the case. Interpretation of 2009/2010 data are conditional on the impact of these proceedings on the rates for early terminations of pregnancy.

**Appendix 2: QHAPDC Trigger codes used to identify early terminations of pregnancy where a congenital anomaly was detected<sup>11</sup>**

Trigger Code ICD-10AM range(s)	Description
0350-0356, 0358, 0359	Maternal care for known/suspected fetal abnormality or damage
0310, 0311, 0312	Complications specific to multiple gestation
0336, 0337	Maternal care for known/suspected disproportion
0360-0366, 0369	Maternal care for other known/suspected fetal problems
0430, 0431, 0438	Placental disorders
<i>At least one ICD-10AM code from:</i>	
0040-0049	Medical abortion
0050-0059	Other abortion
0060-0069	Unspecified abortion
<i>At least one ICD-10AM code from:</i>	
0090	Duration of pregnancy < 5 completed weeks
0091	Duration of pregnancy 5-13 completed weeks
0092	Duration of pregnancy 14-19 completed weeks

**Appendix 3: Coding of selected congenital anomalies**

The congenital anomalies included in this report were coded according to definitions used by the AIHW National Perinatal Statistics Unit (NPSU)<sup>2</sup>

Congenital anomaly	ICD-10-AM codes
Neural tube defects:	Anencephaly + Spina bifida + Encephalocele
Anencephaly	Q000 – Q002
Spina bifida	Q050 – Q059
Encephalocele	Q010 – Q012, Q018, Q019
Trisomy 21	Q900 – Q909
Trisomy 18	Q910 – Q913
Trisomy 13	Q914 – Q917
Microcephaly	Q02
Arinencephaly / Holoprosencephaly	Q041, Q042
Hydrocephaly	Q030, Q031, Q038, Q039
Anophthalmos / Microphthalmos	Q110 – Q112
Microtia	Q172
Transposition of the greater vessels	Q201, Q203, Q205
Tetralogy of Fallot	Q213
Hypoplastic left heart syndrome	Q234
Coarctation of the aorta	Q251
Choanal atresia	Q300
Cleft palate without cleft lip	Q350 – Q359
Cleft lip with or without cleft palate	Q360 – Q369, Q370 – Q375, Q378, Q379
Oesophageal atresia / stenosis	Q390 – Q393
Small intestine atresia /stenosis	Q410 – Q412
Anorectal atresia / stenosis	Q420 – Q423
Hirschsprung's disease	Q431
Epispadias	Q640
Renal agenesis / disgenesis	Q600 – Q606
Cystic kidney	Q610 – Q615
Bladder exstrophy	Q641
Polydactyly	Q690 – Q692, Q699
Limb reduction defects	Q710 – Q719, Q720 – Q729, Q730, Q731, Q738
Diaphragmatic hernia	Q790
Exomphalos	Q792
Gastroschisis	Q793

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