

### SUMMARY STATISTICS ON CONGENITAL ANOMALIES IN QUEENSLAND 1988-2004

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#### Summary

The purpose of this Information Circular is to provide summary statistics on congenital anomalies in Queensland that were identified during the birth episode.

In 2004, 2161 or 4.3% of babies born in Queensland were reported as having a congenital anomaly.

In terms of major diagnostic groups, musculoskeletal anomalies were the most common anomaly reported (16.3 per 1,000 births), followed by cardiovascular (9.5), urogenital (8.0), gastrointestinal (3.4), and chromosomal (1.9).

From 1988 to 2004, rates of neural tube defects decreased by 6.5% per year (95% CI 4.9% 8.0%).

Age-adjusted rates of Down syndrome decreased by 1.3% per year (95% CI: 0.1%, 2.5%) between 1988 and 2004. Since 2000 the Down syndrome rate has decreased by 9.3% per year (95% CI: 1.8%, 16.1%). This trend is probably due to increased screening.

Rates for gastroschisis increased by 7.8% per year (95% CI 4.8% 10.9%) between 1988 and 2004.

Rates for cardiovascular anomalies increased by 6.4% per year (95% CI 5.8% 7.0%) between 1988 and 2004. However this may be due to increased recognition of heart defects during the birth episode.

Lethal congenital anomalies accounted for 15.1% of all stillbirths in Queensland in 1988-2003; 30.2% of all neonatal deaths and 21.3% of post neonatal deaths.

Over the period 1988-2003, there was no statistically significant trend in the death rate due to congenital anomalies (annual percentage change of -0.7%; 95% CI -1.6% +0.2%).

#### Introduction

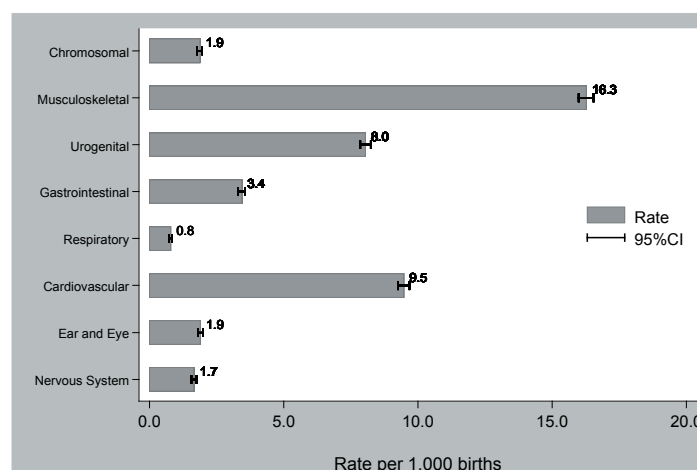
The purpose of this Information Circular is to provide summary statistics on congenital anomalies notified in Queensland between 1988 and 2004. The data were obtained from the population-based Queensland Perinatal Data Collection (QPDC). Only those congenital anomalies identified during the birth episode are captured by the QPDC. Consequently, for the purposes of this Circular, a congenital anomaly is defined as a structural defect or chromosomal abnormality that is present at birth and diagnosed prior to separation from care. Multiple

specific anomalies that make up a particular 'syndrome' are reported as both individual anomalies and as the syndrome.

Unlike some registers for congenital anomalies (The South Australian Birth Defects Register<sup>1</sup>, Birth Defects Registry of Western Australia<sup>2</sup>, Birth Defects Register for Victorian Children<sup>3</sup> and the Birth Defects Registry of New South Wales<sup>5</sup>), the QPDC does not include information on terminations of pregnancy before 20 weeks, or congenital anomalies diagnosed after the birth episode.

#### Types of Congenital Anomalies Notified

Figure 1: Congenital anomalies by major diagnostic grouping, Queensland 1988-2004



**Table 1: Congenital anomalies, numbers and rates, Queensland, 1988-2004**

Year of Birth	Births			Congenital Anomalies			Rate per 1,000 births - Male			Rate per 1,000 births - Female			Rate per 1,000 births - Total		
	Male	Female	Total	Male	Female	Total	Rate	95% CI		Rate	95% CI		Rate	95% CI	
<b>1988-1999</b>	287306	270114	557471	13352	10913	24431	46.5	45.7	47.3	40.4	39.6	41.2	43.8	43.3	44.4
<b>2000</b>	25307	24002	49318	1505	1152	2675	59.5	56.5	62.6	48.0	45.3	50.8	54.2	52.2	56.3
<b>2001</b>	25538	24139	49690	1461	1173	2642	57.2	54.3	60.2	48.6	45.9	51.5	53.2	51.1	55.2
<b>2002</b>	25237	23953	49196	1300	1028	2331	51.5	48.7	54.4	42.9	40.3	45.6	47.4	45.5	49.3
<b>2003</b>	25841	24519	50367	1555	1179	2739	60.2	57.2	63.2	48.1	45.4	50.8	54.4	52.4	56.4
<b>2004</b>	26160	24643	50836	1636	1159	2810	62.5	59.6	65.5	47.0	44.4	49.7	55.3	53.3	57.3
<b>Total</b>	415409	391370	806878	20809	16604	37628	50.1	49.4	50.8	42.4	41.8	43.1	46.6	46.1	47.1

Because a baby can have more than one congenital anomaly the above counts will exceed the number of babies with congenital anomalies. During the period 1988-2004 there were 37628 congenital anomalies among 29062 babies, 17.0% of these babies had more than one anomaly reported.

**Table 2: Specified congenital anomalies by diagnostic category, Queensland 1988-2004**

Diagnostic Category	Year of Birth																											
	1988-1999				2000				2001				2002				2003				2004				1988-2004			
	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI	Count	Rate per 1000 births	Lower 95% CI	Upper 95% CI
Nervous System	949	1.70	1.60	1.81	86	1.74	1.39	2.15	65	1.31	1.01	1.67	64	1.30	1.00	1.66	81	1.61	1.28	2.00	85	1.67	1.34	2.07	1330	1.65	1.56	1.74
Neural Tube Defects	518	0.93	0.85	1.01	33	0.67	0.46	0.94	14	0.28	0.15	0.47	31	0.63	0.43	0.89	26	0.52	0.34	0.76	34	0.67	0.46	0.93	656	0.81	0.75	0.88
Anencephalus	162	0.29	0.25	0.34	7	0.14	0.06	0.29	4	0.08	0.02	0.21	11	0.22	0.11	0.40	9	0.18	0.08	0.34	8	0.16	0.07	0.31	201	0.25	0.22	0.29
Spina bifida	323	0.58	0.52	0.65	23	0.47	0.30	0.70	6	0.12	0.04	0.26	15	0.30	0.17	0.50	12	0.24	0.12	0.42	17	0.33	0.19	0.54	396	0.49	0.44	0.54
Encephalocele	32	0.06	0.04	0.08	3	0.06	0.01	0.18	4	0.08	0.02	0.21	4	0.08	0.02	0.21	0	0.00	0.00	0.00	5	0.10	0.03	0.23	48	0.06	0.04	0.08
Microcephalus	83	0.15	0.12	0.18	6	0.12	0.04	0.26	5	0.10	0.03	0.23	3	0.06	0.01	0.18	5	0.10	0.03	0.23	6	0.12	0.04	0.26	108	0.13	0.11	0.16
Hydrocephalus	199	0.36	0.31	0.41	24	0.49	0.31	0.72	21	0.42	0.26	0.65	14	0.28	0.16	0.48	23	0.46	0.29	0.69	20	0.39	0.24	0.61	301	0.37	0.33	0.42
Arhinencephaly - holoprosencephaly	21	0.04	0.02	0.06	3	0.06	0.01	0.18	4	0.08	0.02	0.21	2	0.04	0.00	0.15	5	0.10	0.03	0.23	1	0.02	0.00	0.11	36	0.04	0.03	0.06
Ear and Eye	916	1.64	1.54	1.75	139	2.82	2.37	3.33	125	2.52	2.09	3.00	113	2.30	1.89	2.76	111	2.20	1.81	2.65	129	2.54	2.12	3.02	1533	1.90	1.81	2.00
Anotia/Microtia	22	0.04	0.02	0.06	6	0.12	0.04	0.26	4	0.08	0.02	0.21	0	0.00	0.00	0.00	2	0.04	0.00	0.14	4	0.08	0.02	0.20	38	0.05	0.03	0.06
Anophthalmos - microphthalmos	37	0.07	0.05	0.09	5	0.10	0.03	0.24	8	0.16	0.07	0.32	4	0.08	0.02	0.21	1	0.02	0.00	0.11	6	0.12	0.04	0.26	61	0.08	0.06	0.10
Cardiovascular	4703	8.44	8.20	8.68	611	12.39	11.43	13.41	575	11.57	10.65	12.56	492	10.00	9.14	10.92	618	12.27	11.32	13.28	651	12.81	11.84	13.83	7650	9.48	9.27	9.70
Transposition of great vessels	211	0.38	0.33	0.43	18	0.36	0.22	0.58	17	0.34	0.20	0.55	14	0.28	0.16	0.48	19	0.38	0.23	0.59	16	0.31	0.18	0.51	295	0.37	0.33	0.41
Tetralogy of Fallot	120	0.22	0.18	0.26	12	0.24	0.13	0.43	17	0.34	0.20	0.55	14	0.28	0.16	0.48	17	0.34	0.20	0.54	14	0.28	0.15	0.46	194	0.24	0.21	0.28
Hypoplastic left heart	119	0.21	0.18	0.26	9	0.18	0.08	0.35	9	0.18	0.08	0.34	9	0.18	0.08	0.35	12	0.24	0.12	0.42	6	0.12	0.04	0.26	164	0.20	0.17	0.24
Coarctation of Aorta	145	0.26	0.22	0.31	14	0.28	0.16	0.48	14	0.28	0.15	0.47	17	0.35	0.20	0.55	15	0.30	0.17	0.49	8	0.16	0.07	0.31	213	0.26	0.23	0.30
Respiratory	449	0.81	0.73	0.88	42	0.85	0.61	1.15	37	0.74	0.52	1.03	20	0.41	0.25	0.63	45	0.89	0.65	1.20	41	0.81	0.58	1.09	634	0.79	0.73	0.85
Choanal atresia	35	0.06	0.04	0.09	4	0.08	0.02	0.21	1	0.02	0.00	0.11	2	0.04	0.00	0.15	2	0.04	0.00	0.14	9	0.18	0.08	0.34	53	0.07	0.05	0.09
Gastrointestinal	1786	3.20	3.06	3.36	176	3.57	3.06	4.14	188	3.78	3.26	4.36	175	3.56	3.05	4.13	223	4.43	3.87	5.05	228	4.49	3.92	5.11	2776	3.44	3.31	3.57
Cleft Palate	338	0.61	0.54	0.67	17	0.34	0.20	0.55	36	0.72	0.51	1.00	27	0.55	0.36	0.80	41	0.81	0.58	1.10	38	0.75	0.53	1.03	497	0.62	0.56	0.67
Cleft Lip with or with out Cleft Palate	492	0.88	0.81	0.96	56	1.14	0.86	1.47	47	0.95	0.69	1.26	44	0.89	0.65	1.20	59	1.17	0.89	1.51	60	1.18	0.90	1.52	758	0.94	0.87	1.01
Oesophageal atresia / stenosis	140	0.25	0.21	0.30	15	0.30	0.17	0.50	12	0.24	0.12	0.42	10	0.20	0.10	0.37	8	0.16	0.07	0.31	14	0.28	0.15	0.46	199	0.25	0.21	0.28
Small intestine atresia / stenosis	121	0.22	0.18	0.26	7	0.14	0.06	0.29	9	0.18	0.08	0.34	10	0.20	0.10	0.37	9	0.18	0.08	0.34	9	0.18	0.08	0.34	165	0.20	0.17	0.24
Anorectal atresia / stenosis	161	0.29	0.25	0.34	18	0.36	0.22	0.58	16	0.32	0.18	0.52	20	0.41	0.25	0.63	13	0.26	0.14	0.44	23	0.45	0.29	0.68	251	0.31	0.27	0.35

4 Specified congenital anomalies by diagnostic category, Queensland 1988-2004, continued

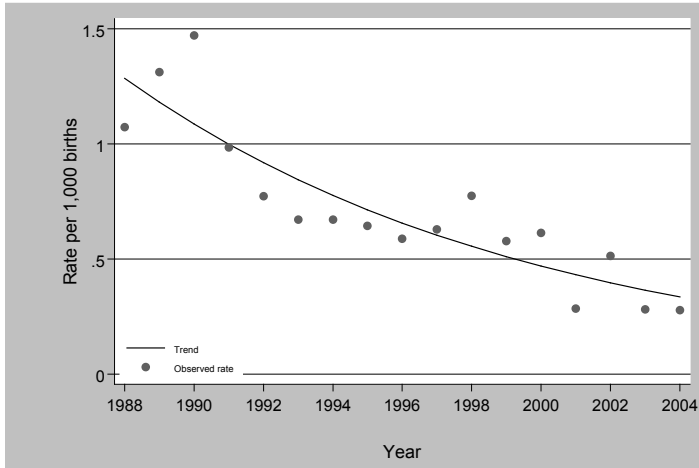
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Urogenital	4120	7.39	7.17	7.62	495	10.04	9.17	10.96	458	9.22	8.39	10.10	421	8.56	7.76	9.42	498	9.89	9.04	10.80	501	9.86	9.01	10.76	6493	8.05	7.85	8.25
Undescended Testicle	991	1.78	1.67	1.89	116	2.35	1.94	2.82	132	2.66	2.22	3.15	102	2.07	1.69	2.52	142	2.82	2.37	3.32	106	2.09	1.71	2.52	1589	1.97	1.87	2.07
Hypospadias	783	1.40	1.31	1.51	119	2.41	2.00	2.89	85	1.71	1.37	2.12	119	2.42	2.00	2.89	121	2.40	1.99	2.87	134	2.64	2.21	3.12	1935	2.40	2.29	2.51
Epispadias	14	0.03	0.01	0.04	1	0.02	0.00	0.11	2	0.04	0.00	0.15	0	0.00	0.00	0.00	4	0.08	0.02	0.20	0	0.00	0.00	0.00	21	0.03	0.02	0.04
Indeterminate sex	87	0.16	0.12	0.19	11	0.22	0.11	0.40	9	0.18	0.08	0.34	7	0.14	0.06	0.29	4	0.08	0.02	0.20	11	0.22	0.11	0.39	129	0.16	0.13	0.19
Renal agenesis	225	0.40	0.35	0.46	21	0.43	0.26	0.65	33	0.66	0.46	0.93	28	0.57	0.38	0.82	28	0.56	0.37	0.80	24	0.47	0.30	0.70	359	0.44	0.40	0.49
Cystic kidney	190	0.34	0.29	0.39	23	0.47	0.30	0.70	15	0.30	0.17	0.50	27	0.55	0.36	0.80	24	0.48	0.31	0.71	29	0.57	0.38	0.82	308	0.38	0.34	0.43
Bladder exstrophy	18	0.03	0.02	0.05	0	0.00	0.00	0.00	1	0.02	0.00	0.11	1	0.02	0.00	0.11	1	0.02	0.00	0.11	2	0.04	0.00	0.14	23	0.03	0.02	0.04
Musculoskeletal	8930	16.02	15.69	16.35	847	17.17	16.04	18.37	919	18.49	17.32	19.73	782	15.90	14.80	17.05	807	16.02	14.94	17.17	846	16.64	15.54	17.80	13131	16.27	16.00	16.55
Polydactyly	419	0.75	0.68	0.83	40	0.81	0.58	1.10	39	0.78	0.56	1.07	46	0.94	0.68	1.25	46	0.91	0.67	1.22	48	0.94	0.70	1.25	638	0.79	0.73	0.85
Reduction Deformity of Limbs	320	0.57	0.51	0.64	18	0.36	0.22	0.58	27	0.54	0.36	0.79	31	0.63	0.43	0.89	35	0.69	0.48	0.97	29	0.57	0.38	0.82	480	0.57	0.52	0.62
Diaphragmatic Hernia	191	0.34	0.30	0.39	12	0.24	0.13	0.43	20	0.40	0.25	0.62	13	0.26	0.14	0.45	15	0.30	0.17	0.49	8	0.16	0.07	0.31	259	0.32	0.28	0.36
Exomphalos	75	0.13	0.11	0.17	6	0.12	0.04	0.26	6	0.12	0.04	0.26	7	0.14	0.06	0.29	14	0.28	0.15	0.47	7	0.14	0.06	0.28	115	0.14	0.12	0.17
Gastroschisis	127	0.23	0.19	0.27	21	0.43	0.26	0.65	21	0.42	0.26	0.65	19	0.39	0.23	0.60	22	0.44	0.27	0.66	13	0.26	0.14	0.44	223	0.28	0.24	0.32
Prune belly sequence	18	0.03	0.02	0.05	0	0.00	0.00	0.00	0	0.00	0.00	0.00	2	0.04	0.00	0.15	2	0.04	0.00	0.14	0	0.00	0.00	0.00	22	0.03	0.02	0.04
Other abdominal wall defects	25	0.04	0.03	0.07	4	0.08	0.02	0.21	5	0.10	0.03	0.23	1	0.02	0.00	0.11	2	0.04	0.00	0.14	2	0.04	0.00	0.14	39	0.05	0.03	0.07
Chromosome	1033	1.85	1.74	1.97	93	1.89	1.52	2.31	103	2.07	1.69	2.51	100	2.03	1.65	2.47	98	1.95	1.58	2.37	83	1.63	1.30	2.02	1510	1.87	1.78	1.97
Down Syndrome	663	1.19	1.10	1.28	69	1.40	1.09	1.77	66	1.33	1.03	1.69	71	1.44	1.13	1.82	60	1.19	0.91	1.53	48	0.94	0.70	1.25	977	1.21	1.14	1.29
Trisomy 13	51	0.09	0.07	0.12	2	0.04	0.00	0.15	3	0.06	0.01	0.18	5	0.10	0.03	0.24	0	0.00	0.00	0.00	6	0.12	0.04	0.26	67	0.08	0.06	0.11
Trisomy 18	100	0.18	0.15	0.22	4	0.08	0.02	0.21	12	0.24	0.12	0.42	7	0.14	0.06	0.29	13	0.26	0.14	0.44	11	0.22	0.11	0.39	147	0.18	0.15	0.21

Note : Figures include all births in Queensland. Babies with multiple anomalies will appear in more than one category, therefore the number of cases in each major category does not necessarily equal the sum of the individual categories listed below it.

## Trends for Selected Congenital Anomalies

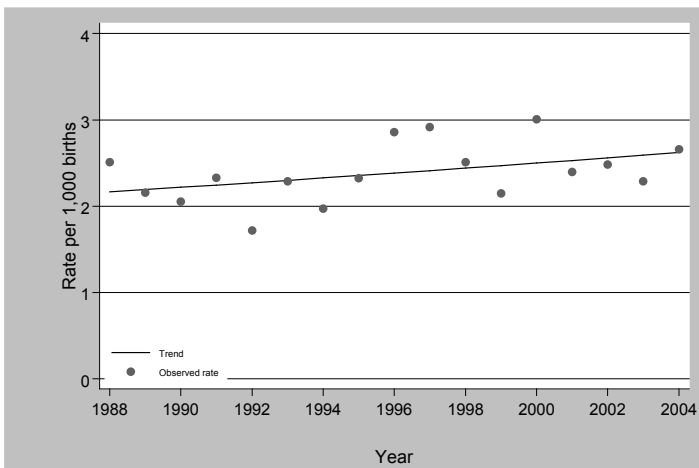
In the trends below the numerator for the rate is comprised of the number of babies with the selected anomalies. For example a baby born with both anencephaly and spina bifida was counted only once in the trend for neural tube defects.

**Figure 2: Trend for neural tube defects, Queensland 1988-2004**



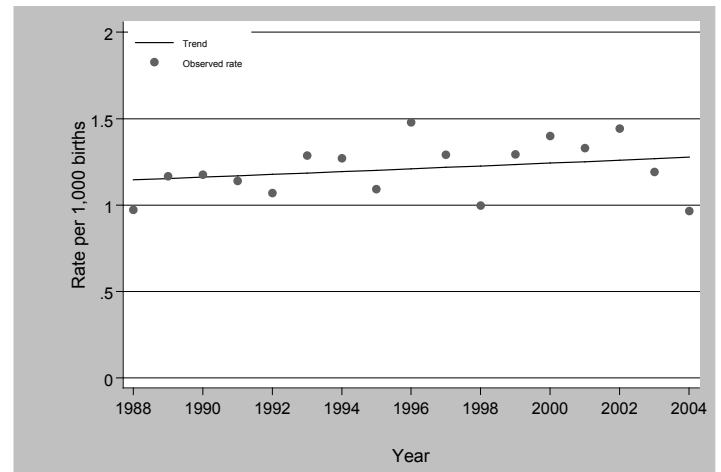
There was a statistically significant decrease in rates of neural tube defects between 1988 and 2004; annual percentage change: 6.5% (95% CI: 4.9%, 8.0%).

**Figure 3: Trend for hypospadias, Queensland 1988-2004**



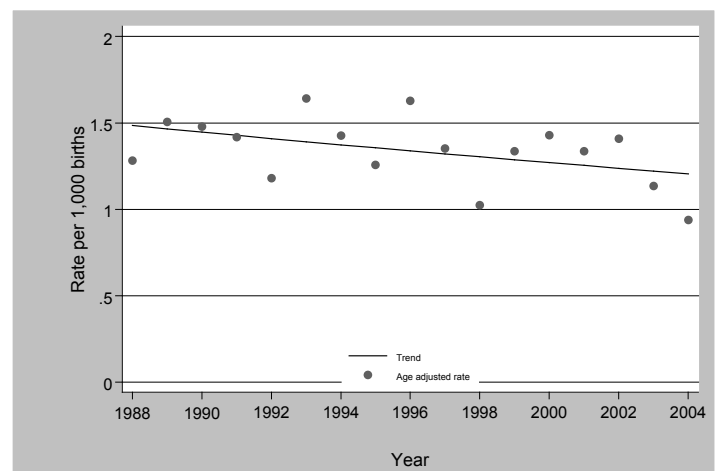
There was a statistically significant increase in rates of hypospadias between 1988 and 2004; annual percentage change: 1.2% (95% CI: 0.3%, 2.1%).

**Figure 4a: Trend for Down Syndrome - not age adjusted, Queensland 1988-2004**



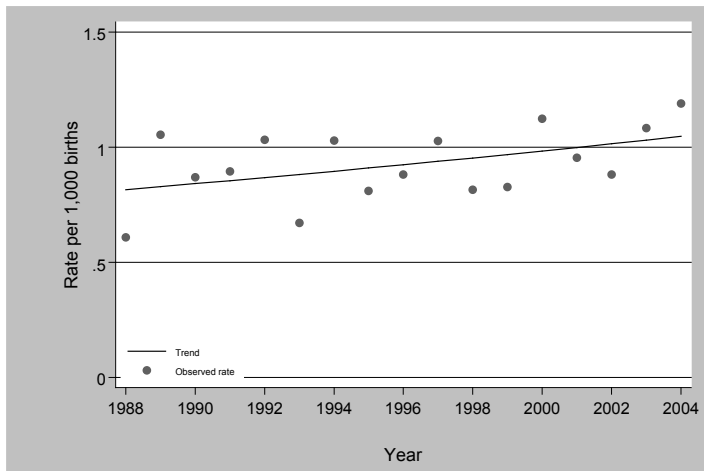
There was no significant trend in the rates of Down syndrome before any age adjustment; annual percentage change: +0.7% (95% CI: -0.6%, +2.0%)

**Figure 4b: Trend for Down Syndrome – age adjusted, Queensland 1988-2004**



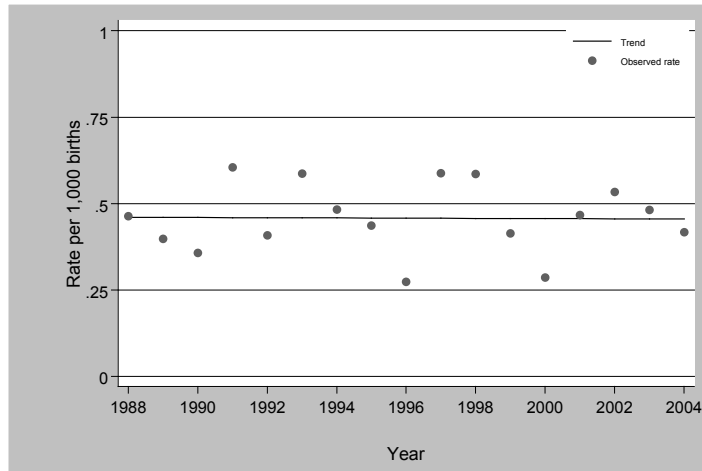
There was a statistically significant decrease in age adjusted rates of Down syndrome between 1988 and 2004; annual percentage change: 1.3% (95% CI: 0.1%, 2.5%). Since 2000 the Down syndrome rate has decreased by 9.3% per year (95% CI: 1.8%, 16.1%). Much of this decrease is probably due to increased screening

**Figure 5: Trend for cleft lip with or without cleft palate, Queensland 1988-2004**



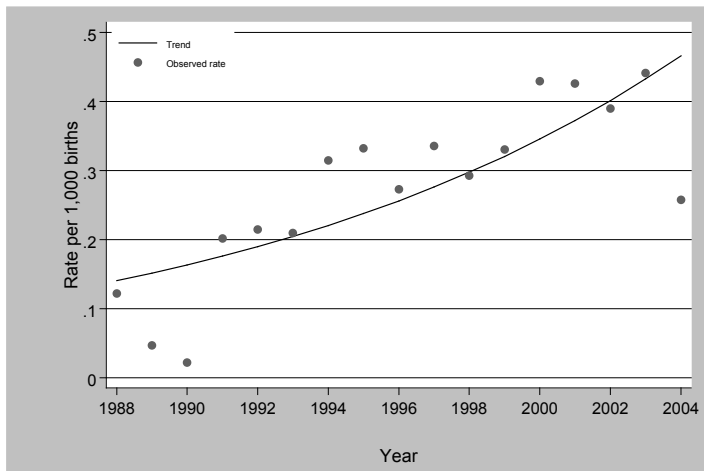
There was a statistically significant increase in the rate of cleft lip with or without cleft palate between 1988 and 2004; annual percentage change: 1.6% (95% CI: 0.1%, 3.1%).

**Figure 7: Trend for reduction deformity of limbs, Queensland 1988-2004**



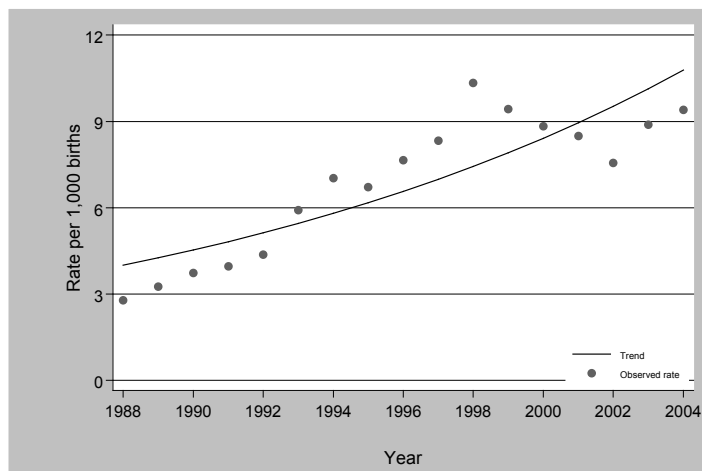
Over the period 1988-2004, there was no statistically significant trend in the rate of reduction deformity of limbs. (annual percentage change: 0.0%; 95% CI: -2.2%, +2.1%).

**Figure 6: Trend for gastroschisis, Queensland 1988-2004**



There was a statistically significant increase in rates of gastroschisis between 1988 and 2004; annual percentage change: 7.8% (95% CI: 4.8%, 10.9%).

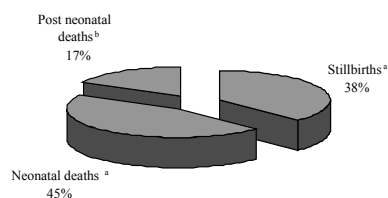
**Figure 8: Trend for cardiovascular anomalies, Queensland 1988-2004**



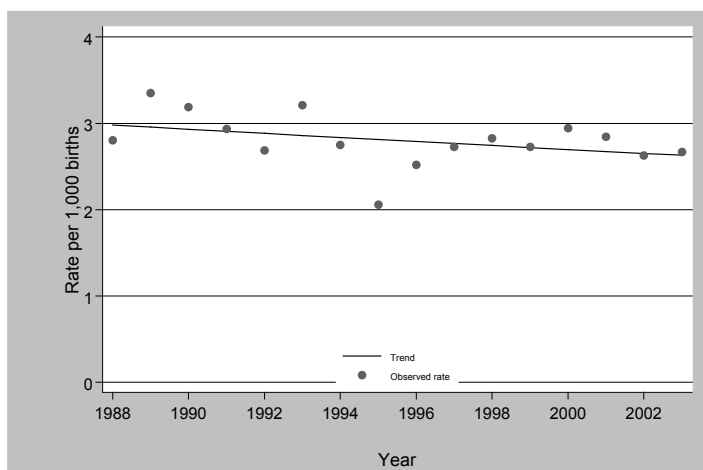
Rates of cardiovascular anomalies increased by 6.4% (95% CI 5.8% 7.0%) per year for the period 1988 – 2004. Much of the increase might be due to increased identification of heart defects during the birth episode.

## Deaths Due To Congenital Anomalies

**Figure 9: Lethal congenital anomalies where death occurred in utero (after 20 weeks); during the neonatal period; during the post neonatal period (28 days to 1 year). Queensland 1988 – 2003**



**Figure 10: Trend for Deaths due to Congenital Anomalies, Queensland 1988-2003**



Note : For each year the numerator is the total number of deaths caused by congenital anomalies up to one year of age and the denominator is the number of births where the mothers residence is in Queensland.

Over the period 1988-2003, there was no significant trend in the death rate due to congenital anomalies (Annual percentage change of -0.7%; 95% CI -1.6% +0.2%).

## Deaths Due To Congenital Anomalies

Table 3: Deaths due to congenital anomalies, Queensland 1988-2003

Year of Birth	Stillbirths <sup>a</sup>	Total still-births	Percentage of still births due to anomalies	Neonatal deaths <sup>a</sup>	Total Neo-natal deaths	Percentage of Neo-natal deaths due to anomalies	Post neonatal deaths <sup>b</sup>	Total Post Neo-natal Deaths	Percentage of Post Neo-natal deaths due to anomalies
1988-1999	538	3927	13.7%	756	2455	30.8%	280	1368	20.5%
2000	67	358	18.7%	57	184	31.0%	22	96	22.9%
2001	65	363	17.9%	51	199	25.6%	25	90	27.8%
2002	61	329	18.5%	46	177	26.0%	25	91	27.5%
2003	67	307	21.8%	54	176	30.7%	14	76	18.4%
Total	798	5284	15.1%	964	3191	30.2%	366	1721	21.3%

(a). Data of stillbirths and neonatal deaths were obtained from the Queensland Perinatal Data Collection, and only included deaths where the mother's state of residence was Queensland.

(b). Data of post neonatal deaths were obtained from the ABS Death Registration Data Set, and only included deaths that occurred in Queensland and where the mother's state of residence was Queensland.

Table 4: Top 10 lethal congenital anomalies responsible for perinatal death, Queensland 2000-2003

Congenital Anomaly	Deaths	Death rate per 10,000 births
Down syndrome	36	1.81
Anencephalus	30	1.51
Trisomy 18	30	1.51
Diaphragmatic hernia	26	1.31
Other congenital anomalies of the peripheral vascular system <sup>(a)</sup>	25	1.26
Hydrocephalus	24	1.21
Hypoplastic left heart	23	1.16
Other congenital anomalies of the brain <sup>(b)</sup>	23	1.16
Other congenital anomalies of the heart <sup>(c)</sup>	21	1.06
Cystic kidney disease	15	0.76
Total deaths – top 10 lethal anomalies	253	12.74
Total deaths - all congenital anomalies	468	23.57

(a) Other congenital malformations of the peripheral vascular system (ICD10<sup>6</sup> codes: Q27.0 – Q27.9) includes congenital absence and hypoplasia of umbilical artery, congenital renal artery stenosis, peripheral arteriovenous malformation, congenital phlebectasia, aberrant subclavian artery, congenital aneurysm, congenital stricture - artery and congenital varix.

(b) Other congenital anomalies of the brain (ICD10<sup>6</sup> codes: Q04.0 – Q04.9) includes agenesis of corpus callosum, arhinencephaly, holoprosencephaly, agyria, hydranencephaly, lissencephaly, microgyria, pachygyria, septo-optic dysplasia, megalencephaly, porencephaly, schizencephaly and macrogyria.

(c) Other congenital anomalies of the heart (ICD10<sup>6</sup> codes: Q24.0 – Q24.9) includes dextrocardia, laevocardia, cor triatriatum, pulmonary infundibular stenosis, congenital subaortic stenosis, congenital heart block, congenital diverticulum of left ventricle, congenital malformation of myocardium/pericardium, malposition of heart and Uhl's disease.

## References

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## Appendix

### Definitions

From January 1988 to July 2002, the Queensland Perinatal Data Collection coded congenital anomalies using the British Paediatric Association Classification of Diseases, a paediatric supplement compatible with the ninth revision of the WHO International Classification of Diseases, 1977. From 1st July 2002 the International Statistical Classification of Diseases and Related Health Problems, Tenth Revision, Australian Modification (ICD-10-AM) was used.

Categories of congenital anomalies used in this information circular are based on the 'sentinel' defects defined by the International Clearinghouse for Birth Defects Monitoring Systems<sup>4</sup>.

The Queensland Perinatal Data Collection defines the following terms:

#### **Baby**

"A product of conception that is born alive or if stillborn is of at least 20 weeks gestation or 400 grams in weight."

#### **Birth**

"The process by which a baby is expelled or extracted from the mother. The number of births per year is equal to the number of livebirths and stillbirths in that year."

#### **Congenital anomaly**

"A structural defect or chromosomal abnormality that is present at birth and diagnosed prior to separation from care."

#### **Neonatal death**

"The death of a live born baby within the first 28 days of life."

#### **Post Neonatal Death**

"The death of a liveborn infant between 28 days of age and the first birthday"

#### **Stillbirth**

"The complete expulsion or extraction from the mother of a product of conception of at least 20 weeks gestation or 400 grams birthweight which, after separation, did not show any signs of life, that is, did not have a heartbeat."