The Consultative Council on Obstetric and
Paediatric Mortality and Morbidity (CCOPMM)

CONGENITAL ANOMALIES IN VICTORIA 2013–2014

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1. Executive summary

The prevention and management of congenital abnormalities in the community relies on population level surveillance and research into the aetiology, diagnosis and prevention of such conditions. Congenital abnormalities are an important cause of child death and disability and are a leading cause of perinatal mortality in Australia[1], affecting 3.1 per cent of all births[2].

A 'congenital anomaly', also called a 'birth defect', 'congenital malformation' or 'congenital disorder', is any abnormality occurring before birth. This includes structural, functional, genetic, chromosomal and biochemical abnormalities. These can be detected before birth, at birth, or later years of life. It is one of the important cause of child death and disability.

Though the causes of congenital anomalies are unknown in half of all the cases, factors that contribute to congenital anomalies include genetic factors, socio-demographic factors (such as ethnicity, maternal age, and socioeconomic status), environmental factors like maternal exposure to alcohol, medications, chemicals, radiation and tobacco during pregnancy, infections like syphilis, rubella and zika virus, maternal nutritional status (such as folate deficiency), obesity and pre-gestational diabetes[3-5].

The Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM), has legislative responsibility to monitor the health outcomes of mothers and babies, and is an expert advisory body to the Victorian Government. CCOPMM maintains the Victorian Congenital Anomalies Register (VCAR) to provide ongoing health surveillance of major congenital anomalies in Victoria. The VCAR also provides data for epidemiological research and investigations into potential clusters of congenital anomalies.

This report highlights the incidence (new cases of major congenital anomalies), trends and risk of major congenital anomalies **in** Victoria between 2013 to 2014. As almost all major congenital anomalies are diagnosed by the age of six years[6], the report focuses on major anomalies in children from before birth up to six years of age. Selected major congenital anomalies of the nervous system and cardiovascular system are reported in more detail.

The reported incidence of major congenital anomalies from the VCAR has decreased from 4.3 per cent between 2007–2009 to 3.2 per cent between 2013–2014. This is likely to reflect a decline in the voluntary notifications to the VCAR rather than a true decline in rates. The department is focussing on improving the ascertainment of congenital anomaly information notified to the register.

The main findings for the reporting period are:

- **Incidence and pregnancies affected:** Major congenital anomalies affected 3.0 per cent of reported pregnancies in 2013 and 3.1 per cent in 2014. In Victoria, one in 32 reported pregnancies resulted in the development of a congenital anomaly in the baby. Among congenital anomaly cases, 15.5 per cent babies were reported to have had more than one congenital anomaly.
- **Most common congenital anomalies:** By diagnostic category, hypospadias was the most common congenital anomaly (4.3 per 1,000 pregnancies) reported between 2013 and 2014, followed by obstructive defects of the renal pelvis (2.8 per 1,000 pregnancies), and developmental dysplasia of the hip and ventricular septal defect (1.7 per 1,000 pregnancies each).
- **Preterm birth:** Babies with a congenital anomaly were three times more likely to be born preterm (birth before 37 weeks gestation) than babies without a congenital anomaly.
- **Maternal age:** Women aged 35 years and older were nine per cent more likely to have a baby with a congenital anomaly than those younger than 35 years of age.
- **Aboriginal women:** Aboriginal women were 31.0 per cent more likely to have a baby with a congenital anomaly as compared to non-Aboriginal women.

- **Obesity:** Obese women (BMI 35 kg/m^2 or more) were almost twice as likely to have a baby with a congenital anomaly as compared to non-obese women. Obesity was found to be associated with major anomalies including hydrocephalus, cleft palate, tetralogy of fallot and exomphalos.
- **Pre-gestational diabetes:** Women with pre-gestational diabetes (development of diabetes before pregnancy) were 69.0 per cent more likely to have a baby with a congenital anomaly as compared to women not having pre-gestational diabetes. Pre-gestational diabetes was found to be associated with major anomalies including hypospadias, tetralogy of fallot, ventricular septal defect, renal agenesis, cleft lip and palate and diaphragmatic hernia.
- **Socioeconomic status:** Lowest socioeconomic quintile increased the risk of having a baby with a congenital anomaly by eight per cent.
- **All neural tube defects:** Incidence of all neural tube defects decreased significantly from 1.1 per 1,000 pregnancies in 2007 and 2009 to 0.6 per 1,000 pregnancies in 2013 and 2014, indicating the impact of interventions such as mandatory folic acid fortification of bread, introduced in Victoria in 2009.

2. Reporting of congenital anomalies in Victoria

2.1 Victorian congenital anomalies register

The Victorian Congenital Anomalies Register (VCAR), formerly known as the Victorian Birth Defects Register (VBDR), was established in 1982 under the *Health Act 1981*[7–12]. The VCAR is a legislative responsibility of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM). CCOPMM also maintains Victorian perinatal data collection (VPDC) for all births in Victoria and mortality database for mothers and children in Victoria.

VCAR is a population based surveillance system that collects information on all major congenital anomalies for live births, stillbirths, terminations. Congenital anomalies notification for children up to six years is collected as it has been reported that nearly 60.0 per cent of all major congenital anomalies are diagnosed during the first week of life, 70.0 per cent by the first month, 90.0 per cent by the first year, and 100.0 per cent by the sixth year[6].

- Data from the VCAR data can assist with:
- planning and provision of healthcare for people with congenital anomalies
- provision of advice to the community/families who are concerned about having a baby with a congenital anomaly
- epidemiological research to increase knowledge in the aetiology and preventability of congenital anomalies
- assessing the effectiveness of primary prevention and screening programs
- responding to concerns about potential clusters or trends in congenital anomalies

2.2 Congenital anomalies: major and minor

A ‘congenital anomaly’, also called a ‘birth defect’, ‘congenital malformation’ or ‘congenital disorder’, is any abnormality of prenatal origin, either present after conception or occurring before the end of pregnancy. This includes structural, functional, genetic, chromosomal and biochemical abnormalities, and can be detected before birth, at birth or in later years of life[3].

This report focuses on major congenital anomalies, which have significant medical, social or cosmetic consequences, typically require a medical intervention and are responsible for most of the deaths, morbidity and disability related to congenital anomalies. These are shown in Appendix A.

Minor congenital anomalies (structural changes without significant health, social or cosmetic consequences) have been excluded from this report. Minor congenital anomalies have mainly been derived from the ‘external minor congenital anomalies list’ provided by the World Health Organization, United States Centers for Disease Control and Prevention (CDC) and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR)[13] (Appendix B).

2.3 Sources of notifications

For the period 2013 to 2014, there were 7,875 congenital anomalies cases notified to VCAR, of which 2,859 were minor anomaly cases (Figure 1). A total of 6,298 major anomalies were notified in 5,016 major anomaly cases. The sources of notifications are shown in Table 1.

Figure 1: Congenital anomaly cases in Victoria in 2013–2014

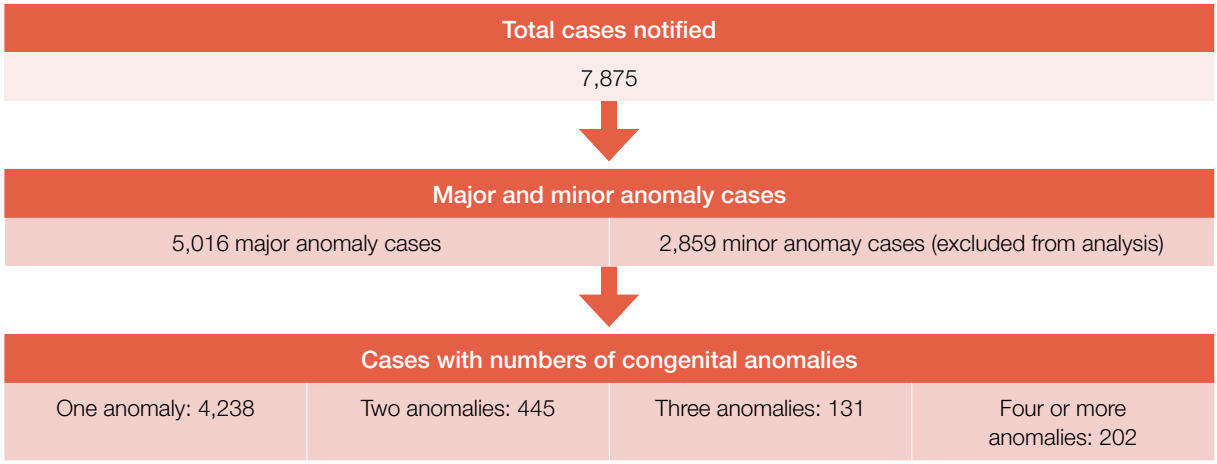


Table 1: Sources of notifications for congenital anomaly cases in Victoria in 2013–2014

Notification source	Number	Percentage
Victorian Perinatal Data Collection (VPDC) – Birth forms	6,451	81.9
Hospital source	1,124	14.3
Maternal and child health nurse	224	2.8
Other professionals (doctor, nurses)	72	0.9
Cytogenetic report	1	0.0
Unknown	3	0.0
Total	**7,875**	**100.0**

Notification about congenital anomalies can be submitted to VCAR through its online form:

https://www2.health.vic.gov.au/hospitals-and-health-services/quality-safety-service/consultative-councils/council-obstetric-paediatric-mortality/congenital-anomalies-register

The same case can be notified through multiple sources and is encouraged as this increases case ascertainment.

2.4 Data items

All notifications of congenital anomalies (excluding terminations of pregnancy before 20 weeks gestation and interstate births) are linked to the VPDC to access the obstetric history for each case. Health services and maternity care providers must provide data to the VPDC for every birth in Victoria. The routinely collected data items included in the VCAR are listed in Appendix C, with additional data items available for each case over 20 weeks' gestation as reported in the VPDC[14].

2.5 Data quality

Data submitted to the VPDC and the VCAR are checked for completeness and validity. Inconsistent or incomplete data is rectified by sending a query to the hospital of birth, or home birth practitioner. Further data cleaning is carried out when all data for the calendar year has been submitted. Validation activities to assess, maintain and improve the quality of the data provided by hospitals to the VPDC and the VCAR are an ongoing commitment of the Department of Health and Human Services (DHHS).

Projects designed to determine the accuracy and completeness of the data submitted to the VPDC and the VCAR are undertaken periodically[15–19], however it is acknowledged that this should be undertaken more regularly. Congenital anomalies reported in termination of pregnancy less than 20 weeks has challenges in ascertainment as all of them are not reported to the VCAR as congenital anomalies notifications is not mandatory.

2.6 Data analysis

The 2013–2014 Victorian congenital anomaly rates are reported per 1,000 pregnancies* for:

- all major congenital anomalies
- major congenital anomalies by maternal and child characteristics
- major congenital anomalies by diagnostic categories.

Trends of major congenital anomalies are reported as well as findings on selected major congenital anomalies of nervous and cardiovascular systems by rate, trends and selected maternal and child characteristics. Relative risk (RR) and its 95 per cent confidence intervals (CI) have been calculated to measure the association of maternal and child characteristics with all congenital anomalies and selected anomalies. A p-value of less than 0.05 was considered significant.

* Includes live birth, stillbirth and termination of pregnancy at any gestation

3. Incidence of congenital anomalies in Victoria

Of all reported pregnancies, 3.0 per cent were affected by at least one congenital anomaly in 2013 and 3.1 per cent in 2014. This translates to an incidence of 32.0 per 1,000 pregnancies in 2013 and 31.4 per 1,000 pregnancies in 2014. This is consistent to the incidence of congenital anomalies in Australia (2002–03). No published national or jurisdictional data was present for congenital anomalies in 2013–2014 for comparison. In Victoria, one in 32 reported pregnancies was affected by one or more congenital anomalies. Of all the congenital anomaly cases reported as terminations of pregnancy before 20 weeks, 48.3 per cent (n = 195) had chromosomal anomalies.

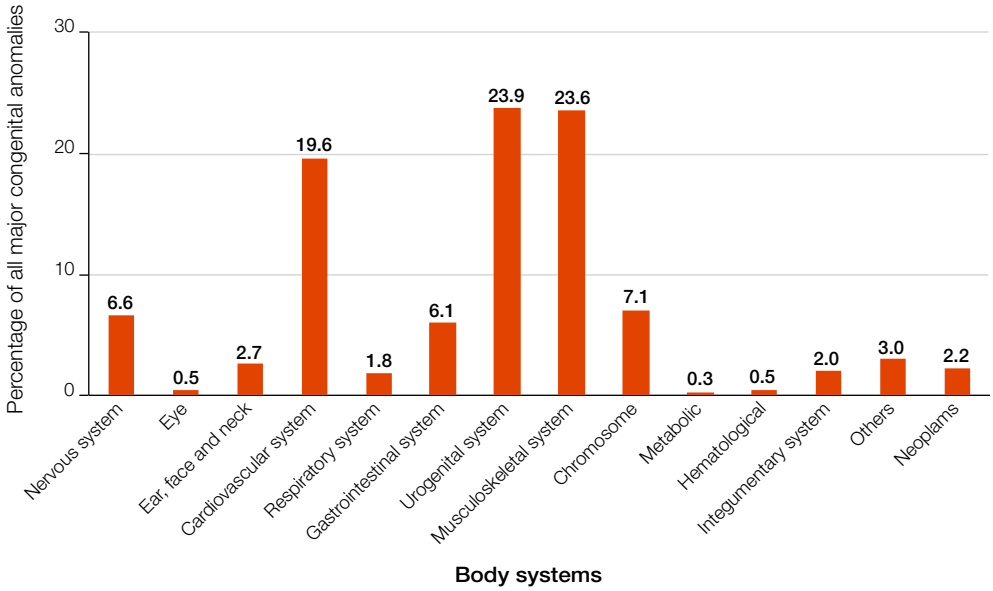
4. Trends of congenital anomalies in Victoria

Incidence of major congenital anomalies have declined from 4.3 per cent of reported pregnancies between 2007 and 2009 to 3.2 per cent of reported pregnancies between 2013 and 2014 (Appendix D). This reflects decrease in notifications of congenital anomalies to VCAR.

5. Congenital anomalies by body systems

Among all the congenital anomalies (not cases), anomalies of the urogenital system were the most common (23.9 per cent), followed by anomalies of the musculoskeletal system (23.6 per cent) and cardiovascular system (19.6 per cent). The proportion of congenital anomaly by body systems is shown in Figure 2. Appendix A provides details on the major anomalies according to each body system.

Figure 2: Congenital anomalies in Victoria by body systems, 2013–2014



6. Congenital anomalies by diagnostic categories

Hypospadias was the most common congenital anomaly reported by diagnostic category, followed by obstructive defects of the renal pelvis and developmental dysplasia of the hip and ventricular septal defect in 2013–2014 (Table 2).

Table 2: Order of incidence of selected congenital anomalies, 2013–2014

Congenital anomaly	N/1,000 pregnancies	1 in x number of pregnancies
Hypospadias	4.3	234
Obstructive defects of the renal pelvis	2.8	357
Developmental dysplasia of the hip	1.7	579
Ventricular septal defect	1.7	588
Trisomy 21	1.2	803
Hydrocephalus	0.8	1,296
All neural tube defects	0.6	1,700
Cystic kidney disease	0.6	1,797
Renal agenesis and dysgenesis	0.5	2,166
Cleft palate	0.5	2,196
Limb reduction defects	0.4	2,635
Cleft lip and palate	0.4	2,680
Tetralogy of fallot	0.4	2,824
Trisomy 18	0.3	3,041
Transposition of great vessels	0.3	3,041
Diaphragmatic hernia	0.3	3,162
Cleft lip	0.3	3,294
Gastroschisis	0.3	3,437
Anencephaly	0.3	3,514
Exomphalos	0.3	3,514
Hypoplastic left heart syndrome	0.3	3,765
Spina bifida	0.2	4,161
Small intestinal atresia and/or stenosis	0.2	5,101
Trisomy 13	0.2	5,856
Anorectal atresia and/or stenosis	0.2	6,082
Coarctation of aorta	0.2	6,588
Microcephalus	0.1	11,294
Encephalocele	0.1	12,163
Oesophageal atresia and/or stenosis	0.1	13,177

7. Congenital anomalies by child's characteristics

7.1 Gender

For all congenital anomaly cases reported between 2013 and 2014, 57.7 per cent were males and 37.7 per cent were females; sex was indeterminate in 0.5 per cent and unknown for 4.2 per cent. The rate of congenital anomalies was 35.7 per 1,000 pregnancies for male babies and 24.6 per 1,000 pregnancies for female babies. Male babies were 45.0 per cent more likely to have a congenital anomaly than female babies (RR 1.45, 95 per cent CI 1.37–1.53, p-value < 0.0001).

7.2 Gestational age

Between 2013 and 2014, babies with a congenital anomaly were three times more likely to be born preterm (less than 37 weeks gestation) than those not having a congenital anomaly (RR 3.18, 95 per cent CI 2.99 to 3.40, p-value < 0.0001). As the gestational age increased, rate of congenital anomalies decreased (Table 3).

Table 3: Congenital anomalies by gestational age

Gestational Age (weeks)*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies/ 1,000 pregnancies
20–27	326	1,725	189.0
28–31	115	1,260	91.3
32–36	499	10,692	46.7
37–41	3,642	142,627	25.5
> 41	23	1,120	20.5

* Excludes terminations less than 20 weeks and those in whom gestational age was not stated

7.3 Birth weight

The rate of congenital anomalies in babies weighing less than 1,000 grams at birth was 185.8 per 1,000 pregnancies and it decreased with increasing birth weight (Table 4). For low birthweight babies, the risk of having a congenital anomaly was three times higher than for babies weighing 2,500 grams or more (RR 3.01, 95 per cent CI 2.81–3.24, p-value < 0.0001).

Table 4: Congenital anomalies by birth weight

Weight in grams*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies/ 1,000 pregnancies
< 1000	334	1,798	185.8
1000–2499	546	9,579	57.0
≥ 2500	3,724	146,033	25.5

* Excludes terminations less than 20 weeks and those cases in whom weight in grams was not stated

7.4 Birth plurality

By birth plurality, 89.0 per cent of congenital anomaly cases were singleton and three per cent were multiple births and plurality was unknown for eight per cent. In 2013–2014, the rate of congenital anomaly in singleton births was 29.0 per 1,000 pregnancies and 39.2 per 1,000 pregnancies for multiple births. The risk of congenital anomaly was 35.0 per cent higher in multiple births than singleton births (RR 1.35, 95 per cent CI 1.14 to 1.58, p-value 0.0003). Assisted reproductive technology (ART) might have contributed to higher rate of congenital anomalies in multiple births. In medical literature, while some studies supported the positive association of ART with major congenital anomalies[20], other studies found no association[21,22].

7.5 Perinatal outcomes

Eight per cent (n = 404) of all reported pregnancies with congenital anomalies terminated before 20 weeks gestation and five per cent (n = 255) terminated at 20 weeks or more. Of babies born with a congenital anomaly, 5.6 per cent (n = 281, 70.4 per cent terminations) were stillbirths, 2.3 per cent (n = 117,53.8 per cent terminations) were neonatal deaths and 0.2 per cent (n = 10) were infant deaths. Appendix E shows outcomes for selected major anomalies.

Perinatal mortality rate in babies with one or more congenital anomalies was 79.3 per 1,000 pregnancies as compare to perinatal mortality rate of 9.4 per 1,000 pregnancies in babies not having a congenital anomaly. When adjusted for terminations, perinatal mortality rate in babies with one or more congenital anomalies decreased to 27.3 per 1,000 pregnancies.

8. Congenital anomalies by maternal characteristics

8.1 Age

Between 2013 and 2014, women aged 35 years and older were nine per cent more likely to have a baby with a congenital anomaly than those younger than 35 years of age (RR 1.09, 95 per cent CI 1.02–1.16, p-value 0.007). The rate of congenital anomalies was highest in women aged 40 to 44 years, followed by those younger than 20 years of age (Table 5).

Table 5: Congenital anomalies by mother's age at birth

Age group*	Congenital anomaly cases	Total pregnancies	Congenital anomalies/ 1,000 pregnancies
< 20	110	3,345	32.8
20–24	526	16,694	31.5
25–29	1,207	41,621	29.0
30–34	1,585	56,767	27.9
35–39	945	31,482	30.0
40–44	294	7,733	38.0

* Excludes those cases in whom neither age nor date of birth was stated

8.2 Aboriginal women

Among all the congenital anomaly cases reported, women identified themselves as Aboriginal in 1.5 per cent cases. Aboriginal women were 31.0 per cent more likely to have a baby with a congenital anomaly as compared to non-Aboriginal women (RR 1.31, 95 per cent CI 1.05 to 1.63, p-value 0.014). The rate of congenital anomalies was 38.2 per 1,000 pregnancies in Aboriginal women as compared to 29.0 per 1,000 pregnancies in non-Aboriginal women.

8.3 Parity

The rate of congenital anomaly was 30.2 per 1,000 pregnancies in primiparous women and 28.5 per 1,000 pregnancies in multiparous women between 2013 and 2014. For primiparous women, the risk of having a baby with congenital anomaly was six per cent higher than for multiparous women (RR 1.06, 95 per cent CI 1.00–1.12, p-value 0.047).

8.4 Country of birth

The rate of congenital anomalies was highest for babies of women born in the North Africa and the Middle East in 2013–2014 (Table 6). Babies of women born in North Africa and the Middle East were 22.0 per cent more likely to develop a congenital anomaly than babies of women born in Australia and other countries (RR 1.22, 95 per cent CI 1.05–1.41, p-value 0.006).

Table 6: Congenital anomalies by mother's country of birth

Mother's country of birth	Number of cases with at least one congenital anomaly	Number of reported pregnancies	Congenital anomalies/1,000 pregnancies
Australia	3,024	102,462	29.5
Oceania and Antarctica (Excludes Australia)	126	4,438	28.4
North-West Europe	140	4,598	30.4
Southern and Eastern Europe	84	2,945	28.5
North Africa and The Middle East	185	5,201	35.6
South-East Asia	244	9,732	25.1
North-East Asia	209	7,538	27.7
Southern and Central Asia	414	14,164	29.2
Americas	53	2,138	24.8
Sub-Saharan Africa	112	3,286	34.1
Not Stated/inadequately described	425	1,617	262.8

8.5 Obesity

Between 2013 and 2014, obese women (BMI ≥ 35 kg/m^2) were almost twice as likely to have a baby with a congenital anomaly as compared to non-obese women (RR 1.96, 95 per cent CI 1.82 to 2.09, p-value < 0.0001). The rate of congenital anomalies in women having a body mass index (BMI) of 35 kg/m^2 and above was 50.5 per 1,000 pregnancies as compared to 25.8 per 1,000 pregnancies in women having BMI of less than 35 kg/m^2. Table 7 shows the rates of selected major anomalies in obese women.

Table 7: Selected major congenital anomalies in obese women

Selected congenital anomaly	Congenital anomalies/ 1,000 pregnancies	RR and 95 per cent CI
Hypospadias	4.5	1.06 (0.69–1.57)
Obstructive defects of renal pelvis	2.3	0.96 (0.64–1.45)
Developmental dysplasia of hip	1.5	0.79 (0.48–1.32)
Ventricular septal defect	1.7	1.31 (0.81–2.10)
Trisomy 21	0.6	1.01 (0.46–2.19)
Hydrocephalus	1.1	2.54 (1.36–4.75)*
All neural tube defects	0.5	1.48 (0.58–3.76)
Cystic kidney disease	0.8	1.91 (0.94–3.86)
Renal agenesis	0.4	1.16 (0.41–3.23)
Cleft palate	1	2.61 (1.36–5.02)*
Limb reduction defect	0.3	1.15 (0.35–3.76)
Cleft lip and palate	0.3	1.04 (0.32–3.41)
Tetralogy of Fallot	0.6	2.97 (1.29–6.80)*
Trisomy 18	0	0.38 (0.02–6.41)
Transposition of great vessels	0.4	1.98 (0.68–5.71)
Diaphragmatic Hernia	0.1	0.41 (0.05–3.01)
Cleft lip	0.1	0.38 (0.05–2.81)
Gastroschisis	0	0.25 (0.01–4.16)
Anencephaly	0.1	0.91 (0.11–6.99)
Exomphalos	0.5	3.96 (1.44–10.90)*
Hypoplastic left heart syndrome	0.4	2.26 (0.77–6.59)
Spina bifida	0.2	2.26 (0.77–6.59)
Small intestinal atresia	0.2	1.18 (0.27–5.08)
Trisomy 13	0	1.32 (0.07–24.54)
Anorectal atresia and/or stenosis	0.5	1.08 (0.13–8.37)
Coarctation of aorta	0.1	0.91 (0.11–6.99)
Microcephaly	0.2	3.39 (0.70–16.35)
Encephalocele	0	0.69 (0.04–12.12)
Oesophageal atresia and/or stenosis	0.1	1.08 (0.13–8.37)

*Shows statistically significant association

8.6 Pre-gestational diabetes

Between 2013 and 2014, women having pre-gestational diabetes were 69.0 per cent more likely to have a baby with a congenital anomaly as compared to women not having pre-gestational diabetes (RR 1.69, 95 per cent CI 1.33 to 2.15, p-value < 0.0001).The rate of congenital anomalies in women having pre-gestational diabetes was 53.6 per 1,000 pregnancies as compared to 31.6 per 1,000 pregnancies in women not having pre-gestational diabetes. Table 8 shows the rates of selected major anomalies in mothers having pre-gestational diabetes.

Table 8: Selected major congenital anomalies in women having pre-gestational diabetes

Selected Congenital anomaly	Congenital anomalies/ 1,000 pregnancies	RR and 95 per cent CI
Hypospadias	16	3.82 (2.05–7.14)*
Obstructive defects of renal pelvis	2.5	0.92 (0.29–2.87)
Developmental dysplasia of hip	0	0.23 (0.01–3.84)
Ventricular septal defect	7.5	4.54 (2.34–8.81) *
Trisomy 21	0.83	0.66 (0.09–4.77)
Hydrocephalus	1.7	2.22 (0.55–8.99)
All neural tube defects	0.8	1.42 (0.19–10.23)
Cystic kidney disease	0	0.74 (0.04–11.93)
Renal agenesis	2.5	5.62 (1.77–17.84) *
Cleft palate	0.8	1.84 (0.25–13.29)
Limb reduction defect	0.8	2.67 (0.37–19.38)
Cleft lip and palate	3.3	9.54 (3.46–26.30) *
Tetralogy of Fallot	3.3	10.09 (3.65–27.87) *
Trisomy 18	0	1.24 (0.07–20.22)
Transposition of great vessels	0.8	2.57 (0.35–18.61)
Diaphragmatic Hernia	1.7	5.46 (1.33–22.48) *
Cleft lip	0	1.38 (0.08–22.38)
Gastroschisis	0	1.41 (0.08–22.87)
Anencephaly	0.8	2.98 (0.41–21.63)
Exomphalos	0.8	3.05 (0.42–22.15)
Hypoplastic left heart syndrome	0.8	3.2 (0.44–23.25)
Spina bifida	0	1.70 (0.10–27.71)
Small intestinal atresia	0.8	4.37 (0.59–32.06)
Trisomy 13	0	2.38 (0.14–39.07)
Anorectal atresia and/or stenosis	0	2.47 (0.15–40.58)
Coarctation of aorta	0	2.67 (0.16–43.99)
Microcephaly	0	4.52 (0.27–75.78)
Encephalocele	0	4.85 (0.28–81.67)
Oesophageal atresia and/or stenosis	0	5.24 (0.31–88.56)

*Shows statistically significant association

8.7 Socioeconomic status

The rate of congenital anomalies for women in the lowest socioeconomic quintile was 31.2 per 1,000 pregnancies as compared to 27.7 per 1,000 pregnancies in the highest quintile (Table 9). Women in the lowest socioeconomic quintile were eight per cent more likely to have a baby with congenital anomaly as compared to the higher quintiles (RR 1.08, 95 per cent CI 1.01 to 1.15, p-value 0.03).

Table 9: Congenital anomalies by mother's socioeconomic status

Socioeconomic status quintiles*	Congenital anomaly cases	Total pregnancies	Congenital anomalies/ 1,000 pregnancies
1	933	29,858	31.2
2	912	29853	30.5
3	843	29,851	28.2
4	877	29,858	29.4
5	826	29,847	27.7

*Socioeconomic status measured as SEIFA index as measured by the Australian Bureau of statistics

9. Selected major congenital anomalies

9.1 Anencephaly

Definition: Anencephaly is the total or partial absence of the cranial vault, covering skin and the brain tissue.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 0.9 per cent (n = 45) of babies had anencephaly, of which 87.0 per cent (n = 39) were isolated and 13.0 per cent (n = 6) occurred with other congenital anomalies.

Trend: From 1998 to 2014 there was no significant change (p-value 0.35) in the incidence of anencephaly (Figure 3).

Risk factors: The risk of anencephaly did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 10).

Figure 3: Anencephaly per 1,000 pregnancies, 1998 to 2014

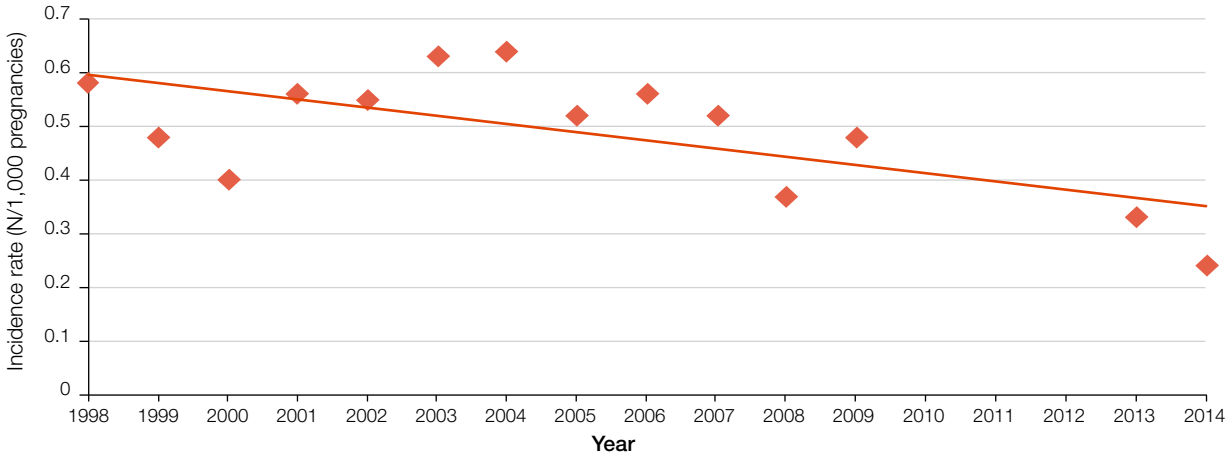


Table 10: Anencephaly by selected maternal and child characteristics, 2013 and 2014

Characteristic		Anencephaly/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.13 0.18	- 1.41 (0.57–3.46)
Plurality	Single (Ref) Multiple	0.1 0.5	- 5.24 (1.20–22.78)
Gender	Male (Ref) Female	0.1 0.1	- 0.60 (0.23–1.55)
Pre-gestational diabetes	No (Ref) Yes	0.3 0.8	- 2.98 (0.41–21.63)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.1	- 0.91 (0.12–6.99)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.1	0.86 (0.24–2.98) -

9.2 Spina Bifida

Definition: Spina bifida is the herniation or exposure of the spinal cord and/or meninges through incomplete closure of the spine. Hydrocephalus may or may not be present.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 0.8 per cent (n = 38) of babies had spina bifida, of which 68.4 per cent (n = 26) were isolated and 31.6 per cent (n = 12) occurred with other congenital anomalies.

Trend: From 1998 to 2009 there was no significant change (p-value 0.578) in the incidence of spina bifida. The rate of spina bifida significantly decreased (p-value 0.01) in 2013 and 2014 (Figure 5), which may partly reflect decrease in ascertainment and notifications to VCAR, but may also indicate the impact of interventions like mandatory folic acid fortification of bread which was introduced in Victoria in 2009.

Risk factors: The risk of spina bifida did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 11).

Figure 4: Spina bifida per 1,000 pregnancies, from 1998 to 2014

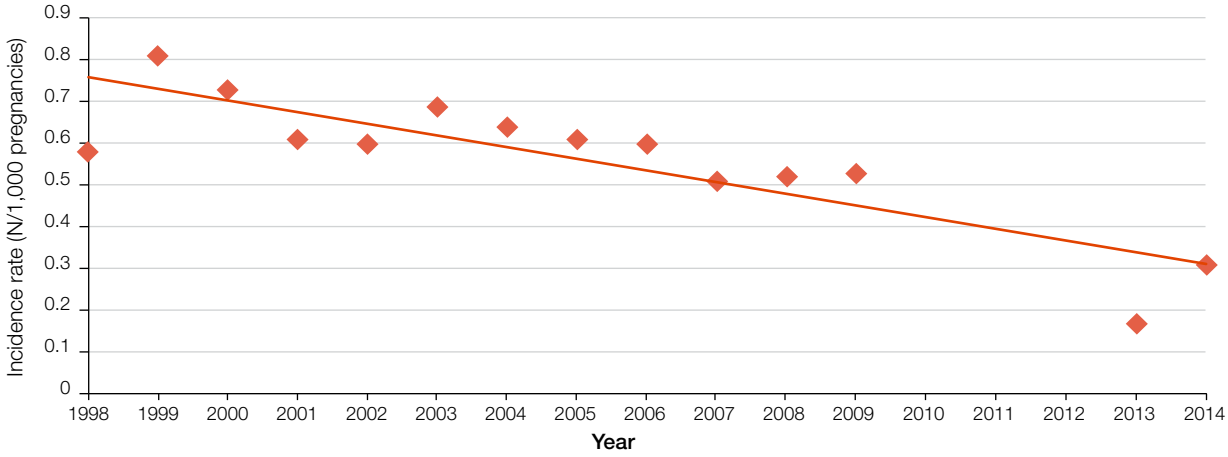


Table 11: Spina bifida by selected maternal and child characteristics, 2013 and 2014

Characteristic		Spina bifida/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.22 0.18	- 1.23 (0.53–2.83)
Plurality	Single (Ref) Multiple	0.2 0.0	- 0.62 (0.03–10.21)
Gender	Male (Ref) Female	0.2 0.2	- 1.01 (0.51–1.99)
Pre-gestational diabetes	No (Ref) Yes	0.0 0.2	- 1.70 (0.10–27.71)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.4 0.2	- 2.26 (0.77–6.59)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.2	0.43 (0.13–1.41) -

9.3 Encephalocele

Definition: Encephalocele is the herniation of the brain and/or meninges through a defect in the skull.

Incidence: Of all congenital anomaly cases reported between 2013 and 2014, 0.3 per cent (n = 13) of babies had encephalocele, of which 69.2 per cent (n = 9) were isolated and 30.8 per cent (n = 4) occurred with other congenital anomalies.

Trend: Incidence rates of encephalocele did not change significantly (p-value 0.35) from 1998 to 2009 (Figure 5).

Risk factors: The risk of encephalocele did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 12).

Figure 5: Encephalocele per 1,000 pregnancies, from 1998 to 2014

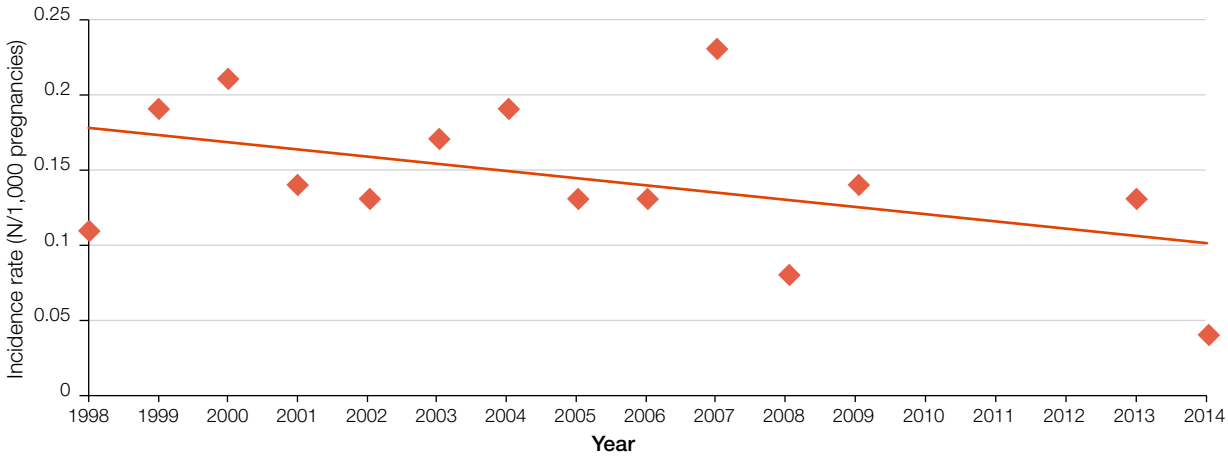


Table 12: Encephalocele by selected maternal and child characteristics, 2013 and 2014

	Characteristic	Encephalocele/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0. 06 0.1	- 1.72 (0.50–5.89)
Plurality	Single (Ref) Multiple	0.1 0.3	- 4.19 (0.53–32.74)
Gender	Male (Ref) Female	0.1 0.1	- 1.42 (0.40–5.04)
Pre-gestational diabetes	No (Ref) Yes	0 0.1	- 4.85 (0.28–81.67)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0	- 0.69 (0.04–12.12)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.1	0.88 (0.19–4.11) -

9.4 All neural tube defects

Definition: All cases of anencephaly, spina bifida and encephalocele.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 1.9 per cent (n = 93) of babies had neural tube defect, of which around 79.6 per cent (n = 74) were isolated and 20.4 per cent (n =19) occurred with other congenital anomalies.

Trend: The incidence of neural tube defects decreased from 1.1 per 1,000 pregnancies in 2007 and 2009 to 0.6 per 1,000 pregnancies in 2013 and 2014 (p-value 0.007) (Figure 6), which may partly reflect decrease in ascertainment and notifications to VCAR before 20 weeks gestation. The decrease in incidence remained significant (p-value 0.006), even for notifications received at 20 weeks and more (Figure 7), indicating reduction due to interventions like mandatory folic acid fortification of bread introduced in Victoria in 2009. This is comparable to the declining trends in South Australia[23], Western Australia[24] and Queensland[25].

Risk factors: The risk of neural tube defects did not did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 13).

Figure 6: All neural tube defects per 1,000 pregnancies from 1998 to 2014 for any gestation

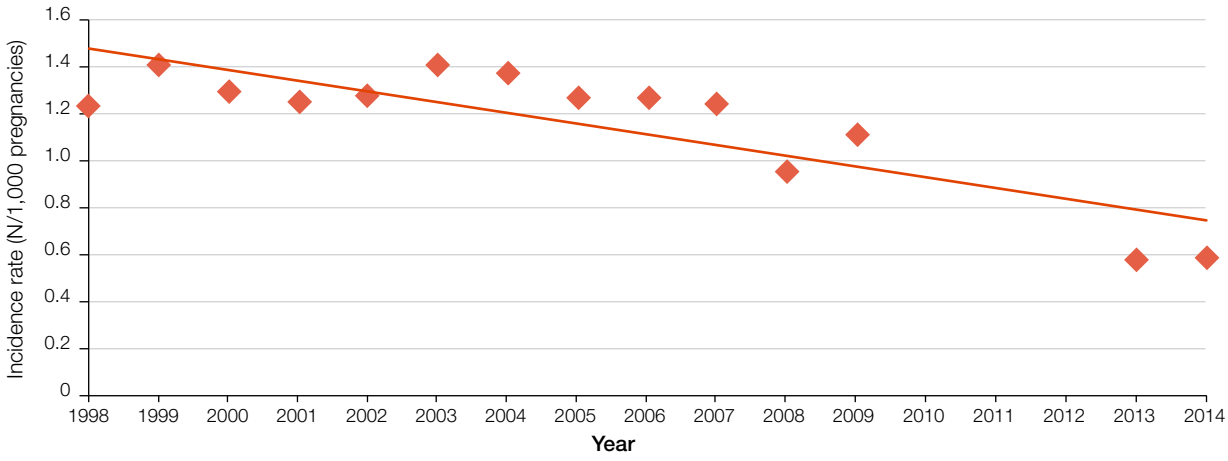


Figure 7: All neural tube defects per 1,000 pregnancies from 1998 to 2014 for gestation ≥ 20 weeks

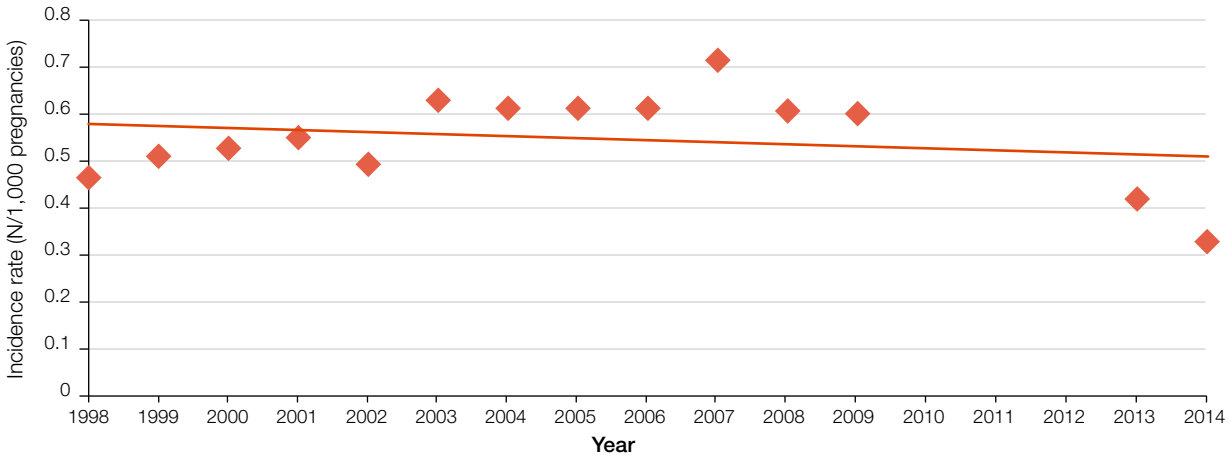


Table 13: All neural tube defects by selected maternal and child characteristics, 2013 and 2014

Characteristic		All neural tube defects/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.4 0.5	- 1.18 (0.68–2.03)
Plurality	Single (Ref) Multiple	0.4 0.8	- 2.20 (0.69–7.04)
Gender	Male (Ref) Female	0.4 0.4	- 1.98 (0.55–1.52)
Pre-gestational diabetes	No (Ref) Yes	0.6 0.8	- 1.42 (0.19–10.23)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.3 0.5	- 1.48 (0.58–3.76)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.4	0.65 (0.30–1.37) -

9.5 Microcephalus

Definition: Microcephalus is the presence of a small cranium defined by an occipito-frontal circumference three standard deviations below the age-sex appropriate distribution curves.

Incidence: Of all congenital anomaly cases reported between 2013 and 2014, 0.3 per cent (n = 14) of babies had microcephalus, of which 50.0 per cent (n = 7) were isolated and 50.0 per cent (n = 7) occurred with other congenital anomalies.

Trend: The incidence of microcephalus significantly decreased from 0.3 per 1,000 pregnancies in 2007 and 2009 to 0.1 per 1,000 pregnancies in 2013 and 2014 (p-value 0.003) (Figure 8), which may reflect decrease in ascertainment and notifications to the VCAR.

Risk factors: The Risk of microcephalus did not did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 14).

Figure 8 : Microcephalus per 1,000 pregnancies from 1998 to 2014

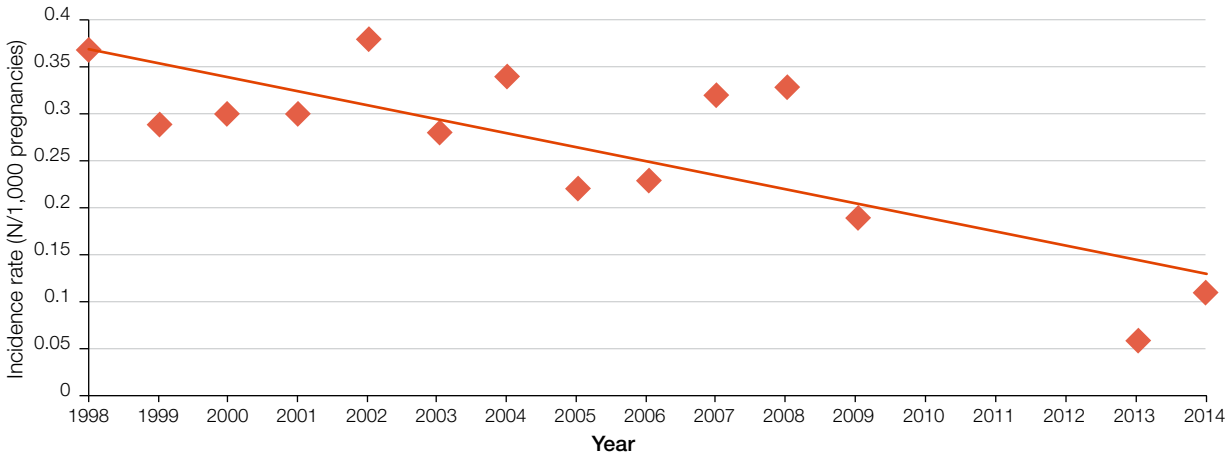


Table 14: Microcephalus by selected maternal and child characteristics, 2013 and 2014

Characteristic		Microcephalus/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.08 0.08	- 1.10 (0.30–4.01)
Plurality	Single (Ref) Multiple	0.1 0	- 1.55 (0.09–26.10)
Gender	Male (Ref) Female	0.1 0.1	- 0.81 (0.27–2.42)
Pre-gestational diabetes	No (Ref) Yes	0 0.1	- 4.52 (0.27–75.78)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.2	- 3.39 (0.70–16.35)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.1	0.72 (0.16–3.28) -

9.6 Hydrocephalus

Definition: Hydrocephalus is the dilatation of the cerebral ventricles (not associated with primary brain atrophy) with or without enlargement of the head. Diagnosed at birth, these cases exclude hydrocephalus associated with spina bifida or encephalocele.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 2.4 per cent (n = 122) of babies had hydrocephalus, of which 55.7 per cent (n = 68) were isolated and 44.3 per cent (n = 54) occurred with other congenital anomalies.

Trend: From 1998 to 2014, the incidence rate of hydrocephalus did not change significantly (p-value 0.138) (Figure 9).

Risk factors: Risk of hydrocephalus increased by 59.0 per cent in women aged 35 years and older, 2.5 times in obese women, twice in women in lowest socioeconomic strata and 4.4 times for multiple births (Table 15).

Figure 9: Hydrocephalus per 1,000 pregnancies from 1998 to 2014

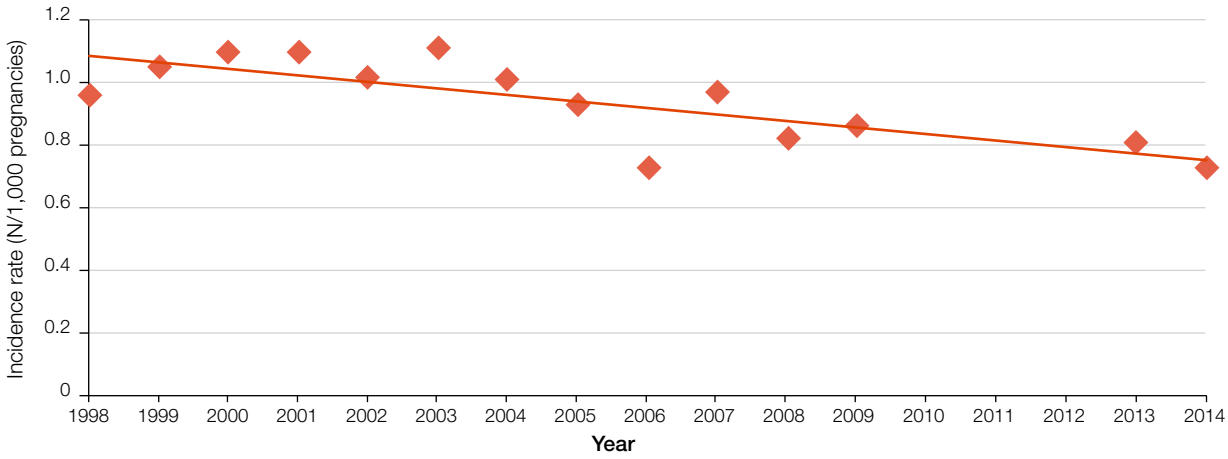


Table 15: Hydrocephalus by selected maternal and child characteristics, 2013 and 2014

Characteristic		Hydrocephalus/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.6 0.9	- 1.59 (1.08–2.34)*
Plurality	Single (Ref) Multiple	0.7 3.0	- 4.48 (2.41–8.34)*
Gender	Male (Ref) Female	0.8 0.6	- 1.41 (0.96–2.06)
Pre-gestational diabetes	No (Ref) Yes	0.8 1.7	- 2.22 (0.55–8.99)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.4 1.1	- 2.54 (1.36–4.75)*
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	1.2 0.6	2.05 (1.38–3.05)* -

*Shows statistically significant association

9.7 Transposition of the great vessels

Definition: In this anomaly, the aorta exits from the right ventricle and the pulmonary artery from the left ventricle, with or without other cardiac defects. It includes double outlet right ventricle, so-called corrected transposition.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 1.0 per cent (n = 52) had transposition of the great vessels, of which 53.9 per cent (n = 28) were isolated and 46.1 per cent (n = 24) occurred with other congenital anomalies.

Trend: There was no significant change (p-value 0.07) in incidence rates from 1998 to 2009 (Figure 10).

Risk factors: Male babies were 2.5 times more likely to develop transposition of the great vessels as compared to the female babies. Risk of transposition of the great vessels did not vary with maternal age, multiple births, pre-gestational diabetes, obesity or socioeconomic status (Table 16).

Figure 10: Transposition of the great vessels per 1,000 pregnancies from 1998 to 2014

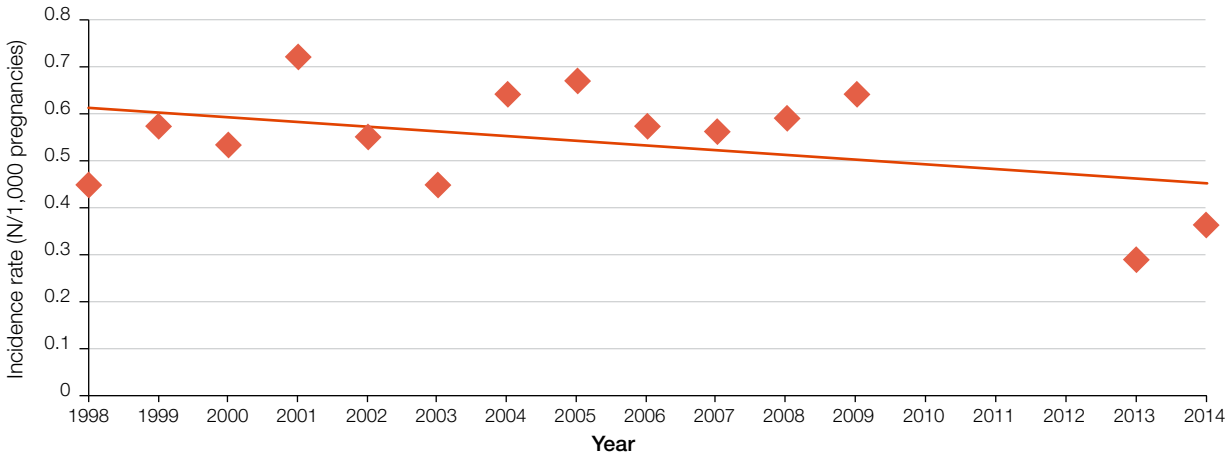


Table 16: Transposition of the great vessels by selected maternal and child characteristics, 2013 and 2014

Characteristic		Transposition of great vessels/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 0.2	- 1.35 (0.68–2.70)
Plurality	Single (Ref) Multiple	0.3 0.3	- 0.83 (0.11–6.06)
Gender	Male (Ref) Female	0.4 0.2	2.27 (1.24–4.16)* -
Pre-gestational diabetes	No (Ref) Yes	0.3 0.8	- 2.57 (0.35–18.61)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.4	- 1.98 (0.68–5.71)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.3	0.97 (0.46–2.01) -

*Shows statistically significant association

9.8 Tetralogy of Fallot

Definition: Tetralogy of fallot is characterised by ventricular septal defect, overriding aorta, infundibular pulmonary stenosis and often right ventricular hypertrophy.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 1.1 per cent (n = 56) of babies had tetralogy of fallot, of which 62.5 per cent (n = 35) were isolated and 37.5 per cent (n=21) occurred with other congenital anomalies.

Trend: Incidence rates from 1998 to 2014 were comparable (Figure 11).

Risk factors: Risk of tetralogy of fallot increased by ten times in women having pre-gestatinal diabetes, about three times in obese women and 82.0 per cent in women in lowest socioeconomic strata (Table 17).

Figure 11: Tetralogy of Fallot per 1,000 pregnancies from 1998 to 2014

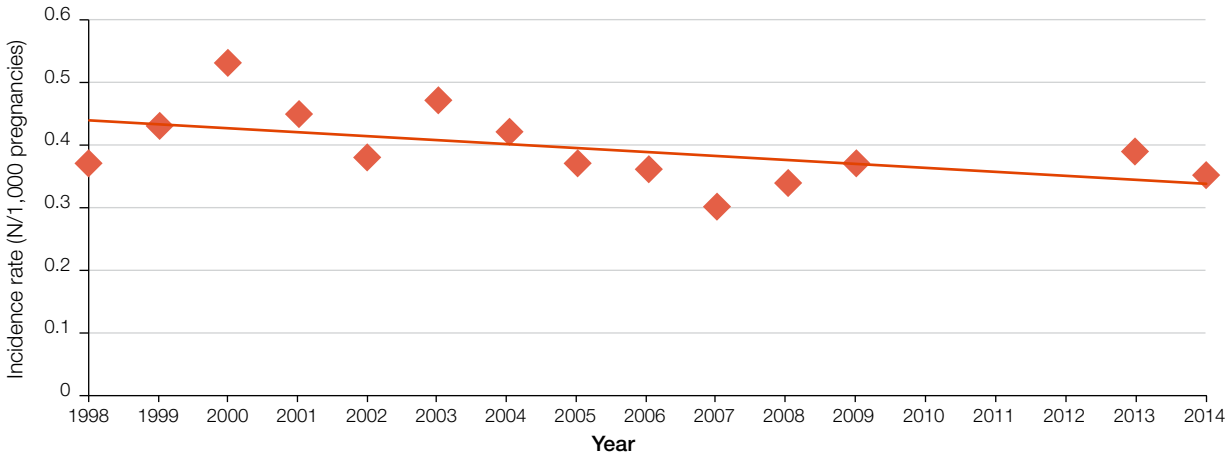


Table 17: Tetralogy of Fallot by selected maternal and child characteristics, 2013 and 2014

Characteristic		Tetralogy of Fallot/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 0.3	- 1.03 (0.56–1.89)
Plurality	Single (Ref) Multiple	0.3 0.5	- 1.58 (0.38–6.48)
Gender	Male (Ref) Female	0.4 0.3	1.42 (0.83–2.44) -
Pre-gestational diabetes	No (Ref) Yes	0.3 3.3	- 10.1 (3.65–27.87)*
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.6	- 2.97 (1.29–6.80)*
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.5 0.3	1.82 (1.01–3.30)* -

*Shows statistically significant association

9.9 Ventricular septal defect

Definition: Ventricular septal defect is characterised by a defect in the septum between the left and right ventricles of the heart, which permits blood to be shunted between them. It excludes ventricular septal defect as part of Tetralogy of Fallot.

Incidence: Of all the congenital anomaly cases reported between 2013 and 2014, 5.4 per cent (n = 269) of babies had ventricular septal defect, of which 49.4 per cent (n = 133) were isolated and 50.6 per cent (n = 136) occurred with other congenital anomalies.

Trend: Incidence rate of ventricular septal defect decreased significantly (p-value < 0.0001) from 1998 to 2014 (Figure 12).

Risk factors: Risk of ventricular septal defect increased by 3.2 times in multiple births and 4.5 times in women having pre-gestational diabetes. Risk of ventricular septal defect did not vary with maternal age, baby's sex, women's obesity or socioeconomic status (Table 18).

Figure 12: Ventricular septal defect per 1,000 pregnancies from 1998 to 2014

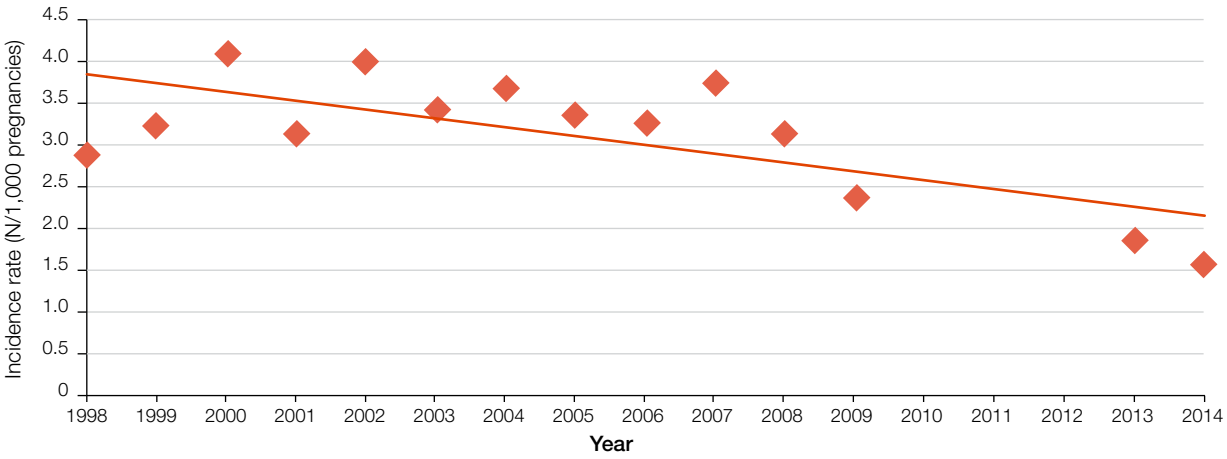


Table 18: Ventricular septal defect by selected maternal and child characteristics, 2013 and 2014

Characteristic		Ventricular septal defect/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	1.6 1.9	- 1.23 (0.94–1.60)
Plurality	Single (Ref) Multiple	1.5 5.1	- 3.26 (2.04–5.19)*
Gender	Male (Ref) Female	1.6 1.7	- 1.07 (0.84–1.37)
Pre-gestational diabetes	No (Ref) Yes	1.7 7.5	- 4.54 (2.34–8.81)*
Obesity	BMI < 35 (Ref) BMI ≥ 35	1.3 1.7	- 1.31 (0.81 to 2.10)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	1.3 1.7	0.75 (0.53–1.05) -

*Shows statistically significant association

9.10 Hypoplastic left heart syndrome

Definition: Hypoplastic left heart syndrome comprises hypoplastic left ventricle associated with aorta and/or mitral valve atresia.

Incidence: Of all the congenital anomaly cases reported in 2013 to 2014, 0.8 per cent (n = 42) of babies had hypoplastic left heart syndrome, of which 50.0 per cent (n = 21) were isolated and 50.0 per cent (n=21) occurred with other congenital anomalies.

Trend: The incidence of hypoplastic left heart syndrome did not change significantly (p-value 0.56) from 1998 to 2014 (Figure 13).

Risk factors: Male babies were 2.4 times more likely to develop hypoplastic left heart syndrome as compared to the female babies. Risk of hypoplastic left heart syndrome did not vary with maternal age, multiple births, pre-gestational diabetes, obesity or socioeconomic status (Table 19).

Figure 13: Hypoplastic left heart syndrome per 1,000 pregnancies from 1998 to 2014

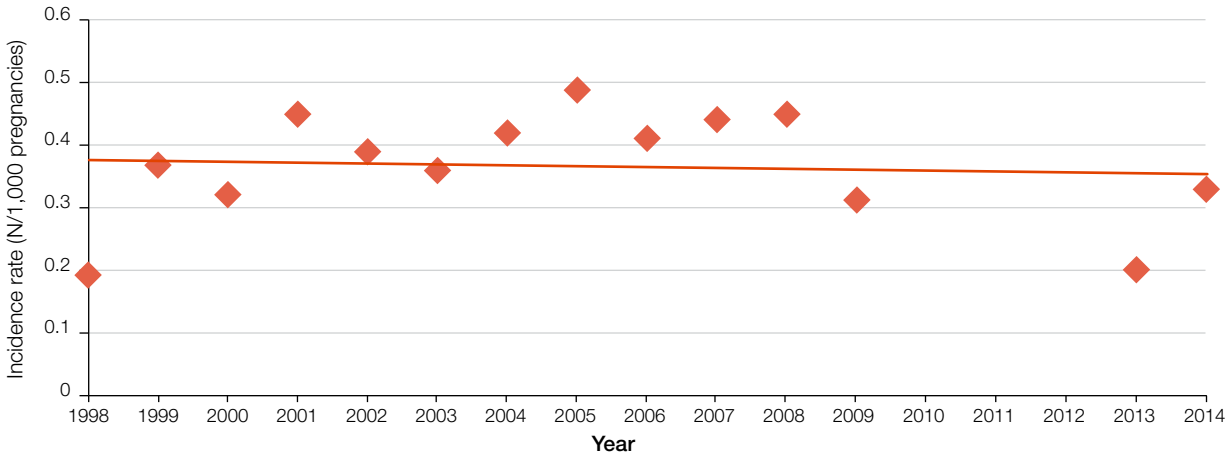


Table 19: Hypoplastic left heart syndrome by selected maternal and child characteristics, 2013 and 2014

Characteristic		Hypoplastic left heart syndrome/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 0.2	- 1.77 (0.56–2.46)
Plurality	Single (Ref) Multiple	0.2 0.3	- 1.10 (0.15–8.03)
Gender	Male (Ref) Female	0.3 0.1	2.41 (1.20–4.85)* -
Pre-gestational diabetes	No (Ref) Yes	0.3 0.8	- 3.20 (0.44–23.25)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.4	- 2.26 (0.77–6.59)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.2	1.44 (0.67–3.0 -

*Shows statistically significant association

9.11 Coarctation of the aorta

Definition: In coarctation of the aorta, there is an obstruction in the descending aorta, almost invariably (98 per cent) at the insertion of the ductus arteriosus.

Incidence: Among all the congenital anomaly cases reported between 2013 and 2014, 0.5 per cent (n = 24) of babies had coarctation of the aorta, of which 37.5 per cent (n = 9) were isolated and 62.5 per cent (n = 15) occurred with other congenital anomalies.

Trend: Incidence rates of coarctation of the aorta did not change significantly (p-value 0.342) from 1998 to 2009 and then decreased significantly (p-value < 0.001) in 2013 and 2014 (Figure 14).

Risk factors: The risk of coarctation of the aorta did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 20).

Figure 14: Coarctation of the aorta per 1,000 pregnancies from 1998 to 2014

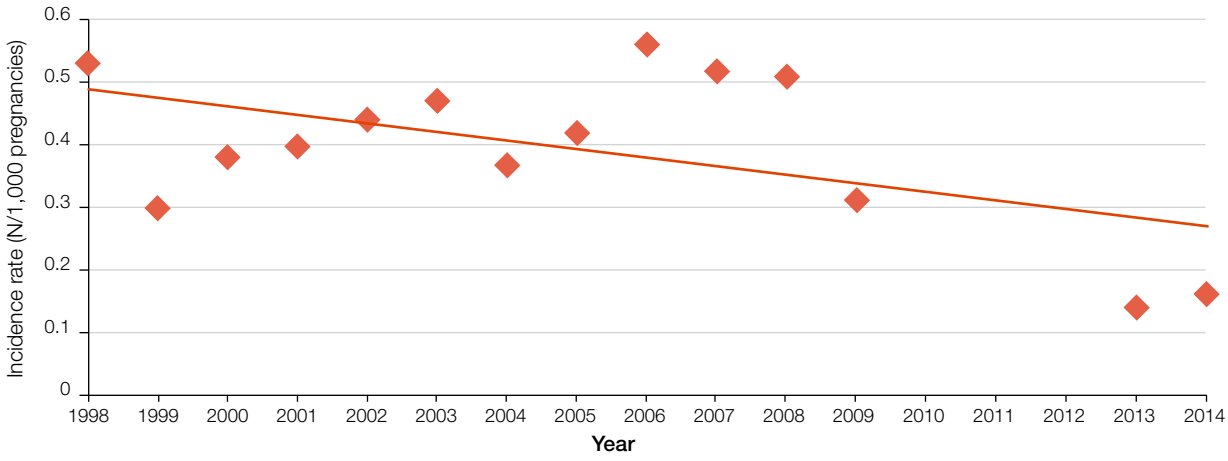


Table 20: Coarctation of the aorta by selected maternal and child characteristics, 2013 and 2014

Characteristic		Coarctation of the aorta/ 1,000 pregnancies	RR and 95 per cent CI
Mother's age	< 35 years (Ref) ≥ 35 years	0.1 0.2	- 1.24 (0.51–2.99)
Plurality	Single (Ref) Multiple	0.2 0	- 0.85 (0.05–14.06)
Gender	Male (Ref) Female	0.1 0.2	- 1.24 (0.56–2.78)
Pre-gestational diabetes	No (Ref) Yes	0 0.2	- 2.67 (0.16–43.99)
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.1	- 0.91 (0.11–6.99)
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.2 0.1	1.17 (0.43–3.18) -

Appendix A: Selected major congenital anomalies by body systems

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Nervous system		
Anencephaly and similar malformations (Q00)	45	0.3
Encephalocele (Q01)	13	0.1
Microcephalus (Q02)	14	0.1
Hydrocephalus (Q03)	122	0.8
Other specified anomalies of brain (Q04)	151	1.0
Spina Bifida (Q05)	38	0.2
Other congenital malformations of spinal cord (Q06)	5	0.0
Other congenital malformations of nervous sytem (Q07)	26	0.2
Congenital and developmental myasthenia (G702)	< 5	0.0
Huntington disease (G10)	< 5	0.0
Total	**417**	**2.6**
Eye		
Congenital malformation of eyelid, lacrimal apparatus and orbit (Q10)	< 5	0.0
Anophthalmos (Q11)	< 5	0.0
Microphthalmos (Q112)	< 5	0.0
Cataract (Q120)	< 5	0.0
Other lens (Q121-Q124, Q129)	< 5	0.0
Congenital malformation s of anterior segment of eye (Q13)	5	0.0
Congenital malformation s of posterior segment of eye (Q14)	< 5	0.0
Other congenital malformations of eye (Q15)	5	0.0
Congenital Nystagmus (H55)	< 5	0.0
Congenital strabismus (H509)	< 5	0.0
Disorder of iris and cilliary body, unspecified (H219)	< 5	0.0
Total	**29**	**0.2**
Ear, face and neck		
Ear-affecting hearing (Q16)		
Absence or stricture of auditory canal (Q161)	< 5	0.0
Absent auricle (Q162)	< 5	0.0
Other specified anomalies of the ear (Q163, Q164, Q165, Q169)	< 5	0.0
Other ear (Q17)		
Accessory auricle (Q170)	96	0.6
Microtia (Q171)	< 5	0.0
Other Ear (Q172-Q175, Q178, Q179)	26	0.2

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Face and neck (Q18)	32	0.2
Deafness (H90)	7	0.0
Total	**169**	**1.1**
Cardiovascular system		
Common truncus (Q200)	8	0.1
Transposition of great vessels complete (Q2031)	52	0.3
Tetralogy of Fallot (Q213)	56	0.4
Double outlet right ventricle (Q201)	22	0.1
Double inlet ventricle (Q204)	< 5	0.0
Discordant atrioventricular connection (Q205)	< 5	0.0
Isomerism of atrial appendages (Q206)	< 5	0.0
Other congenital malformations of cardiac chambers and connections (Q208)	73	0.5
Other congenital malformations of cardiac chambers and connections, unspecified (Q209)	5	0.0
Ventricular septal defect (Q210)	269	1.7
Atrial septal defect (Q211)	38	0.2
Atrioventricular septal defect (Q212)	27	0.2
Congenital malformations of pulmonary and tricuspid valves (Q22)	86	0.5
Congenital malformations of aortic and mitral valves (Q23)	67	0.4
Other congenital malformations of heart (Q24)	340	2.2
Patent ductus arteriosus (Q250)	38	0.2
Coarctation of aorta (Q251)	24	0.2
Stenosis of aorta (Q253)	6	0.0
Other congenital malformations of aorta (Q254)	63	0.4
Pulmonary artery (Q255, Q256, Q257)	5	0.0
Great arteries (Q258, Q259)	9	0.1
Great veins (Q26)	14	0.1
Peripheral vascular (Q27)	16	0.1
Other congenital malformations of circulatory system (Q28)	5	0.0
Total	**1232**	**7.8**
Respiratory system		
Congenital malformations of nose (Q30)	28	0.2
Congenital malformations of larynx (Q31)	< 5	0.0
Congenital malformations of trachea and bronchus (Q32)	< 5	0.0
Congenital malformations of lung (Q33)	71	0.4
Other congenital malformation of respiratory system (Q34)	13	0.1
Total	**114**	**0.7**

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Gastrointestinal tract		
Cleft palate (Q35)	72	0.5
Cleft lip (Q36)	48	0.3
Cleft lip and palate (Q37)	59	0.4
Other congenital malformations of tongue, mouth and pharynx (Q38)	27	0.2
Congenital malformations of oesophagus (Q39)	16	0.1
Other congenital malformations of upper alimentary tract (Q40)	19	0.1
Congenital absence, atresia and stenosis of small intestine (Q41)	31	0.2
Congenital absence, atresia and stenosis of large intestine (Q42)	27	0.2
Other congenital malformations of intestine (Q43)	44	0.3
Congenital malformations of gallbladder, bile ducts and liver (Q44)	21	0.1
Other congenital malformations of digestive system (Q45)	23	0.1
Total	**387**	**2.4**
Urogenital system		
Ovaries, fallopian tubes and broad ligaments (Q50)	< 5	0.0
Uterus and cervix (Q51)	< 5	0.0
Female genitalia (Q52)	29	0.2
Undescended testes (Q53)	287	1.8
Hypospadias (Q54)	354	2.2
Phimosis (N471)	17	0.1
Other congenital malformations of male genital organs (Q55)	52	0.3
Indeterminate sex and pseudohermaphroditism (Q56)	16	0.1
Renal agenesis and other reduction defects of kidneys (Q60)	73	0.5
Cystic kidney disease (Q61)	88	0.6
Obstructive defects renal pelvis/ureter (Q62)	443	2.8
Other congenital malformations of kidneys (Q63)	87	0.6
Other congenital malformations of urinary system (Q64)	52	0.3
Total	**1505**	**9.5**

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Musculoskeletal system		
Congenital deformities of hip (Q65)	284	1.8
Congenital deformities of feet (Q66)	371	2.3
Head, face, spine and chest (Q67)	65	0.4
Other congenital musculoskeletal deformities (Q68)	44	0.3
Polydactyly (Q69)	162	1.0
Syndactyly (Q70)	73	0.5
Reduction defects of upper limbs (Q71)	37	0.2
Reduction defects of lower limb (Q72)	18	0.1
Reduction defects of unspecified limb (Q73)	5	0.0
Other congenital malformations of limbs (Q74)	151	1.0
Other congenital malformations of skull and face bones (Q75)	60	0.4
Spine and bony thorax (Q76)	24	0.2
Osteochondrodysplasia with defects of growth of tubular bones and spine (Q77)	8	0.1
Other osteochondrodysplasias (Q78)	7	0.0
Congenital malformations of musculoskeletal system not elsewhere classified (Q79)	180	1.1
Total	**1489**	**9.4**
Integumentary system		
Congenital Ichthyosis (Q80)	7	0.0
Epidermolysis bullosa (Q81)	8	0.1
Other congenital malformations of skin (Q82)	100	0.6
Congenital malforamtions of breast (Q83)	< 5	0.0
Other disorders of pigmentation (L81)	< 5	0.0
Other congenital malformations of integument (Q84)	6	0.0
Total	**127**	**0.8**
Chromosomal		
Down's syndrome (Q90)	197	1.2
Edward's syndrome and Patau's syndrome (Q91)	79	0.5
Other trisomies and partial trisomies of the autosomes, not elsewhere classified (Q92)	49	0.3
Monosomies and deletions from chromosomes, not elsewhere classified (Q93)	30	0.2
Balanced rearrangements and structural markers, not elsewhere classified (Q95)	< 5	0.0
Turner'S Syndrome (Q96)	20	0.1

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Other sex chromosome abnormalities, female phenotype, not elsewhere classified (Q97)	7	0.0
Other sex chromosome abnormalities, male phenotype, not elsewhere classified (Q98)	17	0.1
Other chromosome abnormalities, not elsewhere classified (Q99)	46	0.3
Total	**448**	**2.8**
Neoplasms		
Malignant (C00-C97)	5	0.0
Benign (D10-D36)		0.0
Haemangioma (D180)	69	0.4
Cystic hygroma (D181)	29	0.2
Benign neoplasm of other and unspecified intrathoracic organs (D15)	< 5	0.0
Uncertain or unknown behaviour (D37-D48)	35	0.2
Total	**140**	**0.9**
Metabolic/endocrine/nutritional		
Congenital Hypothyroidism (E031)	< 5	0.0
Other Endocrine Glands (E20-E35)	< 5	0.0
Neonatal hypoglycemia (P704)	< 5	0.0
Metabolic disorders (E70-E90)	17	0.1
Total	**21**	**0.1**
Haematological		
Haemolytic Anaemia (D55-D59)	11	0.1
Coagulation defects, purpura and other haemorrhagic conditions (D65-D69)	< 5	0.0
Other diseases of blood and blood-forming organs (D70-D77)	< 5	0.0
Haemolytic disease of fetus and newborn (P55)	7	0.0
Other perinatal haematological disorders (P61)	7	0.0
Total	**30**	**0.2**

Defect with ICD-10-AM code	Number	N/1,000 pregnancies
Others		
Phakomatoses, not elsewhere classified (Q85)	< 5	0.0
Spleen (Q890)	< 5	0.0
Adrenal Gland (Q891)	< 5	0.0
Other Endocrine Gland (Q892)	5	0.0
Situs Inversus (Q893)	< 5	0.0
Conjoined Twins (Q894)	< 5	0.0
Pierre Robin sequence (Q8706)	< 5	0.0
Other congenital malformation syndromes predominantly affecting facial appearance (Q8709)	< 5	0.0
Cockayne syndrome (Q8711)	< 5	0.0
Noonan syndrome (Q8713)	< 5	0.0
Prader-Willi syndrome (Q8714)	< 5	0.0
VATER association (Q8727)	< 5	0.0
Marfan's syndrome (Q874)	< 5	0.0
Other specified congenital malformation syndromes, not elsewhere classified (Q8789)	< 5	0.0
Dysmorphic features (Q8971)	15	0.1
Multiple congenital malformations, not elsewhere classified (Q8979)	< 5	0.0
Other specified congenital malformations (Q898)	< 5	0.0
Congenital malformations, unspecified (Q899)	78	0.5
Congenital cytomegalovirus infection (P351)	< 5	0.0
Hydrops fetalis (P832)	57	0.4
Disorders of psychological development (F80-F89)	< 5	0.0
Total	**190**	**1.2**

Denominators do not modify for sex specific conditions.

Appendix B: Excluded minor congenital anomalies

There has been variation in this list of exclusions between 1983–2002. Some excluded conditions may be included in this report if they were previously not excluded AND occur with other congenital anomalies.

Abnormal palmar creases	Macroglossia (large tongue)
Accessory nipples	Meckel's diverticulum
Anal fissure	Meconium ileus
Balanced autosomal translocation (unless occurring with structural defects)	Mental retardations (unless occurring with a syndrome/ structural defect)
Birth injuries	Metatarsus varus
Birth marks (smaller than 4cm, not including giant naevus)	Micrognathia (unless severe)
Bowing of legs (unless severe)	Mongolian spots
Blocked tear ducts (dacrostenosis)	Occiput, flat/prominent
Brushfield spots	Patent ductus arteriosus (< 37 weeks)
Cephalhaematoma	Philtrum, long/short
Cleft gum	Plagiocephaly
Clicky hips	Pre-auricular sinus
Clinodactyly	Prominent forehead
Craniotabes (unless severe)	Protruding tongue
Dermatogplyphic abnormalities	Ptosis
Ear abnormalities (minor)	Retrognathia (unless severe)
Epicanthic folds	Rocker-bottom feet (prominent heels)
Gastro-oesophageal reflux	Sacral pits, dimples, sinuses
Haemangioma (< 4 cm wide)	Short sternum
Hernia – inguinal, umbilical	Simian creases
High-arched palate	Single umbilical artery/2 vessels in cord
Hydrocele	Skin folds/tags
Hypertelorism	Slanting eyes
Imperforate hymen	Small mouth
Laryngeal stridor	Laryngomalacia
Low slung/set ears	

Appendix C: Routine data items contained in the Victorian Congenital Anomalies Register

Maternal data: postcode, date of birth, method of prenatal diagnosis

Child /fetus data: hospital of birth, date of birth (or termination), sex, birth weight, plurality, rank, condition of birth (termination before 20 weeks, termination ≥ 20 weeks, stillbirth, live birth), date of death (if applicable), BPA Codes for congenital defects, position code, source of notification

Other data items available from linkage to the Victorian Perinatal Data Collection (VPDC) data:

Maternal items: UR number, local government area, region, country of birth, aboriginality, discharge date and status, marital status, number of previous pregnancies, date of completion of last pregnancy, outcome of last pregnancy, maternal medical conditions, obstetric complications, indication(s) for operative delivery, complications of labour birth and postnatal, procedures and operations, type of labour, presentation, method of delivery

Child data items: APGAR, time to establish respiration, resuscitation methods, neonatal morbidity

More detail about these data items can be obtained from:

Department of Health and Human Services Victoria. Victorian Perinatal Data Collection Manual Section 3 – Data Definitions, V3.0 Release date 23 April 2013. Available from: https://www2.health.vic.gov.au/hospitals-and-health-services/quality-safety-service/consultative-councils/council-obstetric-paediatric-mortality/perinatal-data-collection

Appendix D: Congenital anomalies by year, 1983 to 2014

Year	Total births, 20 weeks and later	Defects, 20 weeks and later	Defects before 20 weeks (terminations)	N/1,000 Pregnancies (including terminations before 20 weeks)	Percentage
1983	60,628	1,653	< 5	27.3	2.7
1984	60,737	1,691	9	28.0	2.8
1985	61,189	1,591	18	26.3	2.6
1986	61,253	1,604	80	27.5	2.7
1987	61,566	1,620	55	27.2	2.7
1988	63,666	1,874	103	31.0	3.1
1989	64,255	1,930	123	31.9	3.2
1990	66,878	2,164	132	34.3	3.4
1991	65,248	2,235	140	36.3	3.6
1992	66,305	2,295	152	36.8	3.7
1993	64,737	2,250	203	37.8	3.8
1994	64,932	2,295	250	39.0	3.9
1995	63,717	2,444	257	42.2	4.2
1996	62,951	2,217	272	39.4	3.9
1997	62,308	2,306	297	41.6	4.2
1998	62,091	2,332	274	41.8	4.2
1999	62,690	2,555	294	45.2	4.5
2000	62,564	2,614	292	46.2	4.6
2001	62,149	2,394	308	43.3	4.3
2002	63,133	2,520	327	44.9	4.5
2003	63,552	2,690	356	47.7	4.8
2004	63,700	2,774	342	48.7	4.9
2005	66,654	2,500	339	42.4	4.2
2006	69,856	2,836	366	45.6	4.6
2007	72,474	3,013	364	46.4	4.6
2008	72,545	2,630	360	41.0	4.1
2009	73,264	2,664	414	41.8	4.2
2013	78,536	2,336	187	32.0	3.0
2014	79,179	2,276	217	31.4	3.1
Total	**1,902,757**	**66,303**	**6,533**	**38.1**	**3.8**

These figures may differ from the number of births presented elsewhere due to the inclusion of terminations of pregnancy for psychosocial reasons or congenital anomalies at greater than 20 weeks' gestation which are excluded from some analyses in other CCOPMM reports.

Appendix E: Outcomes of selected major congenital anomalies, 2013–2014

Chromosomal anomaly*	Terminations < 20 weeks	Terminations ≥ 20 weeks	Still births†	Neonatal deaths†
Trisomy 21	83	23	17	11
Trisomy 13	15	< 5	5	< 5
Trisomy 18	27	13	20	< 5

Selected major anomalies*	Terminations	Still births	Neonatal deaths
Anencephaly	33	8	7
Spina bifida	27	17	5
Encephalocele	5	5	< 5
All neural defects (combined)	65	32	15
Microcephalus	< 5	< 5	< 5
Hydrocephalus	37	41	10
Transposition of the great vessels	< 5	< 5	< 5
Tetralogy of fallot	8	< 5	< 5
Ventricular septal defect	21	18	10
Hypoplastic left heart syndrome	18	15	5
Coarctation of the aorta	< 5	< 5	< 5
Cleft palate	< 5	< 5	< 5
Cleft lip	5	5	0
Cleft lip and palate	13	9	< 5
Oesophageal atresia and/or stenosis	0	0	0
Small intestinal atresia and/or stenosis	< 5	< 5	0
Anorectal atresia and/or stenosis	0	0	< 5
Hypospadias	0	0	< 5
Renal agenesis and dysgenesis	22	12	7
Cystic kidney disease	18	12	6
Obstructive defects of renal pelvis	5	6	5
Developmental dysplasia of the hip	0	0	< 5
Limb reduction defects	20	12	< 5
Diaphragmatic hernia	9	6	6
Exomphalos	14	9	< 5
Gastroschisis	< 5	< 5	0

*Congenital anomaly can be isolated or can occur with other anomalies

†Includes terminations

Definitions

Birth	Birth refers to both live births and stillbirths.
Birth plurality	Refers to the total number of births resulting from a single pregnancy. Singleton or Single refers to one birth resulting from a single pregnancy and multiple refers to more than one birth from a single pregnancy.
Confinements	These are the number of pregnancies resulting in at least one birth. Number of confinements does not equal number of births. One confinement may result in two births i.e. Twins
Congenital anomaly	Any abnormality of prenatal origin, either present following conception or occurring before the end of pregnancy. This includes structural, functional, genetic, and chromosomal and biochemical abnormalities.
Congenital anomaly cases	Congenital anomaly cases refer to the number of live born or stillborn babies, or terminations at any gestation affected by at least one congenital anomaly.
Isolated congenital anomaly	Anomaly that is not related to any other condition and occurs as a single defect example isolated cleft lip.
Live birth	Complete expulsion or extraction from its mother of a baby of at least 20 weeks gestation or, if gestation is unknown, weighing at least 400g who, after being born, breathes or shows any evidence of life such as a heartbeat.
Neonatal death	It refers to a death occurring within 28 days of live birth in an child whose gestation was at least 20 weeks or, if gestation is unknown, weighing at least 400g.
Obesity	Pre-conception Body mass index (BMI) of 35 kg/m^2 and more.
Parity	Number of pregnancies carried to a viable gestational age. A woman who has never carried a pregnancy beyond 20 weeks gestation is called nulliparous, woman who has carried one pregnancy beyond 20 weeks is called primiparous and a woman who has carried two or more pregnancies beyond 20 weeks is called multiparous.
Perinatal death/mortality	Perinatal death is a stillbirth or neonatal death.
Pre-gestational diabetes	Type I or Type II diabetes that exists before conception
Pregnancy	Includes live birth, stillbirth, and termination of pregnancy at any gestation.
Stillbirth	Complete expulsion or extraction from its mother of a baby of at least 20 weeks gestation or, if the gestation is unknown, weighing at least 400g who did not, at any time after delivery, breathe or show any evidence of life such as a heartbeat.

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