Consultative Council on
Obstetric and Paediatric
Mortality and Morbidity

Congenital anomalies in Victoria

2015–2016

Safer Care
Victoria

To receive this publication in an accessible format phone (03) 9096 0380, using the National Relay Service 13 36 77 if required, or email consultative.councils@safercare.vic.gov.au.

Authorised and published by the Victorian Government, 1 Treasury Place, Melbourne.

Where the term 'Aboriginal' is used it refers to both Aboriginal and Torres Strait Islander people.

ISSN 2207-2764 (pdf/online)

Available at bettersafercare.vic.gov.au

Recommended citation: Victorian Congenital Anomalies Register 2018, *Congenital anomalies in Victoria 2015–2016,* Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM), Melbourne.

VICTORIA State Government

Message from the chair

The release of this report covering major congenital anomalies detected in children born in Victoria marks the return of timely reporting and analyses of these conditions which commenced in 1982.

Collection of the data that underlies this report comes from the Victorian Perinatal Data Collection, from clinicians including the maternal and child health nurses and cytogenetic laboratories and we express our appreciation for their cooperation and facilitation of this collection.

With the expansion in genetic analysis, the understanding of the causation of many anomalies has undergone major developments as the ability to detect changes in the child's DNA is rapidly improving. With this knowledge will come initiatives to prevent and correct at least some of these anomalies that can have such a major impact on the child and their parents and family.

This report is the result of a substantial effort by the staff of the Consultative Councils Unit of Safer Care Victoria that supports the work of the Consultative Council on Obstetric and Paediatric Mortality and Morbidity that has the legislative responsibility for the surveillance and reporting on congenital anomalies of children born in Victoria.

Jeremy J N Oats
Chair
Consultative Council on
Obstetric and Paediatric
Mortality and Morbidity

Acknowledgements

This report would not have been possible without the particular assistance of:

- midwives in Victoria who provided notification of congenital anomalies via their hospital's electronic system or the online e-form to the Victorian Perinatal Data Collection (VPDC)
- maternal and child health nurses who completed the Birth Anomalies Notification form – either via hard copy or online
- health information managers at all hospitals with maternity services and, in particular, those at hospitals with paediatric services
- cytogenetic services in Victoria.

We thank them for their contributions.

This report was developed by the Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM) with support from the following team members from Safer Care Victoria's Consultative Councils Unit:

Dr Shirin Anil	Acting Manager
Ms Joanna Gaston	Senior Project Officer
Ms Taliesin Ryan-Atwood	Research Officer
Dr Sophie Treleaven	Paediatric Epidemiologist
Dr Mary-Ann Davey	Perinatal Epidemiologist; Acting Team Leader, Research and Reporting
Ms Sarah Kenny	Team Leader, Council Program Support
Ms Diana Stubbs	Liaison Midwife
Ms Adelinda Botham	Project Officer
Ms Marina Forte	Administration Officer

We would like to acknowledge the following people for their contributions to the report:

Dr Catherine Rose from Victorian Clinical Genetics Service (VCGS) and POSSUMweb, Murdoch Children's Research Institute and Ms Fiona Norris, from VCGS.

We would like to extend our sincere gratitude and thanks to Professor Jeremy Oats, chairperson of CCOPMM since 2007, for his expertise and passion for improving obstetric and paediatric outcomes for Victorian mothers and babies. His guidance of CCOPMM over the past 11 years has ensured that the Minister for Health and the Department of Health and Human Services have been informed and supported on obstetric and paediatric issues in Victoria. We wish Professor Oats all the best in his retirement, and his many contributions to improving the health and wellbeing of Victorians are greatly appreciated.

Contents

Tables

Figures

Executive summary

Congenital anomalies are a major cause of child death and disability and are the leading cause of perinatal mortality. Three in 10 (31.0 per cent) perinatal deaths in Australia are associated with congenital anomalies.[1] In Victoria, approximately one in every 22 babies have been affected by a congenital anomaly. The prevention, detection and management of congenital anomalies relies on population-level surveillance and research into the aetiology and diagnosis of congenital anomalies and active promotion of primary prevention strategies.

A 'congenital anomaly', also called a 'birth defect', 'congenital malformation', 'congenital abnormality' or 'congenital disorder', is any abnormality occurring before birth. Anomalies can be minor or major. They may be structural, functional, genetic, chromosomal or biochemical. They can be detected before birth, at birth or in early childhood.

Factors that may contribute or increase the chances of a baby being born with a congenital anomaly include genetic factors, socio-demographic factors (such as ethnicity and increasing maternal age), environmental factors (such as maternal exposure to alcohol, medications, chemicals, radiation and tobacco), infections (such as syphilis, rubella and Zika virus), maternal nutritional status (such as folate deficiency), obesity and pre-gestational diabetes.[2–4]

Congenital anomalies can occur during any stage of pregnancy; however, the first three months, when the organs of the baby are developing, is when most anomalies occur. Promotion, implementation and the accessibility of primary prevention strategies before and during early pregnancy will increase the chances of a healthy baby. Ensuring women have been vaccinated for infections known to cause birth defects (such as rubella), encouraging the use of folic acid to mitigate the risk of neural tube defects, managing diabetes particularly before and during early pregnancy, and educating women to avoid exposure to substances such as alcohol, tobacco and some medications, are all well-established strategies that result in positive health outcomes.

The Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM) has legislative responsibility under s. 46 of the *Public Health and Wellbeing Act 2008* 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 prevalence of major congenital anomalies in Victoria in 2015 and 2016. As almost all major congenital anomalies are diagnosed by the age of six years,[5] the report focuses on major anomalies that are identified in children before birth and up to six years of age. Selected major congenital anomalies of the gastrointestinal system and selected chromosomal anomalies are reported in more detail.

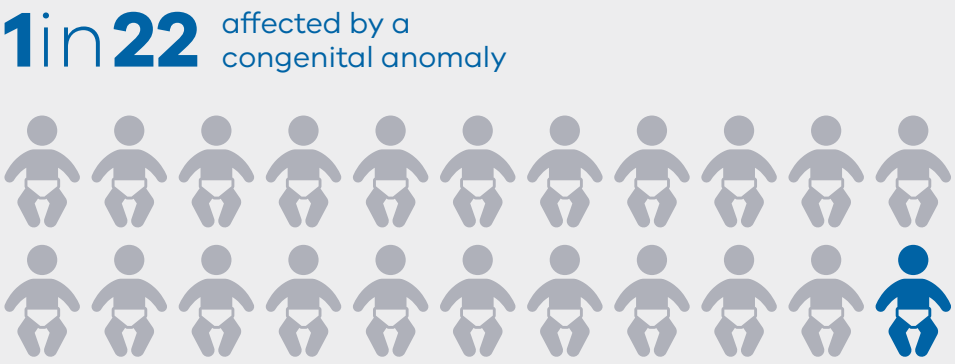
FINDINGS AT A GLANCE

The prevalence of major congenital anomalies has increased from 3.2 per cent of reported pregnancies in 2013 and 2014 to 4.5 per cent of pregnancies in 2015 and 2016. This is attributed to improvement in the notification to the VCAR.

The main findings for the 2015–2016 reporting period are as follows:

- **Prevalence and pregnancies affected:** Major congenital anomalies affected 4.5 per cent of reported pregnancies in 2015 and 2016. In Victoria, one in 22 pregnancies resulted in a congenital anomaly and 12.3 per cent of babies were reported to have had more than one anomaly.
- **Most common congenital anomalies:** By diagnostic category, hypospadias was the most common congenital anomaly (5.3 per 1,000 pregnancies), followed by trisomy 21 (3.0 per 1,000 pregnancies) and obstructive defects of the renal pelvis (2.6 per 1,000 pregnancies).
- **Chromosomal anomalies:** Of all the major anomalies reported, 35.8 per cent were chromosomal anomalies.
- **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 or older were 59.0 per cent more likely to have a baby with a congenital anomaly than those younger than 35 years of age.
- **Aboriginal women:** The rate of congenital anomalies was similar for Aboriginal women and non-Aboriginal women.
- **Obesity:** Obese women (BMI 35 kg/m^2 or more) were 12.8 per cent more likely to have a baby with a congenital anomaly than non-obese women.
- **Pre-gestational diabetes:** Women with pre-gestational diabetes (development of diabetes before pregnancy) were 42.8 per cent more likely to have a baby with a congenital anomaly than women without pre-gestational diabetes.
- **Socioeconomic status:** Lowest socioeconomic status did not increase the risk of having a baby with a congenital anomaly in 2015–2016.

In Victoria, about one in 22 babies is affected by a congenital anomaly



Reporting of congenital anomalies in Victoria

VICTORIAN CONGENITAL ANOMALIES REGISTER

The VCAR, first established in 1982 under the *Health Act 1981,* is a population-based surveillance system that collects information on all congenital anomalies for live births, still births and terminations of pregnancy. More than half the notifications to the VCAR are made directly to the VPDC, with the remaining from sources such as hospitals, maternal and child health nurses, cytogenetic laboratories, death certificates, autopsy reports and private paediatricians. The VCAR includes both suspected and confirmed congenital anomalies.

The VPDC collects and analyses information on the health of mothers and babies in Victoria to contribute to improved maternal and perinatal health outcomes. The VCAR and VPDC are the legislative responsibility of CCOPMM. The principal function of CCOPMM is to identify and report on issues relating to perinatal, maternal and paediatric mortality and morbidity and to provide advice on strategies to improve the safety and quality of paediatric and maternity services in Victoria. Specifically, the Public Health and Wellbeing Act states that CCOPMM is required to identify and monitor trends in perinatal health including birth defects and disabilities and to establish and maintain a register of these.

Data from the VCAR can assist with:

- planning for and providing healthcare to people with congenital anomalies
- information for families concerned about having a baby with a congenital anomaly
- epidemiological research in the aetiology and preventability of congenital anomalies
- assessing the effectiveness of primary prevention and screening programs
- informing the need for additional primary prevention strategies or screening programs
- responding to concerns about potential clusters or trends in congenital anomalies.

The VCAR collects notifications of congenital anomalies in children from before birth to six years of age. This is because approximately 99 per cent of all major congenital anomalies are diagnosed during this period.[5]

Congenital anomalies can be notified to VCAR by completing an online notification form available at bettersafercare.vic.gov.au/ccopmm.

CONGENITAL ANOMALIES: MAJOR AND MINOR

This report focuses on major congenital anomalies, which are generally defined as structural changes that have significant medical, social or cosmetic consequences, typically require medical intervention and are known to significantly contribute to perinatal and childhood morbidity, mortality and disability. These are listed in Appendix 1.

Minor congenital anomalies are structural changes that pose no significant health problem in the neonatal period and have limited social or cosmetic consequences. Minor congenital anomalies have been excluded from this report. Classification of these anomalies have primarily been based on the 'External minor congenital anomalies' table provided by the World Health Organization (WHO), United States Centers for Disease Control and Prevention (CDC) and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) (Appendix 2).[6]

SOURCES OF NOTIFICATION

For the years 2015 and 2016, the primary sources of notifications for congenital anomaly cases in Victoria were the VPDC (62.1 per cent) and cytogenetic reports (26.3 per cent). Notification sources are shown in Table 1.

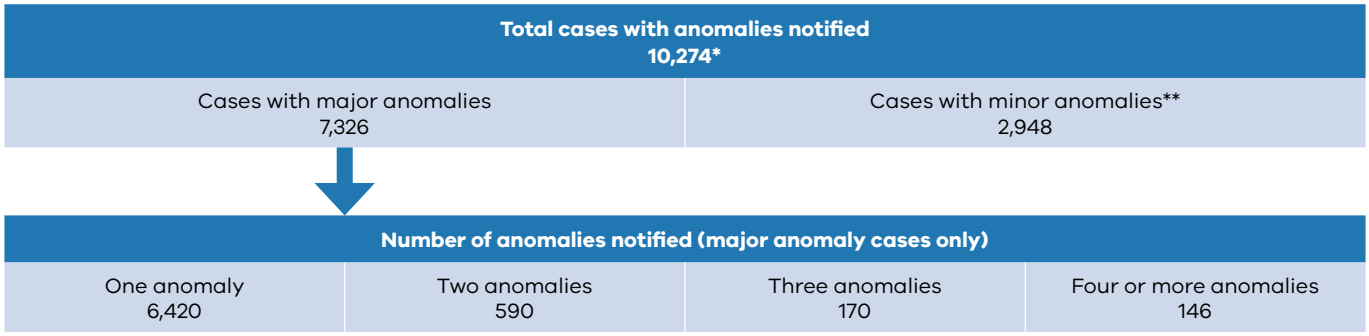
Table 1: Sources of notifications for congenital anomaly cases in Victoria, 2015–2016

Notification source	Number	Percentage
Victorian Perinatal Data Collection	6,835	62.1
Hospital	1,176	10.7
Maternal and child health nurse	79	0.7
Other professionals (doctor, nurse)	19	0.2
Cytogenetic report	2,898	26.3
Total	**11,007***	**100.0**

* Total of all notifications received including duplicates.

In 2015 and 2016 there were 10,274 congenital anomalies cases notified to the VCAR, of which 7,326 were major anomaly cases and 2,948 were minor. There was a 30.4 per cent increase in notifications of congenital anomalies (10,274 notified in 2015–2016 compared to 7,875 in 2013–2014) attributed to a shift from passive to active surveillance and better education. The number of cases reported to have one, two or more anomalies are shown in Figure 1.

Figure 1: Congenital anomaly cases in Victoria, 2015–2016



* Excludes duplicates.

** Excluded from further analysis.

DATA ITEMS

All notifications of congenital anomalies reported to the VCAR (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 occurring after 20 weeks of gestation, or if the gestation is unknown, births of babies weighing more than 400 grams. Appendix 3 provides data items routinely collected in the VCAR and additional data items available for each case (more than 20 weeks' gestation) as reported in the VPDC.[7]

DATA QUALITY

Data submitted to the VPDC and the VCAR are checked for completeness and validity. Further data cleaning is carried out when all data for the calendar year have been submitted.

Projects to determine the accuracy and completeness of data submitted to the VPDC and the VCAR are undertaken periodically.[8–12] Ascertainment of congenital anomalies from terminations of pregnancy less than 20 weeks is incomplete because reporting of these is not mandated by legislation.

DATA ANALYSIS

The 2015–2016 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.

Major congenital anomalies, anomalies of the gastrointestinal system and chromosomal anomalies are reported by prevalence, trends and selected maternal and child characteristics. Relative risk (RR) and its 95 per cent confidence intervals (CI) have been calculated to assess the association of maternal and child characteristics with all congenital anomalies and selected anomalies. A *p*-value of less than 0.05 is considered statistically significant.

During the period 2010–2012 congenital anomaly data was collected but not analysed or reported due to resource limitations. As a result notifications decreased in subsequent years. This was reflected in the reduction in congenital anomalies reported in 2013–2014 and gave rise to possible differences in trends as noted in the 2013–2014 report. Notifications have now returned to higher levels due to improved surveillance.

TRENDS IN PRENATAL TESTING[13]

Advances in prenatal testing, such as the introduction of a non-invasive prenatal test (NIPT) and chromosome microarray (CMA), have resulted in significant changes in prenatal screening and testing in Victoria. The NIPT test, used to screen for common chromosome anomalies such as trisomies 13, 18 and 21, and combined first trimester screening (CFTS) have increased the ability to identify pregnancies at risk of a chromosomal anomaly. While this has led to a decrease in invasive procedures in pregnancies that are low risk for a chromosomal anomaly, pregnancies identified as high risk are confirmed by an invasive procedure such as chorionic villus sampling (CVS) and amniocentesis testing. This has led to an increase in detecting these anomalies through CVS and amniocentesis testing.

NIPT may include screening for a broader range of chromosomal conditions such as trisomies of all chromosomes as well as genetic changes such as deletions and duplications of chromosomes or parts thereof.

The introduction of CMA in 2011 has further improved detection and diagnosis of chromosomal anomalies. CMA, however, has the potential for uncertain or unknown genetic information of unknown significance. We all carry genetic changes, but in most cases these do not affect our health and wellbeing. It can be difficult to interpret results of uncertain or unknown significance.

The increase of chromosomal anomalies notified to VCAR for 2015–2016 may in part be attributed to improved screening and testing for chromosomal anomalies.

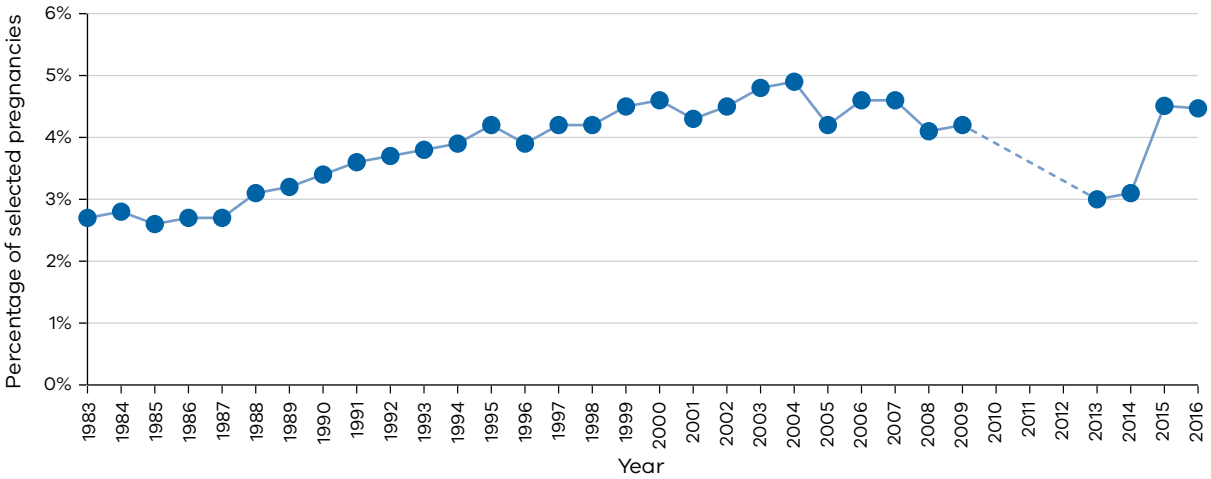
Prevalence of congenital anomalies in Victoria

Of all reported pregnancies, 4.5 per cent, or one in 22 pregnancies, were affected by at least one congenital anomaly in 2015 and 2016. This translates to a prevalence of 45 per 1,000 pregnancies in 2015 and in 2016. Of all the congenital anomaly cases reported as terminations of pregnancy before 20 weeks, 71 per cent (*n* = 191) had chromosomal anomalies.

Congenital anomaly trends in Victoria

The prevalence of major congenital anomalies has increased from 3.2 per cent of reported pregnancies between 2013 and 2014 to 4.5 per cent of reported pregnancies between 2015 and 2016 (Figure 2). This is attributed to an increase in notifications to the VCAR by the cytogenetic labs and education to use the online notification system. Major congenital anomalies by year are provided in Appendix 4.

Figure 2: Prevalence of congenital anomalies, 1983–2016



Note: During the period 2010–2012 congenital anomaly data was collected but not analysed or reported on.

Congenital anomalies by body system

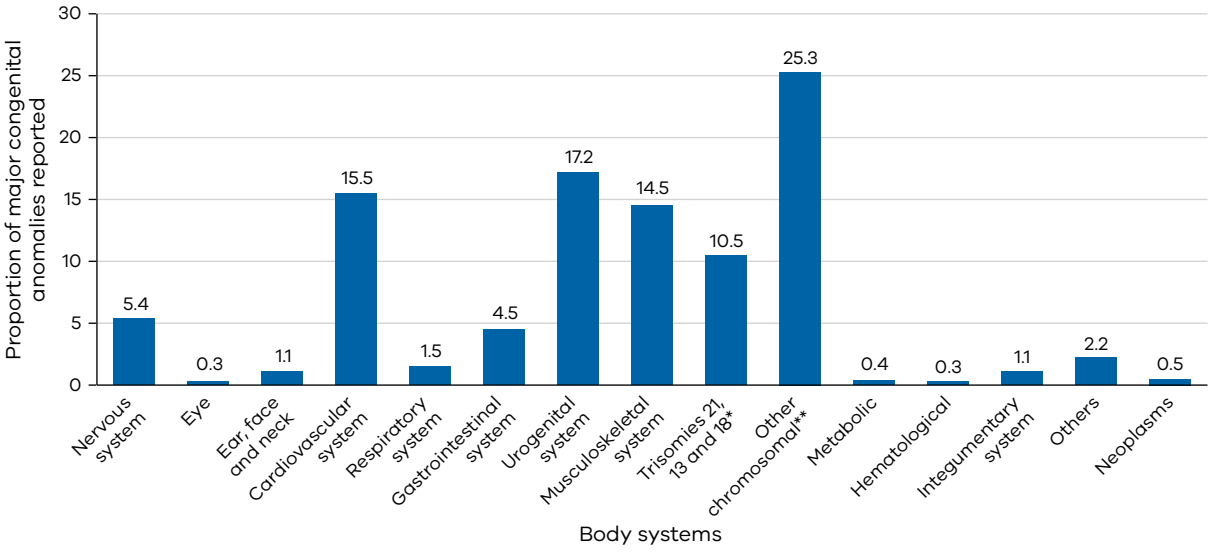
Among all the congenital anomalies reported, chromosomal were the most common (35.8 per cent) followed by anomalies of the urogenital system (17.2 per cent), anomalies of the cardiovascular system (15.5 per cent) and anomalies of the musculoskeletal system (14.5 per cent) (Figure 3).

Of all chromosomal anomalies, Trisomies 21, 13 and 18 accounted for 10.5 per cent. This category includes Down syndrome (trisomy 21), Edwards' syndrome (trisomy 18), Patau's syndrome (trisomy 13), Turner's syndrome (monosomy X, 45,X) and Klinefelter's syndrome (47 XXY).

Other chromosomal anomalies accounted for 25.3 per cent. This includes other trisomies, duplications and deletions of chromosomes of parts thereof, chromosomal anomalies not further specified or those that are not classified elsewhere. The clinical significance of some of these anomalies may be unknown. It is likely that the observed increase in chromosomal anomalies reported during 2015–2016 is in part a result of increased affordability of prenatal screening tests such as the NIPT, used to identify pregnancies with a greater chance of a chromosomal condition.

Appendix 1 provides details on the major anomalies according to each body system.

Figure 3: Congenital anomalies in Victoria by body systems, 2015–2016



* Includes Q95, Q96, Q97 and Q98.

** Includes other trisomies, duplications and deletions of chromosomes of parts thereof, chromosomal anomalies not further specified or those that are not classified elsewhere (Q92, Q93 and Q99). The clinical significance of some of these anomalies may be unknown. The higher rate observed may be due to an increase in notifications from cytogenetic laboratories and an increased uptake of genetic testing in Victoria.

Congenital anomalies by diagnostic category

Hypospadias (5.3 per 1,000 pregnancies) was the most common congenital anomaly reported by diagnostic category, followed by trisomy 21 (3.0 per 1,000 pregnancies) and obstructive defects of the renal pelvis (2.6 per 1,000 pregnancies). Table 2 presents the prevalence of key congenital anomalies.

Table 2: Order of prevalence of key congenital anomalies, 2015–2016

ICD-10-AM	Congenital anomaly	Number in 1,000 pregnancies	One in number of pregnancies
Q54	Hypospadias*	5.3	190
Q90	Trisomy 21†	3.0	331
Q62	Obstructive defects of the renal pelvis	2.6	383
Q210	Ventricular septal defect	1.7	593
Q65	Deformities of hip	1.2	823
Q910–Q913	Trisomy 18†	0.8	1,294
Q00, Q01, Q05	All neural tube defects (combined)	0.5	1,831
Q61	Cystic kidney disease	0.5	1,964
Q35	Cleft palate†	0.5	2,063
Q914–Q917	Trisomy 13†	0.5	2,090
Q37	Cleft lip and palate†	0.4	2,296
Q60	Renal agenesis and dysgenesis	0.4	2,547
Q234	Hypoplastic left heart syndrome	0.4	2,547
Q2031	Transposition of great vessels	0.3	3,075
Q213	Tetralogy of Fallot	0.3	3,196
Q03	Congenital hydrocephalus	0.3	3,326
Q793	Gastroschisis†	0.3	3,396
Q05	Spina bifida	0.3	3,543
Q251	Coarctation of aorta	0.3	3,881
Q790	Diaphragmatic hernia†	0.2	4,075
Q71–Q73	Limb reduction defects	0.2	4,405
Q792	Exomphalos†	0.2	4,657
Q00	Anencephaly	0.2	4,939
Q36	Cleft lip†	0.2	5,258
Q41	Absence, atresia and stenosis of small intestine†	0.2	5,621
Q42	Absence, atresia and stenosis of large intestine†	0.2	6,269
Q39	Malformations of oesophagus†	0.1	7,409
Q02	Microcephaly	0.1	10,867
Q01	Encephalocele	0.1	16,300

* This figure used male babies only as the denominator as hypospadias is a condition that affects only males.
† Anomalies reported in more detail.

Congenital anomalies by the child's characteristics

GENDER

For all congenital anomaly cases reported in Victoria in 2015 and 2016, 47.3 per cent were males, 31.3 per cent were females and 0.4 per cent were indeterminate (Figure 4). Gender data was incomplete in 21.0 per cent of reported cases. Nationally in Australia, the proportion of males diagnosed with a reported congenital anomaly is higher than that of females (59.0 per cent and 41.0 per cent respectively).[14]

The rate of congenital anomalies was 42.1 per 1,000 pregnancies for male babies and 29.4 per 1,000 pregnancies for female babies. Male babies were 43 per cent more likely to have a congenital anomaly than female babies (RR 1.43, 95% CI 1.36–1.51, p = < 0.0001).

When adjusted for terminations, 57.0 per cent were males, 36.4 per cent were female and 0.3 per cent were indeterminate. Gender data was incomplete in 6.3 per cent of reported cases (Figure 5).

Figure 4: Congenital anomalies by gender

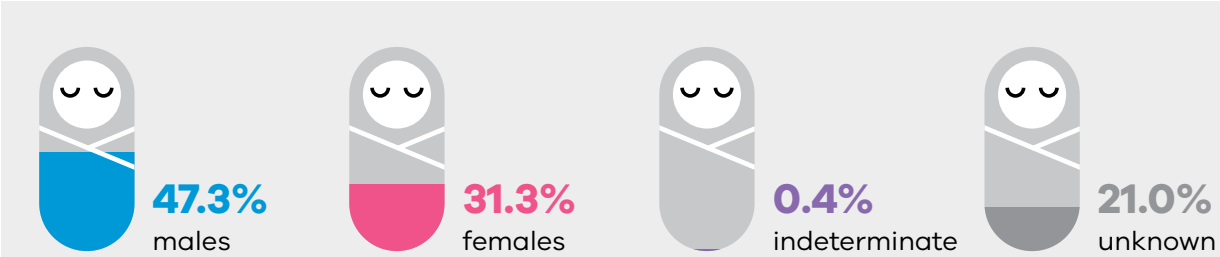
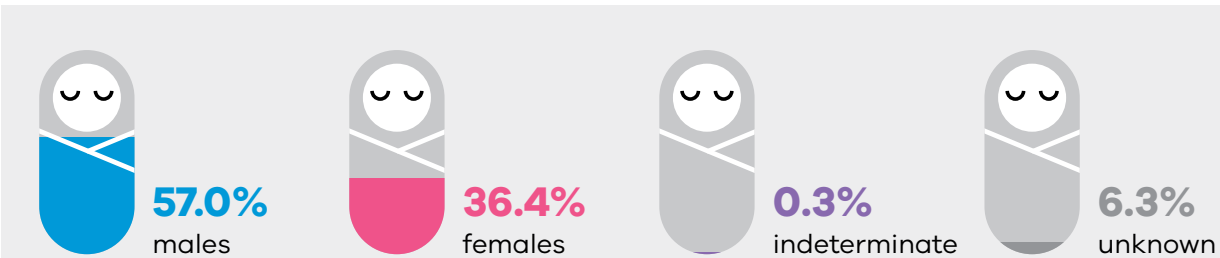


Figure 5: Congenital anomalies excluding terminations by gender*



* Excludes terminations and termination status unknown.

GESTATIONAL AGE

In Victoria in 2015 and 2016, babies with a congenital anomaly were three times more likely to be born preterm (less than 37 weeks' gestation) than those without a congenital anomaly (RR 3.1, 95% CI 2.90–3.31, p = < 0.0001). As the gestational age increased the rate of congenital anomalies decreased (Table 3).

Table 3: Congenital anomalies by gestational age, 2015–2016

Gestational age (weeks)*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
20–27	368	1,620	227.2
28–31	138	1,190	116.0
32–36	556	11,148	49.9
37–41	3,572	145,578	24.5
> 41	12	661	18.2

* Excludes pregnancies where gestation is unknown.

BIRTHWEIGHT

The rate of congenital anomalies in babies weighing less than 1,000 grams at birth was 208.0 per 1,000 pregnancies and this decreased with increasing birthweight (Table 4). For low-birthweight babies, the risk of having a congenital anomaly was three and a half times higher than for babies weighing 2,500 grams or more (RR 3.50, 95% CI 3.27–3.75, p = < 0.0001). In this report, a low birth weight baby is a baby weighing less than 2,500 grams at birth.

The rate of congenital anomalies in babies decreased with increasing birthweight. This is consistent with previous findings (Table 5).

Table 4: Congenital anomalies by birthweight, 2015–2016

Weight in grams*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
< 1,000	352	1,692	208.0
1,000–2,499	630	9,814	64.2
≥ 2,500	3,622	148,640	24.4

* Excludes terminations less than 20 weeks and/or where weight was not stated.

Table 5: Congenital anomalies by birthweight and reporting years

Weight in grams*	Congenital anomalies / 1,000 pregnancies 2015–2016	Congenital anomalies / 1,000 pregnancies 2013–2014	Congenital anomalies / 1,000 pregnancies 2007–2009
< 2,500	85.3	77.3	118.3
≥ 2,500	24.4	25.5	31.9

* Excludes terminations less than 20 weeks and/or where weight was not stated.

BIRTH PLURALITY

By birth plurality, 66.4 per cent (*n* = 4,865) of congenital anomaly cases were singleton and 2.7 per cent (*n* = 195) were multiple births. Plurality data was incomplete for 30.9 per cent (*n* = 2,266) of cases. In 2015–2016, the rate of congenital anomaly in singleton births was 31.2 per 1,000 pregnancies (3.1 per cent) and 42.0 per 1,000 (4.2 per cent) pregnancies for multiple births (Figure/Table 6). The risk of congenital anomaly was 35.0 per cent higher in multiple births than singleton births (RR 1.35, 95% CI 1.17–1.55, *p* = 0.0001).

Fertility treatments such as in vitro fertilization (IVF) and assisted reproduction technology (ART) commonly result in multiple births.[15] Given this, the contribution of ART to congenital anomalies is uncertain. There have been some reports of a positive association between ART and major congenital anomalies,[16] however other studies found no association.[17,18]

Figure 6: Congenital anomalies by birth plurality, 2015–2016

Table 6: Congenital anomalies by birth plurality, 2015–2016

Birth plurality	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
Single births	4,865	156,021	31.2
Multiple births	195	4,640	42.0

PERINATAL OUTCOMES

In 2015–16, the perinatal mortality rate (excluding terminations) in babies with one or more congenital anomalies was 22.0 per 1,000 pregnancies compared with 6.3 per 1,000 pregnancies for babies not having a congenital anomaly.

Pregnancies with congenital anomalies were four times more likely to result in perinatal death (excluding terminations) compared with pregnancies with babies not having a congenital anomaly (RR 4.07, 95% CI 3.45–4.80) p = < 0.0001.

Of all reported pregnancies with congenital anomalies, 17.7 per cent (n = 1,300) resulted in termination – 3.7 per cent (n = 269) before 20 weeks' gestation, 3.9 per cent (n = 284) after 20 weeks and the gestation period was unknown in 10.5 per cent (n = 747) of cases. More than two-thirds (67.1 per cent, n = 4,913) of reported pregnancies with congenital anomalies resulted in a live birth. Of these, 1.3 per cent died before 28 days of age (neonatal death, n = 64). Still birth occurred in 1.3 per cent (n = 97) of reported pregnancies with congenital anomalies. Perinatal outcome data was incomplete for 13.9 per cent of births (n = 1,016). Appendix 5 shows perinatal outcomes for selected major anomalies.

Congenital anomalies by maternal characteristics

MOTHER'S AGE AT BIRTH

Maternal age is a known risk factor for abnormal fetal development and increased risk of chromosomal abnormalities, including Down syndrome.[19] In Australia, perinatal deaths due to congenital abnormalities increased with increasing maternal age.[1]

In 2015 and 2016, women aged 35 years or older were 59 per cent more likely to have a baby with a major congenital anomaly than those younger than 35 years of age (RR 1.59, 95% CI 1.51–1.67, p = < 0.0001). The rate of congenital anomalies was highest in women aged 40–44 years, followed by those aged 35–39 and women younger than 20 years of age (Table 7).

Table 7: Congenital anomalies by mother's age at birth, 2015–2016

Mother's age group*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
< 20	98	2,515	39.0
20–24	557	15,779	35.3
25–29	1,336	41,444	32.2
30–34	2,092	60,696	34.5
35–39	1,563	33,416	46.8
40–44	679	8,109	83.7

* Excludes cases where age or date of birth was not stated.

ABORIGINAL WOMEN

The rate of congenital anomalies was similar for Aboriginal women (31.3 per 1,000 pregnancies) and non-Aboriginal women (28.6 per 1,000 pregnancies) in 2015–2016, indicating comparable risk of having a baby with a major congenital anomaly (RR 1.09, 95% CI 0.87–1.38, p = 0.44) (Table 8).

Table 8: Congenital anomalies by Aboriginality, 2015–2016

Aboriginality*	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
Aboriginal	72	2,300	31.3
Non-Aboriginal	4,505	157,394	28.6

* Aboriginality is inclusive of Aboriginal and Torres Strait Islander people.

PARITY

The rate of congenital anomaly was 29.9 per 1,000 pregnancies in primiparous women and 27.8 per 1,000 pregnancies in multiparous women in 2015 and 2016. For primiparous women, the risk of having a baby with a major congenital anomaly was seven per cent higher than for multiparous women (RR 1.07, 95% CI 1.01–1.14, p = 0.01) (Table 9).

Table 9: Congenital anomalies by parity, 2015–2016

Parity	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
Primiparous	2,105	70,502	29.9
Multiparous	2,494	89,710	27.8

MOTHER'S COUNTRY OF BIRTH

In 2015–2016 the rate of congenital anomalies was highest for babies of women born in North Africa and the Middle East (Table 10). Babies of women born in North Africa and the Middle East were 27 per cent more likely to have a major congenital anomaly than babies of women born in Australia or other countries (RR 1.27, 95% CI 1.11–1.45, p = 0.0004).

Congenital anomalies can be caused by a range of factors including environmental factors (exposure to alcohol, drugs, medications, chemicals and radiation) socio-demographic factors such as maternal age, ethnicity and maternal health prior to pregnancy. Genetics may also result in congenital anomalies. Everyone has variations in their genes which do not usually cause any health impacts. As genetic information is passed down from generation to generation, couples that have a similar genetic make-up are more likely to have a common gene variation that may result in a congenital anomaly. Genetic screening of these parents and others who have a child with a genetic disorder, prior to conception may identify potential inherited disorders.[20]

Table 10: Congenital anomalies by mother's country of birth, 2015–2016

Mother's country of birth	Number of cases with at least one congenital anomaly	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
North Africa and the Middle East	215	5,678	37.9
Sub-Saharan Africa	119	3,545	33.6
Oceania and Antarctica (excludes Australia)	145	4,504	32.2
North-West Europe	150	4,698	31.9
Australia	3,053	100,457	30.4
Southern and Central Asia	494	16,760	29.5
Southern and Eastern Europe	85	2,934	29.0
South-East Asia	273	10,277	26.6
North-East Asia	203	8,328	24.4
Americas	53	2,208	24.0
Not stated / inadequately described	2,536	3,606	703.3

SMOKING

Smoking during pregnancy increases the risk of health problems for mothers and their babies. Babies born to mothers who smoke are more likely to have poorer perinatal outcomes such as low birthweight, are at a greater risk of sudden unexpected death in infancy (SUDI or SIDS) and are at increased risk of certain congenital anomalies such as cleft lip and cleft palate.[21] Avoiding exposure to cigarette smoke during pregnancy will reduce the risk of adverse outcomes for both mothers and babies.

In Australia, 10 per cent of women report smoking at some time during pregnancy.[1] The rate is likely to be as high as 25 per cent because of under-reporting due to the social stigma associated with smoking during pregnancy.[22] Programs that support women to quit smoking during pregnancy are important components of antenatal and postnatal care.

In Victoria during 2015–2016, babies exposed to smoke in utero were 12 per cent more likely to have a major congenital anomaly (RR 1.12, 95% CI 1.02–1.23, p = 0.02). When comparing individual anomalies and maternal smoking status, no statistically significant associations were found.

Table 11: Congenital anomalies by smoking status

Smoked at all during pregnancy	Congenital anomaly cases	Number of reported pregnancies	Congenital anomalies / 1,000 pregnancies
Yes	476	14,683	32.4
No	4007	138,073	29.0

OBESITY

Obesity in pregnancy contributes to increased morbidity and mortality for mothers and babies. Pregnant women who are obese have higher rates of congenital anomalies, stillbirth and neonatal death compared with mothers who are not obese.[1] A body mass index (BMI) of 35 or higher is considered significantly obese. In this report, obesity refers to women with a BMI equal to or greater than 35 kg/m^2 and non-obese refers to a BMI less than 35 kg/m^2.

In 2015 and 2016, women with a BMI of 35 kg/m^2 or higher were 13 per cent likely to have a baby with a congenital anomaly compared with non-obese women (RR 1.13, 95% CI 1.02–1.25, p = 0.02). The rate of congenital anomalies in women having a BMI of 35 kg/m^2 and above is 31.2 per 1,000 pregnancies compared with 27.7 per 1,000 pregnancies in women having a BMI of less than 35 kg/m^2. Table 12 shows the rates of selected key anomalies in obese women.

Table 12: Selected key congenital anomalies in obese women, 2015–2016

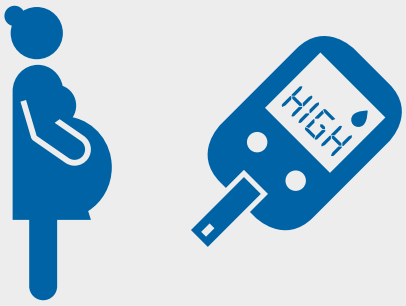
ICD-10-AM	Congenital anomaly	Congenital anomalies / 1,000 pregnancies	RR (95% CI)
Q54	Hypospadias	5.4	1.05 (0.74–1.50)
Q62	Obstructive defects of renal pelvis	1.9	0.79 (0.52–1.20)
Q210	Ventricular septal defect	1.9	1.14 (0.74–1.77)
Q90	Trisomy 21 (Down syndrome)	0.9	1.16 (0.62–2.15)
Q65	Deformities of hip	0.8	0.59 (0.30–1.15)
Q35	Cleft palate	0.8	1.60 (0.80–3.20)
Q03	Hydrocephalus	0.7	3.40 (1.55–7.45)*
Q00, Q01, Q05	All neural tube defects	0.6	2.08 (0.93–4.64)
Q37	Cleft lip and palate	0.6	1.60 (0.73–3.52)
Q234	Hypoplastic left heart syndrome	0.6	2.19 (0.98–4.90)
Q60	Renal agenesis and dysgenesis	0.5	1.43 (0.61–3.33)
Q251	Coarctation of aorta	0.5	2.30 (0.96–5.51)
Q790	Diaphragmatic hernia	0.4	2.20 (0.85–5.72)
Q05	Spina bifida	0.4	2.38 (0.91–6.21)
Q61	Cystic kidney disease	0.3	0.72 (0.26–1.98)
Q71-Q73	Limb reduction defects	0.3	1.49 (0.45–4.94)
Q213	Tetralogy of Fallot	0.3	1.25 (0.45–3.51)
Q910-Q913	Trisomy 18	0.3	2.10 (0.62–7.16)
Q36	Cleft lip	0.3	1.83 (0.64–5.24)
Q792	Exomphalos	0.3	2.16 (0.75–6.27)
Q42	Absence, atresia and stenosis of large intestine	0.3	2.38 (0.81–6.96)
Q2031	Transposition of great vessels	0.2	0.55 (0.13–2.28)
Q00	Anencephaly	0.2	1.98 (0.44–8.85)
Q914-Q917	Trisomy 13	0.2	1.49 (0.34–6.46)
Q793	Gastroschisis	0.1	0.34 (0.05–2.48)
Q41	Absence, atresia and stenosis of small intestine	0	0.25 (0.02–4.17)
Q02	Microcephaly	0	0.57 (0.03–9.66)
Q01	Encephalocele	0	1.70 (0.09–32.88)
Q39	Malformations of oesophagus	0	0.36 (0.02–6.01)

* Statistically significant.

PRE-GESTATIONAL DIABETES

Women who have pre-gestational diabetes are at increased risk of adverse perinatal outcomes.[23] Controlling maternal glycaemia before and particularly in early pregnancy (during fetal organ development) through counselling, weight management, diet and appropriate hypoglycaemic therapy may decrease the frequency of certain congenital anomalies.[24] As these mothers have been found to be at increased risk of neural tube anomalies, prophylactic folate therapy of 1–5mg daily is essential.[24] Aboriginal mothers are also more likely to be obese or to have pre-existing diabetes.[1]

In 2015 and 2016, women having pre-gestational diabetes were 43 per cent more likely to have a baby with a congenital anomaly compared with women not having pre-gestational diabetes (RR 1.43, 95% CI 1.17–1.75, p = 0.0006). The rate of congenital anomalies in babies born to women having pre-gestational diabetes was 64.7 per 1,000 pregnancies compared with 44.8 per 1,000 pregnancies in women not having pre-gestational diabetes. Table 13 shows the rates of selected key anomalies in mothers with pre-gestational diabetes.



Women with pre-gestational diabetes were 43 per cent more likely to have a baby with a congenital anomaly compared to women not having pre-gestational diabetes.

Table 13: Selected key congenital anomalies in women with pre-gestational diabetes, 2015–2016

ICD-10-AM	Congenital anomaly	Congenital anomalies / 1,000 pregnancies	RR (95% CI)
Q54	Hypospadias	16.3	3.22 (1.78–5.83)*
Q210	Ventricular septal defect	5.4	3.04 (1.44–6.43)*
Q62	Obstructive defects of renal pelvis	2.3	0.90 (0.29–2.80)
Q213	Tetralogy of Fallot	2.3	7.48 (2.33–23.99)*
Q2031	Transposition of great vessels	2.3	7.18 (2.24–23.00)*
Q03	Hydrocephalus	1.6	5.33 (1.29–21.93)*
Q61	Cystic kidney disease	1.6	2.96 (0.73–12.02)
Q35	Cleft palate	1.6	3.11 (0.77–12.66)
Q251	Coarctation of aorta	1.6	5.99 (1.45–24.76)*
Q00, Q01, Q05	All neural tube defects	0.8	1.45 (0.20–10.37)
Q37	Cleft lip and palate	0.8	1.71 (0.24–12.32)
Q792	Exomphalos	0.8	1.69 (0.10–27.52)
Q234	Hypoplastic left heart syndrome	0.8	1.97 (0.27–14.17)
Q05	Spina bifida	0.8	2.92 (0.40–21.24)
Q42	Absence, atresia and stenosis of large intestine	0.8	4.80 (0.65–35.37)
Q65	Deformities of hip	0	0.30 (CI 0.02–4.87)
Q90	Trisomy 21 (Down syndrome)	0	0.12 (0.01–1.95)
Q60	Renal agenesis and dysgenesis	0	0.96 (0.06–15.50)
Q71-Q73	Limb reduction defects	0	1.84 (0.11–30.11)
Q910-Q913	Trisomy 18	0	0.47 (0.03–7.62)
Q790	Diaphragmatic hernia	0	1.48 (0.09–24.06)
Q36	Cleft lip	0	1.90 (0.12–31.09)
Q793	Gastroschisis	0	1.24 (0.08–20.04)
Q00	Anencephaly	0	1.79 (0.11–29.19)
Q41	Absence, atresia and stenosis of small intestine	0	2.10 (0.13–34.44)
Q914-Q917	Trisomy 13	0	0.76 (0.05–12.31)
Q02	Microcephaly	0	3.87 (0.23–64.60)
Q01	Encephalocele	0	6.31 (0.37–108.35)
Q39	Malformations of oesophagus	0	2.92 (0.18–48.32)

* Statistically significant.

SOCIOECONOMIC STATUS

Socioeconomic status may be an indirect determinant for congenital anomalies. Pregnant women may not have access to sufficient and nutritious foods, healthcare services and preventive strategies, (such as access to folic acid) and may have increased exposure to smoke, alcohol or other infections.[24]

In 2015–2016 the rate of congenital anomalies for women in the lowest socioeconomic quintile was 28.1 per 1,000 pregnancies compared with 27.7 per 1,000 pregnancies in the highest quintile (Table 14). No significant association between socioeconomic status and the risk of congenital anomaly was found (RR 0.99, 95% CI 0.92–1.07, p = 0.81).

Table 14: Congenital anomalies by mother's socioeconomic status, 2015–2016

Socioeconomic status quintiles*	Congenital anomaly cases	Selected pregnancies	Congenital anomalies / 1,000 selected pregnancies
1	870	30,930	28.1
2	877	28,790	30.5
3	973	34,981	27.8
4	1,009	36,329	27.8
5	705	25,479	27.7

* Socioeconomic status used is SEIFA index, as measured by Australian Bureau of Statistics.

Selected congenital anomalies

CLEFT PALATE (Q35)

Definition: Cleft palate is a closure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip. This definition excludes cleft plate with cleft lip, cleft uvula, functional short palate and high narrow palate.

Trend: The prevalence of cleft palate has declined since 1998, however it has remained relatively stable since 2013. Trend shown in Figure 7. Of all the congenital anomaly cases reported in 2015 and 2016, 1.1 per cent (*n* = 79) had cleft palate.

Risk factors: Multiple births increased the risk of cleft palate almost threefold. The risk of cleft palate in a baby did not vary with maternal age, with the gender of the baby or with pre-gestational diabetes, obesity or socioeconomic status (Table 15).

Figure 7: Cleft palate per 1,000 pregnancies, 1998–2016

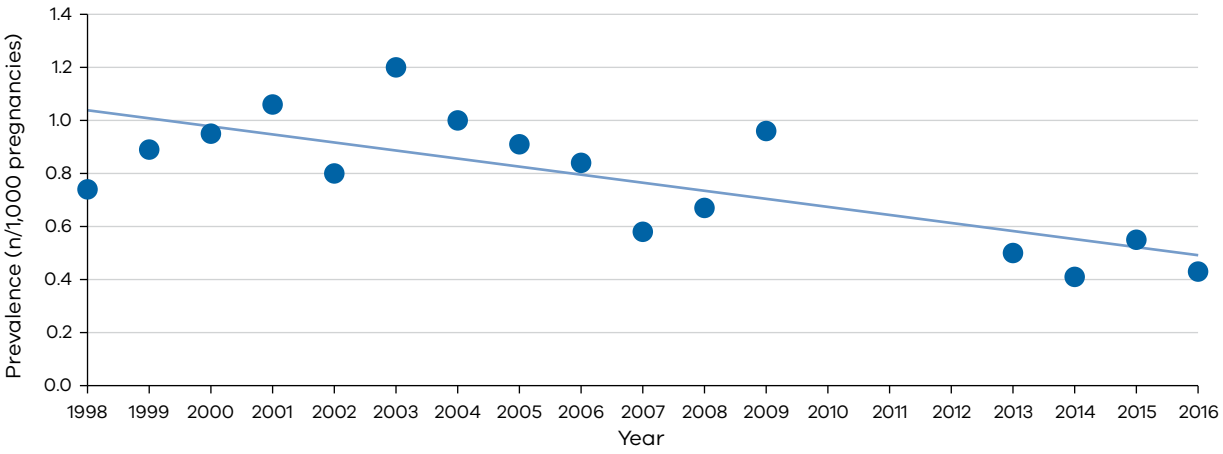


Table 15: Cleft palate by selected maternal and child characteristics, 2015–2016

Characteristic		Cleft palate / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.5 0.5	1.09 (0.66–1.80), 0.73
Plurality	Single (Ref) Multiple	0.5 1.3	2.79 (1.22–6.42)*, 0.02
Gender	Male (Ref) Female	0.4 0.6	1.62 (1.03–2.54)*, 0.04
Pre-gestational diabetes	No (Ref) Yes	0.5 1.6	3.11 (0.77–12.66), 0.11
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.5 0.8	1.60 (0.80–3.20), 0.19
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.5 0.5	0.89 (0.50–1.58), 0.69

* Statistically significant.

CLEFT LIP (Q36)

Definition: Cleft lip is a closure defect (partial or complete) of the upper lip, excluding the alveolar ridge and palate.

Trend: The prevalence of cleft lip has declined from 0.3/1,000 pregnancies in 2013–14 to 0.2/1,000 pregnancies in 2015–16. Trend shown in Figure 8. Of all the congenital anomaly cases reported in 2015 and 2016, 0.4 per cent (*n* = 31) had cleft lip.

Risk factors: Female babies were 73 per cent less likely to develop cleft lip compared with male babies. The risk of cleft lip in a baby did not vary with maternal age, birth plurality, pre-gestational diabetes, obesity or socioeconomic status (Table 16).

Figure 8: Cleft lip per 1,000 pregnancies, 1998–2016

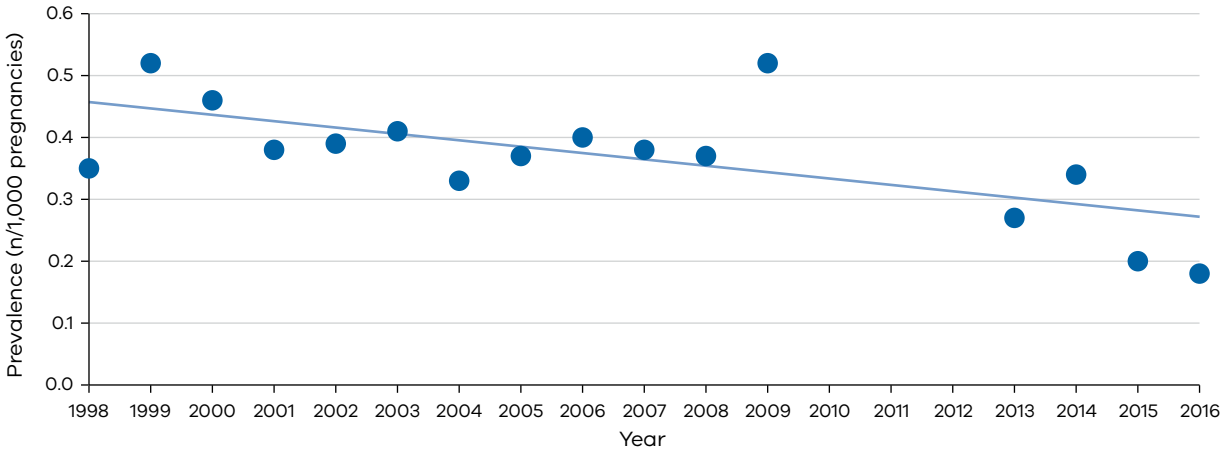


Table 16: Cleft lip by selected maternal and child characteristics, 2015–2016

Characteristic		Cleft lip / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.2 0.2	1.03 (0.46–2.30), 0.94
Plurality	Single (Ref) Multiple	0.2 0.2	1.13 (0.15–8.31), 0.90
Gender	Male (Ref) Female	0.3 0.1	0.27 (0.11–0.67)*, 0.00
Pre-gestational diabetes	No (Ref) Yes	0.2 0.0	1.90 (0.12–31.09), 0.65
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.3	1.83 (0.64–5.24), 0.26
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.2 0.2	1.06 (0.43–2.60), 0.90

* Statistically significant.

CLEFT LIP AND PALATE (Q37)

Definition: Cleft lip and palate is a closure defect (partial or complete) of the upper lip, alveolar ridge and palate.

Trend: The prevalence of cleft lip and palate has declined since 1998, however it has remained relatively stable since 2013. Trend shown in Figure 9. Of all congenital anomaly cases reported in 2015 and 2016, 1.0 per cent (*n* = 71) had cleft lip and palate.

Risk factors: Female babies were 45 per cent less likely to develop cleft lip and palate compared with male babies. The risk of cleft lip and palate in a baby did not vary with maternal age, birth plurality, pre-gestational diabetes, obesity or socioeconomic status (Table 17).

Figure 9: Cleft lip and palate per 1,000 pregnancies, 1998–2016

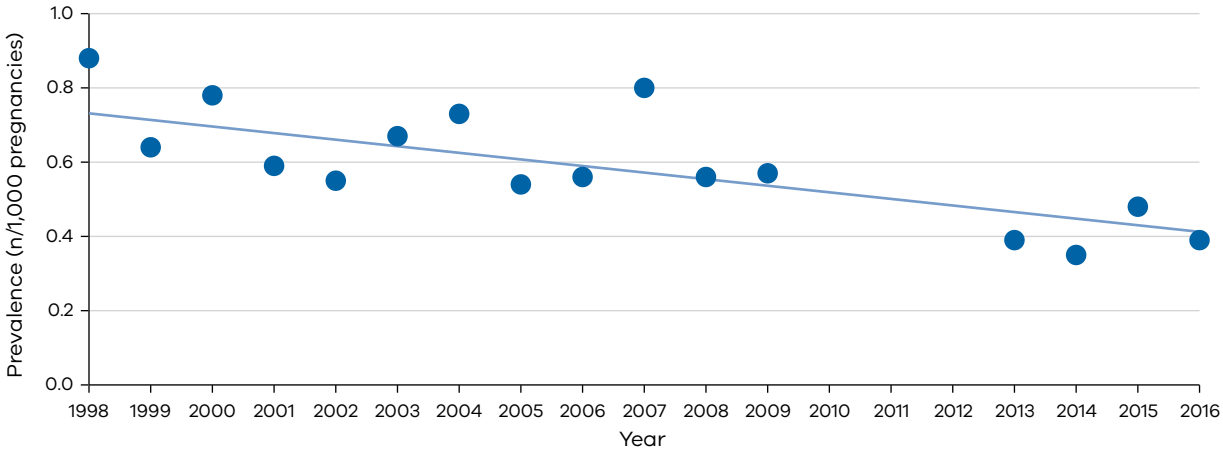


Table 17: Cleft lip and palate by selected maternal and child characteristics, 2015–2016

Characteristic		Cleft lip and palate / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.4 0.6	1.55 (0.94–1.73), 0.08
Plurality	Single (Ref) Multiple	0.4 0.7	1.55 (0.49–4.91), 0.46
Gender	Male (Ref) Female	0.5 0.3	0.55 (0.33–0.92)*, 0.02
Pre-gestational diabetes	No (Ref) Yes	0.5 0.8	1.71 (0.24–12.32), 0.59
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.4 0.6	1.60 (0.73–3.52), 0.24
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.4	0.64 (0.30–1.34), 0.24

* Statistically significant.

MALFORMATIONS OF OESOPHAGUS (Q39)

Definition: Malformations of the oesophagus include oesophageal atresia and/or stenosis, defined as the absence of continuity or narrowing of the oesophagus, with or without tracheal fistula.

Trend: The prevalence of congenital malformations of the oesophagus has declined since 1998, however it has remained relatively stable since 2013 (Figure 10). Of all congenital anomaly cases reported in 2015 and 2016, 0.3 per cent (*n* = 20) had congenital malformations of the oesophagus.

Risk factors: Female babies were 65 per cent less likely to develop congenital malformations of the oesophagus compared with male babies. The risk of congenital malformations of the oesophagus in a baby did not vary with maternal age, birth plurality, pre-gestational diabetes, obesity or socioeconomic status (Table 18).

Figure 10: Malformations of the oesophagus per 1,000 pregnancies, 1995–2016

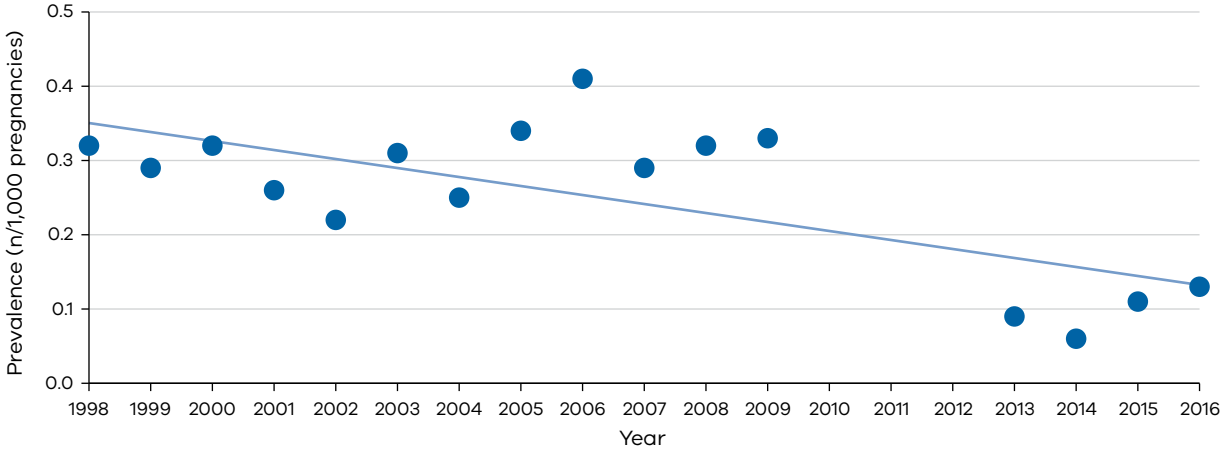


Table 18: Malformations of the oesophagus by selected maternal and child characteristics, 2015–2016

Characteristic		Oesophageal atresia and/or stenosis / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.1 0.2	1.27 (0.49–3.30), 0.63
Plurality	Single (Ref) Multiple	0.1 0.2	1.79 (0.24–13.37), 0.57
Gender	Male (Ref) Female	0.2 0.1	0.35 (0.13–0.96)*, 0.04
Pre-gestational diabetes	No (Ref) Yes	0.1 0.0	2.92 (0.18–48.32), 0.45
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.0	0.36 (0.02–6.01), 0.48
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.1	0.76 (0.22–2.61), 0.66

* Statistically significant.

ABSENCE, ATRESIA AND STENOSIS OF SMALL INTESTINE (Q41)

Definition: Absence, atresia and stenosis of the small intestine is the complete or partial occlusion of the lumen of a segment of the small intestine. It can involve a single area or multiple areas of the duodenum, jejunum or ileum.

Trend: The prevalence of congenital absence, atresia and stenosis of the small intestine has declined since 1998 and continues to decline (Figure 11). Of all the congenital anomaly cases reported in 2015 and 2016, 0.4 per cent (*n* = 28) had congenital absence, atresia and stenosis of the small intestine.

Risk factors: The risk of congenital absence, atresia and stenosis of the small intestine in a baby did not vary with maternal age, birth plurality, the gender of the baby, or with pre-gestational diabetes, obesity or socioeconomic status (Table 19).

Figure 11: Absence, atresia and stenosis of small intestine per 1,000 pregnancies, 1998–2016

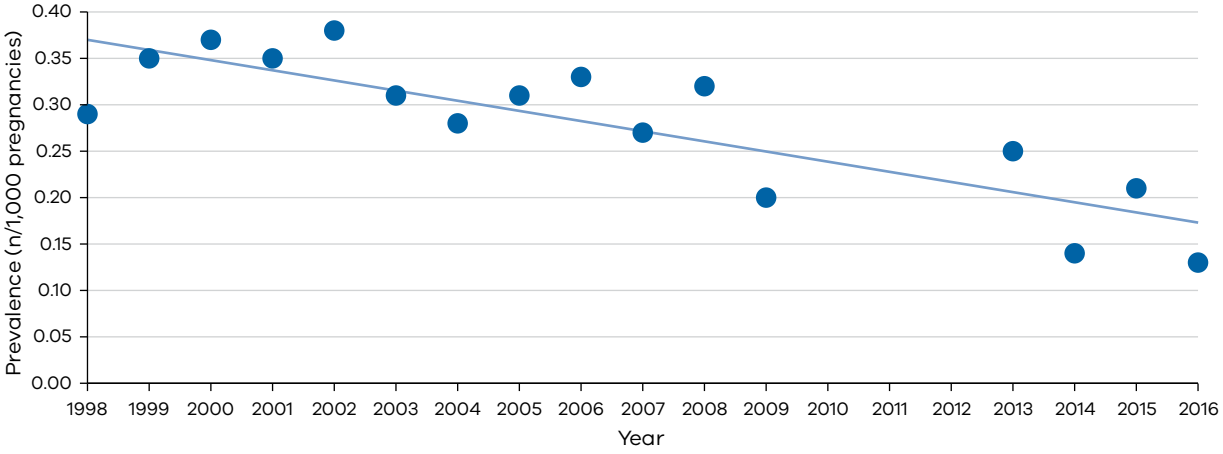


Table 19: Absence, atresia and stenosis of small intestine by selected maternal and child characteristics, 2015–2016

Characteristic		Absence, atresia and stenosis of small intestine / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.2 0.2	0.81 (0.33–2.00), 0.64
Plurality	Single (Ref) Multiple	0.2 0.4	2.62 (0.62–11.01), 0.19
Gender	Male (Ref) Female	0.2 0.1	0.77 (0.35–1.67), 0.50
Pre-gestational diabetes	No (Ref) Yes	0.2 0.0	2.10 (0.13–34.44), 0.60
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.0	0.25 (0.02–4.17), 0.34
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.2	0.85 (0.30–2.51), 0.78

ABSENCE, ATRESIA AND STENOSIS OF LARGE INTESTINE (Q42)

Definition: Absence, atresia and stenosis of the large intestine is the absence of continuity of the anorectal canal or communication between the rectum and anus, or narrowing of the anal canal, with or without fistula to neighbouring organs. Excludes ectopic anus.

Trend: The prevalence of congenital absence, atresia and stenosis of the large intestine has declined since 1998 and continues to decline (Figure 12). Of all the congenital anomaly cases reported in 2015 and 2016, 0.4 per cent (*n* = 26) had congenital absence, atresia and stenosis of the large intestine.

Risk factors: Women aged 35 years or older were two and a half times more likely to give birth to a baby with congenital absence, atresia and stenosis of the large intestine compared with women younger than 35 years of age. The risk of congenital absence, atresia and stenosis of the large intestine did not vary with birth plurality, with the gender of the baby or with pre-gestational diabetes, obesity or socioeconomic status (Table 20).

Figure 12: Absence, atresia and stenosis of the large intestine per 1,000 pregnancies, 1998–2016

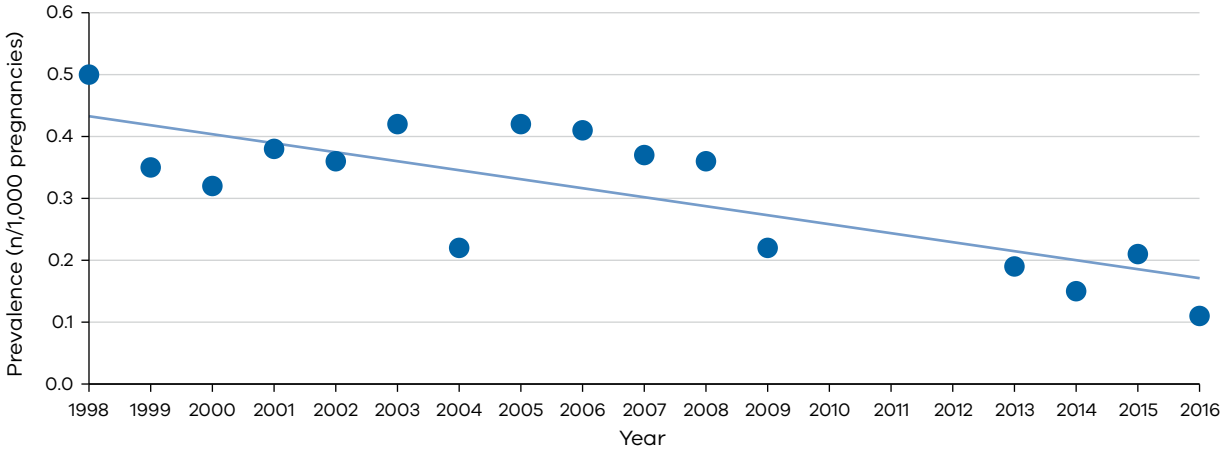


Table 20: Absence, atresia and stenosis of the large intestine by selected maternal and child characteristics, 2015–2016

Characteristic		Absence, atresia and stenosis of large intestine / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.1 0.3	2.54 (1.17–5.49)*, 0.02
Plurality	Single (Ref) Multiple	0.2 0.4	2.83 (0.67–11.98), 0.16
Gender	Male (Ref) Female	0.2 0.2	0.90 (0.41–1.94), 0.28
Pre-gestational diabetes	No (Ref) Yes	0.2 0.8	4.80 (0.65–35.37), 0.12
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.3	2.38 (0.81–6.96), 0.11
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.2 0.2	1.22 (0.49 to 3.03), 0.67

* Statistically significant.

DIAPHRAGMATIC HERNIA (Q790)

Definition: Diaphragmatic hernia is the herniation (protrusion) of abdominal organs into the thorax through a defect of the diaphragm.

Trend: The prevalence of congenital diaphragmatic hernia has reduced slightly since 1998 (Figure 13). Of all the congenital anomaly cases reported in 2015 and 2016, 0.5 per cent (*n* = 40) had congenital diaphragmatic hernia.

Risk factors: The risk of congenital diaphragmatic hernia in a baby did not vary with maternal age, with birth plurality, with the gender of the baby or with pre-gestational diabetes, obesity or socioeconomic status (Table 21).

Figure 13: Diaphragmatic hernia per 1,000 pregnancies, 1998–2016

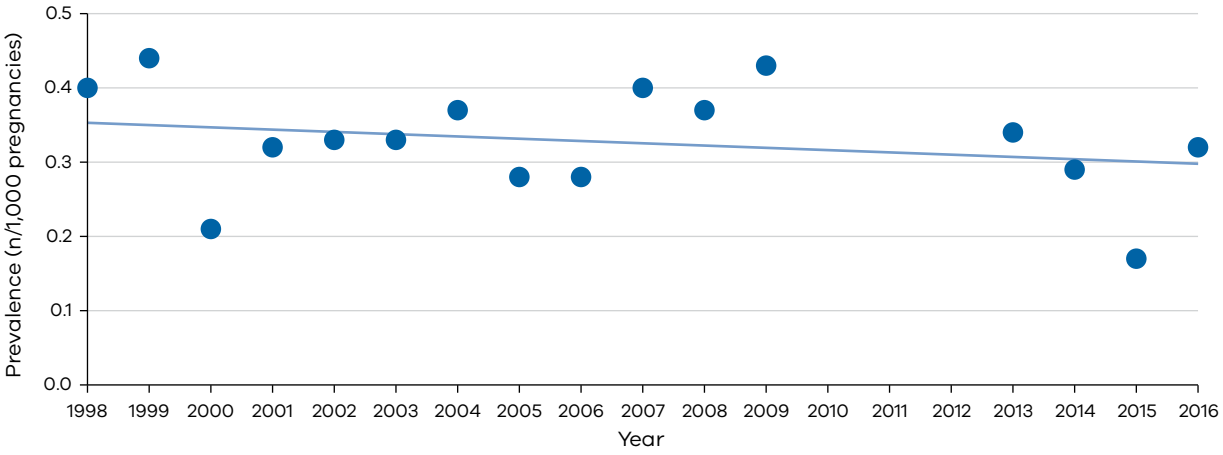


Table 21: Diaphragmatic hernia by selected maternal and child characteristics, 2015–2016

Characteristic		Diaphragmatic hernia / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 0.2	0.89 (0.42–1.87), 0.76
Plurality	Single (Ref) Multiple	0.3 0.0	0.42 (0.03–6.83), 0.54
Gender	Male (Ref) Female	0.2 0.2	1.11 (0.57–2.15), 0.77
Pre-gestational diabetes	No (Ref) Yes	0.3 0.0	1.48 (0.09–24.06), 0.78
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.4	2.20 (0.85–5.72), 0.11
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.2 0.2	1.14 (0.49–2.63), 0.76

GASTROSCHISIS (Q793)

Definition: Gastroschisis is the visceral herniation through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane.

Trend: The prevalence of gastroschisis has remained relatively stable between 1998 and 2016 (Figure 14). Of all the congenital anomaly cases reported in 2015 and 2016, 0.7 per cent (*n* = 48) had gastroschisis.

Risk factors: The risk of gastroschisis in babies varied with maternal age, with women younger than 20 years at nine times greater risk compared with women over 20 years of age (RR 9.26, 95 CI 3.93–21.78, *p* = < 0.0001). The risk of having a child with gastroschisis in women over 35 years was comparable to the risk in women under 35 years (Table 22).

Figure 14: Gastroschisis per 1,000 pregnancies, 1998–2016

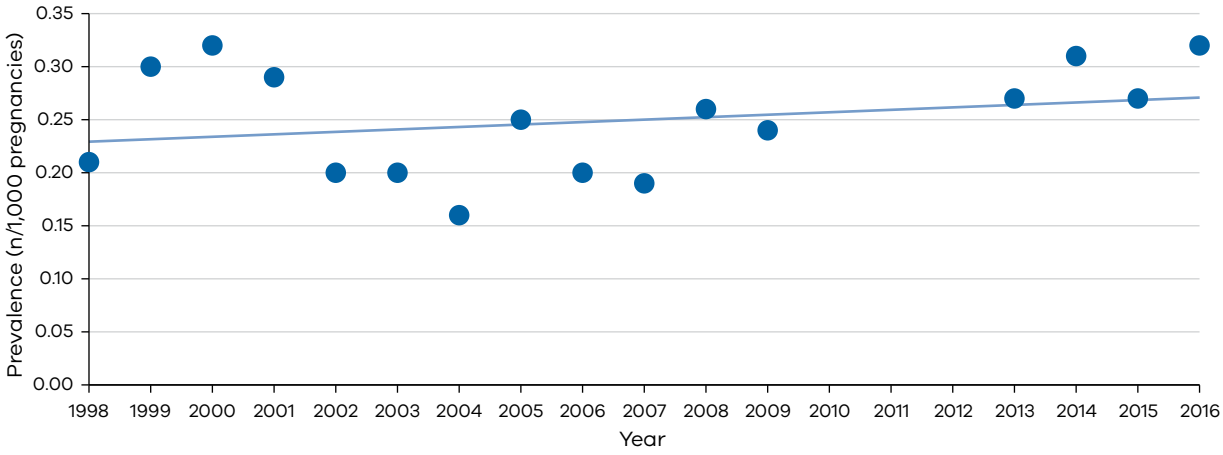


Table 22: Gastroschisis by selected maternal and child characteristics, 2015–2016

Characteristic		Gastroschisis / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 0.2	0.52 (0.23–1.16), 0.11
Plurality	Single (Ref) Multiple	0.3 0.0	0.35 (0.02–5.69), 0.46
Gender	Male (Ref) Female	0.3 0.2	0.73 (0.41–1.32), 0.30
Pre-gestational diabetes	No (Ref) Yes	0.3 0.0	1.24 (0.08–20.04), 0.88
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.1	0.34 (0.47–2.48), 0.29
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.2	1.08 (0.50–2.36), 0.84

EXOMPHALOS (Q792)

Definition: Exomphalos is the herniation of abdominal contents through the umbilical insertion and covered by membrane that may or may not be intact.

Trend: The prevalence of exomphalos has decreased from 0.3/1,000 pregnancies in 2013–14 to 0.2/1,000 pregnancies in 2015–16 (Figure 15). Of all the congenital anomaly cases reported in 2015 and 2016, 0.5 per cent (*n* = 35) had exomphalos.

Risk factors: The risk of exomphalos in a baby did not vary with maternal age, with birth plurality, with the gender of the baby or with pre-gestational diabetes, obesity or socioeconomic status (Table 23).

Figure 15: Exomphalos per 1,000 pregnancies, 1998–2016

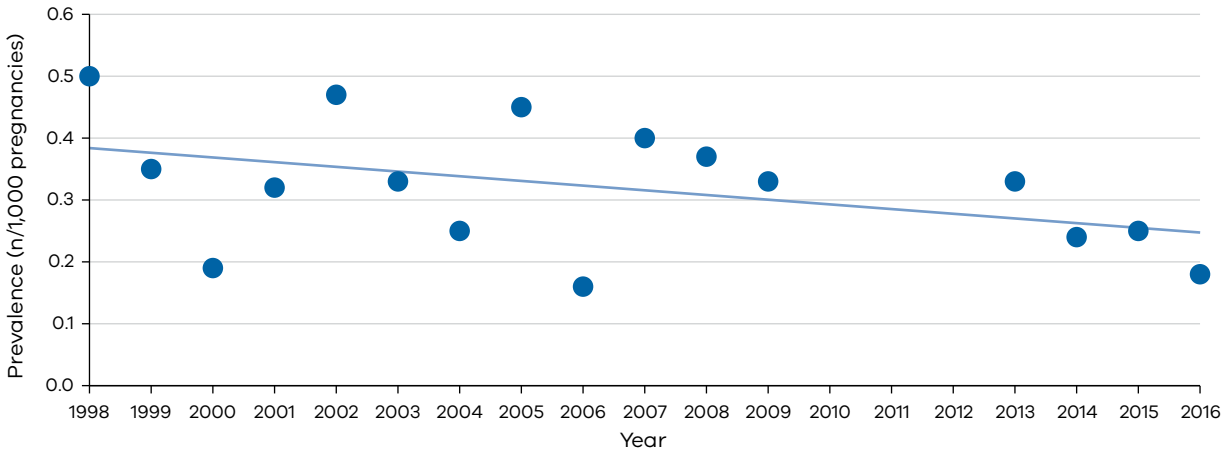


Table 23: Exomphalos by selected maternal and child characteristics, 2015–2016

Characteristic		Exomphalos / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.2 0.2	0.80 (0.35–1.84), 0.60
Plurality	Single (Ref) Multiple	0.2 0.4	2.19 (0.53–9.16), 0.28
Gender	Male (Ref) Female	0.2 0.2	0.74 (0.35–1.54), 0.42
Pre-gestational diabetes	No (Ref) Yes	0.2 0.0	1.69 (0.10–27.52), 0.71
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.2 0.3	2.16 (1.75–6.27), 0.16
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.2	1.62 (0.72–3.69), 0.25

TRISOMY 21 (Q90)

Definition: Trisomy 21, also called Down syndrome, is characterised by an additional chromosome 21.

Trend: The prevalence of trisomy 21 has remained relatively stable over time (Figure 16).[†] Among all the congenital anomaly cases reported in 2015 and 2016, 6.7 per cent (*n* = 492) had trisomy 21.

Risk factors: Mothers aged 35 years or older were five and a half times more likely to give birth to a baby with trisomy 21 than mothers younger than 35 years (Table 24).

Figure 16: Trisomy 21 per 1,000 pregnancies, 1998–2016

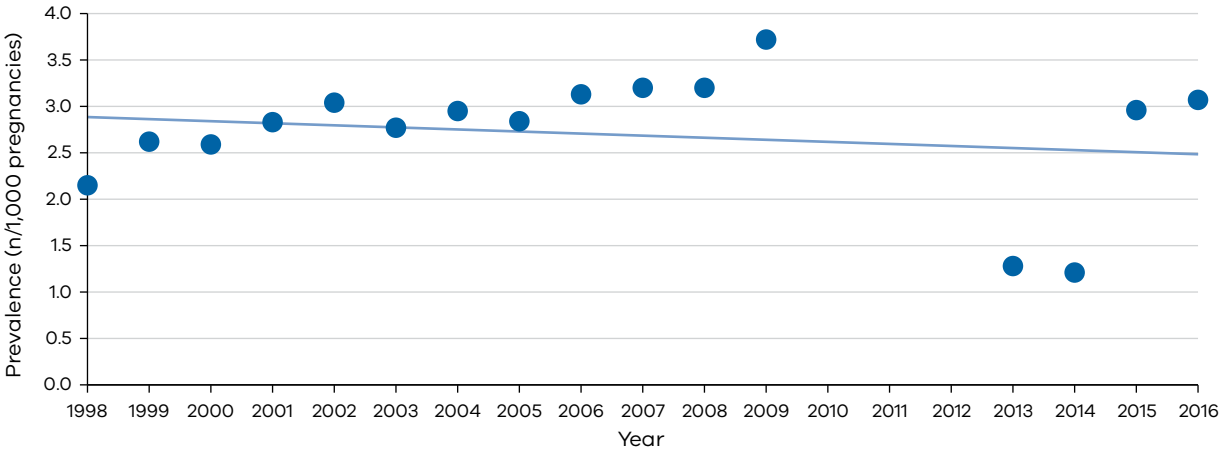


Table 24: Trisomy 21 by selected maternal and child characteristics, 2015–2016

Characteristic		Trisomy 21 / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	1.4 7.6	5.49 (4.54–6.64)*, < 0.0001
Plurality	Single (Ref) Multiple	1.3 0.7	0.51 (0.16–1.58), 0.24
Gender	Male (Ref) Female	1.5 1.3	0.84 (0.65–1.10), 0.20
Pre-gestational diabetes	No (Ref) Yes	3.2 0.0	1.12 (0.01–1.95), 0.14
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.8 0.9	1.16 (0.62–2.15), 0.64
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.9 1.0	0.94 (0.62–1.42), 0.76

* Statistically significant.

[†] Differences in trends in 2013–14 is attributed to a reduction in reporting of congenital anomalies as outlined in 'Data analysis' on page 5 of this report.

TRISOMY 13 (Q914, Q915, Q916, Q917)

Definition: Trisomy 13, also called Patau's syndrome, is characterised by an additional chromosome 13.

Trend: The prevalence of trisomy 13 has increased over time (Figure 17).‡ Among all the congenital anomaly cases reported in 2015 and 2016, 1.1 per cent (*n* = 78) had trisomy 13.

Risk factors: Mothers 35 years or older were four times more likely to give birth to a baby with trisomy 13 than mothers younger than 35 years (Table 25).

Figure 17: Trisomy 13 per 1,000 pregnancies, 1998–2016

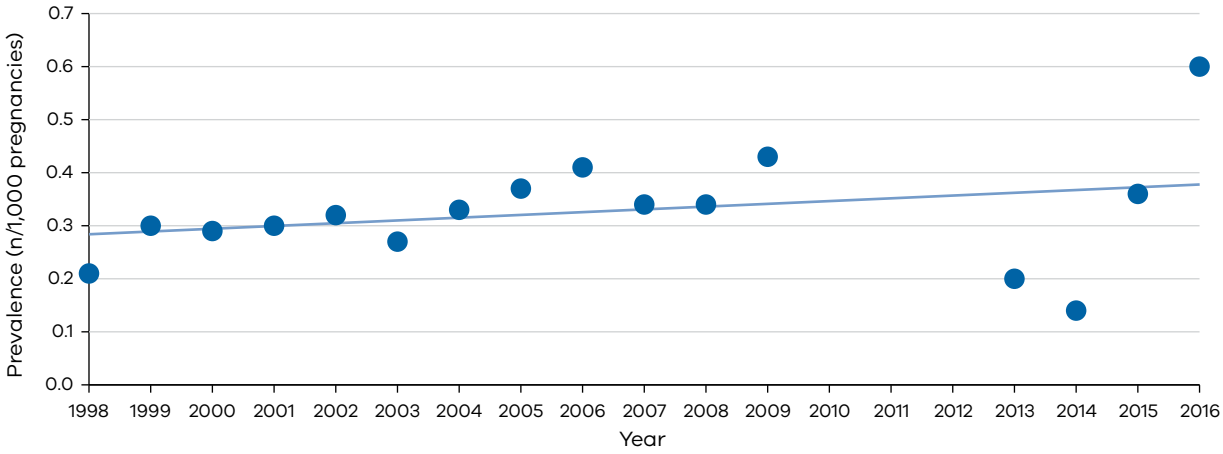


Table 25: Trisomy 13 by selected maternal and child characteristics, 2015–2016

Characteristic		Trisomy 13 / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 1.1	4.20 (2.65–6.65)*, < 0.0001
Plurality	Single (Ref) Multiple	0.2 0.4	2.19 (0.53–9.16), 0.28
Gender	Male (Ref) Female	0.2 0.2	1.11 (0.56–2.20), 0.77
Pre-gestational diabetes	No (Ref) Yes	0.5 0.0	0.76 (0.05–12.32), 0.85
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.2	1.49 (0.34–6.46), 0.60
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.1 0.1	1.01 (0.34–3.04), 0.98

* Statistically significant.

‡ Differences in trends in 2013–14 is attributed to a reduction in reporting of congenital anomalies as outlined in 'Data analysis' on page 5 of this report.

TRISOMY 18 (Q910, Q911, Q912, Q913)

Definition: Trisomy 18, also called Edwards' syndrome, is characterised by an additional chromosome 18.

Trend: The prevalence of trisomy 18 has remained relatively stable over time (Figure 18).[§] Among all the congenital anomaly cases reported in 2015 and 2016, 1.7 per cent (*n* = 128) had trisomy 18.

Risk factors: Mothers 35 years or older were seven times more likely to give birth to a baby with trisomy 18 than mothers younger than 35 years. Mothers in the lowest socioeconomic quintile were two and a half times more likely to give birth to a baby with trisomy 18 than mothers in the highest socioeconomic quintile (Table 26).

Figure 18: Trisomy 18 per 1,000 pregnancies, 1998–2016

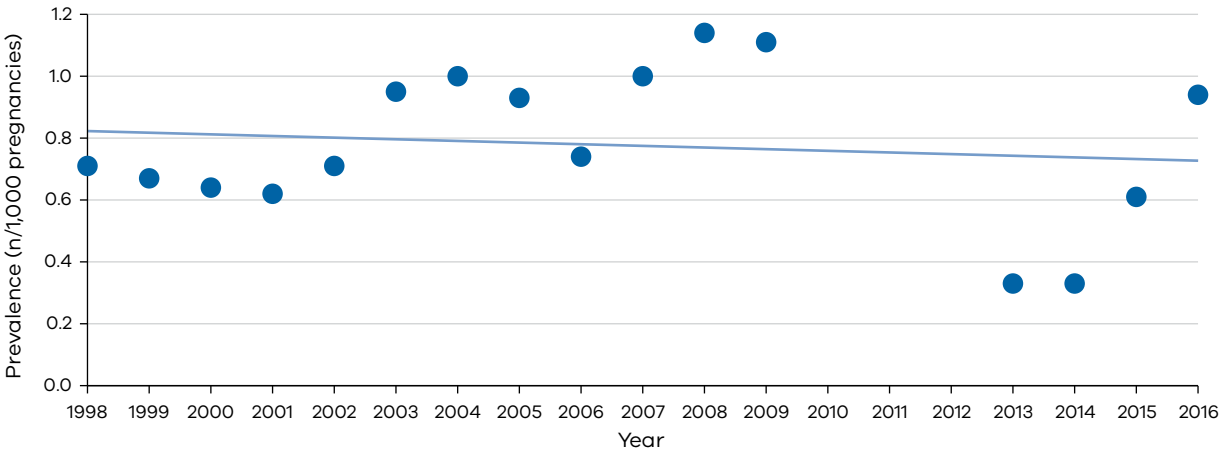


Table 26: Trisomy 18 by selected maternal and child characteristics, 2015–2016

Characteristic		Trisomy 18 / 1,000 pregnancies	RR (95% CI), p-value
Mother's age	< 35 years (Ref) ≥ 35 years	0.3 2.2	7.23 (4.90–10.65)*, < 0.0001
Plurality	Single (Ref) Multiple	0.3 0.4	1.62 (0.39–6.69), 0.51
Gender	Male (Ref) Female	0.3 0.2	0.82 (0.44–1.51), 0.52
Pre-gestational diabetes	No (Ref) Yes	0.8 0.0	0.47 (0.03–7.62), 0.60
Obesity	BMI < 35 (Ref) BMI ≥ 35	0.1 0.3	2.10 (0.62–7.16), 0.24
Socioeconomic status	Lowest quintile Higher quintiles (Ref)	0.3 0.1	2.50 (1.04–6.03)*, 0.04

* Statistically significant.

§ Differences in trends in 2013–14 is attributed to a reduction in reporting of congenital anomalies as outlined in 'Data analysis' on page 5 of this report.

Appendix 1: Selected major congenital anomalies by body system

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Nervous system			
Q00	Anencephaly and similar malformations	0.2	33
Q01	Encephalocele	0.1	10
Q02	Microcephaly	0.1	15
Q03	Congenital hydrocephalus	0.3	49
Q04	Other congenital malformations of brain	1.8	279
Q05	Spina bifida	0.3	46
Q06	Other congenital malformations of spinal cord	0.0	< 5
Q07	Other congenital malformations of nervous system	0.2	33
G702	Congenital and developmental myasthenia	0.0	0
G10	Huntington's disease	0.0	< 5
Total		**3.0**	**470**
Eye			
Q10	Congenital malformation of eyelid, lacrimal apparatus and orbit	0.0	5
Q11	Anophthalmos, microphthalmos and macrophthalamos	0.0	< 5
Q112	Microphthalmos	0.0	< 5
Q120	Congenital cataract	0.0	6
Q121–Q129	Other congenital lens malformations	0.0	< 5
Q13	Congenital malformations of anterior segment of eye	0.0	0
Q14	Congenital malformations of posterior segment of eye	0.0	< 5
Q15	Other congenital malformations of eye	0.0	6
H55	Congenital nystagmus and other irregular eye movements	0.0	0
H509	Congenital strabismus, unspecified	0.0	< 5
H219	Disorder of iris and ciliary body, unspecified	0.0	0
Total		**0.2**	**24**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Ear, face and neck			
Q16	Congenital malformations of ear causing impairment of hearing (excludes congenital deafness)	0.1	8
Q161	Congenital absence, atresia and stricture of auditory canal (external)	0.0	< 5
Q162	Absence of eustachian tube	0.0	0
Q163–Q165, Q169	Other malformations of ear causing impairment of hearing	0.0	5
Q17	Other congenital malformations of ear	0.4	71
Q170	Accessory auricle and ear tag	0.3	54
Q171	Macrotia	0.0	0
Q172–Q175, Q178–Q179	Other specified congenital malformations of ear	0.0	8
Q18	Other congenital malformations of face and neck	0.1	11
H90	Congenital deafness	0.0	< 5
Total		**0.6**	**93**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Cardiovascular system			
Q200	Common arterial trunk	0.0	< 5
Q2031	Transposition of great vessels	0.3	53
Q213	Tetralogy of Fallot	0.3	51
Q201	Double outlet right ventricle	0.1	14
Q204	Double inlet ventricle	0.1	9
Q205	Discordant atrioventricular connection	0.0	< 5
Q206	Isomerism of atrial appendages	0.0	< 5
Q208	Other congenital malformations of cardiac chambers and connections	0.2	25
Q209	Other congenital malformations of cardiac chambers and connections, unspecified	0.0	< 5
Q210	Ventricular septal defect	1.7	275
Q211	Atrial septal defect	0.3	44
Q212	Atrioventricular septal defect	0.1	22
Q22	Congenital malformations of pulmonary and tricuspid valves	0.7	113
Q23	Congenital malformations of aortic and mitral valves	0.6	93
Q24	Other congenital malformations of heart	2.3	362
Q250	Patent ductus arteriosus	0.3	45
Q251	Coarctation of aorta	0.3	42
Q253	Stenosis of aorta	0.1	11
Q254	Other and unspecified congenital malformations of aorta	0.4	65
Q255–Q257	Atresia, stenosis and other and unspecified congenital malformations of pulmonary artery	0.1	22
Q258–Q259	Congenital malformations of great arteries	0.1	13
Q26	Congenital malformations of great veins	0.2	32
Q27	Other congenital malformations of peripheral vascular system	0.3	40
Q28	Other congenital malformations of circulatory system	0.1	8
Total		**8.5**	**1,351**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Respiratory system			
Q30	Congenital malformations of nose	0.3	51
Q31	Congenital malformations of larynx	0.0	7
Q32	Congenital malformations of trachea and bronchus	0.0	< 5
Q33	Congenital malformations of lung	0.3	47
Q34	Other congenital malformation of respiratory system	0.2	24
Total		**0.8**	**132**
Gastrointestinal tract			
Q35	Cleft palate	0.5	79
Q36	Cleft lip	0.2	31
Q37	Cleft lip and palate	0.4	71
Q38	Other congenital malformations of tongue, mouth and pharynx	0.2	30
Q39	Congenital malformations of oesophagus	0.1	22
Q40	Other congenital malformations of upper alimentary tract	0.1	12
Q41	Congenital absence, atresia and stenosis of small intestine	0.2	29
Q42	Congenital absence, atresia and stenosis of large intestine	0.2	26
Q43	Other congenital malformations of intestine	0.2	29
Q44	Congenital malformations of gallbladder, bile ducts and liver	0.1	19
Q45	Other congenital malformations of digestive system	0.3	41
Total		**2.5**	**389**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Urogenital system			
Q50	Congenital malformations of ovaries, fallopian tubes and broad ligaments	0.0	< 5
Q51	Congenital malformations of uterus and cervix	0.0	< 5
Q52	Other congenital malformations of female genitalia	0.1	9
Q53	Undescended testes	1.7	268
Q54	Hypospadias	2.6	418
N47	Phimosis	0.1	10
Q55	Other congenital malformations of male genital organs	0.4	62
Q56	Indeterminate sex and pseudohermaphroditism	0.1	12
Q60	Renal agenesis and other reduction defects of kidneys	0.4	64
Q61	Cystic kidney disease	0.5	83
Q62	Congenital obstructive defects of renal pelvis and congenital malformations of ureter	2.7	426
Q63	Other congenital malformations of kidneys	0.5	78
Q64	Other congenital malformations of urinary system	0.4	57
Total		**9.4**	**1,492**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Musculoskeletal system			
Q65	Congenital deformities of hip	1.3	198
Q66	Congenital deformities of feet	2.1	336
Q67	Congenital musculoskeletal deformities of head, face, spine and chest	0.2	38
Q68	Other congenital musculoskeletal deformities	0.2	32
Q69	Polydactyly	1.0	153
Q70	Syndactyly	0.4	61
Q71	Reduction defects of upper limb	0.2	24
Q72	Reduction defects of lower limb	0.0	7
Q73	Reduction defects of unspecified limb	0.0	6
Q74	Other congenital malformations of limb(s)	0.9	141
Q75	Other congenital malformations of skull and face bones	0.4	57
Q76	Congenital malformations of spine and bony thorax	0.2	29
Q77	Osteochondrodysplasia with defects of growth of tubular bones and spine	0.0	< 5
Q78	Other osteochondrodysplasias	0.1	10
Q79	Congenital malformations of musculoskeletal system not elsewhere classified	1.1	167
Total		**8.0**	**1,263**
Integumentary system			
Q80	Congenital ichthyosis	0.1	8
Q81	Epidermolysis bullosa	0.0	5
Q82	Other congenital malformations of skin	0.5	78
Q83	Congenital malformations of breast	0.0	< 5
L81	Other disorders of pigmentation	0.0	0
Q84	Other congenital malformations of integument	0.0	7
Total		**0.6**	**99**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Chromosomal			
Q90	Down syndrome	3.1	492
Q91	Edwards' syndrome and Patau's syndrome	1.3	211
Q92	Other trisomies and partial trisomies of the autosomes, not elsewhere classified	6.0	951
Q93	Monosomies and deletions from chromosomes, not elsewhere classified	3.9	623
Q95	Balanced rearrangements and structural markers, not elsewhere classified	0.0	< 5
Q96	Turner's syndrome	0.8	128
Q97	Other female sex chromosome abnormalities, not elsewhere classified	0.1	23
Q98	Other male sex chromosome abnormalities, not elsewhere classified	0.3	53
Q99	Other chromosome abnormalities, not elsewhere classified	4.0	629
Total		**19.7**	**3,113**
Neoplasms			
C00–C97	Malignant neoplasms	0.0	< 5
D10–D36	Benign neoplasms	0.2	36
D180	Haemangioma	0.0	5
D181	Lymphangioma	0.2	24
D15	Benign neoplasm of other and unspecified intrathoracic organs	0.0	< 5
D37–D48	Neoplasms of uncertain or unknown behaviour	0.0	8
Total		**0.3**	**46**
Metabolic/endocrine/nutritional			
E031	Congenital hypothyroidism without goitre	0.0	< 5
E20–E35	Disorders of other endocrine glands	0.0	7
P704	Other neonatal hypoglycaemia	0.0	< 5
E70–E90	Metabolic disorders	0.2	24
Total		**0.0**	**37**

ICD-10-AM code*	Congenital anomaly	Number in 1,000 pregnancies	Number
Haematological			
D55–D59	Haemolytic anaemia	0.1	8
D65–D69	Coagulation defects, purpura and other haemorrhagic conditions	0.0	< 5
D70–D77	Other diseases of blood and blood-forming organs	0.0	< 5
P55	Immune haemolytic disease of fetus and newborn	0.0	< 5
P61	Other perinatal haematological disorders	0.0	6
Total		**0.1**	**22**
Other selected major congenital anomalies			
Q85	Phakomatoses, not elsewhere classified	0.0	< 5
Q890	Congenital malformations of spleen	0.0	< 5
Q891	Congenital malformations of adrenal gland	0.0	0
Q892	Congenital malformations of other endocrine glands	0.0	< 5
Q893	Situs inversus	0.0	7
Q894	Conjoined twins	0.0	< 5
Q8706	Pierre Robin sequence	0.0	< 5
Q8709	Other specified congenital malformation syndromes predominantly affecting facial appearance	0.0	0
Q8711	Cockayne's syndrome	0.0	0
Q8713	Noonan's syndrome	0.0	0
Q8714	Prader-Willi syndrome	0.0	< 5
Q8727	VATER association	0.0	0
Q874	Marfan's syndrome	0.0	< 5
Q878	Other specified congenital malformation syndromes, not elsewhere classified	0.1	11
Q897	Multiple congenital malformations, not elsewhere classified†	0.1	18
Q898	Other specified congenital malformations	0.0	< 5
Q899	Congenital malformations, unspecified	0.6	91
P351	Congenital cytomegalovirus infection	0.0	< 5
P832	Hydrops fetalis not due to haemolytic disease	0.2	38
F80–F89	Disorders of psychological development	0.0	< 5
Total		**1.0**	**160**

Denominators do not modify for sex-specific conditions (such as undescended testes).
* ICD-10-AM. The International Statistical Classification of Disease and Related Health Problems, Australian Modification.
† Includes dysmorphic features.

Appendix 2: Excluded minor congenital anomalies

There has been variation in this list of exclusions[¶] between 1983 and 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
- Accessory nipples
- Anal fissure
- Balanced autosomal translocation (unless occurring with structural defects)
- Birth injuries
- Birth marks (< 4 cm, not including giant naevus)
- Bowing of legs (unless severe)
- Brushfield spots
- Cephalhaematoma
- Cleft gum
- Clicky hips
- Clinodactyly
- Craniotabes (unless severe)
- Dacrostenosis (blocked tear ducts)
- Dermatogplyphic abnormalities
- Developmental delays (unless occurring with a syndrome or structural defect)
- Ear abnormalities (minor)
- Epicanthic folds
- Gastro-oesophageal reflux
- Haemangioma (< 4 cm wide)
- Hernia – inguinal, umbilical
- High-arched palate
- Hydrocele
- Hypertelorism
- Imperforate hymen
- Laryngeal stridor
- Laryngomalacia
- Low slung/set ears
- Macroglossia
- Meckel's diverticulum
- Meconium ileus
- Mental retardations (unless occurring with a syndrome/structural defect)
- Metatarsus varus
- Micrognathia (unless severe)
- Mongolian spots
- Occiput, flat/prominent
- Patent ductus arteriosus (< 37 weeks)
- Philtrum, long/short
- Plagiocephaly
- Pre-auricular sinus
- Prominent forehead
- Protruding tongue
- Ptosis
- Retrognathia (unless severe)
- Rocker-bottom feet (prominent heels)
- Sacral pits, dimples, sinuses
- Short sternum
- Simian creases
- Single umbilical artery / two vessels in cord
- Skin folds/tags
- Slanting eyes
- Small mouth
- Spina bifida occulta (without evidence of spinal lesion)
- Sternomastoid tumour
- Subluxating knee joint
- Talipes (positional)
- Toe anomalies – minor
- Tongue tie
- Torticollis
- Ureteric reflux (ultrasound diagnosed)
- Webbing of 2nd and 3rd toes/fingers
- Wide suture lines

[¶] Classification of these anomalies have primarily been based on the 'External minor congenital anomalies' table provided by the World Health Organization (WHO), United States Centers for Disease Control and Prevention (CDC) and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).

Appendix 3: 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, birthweight, plurality, rank, condition of birth (termination before 20 weeks, termination ≥ 20 weeks, stillbirth, live birth), date of death (if applicable), congenital anomalies text, source of notification

Additional data items available from the Victorian Perinatal Data Collection:

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** score, time to establish respiration, resuscitation methods, neonatal morbidity

Additional information on data items can be found in the *Victorian Perinatal Data Collection manual, section 3 – Data definitions* available at bettersafercare.vic.gov.au/ccopmm.

** Appearance, Pulse, Grimace, Activity and Respiration score.

Appendix 4: Major congenital anomalies by year, 1983–2016

Year	Cases with an anomaly, terminations < 20 weeks' gestation	Cases with an anomaly, ≥ 20 weeks' gestation	Total births ≥ 20 weeks' gestation	Total congenital anomalies†	Number in 1,000 pregnancies with an anomaly*	Percentage
1983	< 5	1,653	60,628	1,655	27.3	2.7
1984	9	1,691	60,737	1,700	28.0	2.8
1985	18	1,591	61,189	1,609	26.3	2.6
1986	80	1,604	61,253	1,684	27.5	2.7
1987	55	1,620	61,566	1,675	27.2	2.7
1988	103	1,874	63,666	1,977	31.0	3.1
1989	123	1,930	64,255	2,053	31.9	3.2
1990	132	2,164	66,878	2,296	34.3	3.4
1991	140	2,235	65,248	2,375	36.3	3.6
1992	152	2,295	66,305	2,447	36.8	3.7
1993	203	2,250	64,737	2,453	37.8	3.8
1994	250	2,295	64,932	2,545	39.0	3.9
1995	257	2,444	63,717	2,701	42.2	4.2
1996	272	2,217	62,951	2,489	39.4	3.9
1997	297	2,306	62,308	2,603	41.6	4.2
1998	274	2,332	62,091	2,606	41.8	4.2
1999	294	2,555	62,690	2,849	45.2	4.5
2000	292	2,614	62,564	2,906	46.2	4.6
2001	308	2,394	62,149	2,702	43.3	4.3
2002	327	2,520	63,133	2,847	44.9	4.5
2003	356	2,690	63,552	3,046	47.7	4.8
2004	342	2,774	63,700	3,116	48.7	4.9
2005	339	2,500	66,654	2,839	42.4	4.2
2006	366	2,836	69,856	3,202	45.6	4.6
2007	364	3,013	72,474	3,377	46.4	4.6
2008	360	2,630	72,545	2,990	41.0	4.1
2009	414	2,664	73,264	3,078	41.8	4.2
2013	187	2,336	78,536	2,523	32.0	3.0
2014	217	2,276	79,179	2,493	31.4	3.1
2015	434	2,344	80,650	3,641	45.1	4.5
2016	123	2,302	82,348	3,685	44.7	4.5
Total	**7,090**	**67,605**	**2,065,755**	**80,162**	**385**	**3.8**

* Includes terminations < 20 weeks' gestation. † Includes cases of missing gestation data.
Figures may differ from the number of births presented in other CCOPMM reports due to the inclusion of terminations of pregnancy for psychosocial reasons or congenital anomalies at greater than 20 weeks' gestation.

Appendix 5: Perinatal outcomes of key congenital anomalies, 2015–2016

Chromosomal anomaly*	Total**	Terminations < 20 weeks	Terminations ≥ 20 weeks	Still births	Live birth	Neonatal deaths†
Down syndrome (trisomy 21)	492	72	23	7	107	0
Trisomy 13	78	8	7	< 5	10	5
Trisomy 18	126	25	12	5	6	< 5

Selected key anomaly*	Total**	Terminations (all)	Still births	Live birth	Neonatal deaths†
All neural defects (combined)	89	49	< 5	25	8
Anencephaly	33	22	< 5	7	6
Congenital absence, atresia and stenosis of large intestine	26	< 5	0	24	< 5
Cleft lip	31	< 5	0	28	< 5
Cleft lip and palate	71	12	< 5	51	< 5
Cleft palate	79	< 5	0	77	< 5
Coarctation of the aorta	42	< 5	0	41	< 5
Cystic kidney disease	83	10	< 5	66	< 5
Congenital deformities of hip	198	< 5	0	195	0
Congenital diaphragmatic hernia	40	13	0	26	5
Encephalocele	10	< 5	0	< 5	< 5
Exomphalos	35	< 5	< 5	22	< 5
Gastroschisis	48	6	< 5	39	< 5
Congenital hydrocephalus	49	24	5	16	< 5
Hypoplastic left heart syndrome	64	20	< 5	41	5
Hypospadias	418	0	0	413	0
Limb reduction defects	37	5	< 5	25	< 5
Microcephaly	15	< 5	< 5	12	0
Congenital obstructive defects of renal pelvis and malformations of ureter	426	13	< 5	377	< 5
Congenital malformations of oesophagus	22	< 5	< 5	17	0
Renal agenesis and other reduction defects of kidney	64	6	< 5	52	< 5
Congenital absence, atresia and stenosis of small intestine	29	< 5	0	25	< 5

Selected key anomaly*	Total**	Terminations (all)	Still births	Live birth	Neonatal deaths†
Spina bifida	46	23	< 5	15	< 5
Tetralogy of Fallot	51	9	0	42	< 5
Transposition of the great vessels	53	< 5	0	50	< 5
Ventricular septal defect	275	19	0	256	< 5

* Congenital anomaly can occur in isolation or with other anomaly.

** Includes missing birth outcome data.

† Cases where baby is born live but dies within the first 28 days after birth.

Glossary

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.
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	Refers to the number of live born babies, stillborn babies or terminations at any gestation affected by at least one congenital anomaly.
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 400 g who, after being born, breathes or shows any evidence of life such as a heartbeat.
Major congenital anomaly*	A structural change that has significant medical, social or cosmetic consequences for the affected individual. This type of anomaly typically requires medical intervention.
Minor congenital anomaly*	A structural change that poses no significant health problem and tends to have limited social or cosmetic consequences for the affected individual.
Multiparous	A woman who has carried two or more pregnancies beyond 20 weeks' gestation.
Neonatal death	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 400 g.
Obesity	Pre-conception body mass index (BMI) of 35 kg/m2 or more.
Parity	Number of previous pregnancies resulting in live births or stillbirths, excluding the current pregnancy.
Perinatal death / mortality	A stillbirth or neonatal death.
Pre-gestational diabetes	Type 1 or type 2 diabetes that exists before conception.
Pregnancy	Includes live birth, stillbirth and termination at any gestation.
Primiparous	A woman who has carried one pregnancy only beyond 20 weeks' 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 400 g who did not, at any time after delivery, breathe or show any evidence of life such as a heartbeat.

* WHO/CDC/ICBDSR. Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization; 2014.

ACRONYMS

ART	Assisted reproduction technology
CCOPMM	Consultative Council on Obstetric and Paediatric Mortality and Morbidity
CDC	Centers for Disease Control and Prevention
CFTS	Combined first trimester screening
CMA	Chromosome microarray
CVS	Chorionic villus sampling
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
IVF	In vitro fertilization
NIPT	Non-invasive prenatal test
RR	Relative risk
VCAR	Victorian Congenital Anomalies Register
VCGS	Victorian Clinical Genetics Service
VPDC	Victorian Perinatal Data Collection
WHO	World Health Organization

References

1. Australian Institute of Health and Welfare 2017. Australia's mothers and babies 2015—in brief. Perinatal statistics series no. 33. Cat no. PER 91. Canberra: AIHW.
2. World Health Organization. Congenital anomalies, 2015. Available at: http://www.who.int/mediacentre/factsheets/fs370/en/.
3. Vinceti M, Malagoli C, Rothman KJ, Rodolfi R, Astolfi G, Calzolari E, Puccini A, Bertolotti M, Lunt M, Paterlini L, Martini M. Risk of birth defects associated with maternal pregestational diabetes. European Journal of Epidemiology, 2014;29(6):411–8.
4. Stothard KJ, Tennant PW, Bell R, Rankin J. Maternal overweight and obesity and the risk of congenital anomalies: a systematic review and meta-analysis. JAMA. 2009;301(6):636–50.
5. Bower C, Rudy E, Callaghan A, Quick J, Nassar N. Age at diagnosis of birth defects. Birth Defects Research A, Clinical and Molecular Teratology, 2010;88(4):251–5.
6. World Health Organisation, Centers for Disease Control, International Clearinghouse for Birth Defects Surveillance and Research 2014, Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization.
7. Victorian Perinatal Data Collection. Victorian Perinatal Data Collection (VPDC) Manual version. 6.0, January 2018. Available at: https://www2.health.vic.gov.au/hospitals-and-health-services/quality-safety-service/consultative-councils/council-obstetric-paediatric-mortality/perinatal-data-collection.
8. Lumley J, Palma S, Fischer M, Robertson H. The tip of the iceberg: a validation study of the Victorian Congenital Malformations/Birth Defects Register. Australian Paediatric Journal, 1988;24:244–6.
9. Kilkenny M, Riley M, Lumley J. Follow-up validation study of the Victorian Congenital Malformations Register. Journal of Paediatric and Child Health, 1995;31:323–25.
10. Riley M, Phyland S, Halliday J. Validation study of the Victorian Birth Defects Register, Journal of Paediatric and Child Health, 2004:40:544–8.
11. Riley M, Howard J, Dale K, Palma S, Halliday J. Validating notifications of pregnancy terminations for birth defects before 20 weeks' gestation, Health Information Management, 2001;30: No.2.
12. Davey MA, Sloan ML, Palma S, Riley M, King J. Methodological processes in validating and analysing the quality of population-based data: a case study using the Victorian Perinatal Data Collection. Health Information Management Journal, 2013;42(3):12.
13. VSGS. Impact of recent advances in prenatal testing. The Partumpost, 2018:2(1), April. Available at: https://www.vcgs.org.au/sites/default/files/general/partum_post_v2i1%202018.pdf.
14. 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.
15. CDC ART and Multiple Births. https://www.cdc.gov/art/key-findings/multiple-births.html
16. Wen J, Jiang J, Ding C, Dai J, Liu Y, Xia Y, Liu J, Hu Z. Birth defects in children conceived by in vitro fertilization and intracytoplasmic sperm injection: a meta-analysis. Fertility and Sterility, 2012 Jun 30;97(6):1331–7.
17. Yan J, Huang G, Sun Y, Zhao X, Chen S, Zou S, Hao C, Quan S, Chen ZJ. Birth defects after assisted reproductive technologies in China: analysis of 15,405 offspring in seven centers (2004 to 2008). Fertility and Sterility, 2011 Jan 31;95(1):458–60.
18. Setti PE, Moioli M, Smeraldi A, Cesaratto E, Menduni F, Livio S, Morenghi E, Patrizio P. Obstetric outcome and incidence of congenital anomalies in 2351 IVF/ICSI babies. Journal of Assisted Reproduction and Genetics, 2016 Jun:1–7.
19. World Health Organisation: Congenital anomalies. http://www.who.int/en/news-room/fact-sheets/detail/congenital-anomalies.
20. Centre for Genetics Education: Fact Sheet 18. When parents are related – consanguinity. Available at: http://www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-18-when-parents-are-relatives-consanguinity.
21. Centers for Disease Control and Prevention: Commit to Healthy Choices to Help Prevent Birth Defects. https://www.cdc.gov/ncbddd/birthdefects/prevention.html.
22. The Royal Australian College of General Practitioners. Management of smoking in pregnant women. Available at: https://www.racgp.org.au/afp/2014/januaryfebruary/smoking-in-pregnant-women/.
23. Consultative Council on Obstetric and Paediatric Mortality and Morbidity 2017, Victoria's mothers, babies and children 2016. Melbourne: Department of Health and Human Services.
24. World health Organization, Congenital anomalies, Fact sheet, Updated September 2016. Available at: http://www.who.int/mediacentre/factsheets/fs370/en/.

Consultative Council on
Obstetric and Paediatric
Mortality and Morbidity

Safer Care
Victoria