Congenital anomalies in Australia

Web report | Last updated: **29 Jun 2023** | Topic: Mothers & babies

About

Congenital anomalies are a cause of child death and disability, and a major cause of perinatal death. The AIHW is re-establishing a national congenital anomalies data collection and this report updates last year's release with information on congenital anomalies in babies born in 2017. It is based on data from 7 jurisdictions and for babies diagnosed up to 12 months of age. Over 8,400 (3%) babies were born with a congenital anomaly in 2017 – around 32 babies per 1,000 births.

Final data on congenital anomalies for the 2016 and 2017 birth cohorts and preliminary data for the 2018 birth cohort are available as data tables and can be found under 'Data'.

Cat. no: PER 119

Findings from this report:

- Over 8,400 (3%) babies born in 2017 had a congenital anomaly
- Circulatory system anomalies were found in 33% of babies with an anomaly, and musculoskeletal system anomalies in 24%
- Most (90%) babies with an anomaly survived their first year
- Around 1 in every 32 babies were born with a congenital anomaly in 2017

Summary

Congenital anomalies encompass a wide range of atypical bodily structures or functions that are present at or before birth. They are a cause of child death and disability, and a major cause of perinatal death – where a baby is stillborn or dies within 28 days of birth. In 2017 and 2018, nearly one-third (31%) of perinatal deaths were caused by a congenital anomaly (AIHW 2021). From 2000–2020, there were around 12,500 deaths in Australia due to a congenital anomaly and half (50%) of these were children aged 0–4 (AIHW 2022).

What congenital anomalies are included?

This report presents information on congenital anomalies in babies born in 2017 that were reported to the National Congenital Anomalies Data Collection. It includes data from 7 jurisdictions – New South Wales, Victoria, Queensland, South Australia, Northern Territory, the Australian Capital Territory and Tasmania – and is based on the most recent data available across them. The report focuses on anomalies that:

- have significant medical, social or cosmetic outcomes for an individual
- were diagnosed in babies up to 12 months of age
- have data available across the 7 reporting jurisdictions.

The numbers and rates presented will underestimate the prevalence of congenital anomalies in Australia (see Technical notes for more information).

Key findings

- **Around 3% of babies born in 2017 had a congenital anomaly**
  Over 8,400 (3%) babies were born with a congenital anomaly in 2017 – around 1 in every 32 babies born.
- **Circulatory system anomalies were the most common type of anomaly**
  Circulatory system anomalies (these are anomalies of the heart and major blood vessels) were the most common type of congenital anomaly. These were reported in 33% of babies with any anomaly in 2017. This was followed by musculoskeletal system anomalies (24%) and urinary system anomalies (15%).
- **Most (90%) babies with an anomaly survived their first year**
  Most (90%) babies born with a congenital anomaly survived their first year. However, nearly 1 in 10 babies with an anomaly did not live past their first birthday. Around 7% of babies born with an anomaly were stillborn and 3% died in either the neonatal or post-neonatal period.
- **Rates of congenital anomalies (per 1,000 births) were higher in the following groups**

Babies born pre-term (before 37 weeks' gestation)

4x as high in babies born pre-term

Babies born with low birthweight (less than 2,500 grams)

5x as high for low birthweight babies

Women having a multiple birth

3x as high for multiple births

Congenital anomalies in Australia

Congenital anomalies are a cause of child death and disability, and a major cause of perinatal death – where a baby is stillborn or dies within 28 days of birth. In 2017 and 2018, nearly one-third (31%) of perinatal deaths in Australia were caused by a congenital anomaly (AIHW 2021). From 2000–2020, there were around 12,500 deaths in Australia due to a congenital anomaly and half (50%) of these were children aged 0–4 (AIHW 2022).

About this report

This report presents information on congenital anomalies in babies born in 2017 that were diagnosed up to 12 months of age. It uses data supplied by 7 jurisdictions to the National Congenital Anomalies Data Collection (NCADC). Data for 2017 were provided by all jurisdictions, except Western Australia.

What congenital anomalies are included?

Reporting focuses on anomalies that have significant medical, social or cosmetic outcomes for an individual (as agreed by the NCADC). Over 400 congenital anomalies are counted as inclusions (see Reporting inclusions). The following should be kept in mind when reading this report:

- There are differences in the way cases of congenital anomalies are identified across jurisdictions, and this may affect jurisdictional and national counts (see Technical notes for more information).
- The scope of this report means the numbers and rates presented will underestimate the prevalence of congenital anomalies (see Methods for more information).
- Data presented are not directly comparable to previous reports published in 2006 and 2008 due to differences in scope, including for example the anomalies reported on and the period of notification used for reporting.

What is a congenital anomaly?

Congenital anomalies encompass a wide range of atypical bodily structures or functions that are present at or before birth, although they may not be detected until later in life. Some examples include neural tube defects, heart defects, cleft lip/palate and chromosomal anomalies such as Down syndrome. Some anomalies may be treated surgically or with non-surgical options; others are life-threatening and cannot be treated or may have lifelong impacts (WHO 2020). Congenital anomalies:

- may have significant medical, social or cosmetic outcomes for an individual
- are major causes of fetal, infant and child deaths, chronic illness and disability
- typically require medical intervention (CDC 2020).

How are they diagnosed?

Many congenital anomalies are diagnosed at birth or during the first week of life; some are diagnosed during pregnancy, and some do not become obvious until later in life.

Many anomalies are diagnosed by a physical examination of the baby and observation of their initial health. Other anomalies are diagnosed by blood tests (including chemical analysis or chromosome testing) or imaging (X-ray and ultrasound).

Prior to birth, anomalies may be diagnosed by prenatal ultrasound or invasive testing (amniocentesis and chorionic villus sampling). These tests may be part of routine antenatal care or recommended because of findings on a screening ultrasound or because of maternal characteristics such as age and family history.

Prenatal diagnosis may allow for planning of pregnancy management, including:

- pregnancy surveillance around the growth and welfare of the baby
- the appropriate site of birth near a specific health-care facility
- providing information that assists with decisions around whether to continue a pregnancy.

How are they classified?

Congenital anomalies are classified by body system, such as nervous system, circulatory system, respiratory system and urinary system. While several classifications are available, the NCADC classifies anomalies according to the relevant edition of the *International statistical classification of diseases and related health problems, 10th revision, Australian modification* (ICD-10-AM) (IHPA 2019). For 2017, all data were mapped to the 10th edition of the ICD-10-AM.

References

AIHW (Australian Institute of Health and Welfare) (2021) *Stillbirths and neonatal deaths in Australia 2017 and 2018*, AIHW, accessed 2 May 2022.

AIHW (2022) General Record of Incidence of Mortality (GRIM) books [data set], AIHW, Australian Government, accessed 2 May 2023, doi:10.25816/287k-9w04.

CDC (Centers for disease Control and Prevention) (2020) *Birth Defects Surveillance Toolkit*, CDC website, accessed 23 June 2020.

IHPA (Independent Hospital Pricing Authority) (2019) *Classification of diseases and interventions*, IHPA website, accessed 23 June 2021.

WHO (World Health Organization) (2020) *Congenital anomalies*, WHO website, accessed 23 June 2021.

Congenital anomalies in Australia

In 2019, the AIHW formed the National Congenital Anomaly Advisory Group (NCAAG) to help it to re-establish a national congenital anomalies data collection, or NCADC. The AIHW had not collected congenital anomaly data from jurisdictions since 2008, due to a lack of consistency in the data affecting the quality and utility of the collection. Collecting national congenital anomaly data supports appropriate health-care planning and funding for treatment and diagnosis, as well as supporting research to develop better diagnostic, preventative and treatment measures. Appropriate health-care may prevent death occurring and improve short and long-term health.

The NCAAG agreed the AIHW would commence collection of congenital anomalies data based on the 2016 birth cohort, these being the most complete data available across jurisdictions, as part of a re-established national collection. More information about the NCADC can be found in Technical notes.

What data can be reported?

Congenital anomalies data for 2017 were the most recent data available nationally and could be supplied by all jurisdictions, except Western Australia. The reporting scope includes over 400 congenital anomalies that have significant medical, social or cosmetic outcomes for an individual, that were diagnosed in the 2017 birth cohort up to 12 months of age. Rates are based on all births (live births and stillbirths) reported to the National Perinatal Data Collection (NPDC) for the 2017 birth cohort (excluding births in Western Australia).

It is not possible to provide national data on all anomalies diagnosed in the 2017 birth cohort. Data from Western Australia were not available and data from the 7 reporting jurisdictions needed to be harmonised for reporting. The following cases were excluded to improve consistency in the data reported across jurisdictions:

- terminations of pregnancy before 20 weeks' gestation
- anomalies diagnosed after 12 months of age
- anomalies that did not have significant medical, social or cosmetic outcomes – these are sometimes referred to in other literature as minor congenital anomalies
- anomalies where data were not available across all reporting jurisdictions.

The numbers and rates presented will therefore underestimate the prevalence of congenital anomalies in Australia. It is estimated that around 12–17% of anomalies are diagnosed after 12 months and before 6 years of age (Bower et al. 2010; Gibson et al. 2016). This may be lower or higher depending on the type of anomaly.

A list of the anomalies included in this report (in scope anomalies) and their respective ICD-10-AM codes are provided in:

- Reporting inclusions
- NCADC reporting inclusions and their ICD-10-AM codes (PDF 320kB).

Anomalies are grouped by body system.

List of anomalies excluded from reporting (PDF 365kB) is also available.

Every effort has been made to collect and report data consistently by using common data specifications and reporting on a similar notification period across jurisdictions. There are differences, however, in the scope and methods used to collect congenital anomalies data across jurisdictions. More information about these and their impacts can be found in Technical notes.

References

Bower C, Rudy E, Callaghan A, Quick J and Nassar N (2010) Age at diagnosis of birth defects, *Birth Defects Research. Part A, Clinical and Molecular Teratology*, 88(4): 251–255, doi:10.1002/bdra.20658

Gibson C, Scott H, Haan E and Scheil W (2016) Age range for inclusion affects ascertainment by birth defects registers, *Birth defects research. Part A, Clinical and molecular teratology*, 106(9): 761–766, doi:10.1002/bdra.23534

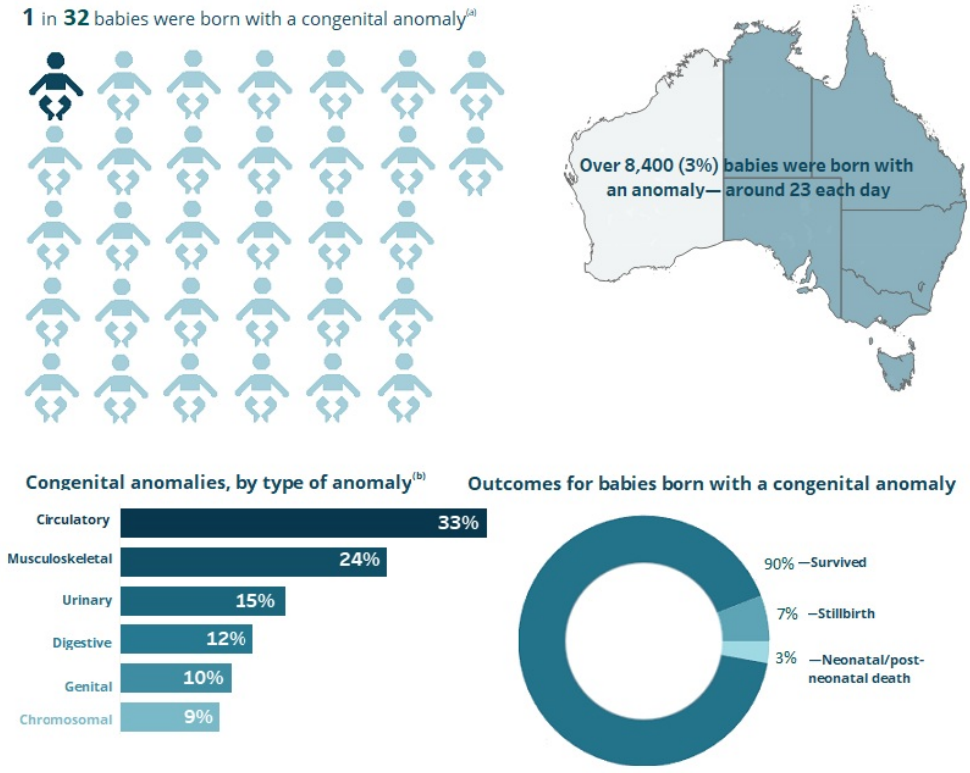
How many babies have a congenital anomaly?

In 2017, over 8,400 (3%) babies were born with a congenital anomaly, or around 23 babies born each day. Over two-thirds of these babies (68%) were diagnosed with one anomaly and just under one-third (32%) had multiple anomalies. Over half of those with more than one anomaly (56%) had these in the same body system; 44% of babies with more than one anomaly had these in different body systems, the most common combination being a circulatory system and chromosomal anomaly.

Circulatory system anomalies (these are anomalies of the heart and major blood vessels) were most commonly reported, with 33% of babies with any anomaly having a circulatory system anomaly. This was followed by musculoskeletal system anomalies (24%) and urinary system anomalies (15%).

Most (90%) babies with an anomaly survived their first year (Figure 1). However, nearly 1 in 10 babies with an anomaly did not live past their first birthday. Around 7% of babies with an anomaly were stillborn and 3% died in the neonatal or post-neonatal period.

Figure 1. Types and outcomes of babies born with congenital anomalies



(a) Includes babies with in scope anomalies in the 2017 birth cohort diagnosed up to 12 months of age in New South Wales, Victoria, Queensland, South Australia, Northern Territory, and the Australian Capital Territory, and those diagnosed during the birthing episode in Tasmania. This will underestimate the total number of babies with an anomaly where these are diagnosed after 12 months of age. In scope anomalies are those that have significant medical, social or cosmetic outcomes. See 'Technical notes' for a list of anomaly inclusions. Data for Western Australia were not available for 2017.
(b) Proportions are based on the number of babies with any in scope anomaly. Congenital anomalies can occur in isolation or with other anomalies, so a baby may be counted in more than one 'Type of anomaly' category. A baby will only be counted once within each category.

Source: AIHW analysis of the NCADC.

What congenital anomalies do babies have?

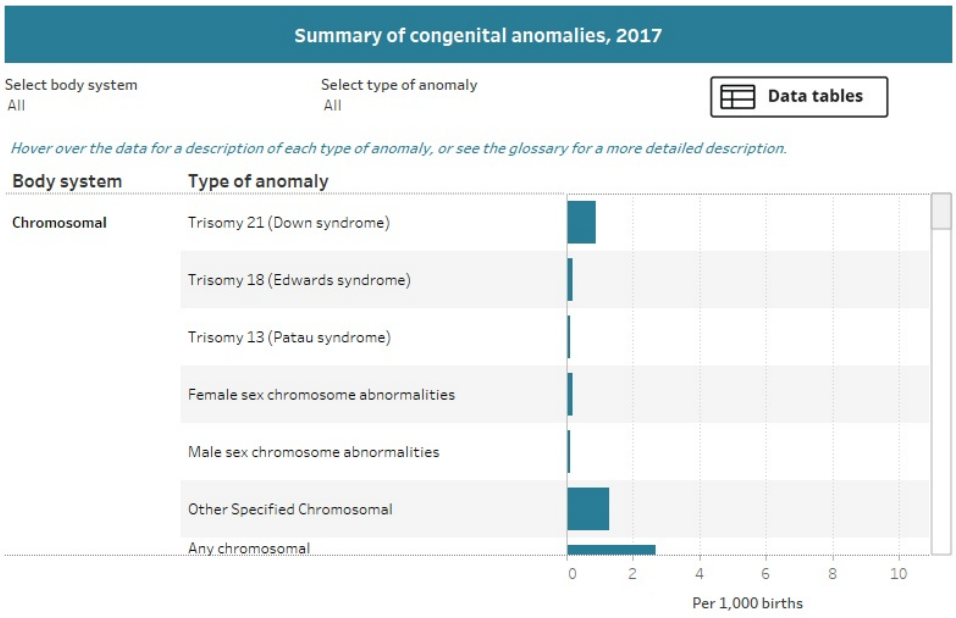
Figure 2 shows the number and rate of babies with different types of anomalies. The anomalies are grouped and can be filtered by body system or type of anomaly. Please note:

- Numbers and rates are based on data from 7 jurisdictions. Data for Western Australia were not available for 2017.
- Out of scope anomalies were excluded from reporting. See Technical notes for information on reporting inclusions and exclusions.
- Numbers and rates are based on anomalies diagnosed in babies up to 12 months of age. This may underestimate the number of cases in some instances, where anomalies are diagnosed after 12 months of age.
- Anomalies can occur in isolation or with other anomalies, so a baby may be counted in more than one body system or type of anomaly category.

Figure 2. Number and rate of congenital anomalies, 2017

Number and rate of selected anomalies, 2017

This data visualisation shows the number and rate of babies with different types of congenital anomalies. Data can be viewed as a table or a figure and filtered by the type of anomaly. It shows that in 2017, over 8,445 babies had a congenital anomaly, or 32 babies per 1,000 births.



Notes
1. Includes babies with in scope anomalies in the 2017 birth cohort for all jurisdictions except Western Australia . Only anomalies that have significant medical, social or cosmetic outcomes are included. See the 'Technical notes' for a list of anomaly inclusions.
2. Counts are based on the number of babies with in scope anomalies diagnosed up to 12 months of age in New South Wales, Victoria, Queensland, South Australia, the Australian Capital Territory, and the Northern Territory, and those diagnosed during the birthing episode in Tasmania. This may underestimate the total number of babies with an anomaly where these are diagnosed after 12 months of age.
3. Congenital anomalies can occur in isolation or with other anomalies, so a baby may be counted in more than one 'Type of anomaly' category. A baby will only be counted once within each category.
4. The rate is calculated using the number of births (live and stillbirths) with a congenital anomaly and the number of total births (live and stillbirths). Births are those of at least 20 or more completed weeks of gestation or with a birthweight of at least 400 grams as collected in the National Perinatal Data Collection. The percentage is the number of babies that have the anomaly as a proportion of babies with any in scope anomaly.
5. There are differences in the way babies with an anomaly are identified across jurisdictions, which may affect counts within jurisdictions and at the national level. Numbers and rates should therefore be interpreted with caution. See the 'Technical notes' for more information.

Source: AIHW analysis of the NCADC and the NPDC.
http://www.aihw.gov.au

Baby characteristics

This section looks at congenital anomalies by various baby characteristics. In 2017, 58% of babies with any anomaly were male and 42% were female. Rates of anomalies per 1,000 births were similar for males and females across many body systems. However, genital organ anomalies were more common in males (5.4 compared with 0.3 per 1,000 births for females), largely due to cases of hypospadias in males. Urinary system anomalies were also more common in males (6.1 compared with 3.1 per 1,000 births for females) due to higher numbers across a range of urinary system anomalies.

Nearly three-quarters of babies with any anomaly were born at term (70%) and had a normal birthweight (72%). However, this varied by type of anomaly; for example, the proportion of babies with a normal birthweight was lower in babies born with a nervous system anomaly (51%) or chromosomal anomaly (51%) and higher for those born with a musculoskeletal or urinary system anomaly (81%).

Congenital anomaly rates were higher in:

- babies born *pre-term* (before 37 weeks' gestation) compared with those born at or after 37 weeks (107 per 1,000 births compared with 24 per 1,000 births)
- babies born with *low birthweight* (less than 2,500 grams) compared with those having a normal birthweight (123 per 1,000 births compared with 24 per 1,000 births)
- babies that were *small for gestational age* (that is with a birthweight below the 10th percentile for their gestational age and sex) compared with those having an appropriate birthweight for gestational age (44 per 1,000 births compared with 26 per 1,000 births).

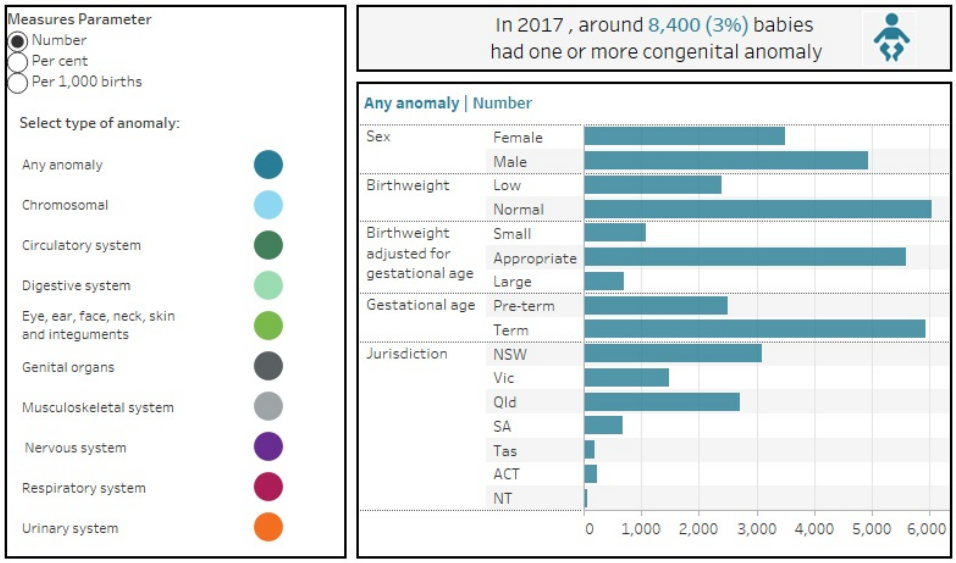
Figure 3 shows the number, proportion and rate of babies with congenital anomalies by various baby characteristics. These include sex, birthweight, birthweight adjusted for gestational age, gestational age and state/territory of birth. The anomalies are grouped and can be filtered by body system (type of anomaly).

Figure 3. Number and rate of congenital anomalies, by baby characteristics, 2017

Number and rate of selected anomalies, by baby characteristics, 2017

This data visualisation shows the number, proportion, and rate of babies with congenital anomalies by various baby characteristics. These include sex, birthweight, birthweight adjusted for gestational age, gestational age and state of birth. Data can be filtered by body system. In 2017, congenital anomaly rates were higher in babies born pre-term, in those born with low birthweight and in those that were small for gestational age.

Summary of congenital anomalies, by baby characteristics, 2017



Notes

1. Is the number of babies with any in scope anomaly.
2. Includes babies with in scope anomalies in the 2017 birth cohort, for all jurisdictions except Western Australia. Only anomalies that have significant medical, social, or cosmetic outcomes for an individual are included. See the 'Technical notes' for a list of anomaly inclusions.
3. Counts are based on cases diagnosed up to 12 months of age in New South Wales, Victoria, Queensland, South Australia, the Australian Capital Territory, and the Northern Territory, and those diagnosed during the birthing episode in Tasmania. This may underestimate the number of cases in some instances, where anomalies may be diagnosed after 12 months of age.
4. There are differences in the way babies with an anomaly are identified across jurisdictions, which may affect counts within jurisdictions and at the national level. Numbers and rates should therefore be interpreted with caution. See 'Data quality, availability and interpretation' for more information.

5. Congenital anomalies can occur in isolation or with other anomalies, so a woman giving birth to a baby with an anomaly may be counted in more than one 'Type of anomaly' category. A woman giving birth to a baby with an anomaly will only be counted once within each category.
6. Major anomalies identified antenatally in the Northern Territory may require the baby to be birthed in a specialist tertiary facility interstate. In the Northern Territory only the primary anomaly associated with a particular syndrome (for example Trisomy 21 for Down syndrome) is reported. The numbers and rates presented will therefore underestimate the prevalence of congenital anomalies in the Northern Territory.

Source: AIHW analysis of the NCADC and the NPDC.
http://www.aihw.gov.au

Additional notes >

Maternal characteristics

This section looks at congenital anomalies by various maternal characteristics. In 2017, around 8,300 (3%) women giving birth had a baby with a congenital anomaly. Most (95%) of these women had a singleton birth; nearly three-quarters (73%) were aged 20–34 years and nearly three-quarters (71%) lived in *Major cities*. Around 6% of women giving birth to a baby with a congenital anomaly were First Nations women. First Nations women refers to women who have identified as being of Aboriginal and/or Torres Strait Islander origin.

Congenital anomaly rates were higher in babies born to:

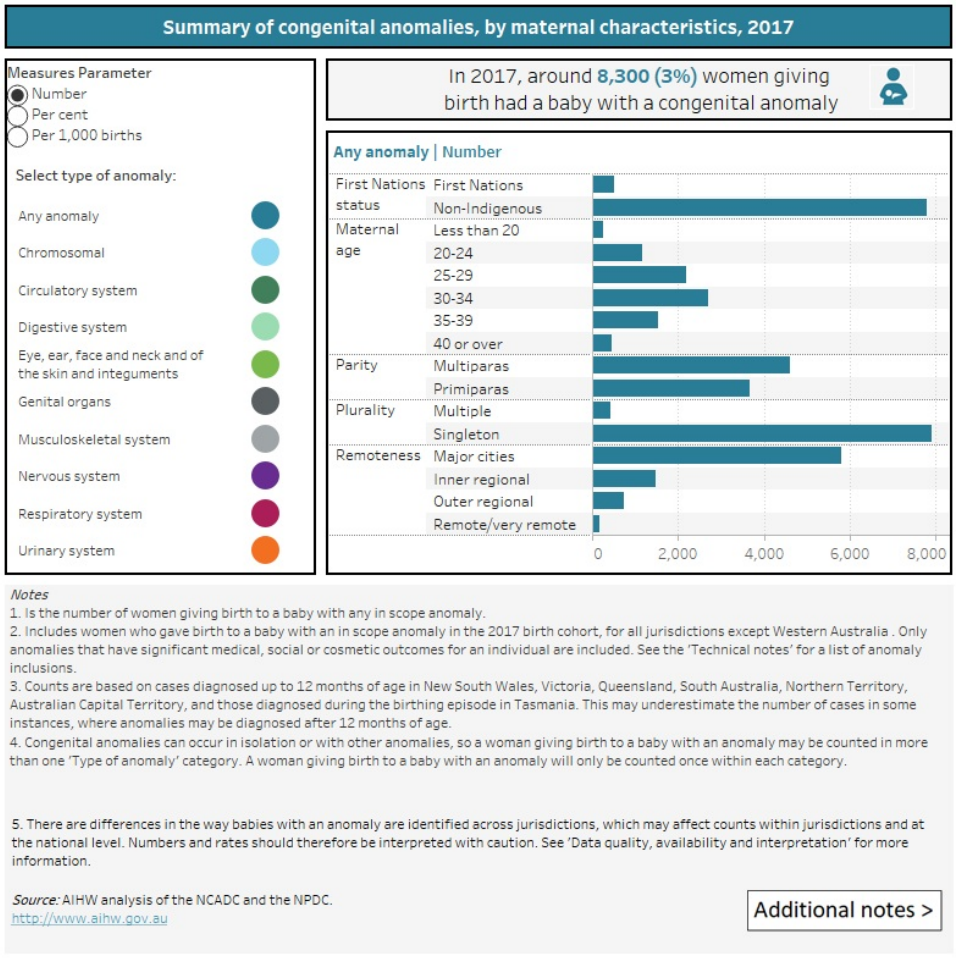
- women having a multiple birth (104 per 1,000 women giving birth), compared with those having a singleton birth (30 per 1,000 women giving birth)
- women aged less than 20, and those aged 40 years and over (43, and 39 per 1,000 women giving birth, respectively, compared with 31 per 1,000 women giving birth aged 20–39 years)
- First Nations women (43 per 1,000 women giving birth), compared with non-Indigenous women (31 per 1,000 women giving birth).

Figure 4 shows the number, proportion and rate of women giving birth to a baby with a congenital anomaly, by various maternal characteristics. These include maternal age at birth, the First Nations status of women giving birth, parity (this indicates whether a woman has had a previous pregnancy), plurality (this indicates the number of births resulting from the pregnancy) and remoteness area of usual residence (where women live). The anomalies are grouped and can be filtered by body system (type of anomaly).

Figure 4. Number and rate of congenital anomalies, by maternal characteristics, 2017

Number and rate of selected anomalies, by maternal characteristics, 2017

This data visualisation shows the number, proportion and rate of women giving birth to a baby with a congenital anomaly by various maternal characteristics. These include maternal age at birth, maternal First Nations status, parity, plurality and remoteness area. Data can be filtered by body system. In 2017 around 8,300 (3%) women giving birth had a baby with an anomaly.



Baby outcomes

Congenital anomalies may be associated with stillbirths, neonatal and post-neonatal deaths. In 2017, most babies (90%) with a congenital anomaly were live births and survived their first year. However, nearly 1 in 10 babies with an anomaly did not live past their first birthday. Around 7% of babies born with an anomaly were stillborn and 3% died in either the neonatal period (within 28 days after birth) or the post-neonatal period (after 28 days and within one year after birth).

The proportion of babies who survived their first year varied by the type of anomaly. A smaller proportion of babies with chromosomal or nervous system anomalies survived their first year (66% and 69% of babies with these anomalies, respectively), while nearly all babies with a genital organ anomaly survived their first year (97%).

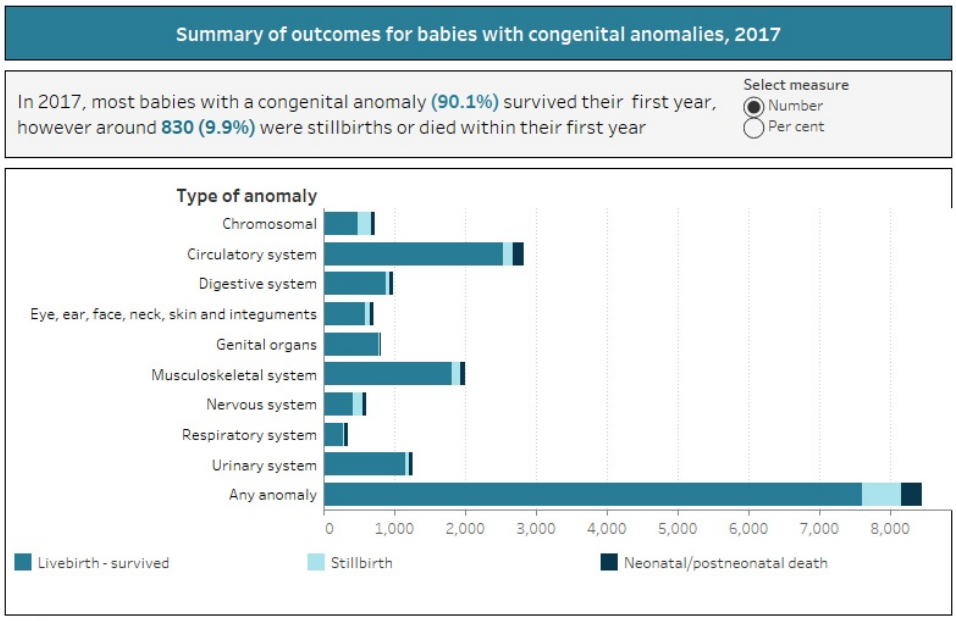
The perinatal mortality rate (stillbirths and neonatal deaths) in babies with one or more congenital anomalies was 92 per 1,000 births compared with the overall perinatal mortality rate for all babies of 9.7 per 1,000 births in 2017 (AIHW 2021).

Figure 5 shows outcomes for babies born with a congenital anomaly by type of anomaly.

Figure 5. Outcome for babies with congenital anomalies, 2017

Outcomes for babies with selected congenital anomalies, 2017

This data visualisation shows the outcomes of babies born with a congenital anomaly by body system (type of anomaly). In 2017, most babies with a congenital anomaly (90%) were liveborn and survived their first year, however, 7% of babies with an anomaly were stillborn and 3% died after birth in either the neonatal or postnatal period.



Notes

1. Includes babies with in scope anomalies in the 2017 birth cohort, diagnosed up to 12 months of age in New South Wales, Victoria, Queensland, South Australia, Northern Territory and Australian Capital Territory and those diagnosed during the birthing episode in Tasmania. Data from Western Australia were not available for 2017. Only anomalies that have significant medical, social or cosmetic outcomes are included. See the Notes tab for a list of anomaly inclusions.
2. Counts are based on the number of babies with in scope anomalies diagnosed up to 12 months of age in New South Wales, Victoria, Queensland, South Australia, the Australian Capital Territory, and the Northern Territory, and those diagnosed during the birthing episode in Tasmania. This may underestimate the total number of babies with an anomaly where these are diagnosed after 12 months of age.
3. Congenital anomalies can occur in isolation or with other anomalies, so a baby may be counted in more than one 'Type of anomaly' category. A baby will only be counted once within each category, even if they have more than one type of anomaly within the same category. As a baby may be counted in more than one 'Type of anomaly' category, the sum of individual categories will be greater than the total number of babies with any anomaly.
4. There are differences in the way babies with an anomaly are identified across jurisdictions, which may affect counts within jurisdictions and at the national level. Numbers should therefore be interpreted with caution. See the 'Technical notes' for more information.
5. Percentages are based on all babies with that type of anomaly.
6. A neonatal death is the death of a liveborn baby within 28 days of birth, a postneonatal death is the death of a liveborn baby occuring between 28 days and 1 year of birth. Stillbirth describes a fetal death prior to birth of a baby of 20 or more completed weeks of gestation or of 400 grams or more birthweight.
7. Data presented for 2017 includes different jurisdictions to previous years of data, and have been mapped to a newer edition of the International Classification of Diseases 10th Edition - Australian Modification. Care should be taken when comparing results across years.
8. Percentages based on small counts (<5) may be unreliable and therefore interpretations and comparisons should be made with caution.

Source: AIHW analysis of the NCADC and the NPDC.
http://www.aihw.gov.au

Reference

AIHW (Australian Institute of Health and Welfare) (2021) *Stillbirths and neonatal deaths in Australia 2017 and 2018*, AIHW, accessed 9 May 2023.

Technical notes

Technical notes

The National Congenital Anomalies Data Collection

The NCADC is a national data collection on babies with a congenital anomaly. It is based on cases of congenital anomalies reported to state and territory data collections, including congenital anomaly registers, perinatal data collections and admitted patient data collections. Data from Western Australia are not yet available as part of this collection. More information about jurisdictional congenital anomaly data sources can be found in State and territory data collections.

Some baby and maternal data elements in the NCADC are sourced from the NPDC. Jurisdictions provide a common identifier so cases with congenital anomalies in the NCADC can be linked to their respective NPDC record.

Collection and reporting scope

The scope of the NCADC includes births (both live births and stillbirths) with a congenital anomaly and terminations of pregnancy due to a congenital anomaly (if this is available). This includes conditions in the tenth edition of the ICD10-AM, including Chapter 17 (Q00-Q99), P35 (congenital viral diseases) and P371 (congenital toxoplasmosis). In practice, what can be supplied varies by jurisdiction and this impacts national reporting. The AIHW has harmonised the data for reporting, so the scope for reporting from the NCADC is narrower than the scope for the collection as a whole.

- Congenital anomaly data supplied using the Royal College of Paediatrics and Child Health's Classification of Diseases (ICD-9-BPA) classification (this included some records from New South Wales) were mapped to the ICD-10-AM (tenth edition) to report data on a single classification.
- The notification period for this report includes anomalies diagnosed up to 12 months of age as all jurisdictions, except Tasmania, could supply this data; Tasmania could supply data for the birthing episode only. Some, but not all, jurisdictions collect data based on notification periods greater than 12 months. For example, South Australia collects anomalies data in children up to 5 years of age. Analysis of the data supplied by South Australia in 2017 indicates around 15% of anomalies in their 2017 birth cohort were diagnosed after 12 months of age and were excluded from this national report.
- Some anomalies were excluded from national reporting because they were not collected across all jurisdictions or because they did not pose significant health concerns for a baby in the first year of life or have major social or cosmetic outcomes. Excluded anomalies were based primarily on those listed by the World Health Organization (WHO), the United States Centers for Disease Control and Prevention (CDC) and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). A list of reporting exclusions (PDF 123kB) is provided for information.
- Terminations of pregnancy before 20 weeks' gestation were excluded as these data were not available across all jurisdictions.
- Only records able to be matched with the NPDC were included for reporting.

Reporting inclusions

Over 400 congenital anomaly condition codes at the 4-character level of the ICD-10-AM are in scope for this report. Around 62% of these are counted in 'other' categories. Table 1 lists these reporting inclusions and their respective ICD-10-AM codes. A list that includes the names of the anomalies in 'other' categories can be found in Data tables.

Table 1: Reporting inclusions and their ICD-10-AM codes, by type of anomaly

Type of anomaly	ICD-10-AM (tenth edition)
Chromosomal	
Trisomy 21 (Down syndrome)	Q90, Q90.0–Q90.2, Q90.9
Trisomy 18 (Edwards syndrome)	Q91.0–Q91.3
Trisomy 13 (Patau syndrome)	Q91.4–Q91.7
Female sex chromosome (including Turner syndrome)	Q96.0–Q96.4, Q96.8–96.9, Q97.0–Q97.3, Q97.8
Male sex chromosome (including Klinefelter syndrome)	Q98, Q98.0–Q98.8
Other chromosomal	Q92, Q92.0–Q92.8, Q93.0–Q93.8, Q95.1–Q95.5, Q95.8, Q99.0-Q99.2, Q99.8
Circulatory system	
Transposition of the great vessels	Q20.1, Q20.3, Q20.5

Cardiac septal anomalies	Q21.0–Q21.2, Q21.4, Q21.8
• Ventricular septal defect	Q21.0
• Atrial septal defect	Q21.1
• Atrioventricular septal defect	Q21.2
• Other cardiac septal defect	Q21.4, Q21.8
Tetralogy of Fallot	Q21.3
Pulmonary valve atresia	Q22.0
Hypoplastic left heart syndrome	Q23.4
Patent ductus arteriosus	Q25.0
Coarctation of aorta	Q25.1
Other circulatory system	Q20.0, Q20.2, Q20.4, Q20.6, Q20.8, Q22, Q22.1–Q22.6, Q22.8, Q23, Q23.0–Q23.3, Q23.8, Q24, Q24.0–Q24.6, Q24.8, Q25.2–Q25.8, Q26.0–Q26.6, Q26.8, Q27.1–Q27.4, Q27.8, Q28.0–Q28.3, Q28.8
Digestive system	
Cleft lip and/or palate	Q35,Q35.1-Q35.3, Q35.9, Q36
Tracheo-oesophageal fistula	Q39.1–Q39.2
Anomalies leading to oesophageal obstruction (without fistula)	Q39.0, Q39.3–Q39.4
Congenital hypertrophic pyloric stenosis	Q40.0
Atresia/stenosis of intestines	Q41.0–Q41.2, Q41.8–Q41.9, Q42.0–Q42.3, Q42.8
Hirschsprung disease	Q43.1
Other digestive system	Q38.7–Q38.8, Q39.5–Q39.6, Q39.8, Q40.1–Q40.3, Q40.8, Q43.2–Q43.4, Q43.6–43.8, Q44.0, Q44.2–Q44.7, Q45.0–Q45.3, Q45.8
Eye, ear, face and neck and integuments	
Anophthalmia and microphthalmia	Q11.1–Q11.2
Microtia (with congenital absence, atresia, and stricture of external auditory canal)	Q17.2 with Q16.1
Other eye, ear, face and neck and integuments	Q10.6-Q10.7, Q11.0, Q12.2, Q13.1, Q13.3-Q13.4, Q13.8, Q15.0, Q16.0, Q16.1 (without Q17.2), Q80, Q80.0-Q80.4, Q80.8, Q81, Q81.0-Q81.2, Q81.8, Q82.0-Q82.4, Q83.0-Q83.1, Q84.0, Q85.0-Q85.1, Q85.8, Q86.0-Q86.2, Q86.8, Q87, Q87.0-Q87.5, Q87.8, Q89.0-Q89.4, Q89.7–Q89.8
Genital organs	
Doubling anomalies of the female genitalia	Q51.1–51.4, Q52.1
Hypospadias	Q54, Q54.0-Q54.3, Q54.8–Q54.9
Anomalies related to indeterminate sex	Q56, Q56.0–Q56.4
Other genital organs	Q50.0–Q50.6, Q51.0, Q51.5–Q51.9, Q52.0, Q52.2, Q52.7, Q53.0, Q55.0, Q55.3–Q55.5, Q55.8

Musculoskeletal system	
Congenital hip dislocation	Q65, Q65.0–Q65.2
Talipes	Q66, Q66.00, Q66.01, Q66.0-Q66.1, Q66.4
Polydactyly	Q69, Q69.0-Q69.2, Q69.9
Syndactyly	Q70, Q70.0–Q70.2, Q70.4, Q70.9
Reduction defect of upper limb(s)	Q71.0–Q71.6, Q71.8–Q71.9
Reduction defect of lower limb(s)	Q72, Q72.0–Q72.9
Congenital diaphragmatic hernia	Q79.0
Exomphalos	Q79.2
Gastroschisis	Q79.3
Other musculoskeletal system	Q65.3–Q65.5, Q65.9, Q67.5, Q73, Q73.0–Q73.1, Q73.8,Q74, Q74.0, Q74.2–Q74.5, Q74.8, Q75, Q75.1, Q76.1–Q76.3, Q76.5, Q76.7, Q77.0- Q77.8, Q78, Q78.0–Q78.6, Q78.8-Q78.9, Q79.1, Q79.4, Q79.6
Nervous system	
Neural tube defects	Q00,Q01, Q05, Q05.0–Q05.9, Q07.0
Anencephaly and related anomalies	Q00, Q00.1-Q00.2
• Craniorachischises	Q001
• Iniencephaly	Q002
Encephalocele	Q01, Q01.0-Q01.2, Q01.8-Q01.9
Spina bifida (including Arnold-Chiari malformation)	Q05, Q05.0–Q05.9, Q07.0
Microcephaly	Q02
Congenital hydrocephalus	Q03, Q03.0-Q-3.1, Q03.8-Q03.9
Other nervous system	Q04, Q04.0–Q04.6, Q04.8, Q06.0–Q06.4, Q06.8, Q07.8
Respiratory system	
Choanal atresia	Q30.0
Hypoplasia and dysplasia of lung	Q33.6
Other respiratory system	Q30.1, Q30.3, Q31.0–Q31.3, Q31.8, Q32.0–Q32.4, Q33, Q33.0, Q33.1–Q33.5, Q33.8, Q34.0–Q34.1, Q34.8
Urinary system	
Renal agenesis/hypoplasia	Q60, Q60.0–Q60.6
Cystic kidney disease	Q61, Q61.0–Q61.5, Q61.8–Q61.9
Exstrophy of urinary bladder	Q64.1
Other urinary system	Q62, Q62.0–Q62.8, Q63, Q63.0–Q63.3, Q63.8, Q64.0, Q64.2–Q64.3, Q64.5-Q64.6, Q64.8

Technical notes

National Perinatal Data Collection

The NPDC is a national collection of data on pregnancy and childbirth. The data are based on births reported to the perinatal data collection in each state and territory in Australia. Midwives and other birth attendants, using information obtained from women and from hospital or other records, complete notification forms for each birth. A standard de-identified extract is provided to the AIHW on an annual basis to form the NPDC.

The NPDC has birthweight and gestational age conditions for records included as live births and stillbirths. This means a very small number of live births occurring before 20 weeks' gestation and weighing less than 400 grams are not included in the NPDC. Data for babies whose gestational age and birthweight were not recorded are also not included. Live births and stillbirths may include terminations of pregnancy after 20 weeks' gestation. There are variations in legislation regarding termination of pregnancy between states and territories, and the recording of terminations is likely to be incomplete.

More information about the NPDC can be found in the data quality statement.

State and territory data collections

Data for the NCADC are sourced from state and territory congenital anomaly registers and perinatal and admitted patient data collections. There are differences in the collection method and scope of these collections, for example the anomalies that are in scope for collection, the availability of and inclusion of terminations of pregnancy data, and the age at which cases can be notified for inclusion.

Collection and reporting may also be affected by the availability and use of prenatal screening programs and diagnostic testing services, and whether the results of these are notified to congenital anomaly collections. Key information about these collections is summarised in Table 2, with more detail available in the information sheet (PDF 120kB).

Table 2: State and territory congenital anomaly data sources, 2017

Jurisdiction[a]	Collection name	Scope	Notification period	More information	Reports
New South Wales[b]	1. NSW Register of Congenital Conditions 2. NSW Admitted Patient Data Collection[b]	Scheduled congenital conditions[c] detected in a fetus during pregnancy or in a child up to 1 year of age. Includes conditions detected in: • stillborn babies or liveborn babies up to 1 year of age • the fetus during pregnancy regardless of whether the pregnancy continues or is terminated.	Before birth to 1 year of age	NSW Register of Congenital Conditions - Reporting Requirements	HealthStats NSW Publications

Victoria	Victorian Congenital Anomalies Register (VCAR)	Notifications of congenital anomalies in children from before birth to 6 years of age. Includes structural, functional, genetic, chromosomal and biochemical abnormalities that can be detected before birth, at birth or days later, in either a liveborn or stillborn baby. All anomalies (major and minor) can be notified to the VCAR, however, reporting is based on major anomalies likely to contribute to perinatal and childhood mortality.	Before birth to 6 years of age	Congenital anomalies | Better Safer Care	Congenital anomalies in Victoria 2015–16 | Better Safer Care Congenital anomalies in Victoria 2013-14 - health.vic
Queensland	Congenital Anomaly Linked File (CALF) Data from the Queensland Perinatal Data Collection is linked with Hospital Admitted Patient Data Collection, ABS Cause of Death and Death Registration Data.	Includes: • terminations of pregnancy at any gestation performed because of a diagnosis of a birth defect • stillbirths and newborn babies with birth defects • children admitted and diagnosed with a birth defect after the neonatal period and prior to their 5th birthday • children who died prior to their 5th birthday, with a birth defect listed on their death record.	Birth to 5 years of age	Statistical Services Branch | Queensland Health	Statistical Services Branch Data Dashboards | Queensland Health
Western Australia[(d)]	Western Australian Register of Developmental Anomalies (WARDA)	A developmental anomaly is defined as: cerebral palsy or a structural or functional anomaly which is present at conception or occurs before the end of pregnancy and is diagnosed during pregnancy, or after stillbirth or termination of pregnancy, or after live birth, but before 6 years of age.	Before birth to 6 years of age	Western Australian Register of Developmental Anomalies (healthywa.wa.gov.au) WA Register of Developmental Anomalies (WARDA) (health.wa.gov.au)	2014 Annual Report of the WA Register of Developmental Anomalies (health.wa.gov.au)

South Australia	South Australian Birth Defects Register (SABDR)	A birth defect is any abnormality, structural or functional, identified up to 5 years of age, provided the condition had its origin before birth. Includes: • terminations of pregnancy at any gestation performed because of a diagnosis of a birth defect • late fetal deaths (stillbirths ≥ 20 weeks' gestation) • newborn babies • children diagnosed after the neonatal period and prior to their 5th birthday.	Before birth to 5 years of age	Women's and Children's Hospital, Birth Defect Register (wch.sa.gov.au)	Birth Defects in South Australia 2016 (wch.sa.gov.au)
Tasmania	1. Tasmanian Perinatal Data Collection 2. Tasmanian Admitted Patient Data Collection	Any structural or anatomical abnormalities of the baby that are present at birth, in either a liveborn or stillborn baby, and diagnosed before separation from care. Only anomalies diagnosed before discharge from the birthing hospital are included in the collection.	Birth to discharge from birthing episode	Tasmania does not routinely collate or report this data but has extracted for the NCADC. Council of Obstetric and Paediatric Mortality and Morbidity | Tasmanian Department of Health	n.a.
Australian Capital Territory	1. ACT Perinatal Data Collection 2. ACT Admitted Patient Data Collection	Includes congenital anomalies identified on the perinatal form and from hospital separations using relevant ICD-10-AM codes.	Birth to 1 year of age	The ACT does not routinely collate or report this data but has extracted for the NCADC. Data collections | Health (act.gov.au)	n.a.
Northern Territory	NT Perinatal registry	The diagnosis of a structural of functional abnormality in a child up to 12 months of age that was present from conception or that occurs before the end of pregnancy. Includes • Diagnosed in pregnancy in the Northern Territory • Stillbirths and newborns Children diagnosed up to 12 months of age	Birth to 1 year of age	Perinatal registry | NT Health	

a. NSW use their register and admitted patient data collection to report on notifications of congenital anomalies to the NCADC.
b. Scheduled congenital conditions include: all structural malformations; chromosomal abnormalities; and 4 medical conditions (cystic fibrosis, phenylketonuria, congenital hypothyroidism and thalassaemia major). Conditions that are not notifiable include: minor anomalies occurring in isolation; birth injuries; congenital infections which do not result in a structural malformation; tumours and cysts; and conditions arising from prematurity or asphyxiation.
c. Western Australia collects congenital anomalies data through the Western Australian Register of Developmental Anomalies, however, data for 2017 were not supplied to the NCADC in time for reporting.

Technical notes

Data are presented on the number of babies with in scope congenital anomalies in the 2017 birth cohort, that were diagnosed up to 12-months of age and the women who gave birth to these babies. The number of babies is higher than the number of women who gave birth due to the occurrence of multiple births.

In scope congenital anomalies are those that have significant medical, social or cosmetic outcomes for an individual. Over 400 congenital anomaly condition codes at the 4-character level of the ICD-10-AM are in scope for this report. These were agreed to by NCAAG, as were out-of-scope anomalies (see Reporting inclusions and Reporting exclusions).

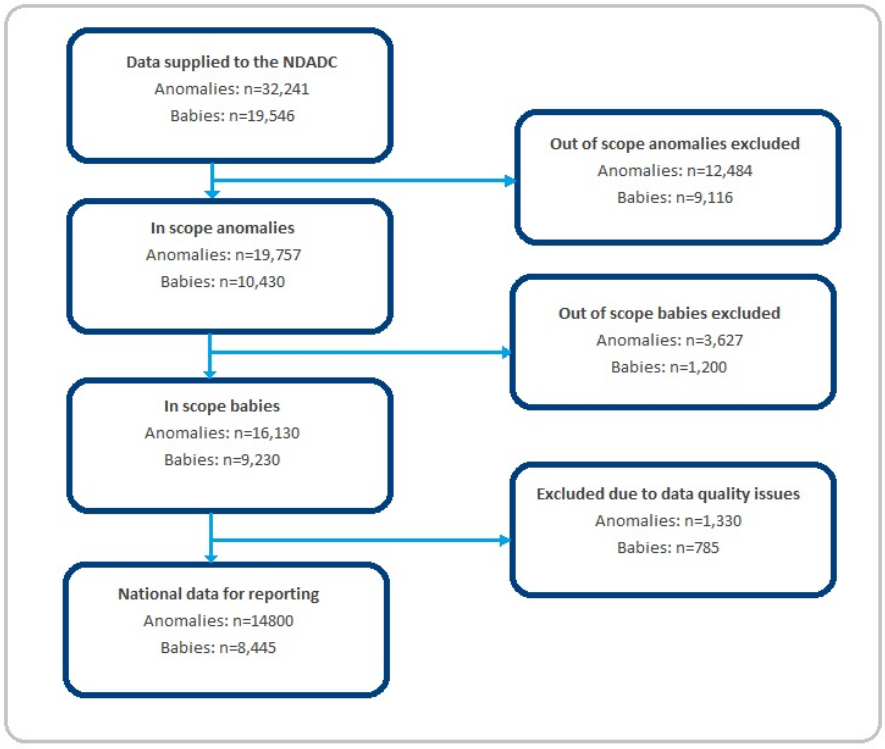
Out of scope anomalies include:

- terminations of pregnancy before 20 weeks' gestation
- anomalies diagnosed after 12 months of age
- anomalies that do not have significant medical, social or cosmetic outcomes – these are sometimes referred to in other literature as minor congenital anomalies (CDC 2020)
- anomalies where data were not available across all reporting jurisdictions.

Some in scope congenital anomalies, 1,226 (4%) were excluded from reporting for data quality reasons. This included where the state of birth was unknown, to ensure duplicate records were not included across jurisdictions, or where records could not be linked to the NPDC.

Figure 6 describes the process for deriving the numbers presented in this report, the cases excluded from national reporting and why these were excluded.

Figure 6. Processing flowchart for 2017 NCADC data



Notes

1. Includes 2017 birth cohort data provided to the National Congenital Anomalies Data Collection from New South Wales, Victoria, Queensland, South Australia, Tasmania, Northern Territory and the Australian Capital Territory. Western Australia were unable to supply 2017 data.
2. Out of scope anomalies were excluded from reporting. These include anomalies that do not have significant medical, social or cosmetic outcomes, and those where data were not available across all reporting jurisdictions.
3. Out of scope babies were excluded from reporting. These include terminations of pregnancy before 20 weeks and those with anomalies diagnosed after 12 months of age.
4. Anomalies excluded due to data quality issues include where: the state of birth was missing; the record was missing a unique identifier; or the record could not be matched to a record in the National Perinatal Data Collection.

Numbers and rates

The NCADC reports estimates per 1,000 total births (live births and stillbirths). This provides information about the rate of the anomaly among all births. Estimates are based on the number of cases (live births and stillbirths) from the NCADC, divided by the total number of births (live births and stillbirths) from the respective NPDC birth cohort. The NPDC includes live births and stillbirths of at least 20 weeks of gestation or at least 400 grams birthweight. Terminations of pregnancy of at least 20 weeks of gestation or at least 400 grams birthweight are included as births. No congenital anomalies data were available for Western Australia in 2017, so this jurisdiction was not included in the numerator or denominator for rate calculations.

It is not possible to identify all cases with a particular anomaly, as a proportion of pregnancies affected with an anomaly will spontaneously miscarry before being diagnosed. This report also excludes terminations of pregnancy before 20 weeks' gestation and cases diagnosed after 12 months of age, so the numbers and rates presented will underestimate the overall prevalence of congenital anomalies in Australia. Moreover, there are differences in the methods used to collect congenital anomalies data between jurisdictions, so the numbers and rates presented should be interpreted with caution. More information about the impacts of these differences can be found in Data quality, availability and interpretation.

Small numbers

Data on specific anomalies are presented at the national level, and any disaggregation by baby and maternal characteristics at the broad body system level, to protect the privacy of individuals. In the baby characteristics and maternal characteristics sections, 'not stated' values were excluded from calculations due to small numbers. These represented a small number (less than 1%) of cases.

Calculation of perinatal mortality rate

The perinatal mortality rate is calculated as the proportion of births in a specified population which are stillbirths or neonatal deaths (perinatal deaths). This proportion is expressed in relation to all births.

The perinatal mortality rate for babies diagnosed with one or more congenital anomalies is calculated as the proportion of births in this population which are stillbirths or neonatal deaths (perinatal deaths). This proportion is expressed in relation to all births in babies with one or more congenital anomalies.

To calculate the perinatal mortality rate:

Perinatal mortality rate = Number of perinatal deaths x 1,000 / Total number of births

Classification system

Anomalies in this report were coded according to the tenth edition of the ICD-10-AM. The classification system used to code congenital anomalies can vary by jurisdiction. In New South Wales, the ICD-9-BPA is used to code anomalies recorded in the New South Wales Register of Congenital Conditions. In Victoria, Queensland, South Australia, Tasmania, the Northern Territory, the Australian Capital Territory, and for the New South Wales admitted patient data collection, anomalies are coded using the ICD-10-AM.

All national congenital anomalies data were standardised to ICD-10-AM. The ICD is hierarchical, with various disease chapters, including one on congenital anomalies that are divided into more specific disease groupings (represented by 3-character codes). These groupings can mostly be divided into more specific disease categories, represented by 4- and 5-character codes. Most NCADC data (98.3%) were supplied and analysed at the 4- and 5-character code level of detail.

Anomaly descriptions

There are no nationally agreed clinical definitions for the anomalies included in this report, so the AIHW worked with the NCAAG to develop simple descriptions for this report. Brief descriptions for a data visualisation and for the glossary were developed. These are based on a range of sources, including for example, the Centers for Disease Control and the WHO birth defect surveillance atlas.

Data elements

Various characteristics of the babies diagnosed with an anomaly and the women giving birth to these babies are presented. Baby characteristics include sex, birthweight, birthweight adjusted for gestational age, gestational age and jurisdiction of birth. Information is also presented on outcomes and whether the baby was liveborn and survived the neonatal and post-neonatal period.

For women giving birth to a baby with an anomaly, information is presented on their First Nations status, age at the birth of their baby, parity, plurality and remoteness area of usual residence. The data elements included in this report are described below.

Baby characteristics

Sex

Data on the sex of each baby were collected as 'male', 'female', 'indeterminate' or 'not stated'. Data for indeterminate or not stated sex were excluded from reporting in the Baby characteristics section due to small numbers.

Birthweight

Birthweight is the first weight of the liveborn, or stillborn baby obtained after birth. In this report babies are defined as being of 'low' birthweight if their weight at birth is less than 2,500 grams. Babies are defined as being a 'normal' birthweight if their weight at birth is 2,500g or more and this includes a small number of babies (around 1%) with a high birthweight (4,500g or more). A small number of records with 'not stated' birthweight were excluded from reporting in the Baby characteristics section due to small numbers.

Gestational age

Gestational age is the duration of pregnancy in completed weeks, calculated from the date of the first day of a woman's last menstrual period and her baby's date of birth; or via ultrasound; or derived from clinical assessment during pregnancy or from examination of the baby after birth. The WHO defines 'pre-term' as less than 37 completed weeks of gestation, 'term' as 37 completed weeks to less than 42 completed weeks of gestation, and 'post-term' as 42 or more completed weeks of gestation. Pre-term birth is associated with morbidity and mortality in newborn babies. In this report babies are defined as being at 'term' if they have 37 completed weeks of gestation. This includes a small number of babies (less than 1%) that were born 'post-term' (at 42 or more completed weeks of gestation). A small number of records with 'not stated' gestational age were excluded from reporting in the Baby characteristics section due to small numbers.

Birthweight adjusted for gestational age

A baby may be small due to being pre-term (born early) or being small for gestational age (either due to genetic factors, or because it is the subject of a growth restriction within the uterus). Adjusting birthweight for gestational age allows for differences in a baby's growth status and maturity to be taken into account when examining their health outcomes at birth. Data on birthweight adjusted for gestational age are limited to liveborn singleton babies. Babies are defined as being 'small for gestational age' if their birthweight is below the 10th percentile for their gestational age and sex, as determined by national percentiles. Babies are defined as being 'large for gestational age' if their birthweight is above the 90th percentile for their gestational age and sex.

Jurisdiction

This is the state or territory where a baby is born. It is not necessarily the state or territory where the woman giving birth usually lives.

Maternal characteristics

Age

The age of a woman at the birth of their baby. A small number of records with 'not stated' age were excluded from reporting in the Maternal characteristics section due to small numbers.

First Nations status

First Nations status refers to whether a woman giving birth has identified as being of Aboriginal, Torres Strait Islander, or both Aboriginal and Torres Strait Islander origin. Non-Indigenous women refers to women giving birth who have not identified as Aboriginal or Torres Strait Islander. A small number of records with 'not stated' First Nations status were excluded from reporting in the Maternal characteristics section due to small numbers.

Parity

Parity refers to the number of previous pregnancies that resulted in live births or fetal deaths. In this report, categories include 'primiparous' for the first pregnancy, or 'multiparous', for one or more previous pregnancies. A small number of records with 'not stated' parity were excluded from reporting in the Maternal characteristics section due to small numbers.

Plurality

Plurality refers to the number of babies resulting from a single pregnancy. In this report, categories include 'singleton' for a single birth, and 'multiple' for the births of twins, triplets, quadruplets, quintuplets, sextuplets and other. A small number of records with 'not stated' plurality were excluded from reporting in the Maternal characteristics section due to small numbers.

Remoteness area

This is the remoteness area of usual residence. This report uses the Australian Statistical Geography Standard (ASGS) that groups geographic areas into 6 classes of Remoteness Area based on their relative access to services using the Accessibility/Remoteness Index of Australia. The 6 classes are: *Major cities*, *Inner regional*, *Outer regional*, *Remote*, *Very remote* and *Migratory* (ABS 2018). Remoteness area is derived by applying the ABS 2011 Australian Statistical Geography Standard to the area of mother's usual residence. It only calculated where geographic area of usual residence was provided. Due to small numbers, remoteness area is presented under 4 categories: *Major cities*, *Inner regional* areas, *Outer regional* areas and *Remote and very remote* areas.

References

ABS (Australian Bureau of Statistics) (2018) *Australian Statistical Geography Standard (ASGS): Volume 5 – Remoteness Structure, July 2016* , ABS website, accessed 5 July 2023.

Technical notes

Due to rounding, percentage totals may not add to 100%.

A baby may have more than one type of anomaly and be counted in more than one type of anomaly category. A baby will only be counted once within each anomaly category. Given this, the sum of individual categories will be greater than the total number of babies with any anomaly.

Every effort has been made to collect and report congenital anomalies data consistently, by using common data specifications and reporting using a similar notification period across jurisdictions. It should be noted, however, that there are differences in the scope and methods used to collect congenital anomalies data across jurisdictions, for example the sources of notification for congenital anomalies varies by jurisdiction (see Table 3).

Table 3. Proportion of congenital anomalies, by jurisdiction and data source

Data source	NSW	Vic	Qld	SA	ACT	Tas	NT
Congenital anomaly register	40.0	5.0	0.0	100.0	0.0	0.0	0.0
Perinatal data collection	0.0	95.0	44.7	0.0	4.5	5.6	93.8
Admitted patient collection	60.0	0.0	55.2	0.0	95.5	94.4	6.2
Other source	0.0	0.0	0.0	0.0	0.0	0.0	0.0
Total	**100.0**	**100.0**	**100.0**	**100.0**	**100.0**	**100.0**	**100.0**

Note: Due to rounding, percentage totals may not add to 100%.

Differences in collection methods may impact national counts and comparability between jurisdictions. For example, in 2017:

- Victoria had relatively more reported cases of chromosomal anomalies and fewer cases of other types of anomalies. This may be due to improved screening and testing for chromosomal anomalies and notifications from cytogenetic laboratories (prenatal diagnosis). Notifications to the VCAR are not mandatory in Victoria, and this may be a factor in the smaller number of notifications for other types of anomalies, compared to jurisdictions with mandated registers or those that use more active surveillance methods.
- Tasmania had a lower number of reported cases compared with other jurisdictions (except Victoria and Northern Territory). This may reflect the narrower notification period of their collection – the perinatal period only, compared with jurisdictions that include cases diagnosed up to 12 months of age.
- The Northern Territory had a relatively low number of reported cases compared with other jurisdictions. This may be due to interstate transfers of babies identified antenatally with major anomalies who require birthing in a specialist tertiary facility. In addition, only the primary anomaly associated with a particular syndrome is reported.
- Queensland had relatively more cases across some body systems (including for example circulatory, urinary, respiratory and musculoskeletal anomalies) and a higher rate for any anomaly overall. This may reflect the multiple sources of notifications and data linkage processes used in this collection (including admitted patient data).
- The Australian Capital Territory perinatal data contains cases of New South Wales residents giving birth in the Australian Capital Territory – the proportion of women giving birth who were non-residents was 15.6% in 2017. In looking at these data it is important to note that births to non-residents may include a disproportionate number of high-risk and multi-fetal pregnancies associated with poorer perinatal outcomes. Women with high-risk pregnancies may be transferred from smaller centres in New South Wales to the Australian Capital Territory to give birth.

More information about the NCADC can be found in the National Congenital Anomaly Data Collection, 2023 Quality Statement.

Comparing NCADC data with jurisdictional reports

The scope for reporting from the NCADC may be different to the scope for reporting from jurisdictional congenital anomaly collections. The AIHW harmonised the national data for reporting around the classification system and the notification period used, and the anomalies included for reporting. This means the numbers and rates in this report may differ from those reported by individual jurisdictions.

Differences between national and jurisdictional reporting may be due to:

- the AIHW reporting on anomalies in babies born in 2017 and diagnosed up to 12 months of age – reports in Victoria are based on anomalies in babies reported in 2017, rather than the year of birth
- the AIHW including data from the admitted patient data collection for New South Wales – in New South Wales, congenital anomalies reporting is based on register data only and does not include data sourced from their admitted patient data collection

- terminations of pregnancy being included in reports in some jurisdictions, regardless of gestational age, for example in reporting in Queensland, South Australia and Victoria
- some jurisdictions, for example, Queensland and South Australia, having a broader scope for reporting with respect to anomaly inclusions or notification periods.

See Table 2 for further information on jurisdictional congenital anomaly reporting.

Terminology

The term 'women giving birth' is used when referring to mothers, whereas 'babies' refers to births.

This report uses the terms 'woman' and 'women' to mean 'female' when referring to data collected in the National Congenital Anomalies Data Collection (NCADC) and the National Perinatal Data Collection (NPDC) as these data sources are based on sex. Information on gender is not recorded in these data collections. 'Woman' and 'women' typically refers to groups of people aged 18 years and over, however in this report people who were pregnant or gave birth aged less than 18 are included.

It is acknowledged that this report includes people who do not identify as women or mothers, and that individual parents and families may use different words to those used in this report. This may include women, transgender men, intersex people, non-binary and gender diverse people.

Data availability and timeliness

Data are presented on babies with in scope congenital anomalies in the 2017 birth cohort, that were diagnosed up to 12 months of age and the women who gave birth to these babies (see Reporting inclusions). It is not possible to provide national data on all cases diagnosed in the 2017 birth cohort, due to differences in collection methods across jurisdictions. This report does not include anomalies that were:

- terminations of pregnancy before 20 weeks' gestation
- diagnosed after 12 months of age, and for Tasmania after the birthing episode
- diagnosed in Western Australia, as these data were not available.

Anomalies were also excluded where data were not available across all reporting jurisdictions, or if they did not have significant medical, social or cosmetic outcomes – these are sometimes referred to in other literature as minor anomalies (see Reporting exclusions).

The numbers and rates presented will therefore underestimate the overall prevalence of congenital anomalies in Australia. It is estimated that around 12–17% of anomalies are diagnosed after 12 months and before 6 years of age (Bower et al. 2010; Gibson et al. 2016). This may be lower or higher depending on the type of anomaly. Analysis of the data supplied in 2017 by South Australia indicates around 15% of anomalies in their 2017 birth cohort were diagnosed after 12 months of age and would be excluded from this national report.

Timeliness

This report is based on the most recent data available across reporting jurisdictions. Data in the NCADC are sourced from various state and territory data collections and collection methods vary. Each collection involves data entry, classification and validation and may include data linkage processes. Some congenital anomalies data collections have long notification periods (up to 5 or 6 years of age), which means the data for a particular birth cohort may take years to finalise. Jurisdictions have noted the following issues in the supply of congenital anomalies data:

1. **Resources and funding**
   The collection of anomalies data is mandated in 4 jurisdictions. There are resource implications for jurisdictions to develop and maintain a congenital anomalies collection.
2. **Complexity of case ascertainment**
   Cases are difficult to ascertain due to their low prevalence rates. Jurisdictions have different methods of case ascertainment and rely on multiple data sources to identify cases of congenital anomalies. There may be timeliness issues around getting data from these different sources and following up sources for case ascertainment. Linkage with other data collections, while improving case ascertainment, may also affect the timely supply of anomalies data as data will need to go through de-duplication and validation processes.
3. **Notification period**
   Timeliness is also dependent on the notification period used. For some congenital anomaly collections (for example, those that collect data until 6 years of age), birth cohort data are incomplete for the first few years.

Previous reports

The Australian Congenital Anomalies Monitoring System (ACAMS) is an AIHW collection containing data on babies with a diagnosed congenital anomaly between 1981 and 2003. The most recent reports from this collection were published in 2007 (Abeywardana et al. 2007), 2008 (Abeywardana and Sullivan 2008) and 2011 (Macaldowie and Hilder 2011). Data in this report are not directly comparable with previous ACAMS reports, due to differences in scope and the period of notification used.

References

Abeywardana S, Karim M, Grayson N and Sullivan EA (2007) Congenital anomalies in Australia 1998–2001, Sydney: AIHW National Perinatal Statistics Unit.

Abeywardana S and Sullivan EA (2008) Congenital anomalies in Australia 2002–2003, Sydney: AIHW National Perinatal Statistics Unit.

Bower C, Rudy E, Callaghan A, Quick J and Nassar N (2010) Age at diagnosis of birth defects, *Birth Defects Research. Part A, Clinical and Molecular Teratology*, 88(4), 251–255. doi:10.1002/bdra.20658

Gibson C, Scott H, Haan E and Scheil, W (2016) Age range for inclusion affects ascertainment by birth defects registers, *Birth Defects Research. Part A, Clinical and Molecular Teratology*, 106(9), 761–766. doi:10.1002/bdra.23534

Macaldowie A and Hilder L (2011). Neural tube defects in Australia: prevalence before mandatory folic acid fortification, Canberra: AIHW.

Data gaps and future directions

The AIHW is working to re-establish a national congenital anomalies data collection. The collection and reporting of congenital anomalies data will assist in monitoring prevalence and trends in congenital anomalies and in planning services for these conditions. There are differences in the way cases are identified across jurisdictions and this will affect both jurisdictional and national counts. Further work is needed to standardise congenital anomalies data across jurisdictions. The next phase of work includes improving both the consistency and timeliness of data provided to the NCADC, supporting the inclusion of data from Western Australia into the collection, and embedding collection and reporting processes.

Technical notes

Birthweight: The first weight of a liveborn or stillborn baby obtained after birth.

Birthweight adjusted for gestational age: A baby may be small due to being pre-term (born early), or due to being small for gestational age (either due to genetic factors, or because it is the subject of a growth restriction within the uterus). Adjusting birthweight for gestational age allows for differences in a baby's growth status and maturity to be taken into account when examining their health outcomes at birth. Data on birthweight adjusted for gestational age are limited to liveborn singleton babies. Babies are defined as being small for gestational age if their birthweight is below the 10th percentile for their gestational age and sex, as determined by national percentiles. Babies are defined as being large for gestational age if their birthweight is above the 90th percentile for their gestational age and sex.

Congenital anomaly: An atypical bodily structure or function that exists at or before birth, although it may not be detected until later in life. It may have significant medical, social or cosmetic outcomes for an individual and typically requires medical intervention. See Table 4 for brief descriptions of the anomalies included in this report.

Fetal death (stillbirth): Death prior to the complete expulsion or extraction from its mother of a product of conception of 20 or more completed weeks of gestation or of 400 grams or more birthweight. The death is indicated by the fact that after such separation the fetus does not breathe or show any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles.

Gestational age: The duration of pregnancy in completed weeks calculated from the date of the first day of a woman's last menstrual period and her baby's date of birth, or via ultrasound, or derived from clinical assessment during pregnancy or from examination of the baby after birth.

Infant death: Death of a liveborn child under 1 year of age.

Live birth: A live birth is defined by the World Health Organization to be the complete expulsion or extraction from the mother of a baby, irrespective of the duration of the pregnancy, which, after such separation, breathes or shows any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of the voluntary muscles, whether or not the umbilical cord has been cut or the placenta is attached. Each product of such a birth is considered liveborn. The NPDC has birthweight and gestational age conditions for those included in the collection as a 'live birth' or 'stillbirth'. The very small number of live births occurring before 20 weeks' gestation and weighing less than 400 grams are not included in the NPDC. Data for babies whose gestational age and birthweight were not recorded are also not included. Live births and stillbirths may include termination of pregnancy after 20 weeks. Terminations of pregnancy performed at 20 or more weeks of gestation may be included and recorded either as stillbirths or, in the event of showing evidence of life, as live births. There are variations in legislation regarding termination of pregnancy between states and territories, and recording of terminations is likely to be incomplete.

Low birthweight: Weight of a baby at birth that is less than 2,500 grams.

Maternal age: Mother's age in completed years at the birth of her baby.

Minor congenital anomalies: These are congenital anomalies that do not pose significant health issues to the baby and have limited social or cosmetic consequences. These types of anomalies were excluded from NCADC reporting.

Neonatal death: Death of a liveborn baby within 28 days of birth.

Parity: Number of previous pregnancies resulting in live births or fetal deaths, excluding the current pregnancy. In this report, categories include 'primiparous' for the first pregnancy, or 'multiparous', for one or more previous pregnancies.

Perinatal death: A fetal or neonatal death of at least 20 weeks' gestation or at least 400 grams birthweight.

Plurality: The number of births resulting from a pregnancy. In this report, categories include 'singleton' for single births, and 'multiple' for the births of twins, triplets, quadruplets, quintuplets, sextuplets and other.

Post-neonatal death: Death of a liveborn baby more than 28 days after birth and less than one year after birth.

Post-term birth: Birth at 42 or more completed weeks of gestation.

Pre-term birth: Birth before 37 completed weeks of gestation.

Stillbirth: A fetal death prior to the complete expulsion or extraction from its mother of a product of conception of 20 or more completed weeks of gestation or of 400 grams or more birthweight.

Termination of pregnancy: The intentional expulsion of a product of conception from the uterus either by medication or instrumentation, with the intention being the death of the embryo or fetus. This includes induction of labour without expectation of fetal survival, for example, in the case of severe pre-eclampsia at pre-viable gestations or prolonged rupture of membranes with severe infection.

Technical notes

Descriptions of the anomalies included in this report are outlined below. These were developed in consultation with the NCAAG and clinical experts using a range of information sources.

Table 4. Congenital anomaly inclusions and descriptions

Type of anomaly	Description
Chromosomal	
Trisomy 21 (Down syndrome)	Trisomy 21, otherwise known as Down syndrome, is a chromosomal anomaly in which there is an excess of chromosome 21 material. In most cases this is due to an extra copy of chromosome 21 in each cell (trisomic Down syndrome); less frequently it is due to an extra copy of chromosome 21 being present in only some cells (mosaic Down syndrome) or a piece of an extra copy of chromosome 21 being attached to another chromosome (translocation Down syndrome). Down syndrome varies in severity causing intellectual disability and developmental delays. It is the most common genetic chromosomal disorder and cause of learning disabilities in children. Babies born with Down syndrome will usually have other anomalies.
Trisomy 18 (Edwards syndrome)	Trisomy 18, otherwise known as Edwards syndrome, is a chromosomal anomaly in which there is an excess of chromosome 18 material. In most cases this is due to an extra copy of chromosome 18 in each cell (trisomic Edwards syndrome); less frequently it is due to an extra copy of chromosome 18 being present in only some cells (mosaic Edwards syndrome) or a piece of an extra copy of chromosome 18 being attached to another chromosome (translocation Edwards syndrome). Babies with Edwards syndrome are usually small at birth due to intrauterine growth delay and may be stillborn; most liveborn babies with Edwards syndrome have a limited life expectancy. All babies with Edwards syndrome have learning disabilities and may have other anomalies.
Trisomy 13 (Patau syndrome)	Trisomy 13, otherwise known as Patau syndrome, is a chromosomal anomaly in which there is an excess of chromosome 13 material. In most cases this is due to an extra copy of chromosome 13 in each cell (trisomic Patau syndrome); less frequently it is due to an extra copy of chromosome 13 being present in only some cells (mosaic Patau syndrome) or a piece of an extra copy of chromosome 13 being attached to another chromosome (translocation Patau syndrome). Patau syndrome babies may miscarry, be stillborn or be small for gestational age due to intrauterine growth restriction, and for liveborn babies it is a life-limiting condition. All babies with Patau syndrome have a range of cardiac, central nervous system and skeletal anomalies.
Female sex chromosome anomalies (including Turner syndrome)	Turner syndrome is a female only chromosomal congenital anomaly. The cause is either a missing X-chromosome (monosomic 45, X) or a chromosomal rearrangement involving the X chromosome (translocation Turner syndrome). The chromosomal anomaly may only be present in some cells (mosaic Turner syndrome). Women with Turner syndrome are short in stature and many have non-functioning ovaries leading to an absence of periods, infertility and premature menopause.
Male sex chromosome anomalies (including Klinefelter syndrome)	Klinefelter syndrome is a male only chromosomal congenital anomaly in which there is an extra copy of the X chromosome (47, XXY). Men with Klinefelter syndrome may not present any symptoms, though learning and behavioural disorders and infertility are common.
Other chromosomal	Other chromosomal anomalies include: • other autosomal trisomies • monosomy and rearrangements • Fragile X-associated conditions • Chimerism • true hermaphrodite.
Any chromosomal	Includes anomalies related to the addition to, absence of and rearrangement of chromosomes. Humans have 23 pairs of chromosomes (thread-like structures in the nucleus of cells).
Circulatory system	

Transposition of the great vessels	Transposition of the great vessels is an anomaly where the 2 main arteries carrying blood out of the heart—the pulmonary artery and the aorta—are switched in position. This means oxygen-poor blood is circulated to the body instead of the lungs and oxygen-rich blood returns to the lungs instead of the body. It is considered a critical congenital heart defect because a baby with this anomaly will need surgery or other procedures soon after birth.
Cardiac septal anomalies	Cardiac septal anomalies are a group of heart conditions in which there is a hole in the wall that divides the chambers of the heart. This includes: • atrial septal defects • atrioventricular septal defects • ventricular septal defects • other cardiac septal defects. Many of these holes are found by chance and never cause any concerns. Small holes may close on their own and medium to large holes may need surgical closure during childhood.
Atrial septal defect	An atrial septal defect is an anomaly in which there is a hole in the wall that separates the 2 upper chambers (right and left atria) of the heart. Before birth this opening allows blood to detour away from the lungs. After birth the opening is no longer needed and closes or becomes very small within several weeks or months. An atrial septal defect results when the opening is larger than normal and does not close after birth. Small defects may be found by chance, may never cause a problem and close during infancy or early childhood. Large defects may require surgical closure later in life.
Atrioventricular septal defect	An atrioventricular septal defect is an anomaly in which there is a hole in the centre of the heart and the valves (mitral and tricuspid) which control the blood flow between the chambers may not have developed typically. This leads to blood flowing back through the hole from the left chamber (which normally pumps blood only to the body) to the right chamber and into the lungs, which causes the lungs and heart to work harder than they should. These defects can be classified into 3 categories: complete, partial (or incomplete) and transitional. Babies born with an atrioventricular septal defect require surgery and may have life-long complications. With regular follow-up and treatment, most grow up to lead healthy lives.
Ventricular septal defect	A ventricular septal defect is a hole in the wall that separates the 2 lower chambers of the heart (right and left ventricles). This means that oxygen-rich blood passes from the left lower chamber through the hole where it mixes with oxygen-poor blood in the right lower chamber. Large defects may need surgical repair early in life to prevent complications. Small defects may not cause problems and close on their own.
Other cardiac septal defects	Includes other cardiac septal defects not already specified.
Tetralogy of Fallot	Tetralogy of Fallot is an anomaly in which 4 cardiac malformations are present: • narrowing of the pulmonary artery which carries blood from the heart to the lungs for oxygen • a ventricular septal defect or opening in the wall between the 2 lower chambers of the heart (right and left ventricles) • an overriding aorta such that the artery that carries oxygen-rich blood to the body is shifted toward the right side of the heart (it should be on the left side) • enlargement of the right ventricle. As a result, oxygen-poor blood is returned to the body rather than the lungs. Tetralogy of Fallot may be associated with a chromosomal anomaly. It is considered a critical congenital heart defect as a baby will need surgery or other procedures soon after birth.
Pulmonary valve atresia	Pulmonary valve atresia is an anomaly in which the pulmonary valve is not formed, so blood cannot get from the right ventricle of the heart to the lungs via the pulmonary artery. Blood must use other routes to bypass the unformed pulmonary valve to allow the oxygenation of the blood. This anomaly is considered a critical congenital heart defect because a baby may need surgery or other procedures soon after birth.
Hypoplastic left heart syndrome	Hypoplastic left heart syndrome is an anomaly in which a number of structures on the left side of the heart are underdeveloped, including the left ventricle, the mitral valve, the aortic valve and the ascending portion of the aorta. Babies with hypoplastic left heart syndrome frequently also have an atrial septal defect. This means the left side of the heart cannot pump oxygen-rich blood to the body properly. This anomaly is considered a critical congenital heart defect because a baby may need surgery or other procedures soon after birth.

Patent ductus arteriosus	Patent ductus arteriosus is an anomaly in which a vascular connection between the aorta and the pulmonary artery persists after a baby is born. During pregnancy, this connection allows blood to bypass circulation to the lungs. After birth, blood must receive oxygen in the lungs and the connection should close. This anomaly results when the hole remains open and blood may skip oxygenation. If left untreated, it can result in pulmonary hypertension and heart failure. It can be treated with surgery or other procedures.
Coarctation of aorta	Coarctation of aorta is an anomaly in which a segment of the aorta (the main artery leaving the heart to supply blood to the body) is narrowed and the blood which typically flows through the aorta is obstructed. It is indicated by weak pulses in the legs or groin and a heart flow murmur found by examination of the baby. If severe, the heart must work harder and high blood pressure and heart failure may result, though symptoms may not appear for some time. Coarctation of aorta often occurs with other cardiac anomalies and may require surgery.
Other circulatory system	Other circulatory system anomalies include anomalies of the: • cardiac chambers and their connections • cardiac septa and the cardiac valves • circulatory system that are rare, such as the heart being on the opposite side of the body (dextrocardia).
Any circulatory system	Includes anomalies of the circulatory system, i.e. the heart and the major blood vessels. The circulatory system (often called the cardiovascular system) is the system that allows blood containing oxygen and nutrients to reach the cells and tissues.
Digestive system	
Cleft lip and/or palate	Cleft lip and/or palate is an anomaly characterised by partial or complete splitting of the upper lip, with or without splitting of the hard palate behind the upper front teeth. Babies with cleft lip and/or palate often have challenges with feeding, hearing and speech. Treatment depends on the severity of the cleft and surgery may be required.
Tracheo-oesophageal fistula	Tracheo-oesophageal fistula is an anomaly in which there is an atypical connection between the oesophagus and the trachea. Normally, the oesophagus and trachea are not connected. It may be associated with oesophageal atresia or the incomplete development of the oesophagus. When a baby with trachea-oesophageal fistula swallows, liquid can pass into the lungs resulting in symptoms such as frothy, white bubbles in the mouth, coughing and choking. Surgical closure of the fistula is often required to allow feeding and to avoid lung damage.
Anomalies leading to oesophageal obstruction (without fistula)	Anomalies which lead to a partial or complete blockage of the oesophagus, causing swallowing difficulties. These include: • atresia of oesophagus without fistula • stenosis and stricture of oesophagus (narrowing) • oesophageal web (a thin web partially obstructing the oesophagus). Oesophageal atresia is an anomaly in which the lower end of the oesophagus is closed and does not connect with the stomach, and as such ends in a blind pouch. It is commonly diagnosed after birth when the baby first tries to feed and has choking or vomiting, or when a tube inserted in the baby's nose or mouth cannot pass down into the stomach. Oesophageal stenosis, stricture and web present with feeding difficulties in a baby and may not be obvious in the newborn.
Congenital hypertrophic pyloric stenosis	Congenital hypertrophic pyloric stenosis is an anomaly caused by the thickening of the lower part of the stomach which connects to the small intestine. This can lead to near complete obstruction of the digestive system preventing food from reaching the small intestine. Pyloric stenosis usually appears within 3 to 5 weeks after birth. It can lead to projectile vomiting, dehydration and weight loss and difficulty feeding. Surgery is required.
Atresia/stenosis of intestines	Atresia of the intestines occurs when there is a complete closure of the intestine. Stenosis is a partial obstruction which causes the opening of the intestine to become narrower. Intestinal atresia and stenosis generally occur at the small intestine but can occur at any point in the gastrointestinal tract. These obstructions prevent proper absorption of food and may lead to vomiting, dehydration, swelling of the abdomen and difficultly in putting on weight. Babies with intestinal atresia or stenosis will require surgery.
Hirschsprung disease	Hirschsprung disease is an anomaly characterised by the absence of the normal movement (peristalsis) in a segment of the bowel due to a lack of nerve supply in that section. As a result, the muscles in the bowel lose their ability to move the bowel contents through the intestine, leading to constipation and/or partial or complete bowel obstruction. Surgery is usually required.

Other digestive system	Other digestive system anomalies include anomalies of the: • mouth and oesophagus, such as obstructive webs and pouches (oesophageal diverticulum) • junction between the oesophagus and stomach (congenital hiatus hernia) • intestine, including twisting and duplication • liver (such as cystic liver disease) and bile ducts (biliary atresia) • pancreas (such as absence, cyst or underdevelopment).
Any digestive system	Includes anomalies of the mouth, oesophagus, stomach, intestines, liver and pancreas.
Eye, ear, face and neck and integuments	
Anophthalmia and microphthalmia	Anophthalmia and microphthalmia are anomalies of the eye. Anophthalmia describes the absence of a recognisable eyeball and microphthalmia an unusually small eyeball. Both conditions may affect one or both eyes. These anomalies are frequently associated with other anomalies.
Microtia with congenital absence, atresia and stricture of external auditory canal	Microtia is characterised by a small and atypically shaped external ear. In most severe cases, the external ear may be absent. Microtia is often associated with the absence or narrowing of the external ear canal. These anomalies are frequently one-sided but may be two-sided and are often associated with other anomalies.
Other eye, ear, face and neck and the integuments	Other eye, ear, face and neck and integumentary system anomalies include anomalies of: • the eyes (including the lens, sclera, iris, cornea and retina) • the ears (including the external ear, ear canal and inner ear mechanisms) • skin and breast • cyst and cleft anomalies of the neck.
Any eye, ear, face and neck and the integuments	Includes anomalies of the eyes, the ears, the face and neck and the integumentary system (skin, hair, nails, sweat glands and breasts).
Genital organs	
Doubling anomalies of the female genitalia	Doubling anomalies of the female genitalia are anomalies where there is a doubling of either the uterus (from a dimple in the top of the uterus to a completely double uterus) or other genital organs such as the vagina. The uterus normally develops from the fusion of two cords of tissue (Mullerian ducts), and this group of anomalies occur when that fusion is incomplete. A duplicate uterus may or may not also develop duplicate cervixes and vaginas. There may be no apparent symptoms or symptoms may become apparent during puberty and treatment, if required, may be undertaken in adulthood.
Hypospadias	Hypospadias is an anomaly where the opening of the urethra is on the underside of the penis instead of at the tip. The urethra is the tube through which urine drains from the bladder and passes through the penis in a male. Hypospadias does not cause difficultly for an infant. Surgery is undertaken at 6 to 12 months of age and aims to restore the regular appearance of the penis and functions such as typical urination and reproduction.
Anomalies related to indeterminate sex	Congenital anomalies related to indeterminate sex are anomalies where there is ambiguity of a baby's genitalia and the observable genital characteristics do not readily allow for sex determination at birth.
Other genital organs	Other genital organ anomalies include: • anomalies of the development of the ovaries, uterus, cervix or vagina • congenital connection between rectum and vagina (rectovaginal fistula) • anomalies of the development of the penis or testes • development of testes in an abnormal site (ectopic testis) • other congenital anomalies of the vas deferens, epididymis, seminal vesicles and prostate.
Any genital organs	Includes anomalies of the genital organs (those involved in human reproduction), including the uterus, ovaries, Fallopian tubes, cervix and vagina; penis, testes and scrotum.

Musculoskeletal system	
Congenital hip dislocation	Congenital hip dislocation (otherwise known as developmental dysplasia of the hip) is an anomaly of the hip joint. The ball at the top of the femur is not stable within the hip socket as the socket of the hip is too shallow, and the ligaments of the hip joint that hold it together may also be loose. In severe cases, the femur may also dislocate. Congenital hip dislocation may affect one or both hips. It is usually treated by a brace or plaster case to position the baby's hips correctly and surgery may be needed as the baby grows.
Talipes	Talipes is a group of anomalies in which the position of the foot is not typical. In talipes equinovarus, the foot turns inward and the front half of the foot points down. In talipes calcaneovalgus, the foot is pushed up and the front half of the foot turns outwards. Talipes is treated by combinations of stretching, casting and bracing, and surgery.
Polydactyly	Polydactyly is an anomaly where a baby is born with extra fingers or toes. It can affect the hand, the foot, or both and is usually treated surgically.
Syndactyly	Syndactyly is an anomaly where a baby is born with webbed or conjoined digits (fingers and/or toes). It may occur in isolation or be part of an inherited syndrome. It is usually treated surgically.
Reduction defect of upper limb(s)	Reduction defect of upper limb(s) is an anomaly where a part of, or the entire arm (upper limb) does not form completely during pregnancy. This is referred to as a 'limb reduction' because a limb is reduced from its expected size or is missing. Treatment is individualised to each child as each anomaly causes unique challenges.
Reduction defect of lower limb(s)	Reduction defect of lower limb(s) is an anomaly where a part of, or the entire leg (lower limb) does not form completely during pregnancy. This is referred to as a 'limb reduction' because a limb is reduced from its expected size or is missing. Treatment is individualised to each child as each anomaly causes unique challenges.
Congenital diaphragmatic hernia	Congenital diaphragmatic hernia is an anomaly in which the diaphragm does not develop typically. The diaphragm usually separates the organs in the abdomen from those in the chest. Severity ranges from a thinned area in part of the diaphragm to its complete absence. It may allow the stomach and intestines to protrude into the chest cavity, crowding the heart and lungs. This can lead to the underdevelopment of the lungs. Congenital diaphragmatic hernia is a critical condition and requires surgery after birth.
Exomphalos	Exomphalos (also known as Omphalocele), is an anomaly where there is incomplete development of the baby's abdominal wall where it meets the umbilical cord. This leads to the intestine, and at times liver and bowel, protruding outside the abdomen in a sac surrounding the umbilical cord. A baby with exomphalos will require surgery after birth to place the organs within the abdominal cavity and close the abdominal wall.
Gastroschisis	Gastroschisis is an anomaly where an opening forms in the abdominal wall, generally at the right side of the joining of the umbilical cord. This leads to the bowel protruding outside the body and developing in the amniotic fluid, causing the intestines to become irritated, shortened, or swollen. Surgery is required after birth to place the organs inside the baby's abdomen and to close the abdominal wall.
Other musculoskeletal system	Other musculoskeletal system anomalies include anomalies such as: • arthrogryposis multiplex congenita • prune belly syndrome • Klippel-Feil syndrome • achondrogenesis • thanatophoric short stature • osteogenesis imperfecta.
Any musculoskeletal	Includes any anomalies of the bones, muscles, tendons, ligaments and soft tissues.
Nervous system	
Neural tube defects	Neural tube defects occur when brain and spinal development is disrupted. During gestation, a flat neural plate 'rolls up' into a tube to form the head and back. If the closure of the neural tube is incomplete, a neural tube defect results. A number of nervous system anomalies are grouped together as neural tube defects including: • anencephaly and related major cephalad neural tube defects • encephalocele • spina bifida.

Anencephaly and related anomalies	Anencephaly is an anomaly characterised by the total or partial absence of the cranial vault, the covering skin, and the brain is missing or reduced to a small mass. The related conditions of craniorachischisis, iniencephaly and acrania are included. Babies with anencephaly are usually either stillborn or die shortly after birth.
Encephalocele	Encephalocele is an anomaly in which there is a sac-like protrusion of the brain and the membranes that cover it through an opening in the skull. It occurs when a portion of the developing skull fails to close properly (a 'neural tube defect'). It is treated surgically by placing the protruding part of the brain and the membranes covering it back into the skull and closing the opening in the skull. It can cause long-term neurological complications and multiple surgical treatments may be necessary.
Spina bifida	Spina bifida describes a group of anomalies in which the developing spinal cord does not develop or close properly (a 'neural tube defect'). This leads to the exposure or protrusion of the spinal cord, with or without its coverings, through the spine. It is usually associated with a degree of paralysis or other nervous system dysfunction. Also includes Arnold-Chiari malformation, an anomaly in which part of the brain at the back of the skull bulges through the opening where the skull joins the spinal canal. This places pressure on part of the brain and spinal cord and can cause dizziness, muscle weakness, numbness, vision problems, headache and challenges with balance and coordination.
Microcephaly	Microcephaly is an anomaly in which a baby's head is significantly smaller than expected when compared to other babies of the same sex and gestational age. It occurs when a baby's brain has not developed typically during pregnancy and has been linked to a range of infections and exposure to harmful substances during pregnancy. It is associated with developmental delay and intellectual disability, seizures and difficulties with vision, hearing and feeding. It is a life-long condition with no known treatments or cure.
Congenital hydrocephalus	Congenital hydrocephalus is an anomaly where there is an excess of the fluid that surrounds and protects the brain and spinal cord. This increases pressure on the brain and causes brain injury and other outcomes. If left untreated, blindness and ongoing cognitive deterioration may occur. Surgical treatments are available to divert the cerebrospinal fluid to another part of the body to reduce pressure on the brain.
Other nervous system	Other nervous system anomalies include anomalies of the: • brain, such as the absence or reduced development of sections of the brain and developmental cysts in the brain • spinal cord, such as the absence, splitting and central canal dilatation.
Any nervous system	Includes anomalies of the central nervous system (brain and spinal cord) and the peripheral nervous system (body nerves).
Respiratory system	
Choanal atresia	Choanal atresia is an anomaly of the rear nasal airway characterised by the obstruction of one or both airways. The effects of this obstruction vary from acute respiratory distress preventing breast feeding because the baby cannot nose breath to chronic nasal obstruction. Endoscopic surgical treatment can treat this issue.
Hypoplasia and dysplasia of lung	Hypoplasia and dysplasia of the lung is an anomaly that occurs when lung tissue and airways are underdeveloped. If severe, lung function may be unable to sustain life. Pulmonary hypoplasia is often associated with other anomalies and the type of treatment will vary. Babies that survive will often have life-long lung issues. During pregnancy, pulmonary hypoplasia is frequently associated with a reduced volume of amniotic fluid surrounding the baby.
Other respiratory system	Other respiratory system anomalies include anomalies of the: • upper airways (the larynx and throat) such as web-like tissue in the larynx (laryngeal web) and narrowing of a portion of the windpipe (subglottic stenosis) • major airways in the lungs, such as when the cartilage that keeps the airway open (trachea and/or bronchi) is soft which makes it difficult to keep the airways open (congenital tracheomalacia and bronchomalacia) • lungs, such as a mass in the chest (congenital cystic lung).
Any respiratory system	Includes anomalies of the respiratory system, i.e. the airways and lungs.
Urinary system	
Renal agenesis/hypoplasia	Renal agenesis is an anomaly where a baby is missing one or both kidneys and renal hypoplasia is where a baby has small and poorly functioning kidneys. Renal agenesis or hypoplasia is usually indicated by an insufficient amount of amniotic fluid during pregnancy which places pressure on the baby. In severe cases of reduced amniotic fluid, compression may lead to Potter syndrome. Potter syndrome refers to a group of features including a flattened nose, recessed chin, skin folds covering the corners of the eyes and low-set ears. Low amniotic fluid can also lead to underdeveloped lungs and severe breathing difficulties for the baby.

Cystic kidney disease	Cystic kidney disease is a group of anomalies where there are one or more cysts in the kidneys. Polycystic kidney disease is a genetic disease that causes cysts to grow inside the kidneys and the kidneys grow larger than they should. It can also cause cysts to develop in the liver and elsewhere in the body. Cystic kidney disease may cause high blood pressure, blood in urine, kidney infections, kidney stones or kidney failure. Early detection and treatment can reduce or prevent complications.
Exstrophy of urinary bladder	Exstrophy of urinary bladder is an anomaly where the bladder opens into the abdominal wall between the umbilicus and the pubic bone. It is often associated with epispadias and structural anomalies of the pubic bones. Without treatment, babies born with bladder exstrophy will be unable to hold urine. Surgery is undertaken to prevent kidney damage and to correct the functioning and appearance of the urinary system and genitals.
Other urinary system	Other urinary system anomalies include: • accessory kidneys (one or more extra kidneys) • blockage of the ureter (the tube leading from the kidney to the bladder) leading to a swollen kidney (congenital hydronephrosis) • atypical development of the ureter (agenesis, atresia or stenosis of ureter) • urinary flow towards, rather than away from, the kidney (congenital vesico-uretero-renal reflux) • atypically placed (ectopic) kidney • over-developed kidney (hyperplastic or giant kidney) • tissue flaps in the bladder of male babies obstructing urine outflow (congenital posterior urethral valves).
Any urinary system	Includes anomalies of the ureters, kidneys and bladder.

Sources: Abeywardana and Sullivan 2008; GARD 2020; ICBDSR 2014; Mayo Clinic 2020; NCBDDD 2020; NHS 2020; NORD 2020; OMIM 2020; Orphanet 2020; RCH 2020; Shaun and Shen 2006; WHO et al. 2014.

The following resources have more detailed clinical information on specific congenital anomalies and rare diseases.

- Genetic and Rare Diseases Information Center
- Human Phenotype Ontology

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Technical notes

Abbreviations

Abbreviation	Full name
ABS	Australian Bureau of Statistics
ACAMS	Australian Congenital Anomalies Monitoring System
ACT	Australian Capital Territory
AIHW	Australian Institute of Health and Welfare
ASGS	Australian Statistical Geography Standard
CALF	Congenital Anomaly Linked File
CDC	Centers for Disease Control
GARD	Genetic and Rare Disease Information Center
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
ICD-10-AM	International statistical classification of diseases and related health problems, 10th revision, Australian modification
ICD-9-BPA	International statistical classification of diseases and related health problems, 9th edition, British Paediatric Association
NSW	New South Wales
NT	Northern Territory
NCADC	National Congenital Anomalies Data Collection
NCAAG	National Congenital Anomaly Advisory Group
NPDC	National Perinatal Data Collection
Qld	Queensland
SA	South Australia
SABDR	South Australian Birth Defects Register
Tas	Tasmania
VCAR	Victorian Congenital Anomalies Register
Vic	Victoria
WA	Western Australia
WARDA	Western Australian Register of Developmental Anomalies
WHO	World Health Organization

Symbols

Symbol	Meaning
. .	not applicable
n.a.	not available
n.p.	not published

Notes

Amendments

02 May 2022 - A correction was made to supplementary data Table 1.1: Number, rate and per cent of congenital anomalies, 2016.

Acknowledgements

This web report was prepared by the Maternal and Perinatal Health Unit at the AIHW and was made possible by the contribution of many stakeholders. The AIHW would especially like to thank members of NCAAG, NPDDC, the National Maternity Data Development Project Advisory Group (NMDDPAG) and clinical consultants Michael Humphrey AM, and Jeremy Oats AM, for their expertise, guidance, and support in re-establishing the NCADC and in developing the first web report released in 2022.

The AIHW gratefully acknowledges the continued time, effort and expertise of all clinical staff in collecting and providing data for both the NCADC and the NPDC and thanks state and territory data providers for their ongoing contribution of congenital anomalies data to the NCADC. Visit Maternal and Perinatal Health Committees to see current members of NCAAG, NPDDC and NMDDPAG.

Data quality statement

National Congenital Anomalies Data Collection, 2023 Quality Statement

Data

Report editions

This release

Congenital anomalies in Australia | 29 Jun 2023

Previous releases

- Congenital anomalies in Australia 2002-2003 |
  Publication | 14 May 2008
- Congenital anomalies in Australia 1998-2001 |
  Publication | 08 Oct 2007
- Congenital malformations, Australia 1997 |
  Publication | 21 Jan 2001
- Congenital malformations Australia 1995 and 1996 |
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