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## Birth defects explained

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### Summary

- A birth defect, also called a 'congenital anomaly', 'congenital malformation', 'congenital abnormality' or 'congenital disorder' is any abnormality occurring before birth. Birth defects are usually detected during pregnancy, at birth or in early childhood.
  - Birth defects can be major or minor.
  - Most birth defects are structural (how the body is built) or functional (how the body works) or both.
  - Although the cause of many birth defects is unknown, there are things you can do to increase the chance of having a healthy baby.
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Birth defects (also known as congenital anomalies, congenital malformations, congenital abnormalities or congenital disorders) are a major cause of death and disability in children. They are the leading cause of death around the time of birth (perinatal mortality). In Victoria, approximately one in every 22 pregnancies is affected by a birth defect.

A birth defect is any abnormality occurring during pregnancy. Birth defects may be detected at any time throughout a person's life or not at all. They are commonly detected during pregnancy, at the time of birth or in early childhood. Birth defects may be structural, functional, genetic, chromosomal or biochemical and may be minor or major in nature. In some cases, they may be fatal.

The most commonly reported birth defects are chromosomal. This means there are missing, extra or other changes to chromosomes, which contain our genetic information. Structural birth defects are less common and include anomalies of the urinary tract and the heart. Some structural birth defects can be treated surgically.

### Types of congenital anomalies

Birth defects can be broadly classified as:

- **structural:** birth defects affecting the way the body is built. For example, a body part is absent or improperly formed. Examples include:
  - **spina bifida** – incomplete closing of the backbone and membranes around the spinal cord
  - **hypospadias** – where the tube from the bladder is in an abnormal position in male babies
  - **cleft lip** – an abnormal gap in the upper lip
  - **ventricular septal defect** – an abnormal connection between the left and right ventricles of the heart
- **functional:** anomalies affecting the way the body works or develops. Examples include those affecting the nervous system or brain such as:
  - **Down syndrome**
  - **phenylketonuria** – a metabolic disorder where the body can't break down the amino acid phenylalanine
  - **haemophilia** – where the body lacks a specific factor needed for blood clotting
  - **Tay-Sachs disease** – a degenerative disorder affecting nerve cells in the brain and spinal cord.

### Causes of birth defects

Birth defects can occur during any stage of pregnancy, however most occur in the first three months when the organs of the baby are developing.

Factors that may contribute to or increase the chances of a birth defect include:

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- **genetics** – an altered or ‘faulty’ gene or set of genes usually means that the information contained in the particular gene is either changed or missing. Genetic testing and screening for the most common disorders can help identify any issues prior to conception and birth
- **socio-demographic factors** – maternal age and some ethnic groups have been associated with an increased risk of birth defects. Consanguinity (where parents descend from the same kinship and have similar genetic make-up) is also associated with an increased risk of birth defects. Couples that have a similar genetic make-up are more likely to have a common gene variation that may result in a birth defect. Genetic screening of these couples before trying for a baby may identify potential inherited disorders
- **drugs and alcohol** – drugs such as alcohol, tobacco, illegal drugs and certain prescription and over-the-counter medications are known to cause congenital anomalies if taken during pregnancy. This includes some complementary and Ayurvedic (traditional Indian) medicines. **Alcohol** and **tobacco** should be avoided during pregnancy. Consult your doctor if you have concerns regarding medication.
- **environmental factors** – exposure to pesticides and other chemicals, such as lead and mercury (including some cleaning products) and radiation (from x-ray machines or other sources) may increase the risk of birth defects and should be avoided where possible. If you have any concerns regarding environmental exposures during pregnancy speak with your doctor
- **vaccine preventable infections** – during pregnancy infections such as **rubella** and **chickenpox** can cause brain, heart, limb and eye anomalies. Some infectious diseases can be prevented by immunisation. Women planning a pregnancy should ensure they have been **immunised**. Talk with your doctor regarding recommended vaccinations.
- **other infections** – some infectious diseases can cause serious harm to unborn babies:
  - **cytomegalovirus (CMV)**, a virus spread through bodily fluids, can be passed from mothers to their unborn baby and may cause disability. Pregnant women should wash their hands after handling bodily fluids from babies or children, for example, after changing nappies or wiping noses
  - **toxoplasmosis** is caused by a parasite. It can be transmitted by ingestion of raw or undercooked contaminated meat; by drinking contaminated unpasteurised milk or by exposure to infected animal faeces, mainly cats. To avoid toxoplasmosis, ensure meat is properly stored and prepared and don't consume raw or uncooked meat – even in restaurants. Avoid changing cat litter while pregnant. If no one else can perform the task, wear disposable gloves and wash your hands thoroughly with soap and warm water afterwards.
- **inadequate nutrition** – adequate folate (or synthetic supplement ‘folic acid’) before and during pregnancy may prevent some neural tube (spinal cord) defects. Foods rich in folate include asparagus, broccoli, oranges, bananas and chickpeas. Most women do not get enough folate from their diet, so a folic acid supplement is recommended. A deficiency in iodine can also lead to intellectual disability in children. Ask your doctor about your dietary requirements during pregnancy
- **pre-gestational diabetes** – women who have pre-gestational diabetes are at increased risk of poor health outcomes. Management and control of blood sugar levels before and during pregnancy may decrease the frequency and risk of birth defects
- **obesity** – obesity in pregnancy has been associated with increased risk of congenital anomaly, stillbirth and infant death. If you are concerned about your weight or diabetes, speak with your doctor about a management plan
- **diet** – some fish may contain high levels of mercury. Exposure to mercury can be harmful to the nervous system of the developing fetus. If you are pregnant, avoid consumption of fish that contain high levels of mercury such as shark, orange roughy, swordfish and ling. For **information on safe levels of mercury** refer to the Food Standards Australia New Zealand (FSANZ) guidelines
- individual pregnancy factors – for example, the mother's blood and the baby's blood may be incompatible (known as **Rhesus or Rh disease**). Discuss individual risk factors with your doctor
- **fertility treatment** – the contribution of **assisted reproductive technology** (ART) to birth defects is uncertain. Some studies have shown a positive association between ART and certain birth defects, such as cleft lip/cleft palate, however other studies found no association. Discuss individual risk factors and concern with your doctor.

## Diagnosis of birth defects during pregnancy

Some women may be at increased risk of having a baby with a birth defect because of maternal risk factors such as age, general health and medical or family history. Testing during pregnancy, especially for women most at risk, can assist in the diagnosis of birth defects before birth.

Some of the tests used during pregnancy include:

- **combined first trimester screening (CFTS)** – a maternal serum screening test performed before 12 weeks of gestation. It combines information from ultrasound and maternal serum markers to calculate the risk of certain chromosomal anomalies (trisomies 13, 18 and 21) in the baby
- **non-invasive prenatal testing (NIPT)** – a sample of the mother's blood is taken to predict the chance of trisomies 13, 18 and 21 in the baby. This test can also detect other chromosomal anomalies (including sex chromosome anomalies) with less accuracy
- **second trimester maternal serum screening (2TMSS)** – a single blood test performed between 14 and 20 weeks of gestation to provide an estimate of the chance of chromosomal anomalies and neural tube defects
- **ultrasound** – a non-invasive early screening scan that uses soundwaves to create a picture of the baby within the womb. It can identify most major structural fetal anomalies and is often used together with other tests to diagnose a birth defect
- **amniocentesis** – a test involving a needle being inserted through the mother's abdominal wall to take a sample of amniotic fluid for genetic testing
- **percutaneous** (through the skin) umbilical blood sampling – the doctor inserts a needle through the mother's abdominal wall into the umbilical cord. A sample of the baby's blood is taken from the cord for genetic testing
- **chorionic villus sampling (CVS)** – a test where a small sample of cells is taken from the placenta. CVS is commonly used to check for Down syndrome and other genetic conditions.

### Reduce the risk of congenital anomalies

The prevalence of major birth defects in Victoria in 2015–2016 was 4.5 per cent. This means approximately one in every 22 pregnancies in Victoria was affected by a major birth defect.

There are some things you can do to reduce the risk of congenital anomalies.

If you are planning to become pregnant, speak to your doctor about:

- vaccinations that are recommended before you become pregnant, during pregnancy or following birth
- your risk factors for birth defects, and management options. Some conditions, such as diabetes and obesity, increase the risk of birth defects when not managed appropriately. Maintaining a healthy weight and adequate blood sugar control are important for a healthy pregnancy
- your travel plans, if you are planning to travel overseas
- managing any ongoing conditions
- treatment for any medical conditions you have not previously discussed with your doctor, such as sexually transmitted infections (STIs)
- folic acid – it is recommended that women take a folic acid supplement for at least one month prior to conception and during the first trimester as directed by their doctor. This can be supplemented by a healthy diet rich in folate or vitamin B
- any medications you are taking including prescription and over-the-counter medications, dietary supplements and complementary or Ayurvedic medicines. Consult with your doctor before stopping or starting any medications. Only take medicines that are registered by the Therapeutic Goods Administration (TGA). If you have any other concerns speak to your doctor.

During pregnancy it is also recommended that you:

- see your doctor or midwife regularly for care
- do not smoke
- avoid exposure to tobacco smoke and other harmful chemicals
- do not use illegal drugs or drink alcohol during pregnancy
- avoid unnecessary x-ray examinations, and inform the radiographer that you are (or might be) pregnant if

there are any x-ray examinations that you cannot avoid

- wash your hands often with soap and running water, especially after contact with body fluids, changing nappies and blowing noses to minimise the risk of infection from **cytomegalovirus (CMV)**
- avoid changing cat litter while pregnant to reduce the risk of **toxoplasmosis**.

#### Where to get help

- Your **GP (doctor)**
- Your **midwife**
- Your **obstetrician**
- **Paediatrician**
- **Royal Women's Hospital Medicines Information Service** Tel. **(03) 8345 3190**
- **Maternal and Child Health Line** (24 hours, 7 days) Tel. 13 22 29
- Your local council immunisation service
- **NURSE-ON-CALL** Tel. **1300 60 60 24** (24 hours, 7 days)
- **National Immunisation Hotline** Tel. **1800 671 811**
- **Victorian Clinical Genetics Services**, Royal Children's Hospital Tel. **(03) 8341 6201**
- Genetic counselling services – available at most large public maternity hospitals
- **Therapeutic Goods Administration information line** Tel. **1800 020 653**
- **Food Standards Australia New Zealand**

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