Birth defects explained

Summary

- A birth defect is an abnormality that may be detected during pregnancy, at birth or in early childhood.
- The abnormality may affect the function or structure of a body part or alter the baby’s metabolism.
- The cause is unknown in about six in every 10 cases.

A birth defect (also known as congenital anomaly) is an abnormality that may be detected during pregnancy, at birth or in early childhood. The abnormality may affect the function or structure of a body part or alter the baby’s metabolism.

In Victoria, about one baby in every 22 is affected by a birth defect. The 4,000 or so different birth defects range in severity from minor to fatal. In Victoria, most birth defects are not severe or life threatening. For example, the most common involve the urinary tract and can be treated with surgery.

Types of birth defects

There are two main types of birth defects:

- structural defects – a particular body part is absent or improperly formed. Some examples include spina bifida, hypospadias and ventricular septal defect
- metabolic defects – one of the body’s chemicals, such as an enzyme, is missing or doesn’t form correctly. Some examples include phenylketonuria (PKU), galactosaemia and Tay-Sachs disease.

Causes of birth defects

Some of the factors that may cause a birth defect include:

- genetic disorders – an altered or ‘faulty’ gene or set of genes usually means that the information contained in the particular gene is either changed or missing
- maternal illness during pregnancy – for example, some infectious diseases can cause serious harm to a pregnant woman or her unborn baby
- alcohol or drugs – drugs such as alcohol, tobacco, some illegal drugs and certain prescription and over-the-counter medications are known to cause birth defects if taken during pregnancy
- individual pregnancy factors – for example, the mother’s blood and the baby’s blood may be incompatible (Rh disease), or there could be too little amniotic fluid in the womb
- radiation exposure – from x-ray machines or other sources can damage the genes of the developing baby, particularly during the first trimester (first three months of pregnancy)
- unknown causes – the cause of birth defects is unknown in about six in every 10 cases.

Prenatal diagnosis of birth defects

Some women may be at increased risk of having a baby with birth defects because of maternal factors such as age, general health, medical history or family history.

Some of the prenatal tests used in the diagnosis of birth defects may include:

- prenatal ultrasound – a non-invasive scan that uses soundwaves to create a picture of the baby within the womb
- maternal blood tests – to check for certain substances such as hormones or particular proteins made by the baby
- amniocentesis – the doctor inserts a needle through the mother’s abdominal wall and takes a sample of
amniotic fluid for testing

- chorionic villus sampling – a sample of the placenta is taken using a small needle and tube (catheter) inserted through the mother’s cervix or through her abdominal wall
- percutaneous 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 testing.

Are you at higher risk of having a baby with a birth defect?

If you are planning a family, make an appointment with your GP (doctor) to find out if you are at increased risk of having a child with a birth defect. There are preventive measures you can take to improve your chances of having a healthy baby.

The cause of most birth defects is unknown. However, you may be at increased risk of having a baby with a birth defect if you:

- are over the age of 35
- are obese (your BMI is greater than 35 kg/m²)
- have pre-gestational diabetes (diabetes before getting pregnant)
- are from North Africa or Middle Eastern countries.

If you are at higher risk, speak with your GP about what you can do to minimise harm to your child, including pre-conception genetic screening.

When you are pregnant, it is important to see your GP regularly to check on your health and the health and development of your child.

Reduce the risk of birth defects

The risk of a birth defect for any baby is about four per cent, regardless of the circumstances during pregnancy. This means that a baby may have a birth defect even when the mother and father did everything right. Keeping this in mind, there are some things you can do to reduce the risk of birth defects.

General suggestions include:

- Ideally, make sure you are up to date with your immunisations (especially for rubella) before you get pregnant.
- Seek treatment for any sexually transmissible infection (STI) before you get pregnant.
- Take folic acid supplements prior to conception and during the first trimester as directed by your GP.
- See your GP regularly for prenatal care.
- Do not smoke.
- Avoid illegal drugs and alcohol during pregnancy.
- If you take medication to manage a chronic illness, don’t stop or alter the dose without the knowledge and consent of your doctor. If you are concerned about your long-term medication, the doctor may, in some cases, be able to prescribe a similar medication that does not have any known effects on the baby.
- Eat a healthy diet rich in vitamin B.
- Avoid unnecessary x-ray examinations.

Where to get help

- Your GP (doctor)
- Midwife
- Obstetrician
- Paediatrician
- The Royal Women’s Hospital Drug Information Centre Tel. (03) 8345 3190
- The Maternal and Child Health Line (24 hours, 7 days) Tel. 132 229
- Your local council immunisation service
- Nurse-on-Call Tel. 1300 60 60 24 – for expert health information and advice (24 hours, 7 days)

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