
Trisomy disorders

Summary

- Humans have 23 pairs of chromosomes.
 - A trisomy is a chromosomal condition characterised by an additional chromosome.
 - A person with a trisomy has 47 chromosomes instead of 46.
 - Down syndrome, Edward syndrome and Patau syndrome are the most common forms of trisomy.
-

Genes are the blueprint for our bodies. Almost every cell in the body has a copy of the blueprint, stored inside a sac called the nucleus. Genes are beaded along chromosomes, which are tightly bundled strands of the chemical substance deoxyribonucleic acid (DNA). Humans usually have 23 pairs of chromosomes, with two sex chromosomes that determine sex and 44 chromosomes that direct other factors, such as growth and function.

A chromosome condition is caused by an alteration in the number or genetic structure of chromosomes. Trisomy ('three bodies') means the affected person has three copies of one of the chromosomes instead of two. This means they have 47 chromosomes instead of 46.

Down syndrome, Edward syndrome and Patau syndrome are the most common forms of trisomy. Children affected by trisomy usually have a range of birth anomalies, including delayed development and intellectual disabilities.

Risk factors for trisomy conditions

The addition of an extra chromosome usually occurs spontaneously during conception. The cause of this is unknown and prevention is not possible. The most important risk factor for trisomy conditions is maternal age. Women in their late 30s and 40s have a higher chance of trisomy conditions occurring.

Trisomy 21 – Down syndrome

In Victoria, Down syndrome affects about one in 300 pregnancies. Down syndrome is also known as Trisomy 21, because the person has three copies of chromosome 21 instead of two.

There are three types of Down syndrome. The most common is Standard Trisomy 21, in which the father's sperm or the mother's egg cell contains the extra chromosome. In Mosaic Down syndrome, the extra chromosome spontaneously appears as the embryo develops. Translocation Down syndrome, which accounts for approximately five per cent of cases, is inheritable.

Some of the physical characteristics of Down syndrome may include:

- slight upward slant of the eyes – nearly all people with Down syndrome have a slight upward slant of the eyes. There can also be a small fold of skin on the inside of the eye (called an 'epicanthic fold') and small white patches on the edge of the iris of the eye (known as Brushfield spots)
- characteristic facial shape – the face of a person with Down syndrome is often rounded and tends to have a flat profile
- smaller stature – babies with Down syndrome are usually smaller and weigh less at birth than others. Children with Down syndrome tend to grow more slowly and are commonly smaller than other children their age. Adults with Down syndrome are commonly smaller than adults who do not have Down syndrome.

All people with Down syndrome will experience some delay in their development and some level of learning disability.

Learn more about [Down syndrome](#).

Trisomy 18 – Edward syndrome

In Victoria, Edward syndrome affects about one in 1,100 pregnancies. Edward syndrome is also known as Trisomy 18, because the person has three copies of chromosome 18 instead of two.

Some of the characteristics of Edward syndrome may include:

- physical irregularity of the kidneys, ureters, heart, lungs and diaphragm
- cleft lip or cleft palate
- small skull (microcephaly)
- malformations of the hands and feet – including missing thumbs, club feet and webbing between the fingers and toes (syndactyly)
- neural tube defect, where the spinal cord, meninges and blood vessels protrude through a gap in the vertebrae (myelomeningocele)
- malformations of the sex organs.

Survival beyond the neonatal period is uncommon for babies with Edward syndrome.

Trisomy 13 – Patau syndrome

In Victoria, Patau syndrome affects around one in 3,000 pregnancies. Patau syndrome is also known as Trisomy 13, because the person has three copies of chromosome 13 instead of two.

Some of the characteristics of Patau syndrome may include:

- small skull (microcephaly)
- an abnormal opening in the skull
- malformations of part of the brain
- structural defects of the eyes
- cleft lip or cleft palate
- additional toes or fingers (polydactyly)
- congenital heart disorders, such as ventricular septal defect
- neural tube defect, where the spinal cord, meninges and blood vessels protrude through a gap in the vertebrae (myelomeningocele)
- malformations of the sex organs.

Survival beyond the neonatal period is uncommon for babies with Patau syndrome.

Signs of trisomy conditions during pregnancy

Sometimes, signs of trisomy conditions may be evident during the pregnancy. Some of these signs may include:

- too much amniotic fluid surrounding the baby (polyhydramnios)
- only one umbilical cord artery
- a smaller than expected placenta
- the baby is small for its gestational date
- the baby is less active than expected
- congenital defects, including cleft palate or heart irregularities, are picked up during ultrasound scans.

Diagnosis of trisomy conditions

Prenatal tests that can help detect trisomy disorders include:

- **ultrasound scans** – sound waves are used to create a picture
- **maternal serum screening** – a specialised blood test
- **amniocentesis** – a sample of the amniotic fluid is taken and examined
- **chorionic villus sampling** – a sample of cells from the chorion, the tissue that will ultimately become the

placenta, is taken and examined

- **non-invasive prenatal testing (NIPT)** – a screening test that measures fetal DNA circulating in the mother's blood.

Genetic counselling and trisomy conditions

If your child has been diagnosed with a trisomy condition, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand your child's condition, what causes it, and what a diagnosis means for your child's health and development. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by trisomy conditions.

Where to get help

- Your **GP (doctor)**
- **Paediatrician**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**
- **SOFT Australia** – Support Organisation for Trisomy in Australia Tel. 0407 820 560 or **0418 479 623**
- **Genetic Support Network of Victoria** Tel **(03) 8341 6315**

This page has been produced in consultation with and approved by:

Victorian Clinical Genetics Services (VCGS)

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

Copyright © 1999/2021 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.