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## Trisomy disorders

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### Summary

- Humans have 23 pairs of chromosomes.
  - A trisomy is a chromosomal disorder characterised by an additional chromosome, so the person has 47 instead of 46.
  - Down syndrome, Edward syndrome and Patau syndrome are the most common forms of trisomy.
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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 decide gender and 44 chromosomes that dictate other factors, such as growth and function.

A chromosome disorder is caused by an alteration in the number or genetic structure of chromosomes. Trisomy ('three bodies') means the affected person has 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 defects, including delayed development and intellectual disabilities.

### Risk factors for birth defects

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 disorders is maternal age. Women in their late 30s and 40s are more likely to have babies with trisomy than younger women.

### 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 characteristics of Down syndrome may include:

- outwardly slanted eyes
- exaggerated fold of skin on the inside of the eye
- flat ears set low on the head
- flattened face
- small teeth
- relatively short arms and legs.

### 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:

- defects 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.

### **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.

### **Signs of trisomy disorders during pregnancy**

Sometimes, signs of trisomy disorders 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 abnormalities, are picked up during ultrasound scans.

### **Diagnosis of trisomy disorders**

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 villi 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 new type of screening test that measures fetal DNA circulating in the mother's blood.

### **Where to get help**

- Your doctor
- Paediatrician
- Victorian Clinical Genetics Services Tel. (03) 8341 6201
- SOFT. Australia – Support Organisation for Trisomy in Australia Tel. (02) 9521 6039

## Things to remember

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

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

Victorian Clinical Genetics Services (VCGS)

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