Trisomy disorders
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.

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.

References

- Genetics fact sheets, NSW Genetics Education Program, Royal North Shore Hospital, St Leonards, Australia. More information here.

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- talk to your doctor or pharmacist
Birth defects explained

The cause of birth defects is often unknown, speak to your GP if you are at increased risk of having a baby with a congenital anomaly...

Disease clusters

Disease clusters are rare but can cause community concern about the possible effects of exposure to environmental hazards...

Birth defect conditions

Abdominal birth defects

During fetal development, the diaphragm or abdominal wall may fail to properly fuse, allowing the abdominal organs to protrude...
**Ambiguous genitalia**

The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.

**Autism spectrum disorder (ASD)**

ASD is a complex disorder that affects a person's ability to interact with the world around them.

**Central nervous system birth defects**

Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of ten cases of neural tube defects.

**Cleft palate and cleft lip**

Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

**Congenital adrenal hyperplasia (CAH)**

CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

**Cri du chat syndrome**

Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.

**Developmental dysplasia of the hip (DDH)**

Around 95 per cent of babies born with developmental dysplasia of the hip can be successfully treated.

**Digestive tract birth defects**

Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

**Down syndrome**

With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

**Dwarfism**

Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

**Fetal alcohol spectrum disorder (FASD)**

The World Health Organization recommends that pregnant women should avoid alcohol.

**Fragile X syndrome**

The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

**Haemophilia**

All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

**Kabuki syndrome**

Kabuki syndrome affects males and females equally and there is no cure.

**Neurofibromatosis**

Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

**Noonan syndrome**

Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

**Phenylketonuria (PKU)**

PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

**Prader-Willi syndrome**

A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

**Spina bifida**

Folate can prevent up to 70 per cent of spina bifida cases if taken daily for one month before conception and during the first three months of pregnancy.

**Syringomyelia**

Syringomyelia is the growth of a cyst in the spinal cord that may result in paraplegia or quadriplegia if not treated.

**Tay-Sachs disease**

betterhealth.vic.gov.au
Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- **Thalassaemia**
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- **Trisomy disorders**
  Children affected by trisomy usually have a range of birth defects, including delayed development and intellectual disabilities.

- **Urinary system birth defects**
  Common birth defects of the urinary system include hypospadias, obstructive defects of the renal pelvis and renal agenesis.

- **Williams syndrome**
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

**Birth defect risks**

- **Drugs, medication and birth defects**
  It can be dangerous for a pregnant woman to stop taking prescription drugs if she has a medical condition or becomes ill.

- **Fetal alcohol spectrum disorder (FASD)**
  The World Health Organization recommends that pregnant women should avoid alcohol.

- **Rubella**
  Rubella is a mild illness for most people, but very dangerous for pregnant women and their babies.

- **Toxoplasmosis**
  Problems only occur if a woman becomes infected with parasites that cause toxoplasmosis for the first time while pregnant.

**Birth defects screening and protection**

- **Egg freezing**
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances.

- **Folate for pregnant women**
  Even women who aren't planning to have a baby should increase their folate intake in case of unplanned pregnancy.

- **Genetic services in Victoria**
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

- **Immunisation and pregnancy**
  Immunisation can protect a woman and her unborn baby against many infectious diseases.

- **Newborn bloodspot screening**
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions.

- **Pregnancy tests - chorionic villus sampling**
  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities.

- **Pregnancy tests - maternal serum screening**
  Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis.

- **Pregnancy tests - ultrasound**
  Ultrasound is used during pregnancy to check the baby's development and to help pick up any abnormalities.

**Related Information**

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- **Heart abnormality birth defects**
  Some congenital heart defects are mild and cause no significant disturbance to the way the heart functions...

- **Ambiguous genitalia**
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals...

**Related information on other websites**
- [Down Syndrome Victoria](#)
- [Genetic support network victoria](#)

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