Genetic disorders
A genetic disorder is caused by an altered gene or set of genes. The four broad groups of genetic disorders include single gene disorders, chromosome abnormalities, mitochondrial disorders and multifactorial disorders.

**Summary**

A genetic disorder is caused by an altered or faulty gene or set of genes. The four broad groups of genetic disorders are single gene disorders, chromosome abnormalities, mitochondrial disorders and multifactorial disorders.

**Single gene disorders**

Genes are paired – one copy of each gene pair is inherited from the mother and the other copy from the father. Around 6,000 known genetic disorders are caused by inheriting an altered gene.

Generally, the alteration (mutation) means that the information contained in the particular gene is either changed or absent. The four main ways of inheriting an altered gene are:

- **Autosomal dominant** – the alteration is present in every generation and may cause the condition in every person who has the alteration. This is because the altered copy of the gene is dominant over the healthy copy. Examples include Huntington’s disease and familial hypercholesterolaemia (genetically linked high cholesterol levels).
- **Autosomal recessive** – the affected person has two copies of the altered gene (they have inherited an altered copy of the gene from both parents). They develop the disorder because they do not have a functioning copy of the gene. Examples of autosomal recessive genetic disorders include cystic fibrosis, phenylketonuria (PKU) and sickle cell anaemia.
- **X-linked dominant** – this type of disorder generally occurs in females. The ‘X’ refers to one of the sex chromosomes that decide gender. The mother always provides an X, while the father provides either X (female child) or Y (male child). Women with an X-linked dominant disorder have one altered copy and one normal copy of a gene that is on the X chromosome. An example of an X-linked dominant genetic disorder is a rare form of rickets known as hypophosphataemic or vitamin D resistant rickets.
- **X-linked recessive** – this type of disorder is more common in males. It is caused by an alteration in a gene on the X chromosome. Since a male has one X and one Y (XY), he does not have a second ‘healthy’ copy of the gene. Examples of X-linked recessive genetic disorders include Duchenne muscular dystrophy and haemophilia.

**Chromosome abnormalities**

Genes are the body’s instructions for making different molecules (such as proteins or hormones). The estimated 23,000 genes that make up a human being are arranged along tightly bundled strands of a chemical substance called deoxyribonucleic acid, or DNA. The DNA strands are tightly packed into structures called chromosomes. Over 1,000 known disorders are caused by chromosome abnormalities.

A chromosome disorder means there is a change in either the structure or the number of chromosomes. This can happen in three main ways:

- The altered chromosome is passed from the parent to the child
- The abnormality happens when either the sperm or egg (germ cells) is created
- Soon after conception.

Chromosome abnormalities can occur in various ways, including changes in the number or structure of chromosomes, or how they are inherited.

**Changes in number of chromosomes**

Most people have 23 pairs of chromosomes, or 46 chromosomes in all. When the egg or sperm is made, the pairs split so that each egg or germ cell only contains 23 chromosomes.

Occasionally an error occurs during the division: for example, the egg or sperm might be missing a chromosome (22 chromosomes) or have an extra one (24 chromosomes), so at conception the baby has either too few (45) or too many (47) chromosomes. A well-known example of this type of genetic disorder is Down syndrome, where a person has 47 chromosomes rather than 46.

Babies are rarely born with changes in chromosome numbers because most of these pregnancies end in miscarriage.

**Changes in chromosome structure**
Sometimes the information contained in a chromosome breaks up and the pieces reform in a different pattern. For example, a fragment of chromosome may break off and be lost during the formation of either the egg or sperm cell. A section of chromosome might also break away and ‘stick’ to another chromosome.

In other cases, a fragment of chromosome may copy itself or the ends of the chromosome may join to form a ring. Some changes in structure are “balanced” (chromosome material is not lost or gained) and are unlikely to result in a genetic disorder.

**Uniparental disomy**

Uniparental disomy means the child inherited a particular gene pair (both copies of the gene) from one parent only. This can cause a disorder if it is necessary for the child to have inherited one such gene from each parent.

**Chromosomal mosaicism**

Normally every cell in the body contains the same genetic information – all 46 chromosomes, designated as 46XX (female) or 46XY (male). A person who has chromosomal mosaicism has different numbers of chromosomes in different cells; for example, 46 in some cells and 47 in others.

**Mitochondrial disorders**

Mitochondria are like little batteries that make energy within each cell. The energy source is a chemical called adenosine triphosphate (ATP). Organs like the brain, heart and liver can’t survive without ATP.

Genes within the mitochondria, as well as in the nucleus of the cell, instruct the cell on how to make the enzymes that are crucial to ATP production. If any of these genes are altered, this can affect enzyme production and interfere with the production of ATP. If one of the genes in the mitochondria is altered, then the condition is inherited only from the mother. This is because each person inherits their mitochondria only from their mother, and not from their father.

The symptoms of a mitochondrial disorder, depending on the genes involved, can affect the:
- brain and spinal cord – intellectual disabilities, deafness, vision problems and seizures
- heart – cardiomyopathy (heart failure) and irregular heartbeat disorders
- musculoskeletal (locomotor) system – poor muscle tone and flappiness.

**Multifactorial disorders**

Multifactorial (involving several factors) disorders, such as many common birth defects or diseases like high blood pressure, are disorders caused by the environment interacting with the action of several genes. (This is also sometimes called polygenic inheritance.) For example, the birth defect spina bifida is caused by the action of several genes and also depends on the amount of folic acid in the mother’s diet during pregnancy (the environment). High blood pressure is influenced by a large number of genes, but also is influenced by a person's diet and salt intake.

**Where to get help**

- Your doctor
- Victorian Clinical Genetics Services (VCGS), Royal Children's Hospital Tel. (03) 8341 6201
- Cancer Council Victoria, Information and Support Service Tel. 13 11 20

**Things to remember**

- A genetic disorder is caused by an altered gene or set of genes.
- The four broad groups of genetic disorders include single gene disorders, chromosome abnormalities, mitochondrial disorders and multifactorial disorders.

**References**

- Changes to chromosomes’ number, size and structure (Fact Sheet 6), Centre for Genetics Education, NSW, Australia. More information here.
- Mitochondrial inheritance – complex patterns of inheritance 2 (Fact Sheet 12), Centre for Genetics Education, NSW, Australia. More information here.
- Genetic conditions – overview (Fact Sheet 2), Centre for Genetics Education, NSW, Australia. More information here.

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More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab
A-Z of genetic conditions

- 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.

- Angelman syndrome
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.

- Ankylosing spondylitis
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.

- Barrett's oesophagus
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.

- Bipolar disorder
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.

- 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 10 cases of neural tube defects.

- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

- 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.

- Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

- 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.

- 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.

- Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

- Essential tremor
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

- 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.

- Friedreich's ataxia
  To the casual observer, a person with Friedreich ataxia may seem to be drunk.

- Genetic factors and cholesterol

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Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- Haemochromatosis

Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- Haemophilia

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

- Hair

Human hair grows one centimetre every month.

- Hearing problems in children

The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

- Huntington's disease

The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

- Kabuki syndrome

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

- Kennedy's disease

Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

- Kidneys - medullary cystic kidney disease

Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- Kidneys - polycystic kidney disease (PKD)

Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- Klinefelter syndrome

Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- Leukodystrophy

Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- Long QT syndrome

You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

- Marfan syndrome

Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

- McCune-Albright syndrome

The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

- Muscular dystrophy

People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

- 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.

- Osteoporosis in children

Osteoporosis in children is rare and usually caused by an underlying medical condition.

- 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.

- Premature and early menopause
The symptoms of premature or early menopause are the same as for menopause at any age...

- **Rett syndrome**
  People with Rett syndrome have a keen desire to communicate...

- **Spinal muscular atrophy (SMA)**
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age...

- **Tay-Sachs disease**
  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...

- **Tourette syndrome**
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders...

- **Treacher Collins syndrome**
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss...

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

- **Tuberous sclerosis**
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity...

- **Turner's syndrome**
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility...

- **Usher syndrome**
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing...

- **Von Willebrand disease**
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth...

- **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...

**Genes and genetics explained**

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  Children inherit physical characteristics such as eye colour from their parents through their genes...

- **Gene therapy**
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases...

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  genetic disorder is caused by an altered or faulty gene or set of genes...

- **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...

**Genetic testing**

- **DNA profiling**
  DNA profiling is a way of establishing identity and is used in a variety of ways...

- **Egg freezing**

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

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- **Genetic testing for inherited cancer**
  A predisposition to certain cancers can be inherited via altered genes...

- **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...

**Related Information**

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**Related information on other websites**

- Centre for Genetics Education - Fact Sheets
- genetic support network victoria (GSNV)
- University of Utah, Genetic Science Learning Centre

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