

Genetic disorders

Genes are the instructions for the growth and development of our bodies. 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 of each pair is inherited from the mother and the other from the father. Around 6,000 known genetic disorders are caused by inheriting an altered gene.

Generally, the alteration 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 (mutation) 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

Over 1,000 known disorders are caused by chromosome abnormalities. The estimated 23,000 genes that make up a human being are beaded along tightly bundled strands of a chemical substance called deoxyribonucleic acid, or DNA. Genes are the body's instructions for making different chemicals and each gene can make several different variations of these chemicals. The genes are, in turn, assembled on long strands called chromosomes.

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 made.

It occurs soon after conception.

Characteristics of chromosome abnormalities include:

- **Changes in number** – a person has 23 pairs of chromosomes, or 46 chromosomes in all. When the egg or sperm are made, the pairs split up 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 either missing a chromosome (22 chromosomes) or have an extra one (24 chromosomes). 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. Changes in chromosome number are common at conception but relatively rare at birth, because most pregnancies with changes in chromosome number end in miscarriage.
- **Changes in structure** – occasionally, the information contained in a chromosome breaks up and the pieces re-form 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' and unlikely to result in a genetic disorder.
- **Uniparental disomy** – this means the child inherited a particular gene pair 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.
- **Mosaicism** – normally, every cell in the body contains the same blueprint; for example, all 46 chromosomes normal, designated as 46XX or 46XY. A person who is a chromosomal mosaic 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 it.

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, depending on the genes affected, can include:

- **Brain and spinal cord problems** – intellectual disabilities, deafness, vision problems and seizures.
- **Heart** – cardiomyopathy (heart failure) and irregular heartbeat disorders.
- **Locomotor system** – poor muscle tone and floppiness.

Multifactorial disorders

Multifactorial 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, but the frequency of spina bifida also depends on the amount of folate in the diet. 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
- Genetic Health Services Victoria – Genetic Clinical Coordinator, Royal Children's Hospital Tel. (03) 8341 6201
- Genetic Health Services Victoria – Genetic Clinical Coordinator, Royal Women's Hospital Tel. (03) 8345 2000
- Genetic Health Services Victoria – Monash Medical Centre Tel. (03) 9594 2026
- The Murdoch Children's Research Institute Tel. (03) 8341 6200
- 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.

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Genetic Health Services Victoria

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