How do genetic conditions occur?

Genetic conditions occur in one of three main ways:

1. There is a problem with a chromosome.
2. There is an alteration in a gene. The alteration can be inherited from one or both parents, or can occur for no obvious reason for the first time in the family (this is known as a new mutation). There are three main patterns of inheritance – autosomal dominant, autosomal recessive and X-linked.
3. Alterations to one or more genes combine with factors in the environment – such as diet, lifestyle and chemical exposure – to cause a problem. This is known as multifactorial inheritance.

Chromosomal problems

Chromosomes are important parts of a cell. They are the parts which carry the genes.

We are meant to have 23 pairs of chromosomes, a total of 46 chromosomes, with one in each pair inherited from each parent.

Occasionally, things go wrong and a child is conceived with 45 chromosomes, or 47 chromosomes, or perhaps the right number but the wrong mix of chromosomes. Also, a child may be born with a change to the way the chromosomes are structured.

Chromosomal problems are not usually inherited, although occasionally they can be.

Autosomal dominant

In an autosomal dominant condition, an alteration in only one gene of a pair of genes is usually enough to cause the condition.

So if a person has an autosomal dominant condition, each of their children has a one in two chance of inheriting the altered gene. Each child also has a one in two chance of not inheriting the altered gene.

Autosomal dominant conditions can run in the family.

New problems with genes can come up at any time; someone with the condition may be the first affected person in the family, with no-one else in previous generations having the gene alteration. These new gene alterations, which occur at the time of conception, are called new mutations.

Autosomal recessive

In an autosomal recessive condition, alterations in both copies of a pair of genes are needed to cause the condition. Therefore, somebody with one altered gene will be a carrier (and is usually unaffected because the normal gene in that pair dominates) and somebody with two altered genes will have the condition.

A person who is a carrier for the condition has a one in two chance of any child being a carrier and a one in two chance of any child not being a carrier.

Carriers are unlikely to have a partner who is also a carrier, unless their partner is a blood relative. But if they do, then the chance of any child of two carriers having the condition is one in four.

Autosomal recessive conditions tend to occur only occasionally in the families in which they appear, but there will probably be quite a few carriers.

X-linked recessive

Most babies are born with 23 pairs of chromosomes – 22 standard pairs and then the pair that determines the sex of the baby. Girls have two X chromosomes and boys have an X and a Y chromosome.

Fathers pass their X chromosome to their daughters and their Y chromosome to their sons. Mothers pass one of their X chromosomes to both their sons and daughters.

In X-linked recessive conditions, the altered gene is found on the X chromosome. There is no equivalent gene on the Y chromosome.

So a woman with an altered gene on one of her two X chromosomes is a carrier and is usually not affected (because the normal gene dominates). But sometimes she may be mildly affected by the condition.

Any son of a woman who is a carrier has a one in two chance of having the altered gene, and a one in two chance of not having it. Any daughter of a woman who is a carrier has a one in two chance of being a carrier, and a one in two chance of not being a carrier.

A man with the condition will have daughters who are carriers and sons who are not affected.

X-linked recessive conditions can run in the family.

New problems with genes can come up at any time; a man with the condition may be the first affected person in the family, with no-one else in previous generations having the gene alteration. These new gene alterations, which occur at the time of conception, are called new mutations.

Multifactorial inheritance

Multifactorial inheritance is inheritance due to an interaction between an individual's genetic make-up and environmental factors. Although these conditions tend to run in families, the patterns are less predictable than other forms of genetic condition.

Many common health problems have multifactorial inheritance. They include some forms of cancer, some forms of heart disease, diabetes and mental illness such as schizophrenia and manic depression.

Note

Things are not quite as straightforward as this. What we have described here is the general pattern, but there are two things to consider:

- The knowledge of genetics is growing, but is incomplete.
- Genes can behave in unpredictable ways, so all advice and testing is provided with the consideration that unexpected things can happen.

Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Australasian Genetic Alliance at **http://www.australasiangeneticalliance.org.au**
- MyDr at **http://www.mydr.com.au**
- The Centre for Genetics Education at **http://www.genetics.edu.au**
- HealthInsite at **http://www.healthinsite.com**
- MedicineNet at **http://www.medicinenet.com**
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email **gtis-australia@unimelb.edu.au** or visit Biotechnology Australia's website at **http://www.biotechnology.gov.au**