Summary

- Genes are the blueprint for our bodies.
- If a gene contains a change, it disrupts the gene message.
- Changes in genes can cause a wide range of conditions.
- Sometimes a changed gene is inherited, which means it is passed on from parent to child.
- Changes in genes can also occur spontaneously.
- Parents who are related to each other are more likely have children with health problems or genetic conditions than unrelated parents (although most related parents will have healthy children).

Your chromosomes contain the blueprint for your body – your genes. Almost every cell in the human body contains a copy of this blueprint, mostly stored inside a special sac within the cell called the nucleus. Chromosomes are long strands of a chemical substance called deoxyribonucleic acid (DNA).

A DNA strand looks like a twisted ladder. The genes are like a series of letters strung along each edge. These letters are used like an instruction book. The letter sequence of each gene contains information on building specific molecules (such as proteins or hormones – both essential to the growth and maintenance of the human body).

Although every cell has two copies of each gene, each cell needs only certain genes to be switched on in order to perform its particular functions. The unnecessary genes are switched off.

Sometimes, a gene contains a change that disrupts the gene’s instructions. A change in a gene can occur spontaneously (no known cause) or it can be inherited. Changes in the coding that makes a gene function can lead to a wide range of conditions.

Chromosomes

Humans typically have 46 chromosomes in each cell of their body, made up of 22 paired chromosomes and two sex chromosomes. These chromosomes contain between 20,000 and 25,000 genes. New genes are being identified all the time.

The paired chromosomes are numbered from 1 to 22 according to size. (Chromosome number 1 is the biggest.) These non-sex chromosomes are called autosomes.

People usually have two copies of each chromosome. One copy is inherited from their mother (via the egg) and the other from their father (via the sperm). A sperm and an egg each contain one set of 23 chromosomes. When the sperm fertilises the egg, two copies of each chromosome are present (and therefore two copies of each gene), and so an embryo forms.

The chromosomes that determine the sex of the baby (X and Y chromosomes) are called sex chromosomes. Typically, the mother’s egg contributes an X chromosome, and the father’s sperm provides either an X or a Y chromosome. A person with an XX pairing of sex chromosomes is biologically female, while a person with an XY pairing is biologically male.

As well as determining sex, the sex chromosomes carry genes that control other body functions. There are many genes located on the X chromosome, but only a few on the Y chromosome. Genes that are on the X chromosome are said to be X-linked. Genes that are on the Y chromosome are said to be Y-linked.

How we inherit characteristics

Parents pass on traits or characteristics, such as eye colour and blood type, to their children through their genes. Some health conditions and diseases can be passed on genetically too.

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Sometimes, one characteristic has many different forms. For example, blood type can be A, B, AB or O. Changes (or variations) in the gene for that characteristic cause these different forms.

Each variation of a gene is called an allele (pronounced 'AL-eel'). These two copies of the gene contained in your chromosomes influence the way your cells work.

The two alleles in a gene pair are inherited, one from each parent. Alleles interact with each other in different ways. These are called inheritance patterns. Examples of inheritance patterns include:

- **autosomal dominant** – where the gene for a trait or condition is dominant, and is on a non-sex chromosome
- **autosomal recessive** – where the gene for a trait or condition is recessive, and is on a non-sex chromosome
- **X-linked dominant** – where the gene for a trait or condition is dominant, and is on the X-chromosome
- **X-linked recessive** – where the gene for a trait or condition is recessive, and is on the X-chromosome
- **Y-linked** – where the gene for a trait or condition is on the Y-chromosome
- **co-dominant** – where each allele in a gene pair carries equal weight and produces a combined physical characteristic
- **mitochondrial** – where the gene for a trait or condition is in your mitochondrial DNA, which sits in the mitochondria (powerhouse) of your cells.

**Dominant and recessive genes**

The most common interaction between alleles is a dominant/recessive relationship. An allele of a gene is said to be dominant when it effectively overrules the other (recessive) allele.

Eye colour and blood groups are both examples of dominant/recessive gene relationships.

**Eye colour**

The allele for brown eyes (B) is dominant over the allele for blue eyes (b). So, if you have one allele for brown eyes and one allele for blue eyes (Bb), your eyes will be brown. (This is also the case if you have two alleles for brown eyes, BB.) However, if both alleles are for the recessive trait (in this case, blue eyes, bb) you will inherit blue eyes.

**Blood groups**

For blood groups, the alleles are A, B and O. The A allele is dominant over the O allele. So, a person with one A allele and one O allele (AO) has blood group A. Blood group A is said to have a dominant inheritance pattern over blood group O.

If a mother has the alleles A and O (AO), her blood group will be A because the A allele is dominant. If the father has two O alleles (OO), he has the blood group O. For each child that couple has, each parent will pass on one or the other of those two alleles. This is shown in figure 1. This means that each one of their children has a 50 per cent chance of having blood group A (AO) and a 50 per cent chance of having blood group O (OO), depending on which alleles they inherit.

**Figure 1 - Father’s blood group (OO, group O)**

<table>
<thead>
<tr>
<th>Mother’s blood group</th>
<th>O</th>
<th>O</th>
</tr>
</thead>
<tbody>
<tr>
<td>AO (group A)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(AO, group A)</td>
<td>O</td>
<td>OO (group O)</td>
</tr>
<tr>
<td>O</td>
<td></td>
<td>OO (group O)</td>
</tr>
</tbody>
</table>

The combination of alleles that you have is called your genotype (e.g. AO). The observable trait that you have – in this case blood group A – is your phenotype.

**Recessive genetic conditions**
If a person has one changed (q) and one unchanged (Q) copy of a gene, and they do not have the condition associated with that gene change, they are said to be a **carrier** of that condition. The condition is said to have a **recessive** inheritance pattern – it is not expressed if there is a functioning copy of the gene present.

If two people are carriers (Qq) of the same recessive genetic condition, there is a 25 per cent (or one in four) chance that they may both pass the changed copy of the gene on to their child (qq, see figure 2.) As the child then does not have an unchanged, fully functioning copy of the gene, they will develop the condition.

There is also a 25 per cent chance that each child of the same parents may be unaffected, and a 50 per cent chance that they may be carriers of the condition.

**Figure 2 - Father (carrier)**

<table>
<thead>
<tr>
<th></th>
<th>Q</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mother (carrier)</td>
<td>Q</td>
<td>QQ (unaffected)</td>
</tr>
<tr>
<td></td>
<td>q</td>
<td>Qq (carrier)</td>
</tr>
</tbody>
</table>

Recessive genetic conditions are more likely to arise if two parents are related, although they are still quite rare. Examples of autosomal recessive genetic conditions include **cystic fibrosis** and **phenylketonuria (PKU)**.

**Co-dominant genes**

Not all genes are either dominant or recessive. Sometimes, each allele in the gene pair carries equal weight and will show up as a combined physical characteristic. For example, with blood groups, the A allele is as ‘strong’ as the B allele. The A and B alleles are said to be **co-dominant**. Someone with one copy of A and one copy of B has the blood group AB.

The inheritance pattern of children from parents with blood groups B (BO) and A (AO) is given in figure 3.

Each one of their children has a 25 per cent chance of having blood group AB (AB), A (AO), B (BO) or O (OO), depending on which alleles they inherit.

**Figure 3 - Father’s blood group - (group B)**

<table>
<thead>
<tr>
<th></th>
<th>B</th>
<th>O</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mother’s blood group</td>
<td>A</td>
<td>AB (group AB)</td>
</tr>
<tr>
<td>(group A)</td>
<td>O</td>
<td>OB (group B)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>OO (group O)</td>
</tr>
</tbody>
</table>

**Gene changes in cells**

A cell reproduces by copying its genetic information then splitting in half, forming two individual cells. Occasionally, an alteration occurs in this process, causing a genetic change.

When this happens, chemical messages sent to the cell may also change. This spontaneous genetic change can cause issues in the way the person’s body functions.

Sperm and egg cells are known as ‘germ’ cells. Every other cell in the body is called ‘somatic’ (meaning ‘relating to the body’).

If a change in a gene happens spontaneously in a person’s somatic cells, they may develop the condition related to...
that gene change, but won’t pass it on to their children. For example, skin cancer can be caused by a build-up of spontaneous changes in genes in the skin cells caused by damage from UV radiation. Other causes of spontaneous gene changes in somatic cells include exposure to chemicals and cigarette smoke. However, if the gene change occurs in a person’s germ cells, that person’s children have a chance of inheriting the altered gene.

Genetic conditions

About half of the Australian population will be affected at some point in their life by a condition that is at least partly genetic in origin. Scientists estimate that more than 10,000 conditions are caused by changes in single genes.

The three ways in which genetic conditions can arise are:

- a change in a gene occurs spontaneously in the formation of the egg or sperm, or at conception
- a changed gene is passed from parent to child that causes health issues at birth or later in life
- a changed gene is passed from parent to child that causes a ‘genetic susceptibility’ to a condition.

Having a genetic susceptibility to a condition does not mean that you will develop the condition. It means that you are at increased risk of developing it if certain environmental factors, such as diet or exposure to chemicals, trigger its onset. If these triggering conditions do not occur, you may never develop the condition.

Some types of cancer are triggered by environmental factors such as diet and lifestyle. For example, prolonged exposure to the sun is linked to melanoma. Avoiding such triggers means significantly reducing the risks.

Genes and genetics – related parents

Related parents are more likely than unrelated parents to have children with health problems or genetic conditions. This is because the two parents share one or more common ancestors and so carry some of the same genetic material. If both partners carry the same inherited gene change, their children are more likely to have a genetic condition.

Related couples are recommended to seek advice from a clinical genetics service if their family has a history of a genetic condition.

Genetic counselling and testing

If a family member has been diagnosed with a genetic condition, or if you know that a genetic condition runs in your family, it can be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand a genetic condition and what causes it, how it is inherited (if it is), and what a diagnosis means for you and your family.

Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

Genetic services in Victoria provide genetic consultation, counselling, testing and diagnostic services for children, adults, families, and prospective parents. They also provide referral to community resources, including support groups, if needed.

Where to get help

- Your GP (doctor)
- Paediatrician
- Genetic counsellor service
- Victorian Clinical Genetics Services (VCGS) Tel. 1300 118 247 or (03) 8341 6212
- Genetic Support Network Victoria Tel. (03) 8341 6315

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