Genes and genetics

Genes are the blueprint for our bodies. 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. Genes are part of chromosomes, which are long strands of a chemical substance called deoxyribonucleic acid (DNA). Therefore, genes are made up of DNA.

The genes contain the information used by our cells to control our growth, development and functioning. This genetic information comes in the form of chemical messages or codes that instruct the cells to perform their tasks. The chromosomes contain some coding DNA (genes) well as non-coding DNA. All of the DNA packaged in the cell makes up the human genome.

Sometimes, a gene contains a variation or 'spelling mistake' that disrupts the gene's coded message. A variation in a gene can occur spontaneously or it can be inherited. Variations in the coding that make a gene not work properly (faulty) are called mutations and can directly or indirectly lead to a wide range of conditions.

Chromosomes and sperm and egg cells

Humans have 46 paired chromosomes, with an estimated 20,000 or so genes that are 'beaded' along these tightly bundled strands. The 46 chromosomes in the human cell are made up of 22 paired chromosomes. These are numbered from one to 22 according to size, with chromosome number one being the biggest. These numbered chromosomes are called autosomes. Cells in the body of a woman also contain two sex chromosomes called 'X' chromosomes, in addition to the 44 autosomes. Body cells in men contain an X and a Y chromosome and 44 autosomes.

Human sperm and egg cells contain one set of 23 chromosomes each – one sex chromosome and 22 autosomes. Sperm cells can contain either an X chromosome and 22 autosomes or a Y chromosome and 22 autosomes. All egg cells contain an X chromosome and 22 autosomes. At the moment of conception, the sperm's chromosomes merge with those of the egg, creating an embryo (new organism) with a complete paired set of 46.

The gender of the developing baby is decided by the sperm. The egg always provides an X, while the sperm contributes either an X or Y chromosome. A pairing of XX sex chromosomes means the baby is a girl, while XY means the baby is a boy. There are many genes located on the X chromosome, but only a few on the Y chromosome.

Dominant and recessive genes

As chromosomes come in pairs, there are two copies of each gene in the cell.

The two copies of the genes contained in each set of chromosomes both send coded messages to influence the way the cell works. The actions of some of these genes, however, appear to be 'dominant' over others. Generally, for example, the coded message from the genes that tells the eye cells to make brown colour is dominant over blue eye colour. However, there are a number of different genes involved in determining eye colour and so blue-eyed parents can have a child with brown eyes.

Chemical communication

While every cell has two copies of the 20,000 or so genes, each cell needs only some specific genes to be switched on in order to perform its particular functions. The unnecessary genes are switched off. Genes communicate with the cell in chemical code, known as the genetic code. The cell carries out its instructions to the letter.

A cell reproduces by copying its genetic information then splitting in half, forming two individual cells. Occasionally, a mistake is made, causing a variation (genetic mutation) that means the wrong chemical message is sent to the cell. This spontaneous genetic mutation can cause problems in the way the body functions.

Genetic mutations are permanent. Some of the factors that can cause a spontaneous genetic mutation include exposure to radiation, chemicals and cigarette smoke. Genetic mutations also build up in our cells as we age.

Variations in the genes in the cells

Sperm and egg cells are known as 'germ' cells, while every other cell in the body is called 'somatic'. If a variation in the information in a gene (mutation) happens spontaneously (for no known reason) 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 mutations in genes in the skin cells caused by damage from UV radiation.

However, if the mutation occurs in the germ cells, the person's children each have a 50 per cent chance of inheriting the faulty (mutated) gene. Sometimes, a parent may have one copy of a gene that is faulty and the other copy containing the correct information. They are said to 'carry' the faulty gene and will not have the condition caused by the faulty gene themselves – they are a genetic carrier for the condition.

The correct copy of a gene overrides the faulty copy. For example, the gene controlling red-green colour recognition is located on the X chromosome. A mother who carries the faulty gene causing red-green colour blindness on one of her X chromosome copies will have perfectly normal vision, as she still has a functioning gene copy for red-green colour recognition on her other X chromosome. However, her sons have a 50 per cent chance of being colour blind.

This is because there is a 50 per cent chance that they will inherit the X chromosome from their mother that contains the faulty gene. There is also a 50 per cent chance that they will inherit the X chromosome containing the correct copy of the gene and so will have normal vision.

Genetic conditions

To date, scientists have identified around 1,700 conditions directly or indirectly caused by changes in the genes. Around half of all miscarriages are due to changes in the total number of genes in the developing baby. Similarly, about half of the Australian population will be affected at some point in their life by an illness that is at least partly genetic in origin.

Genetic conditions can happen in three ways:

- The variation in the gene that makes it faulty (a mutation) happens spontaneously in the formation of the egg or sperm, or at conception.
- The faulty gene is passed from parent to child and may directly cause a problem that affects the child at birth or later in life.
- The faulty gene is passed from parent to child and may cause a genetic susceptibility. Environmental factors, such as diet and exposure to chemicals, combine with this susceptibility to trigger the onset of the disorder.

Faulty gene inheritance

Conditions due to faulty genes can be inherited in four key ways:

- **Autosomal recessive** – the child must receive the faulty gene located on one of the numbered chromosomes (the autosomes) from both parents (who are each unaffected) in order to develop the condition. Examples include cystic fibrosis, thalassaemia and Tay-Sachs disease.

- **Autosomal dominant** – the child receives the faulty gene located on one of the numbered chromosomes (the autosomes) from one parent and the working copy from the other. For some conditions, having only one copy of the gene containing this dominant variation making it faulty and having the other copy working correctly, still means that they will be directly affected by the condition, at birth or in later life. Huntington's disease is an adult-onset neurological condition that works in this way. In other conditions, the presence of the dominant faulty gene means only that the person is at increased risk of developing (is predisposed to) the condition. They may never develop the condition unless other environmental factors are present to 'trigger' the condition. An example of this would be familial breast cancer.
- **X-linked recessive** – the faulty gene is on the X chromosome. Females are usually unaffected by the condition because they are genetic carriers of the faulty gene – they have a faulty gene copy on one X chromosome and a correct copy on their other X chromosome. The resulting condition generally affects males as they have only one X chromosome copy. Examples include muscular dystrophy and haemophilia.
- **X-linked dominant** – if the mother has the condition due to a dominant faulty gene on one of her X chromosome copies, there is a 50 per cent chance that her male and female children will be affected. Males are generally more severely affected. An example is a rare form of Charcot-Marie Tooth disease, a condition of the peripheral nervous system, where males are more severely affected.

Genetic predisposition (inherited susceptibility)

In many cases, being born with a faulty gene associated with a particular disease doesn't mean you are destined to develop it. It simply means you are at increased risk for developing the condition.

Many conditions involving genetic susceptibility, such as some types of cancer, need to be triggered by environmental factors such as diet and lifestyle. For example, prolonged exposure to the sun is linked to melanoma. Avoiding the triggers means significantly reducing the risks.

Where to get help

- Your doctor
- Paediatrician
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Genetic counselling services – available at most large public maternity hospitals

Things to remember

- Genes are the blueprint for our bodies.
- A genetic mutation means that a gene contains a change or 'spelling mistake' that disrupts the gene message (makes the gene faulty).
- Genetic mutations can occur spontaneously.
- Sometimes, a faulty gene is inherited, which means it is passed on from parent to child.
- Genetic changes that make a gene faulty can cause a wide range of conditions.

This page has been produced in consultation with, and approved by:

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