Genes and genetics explained

Summary

- Genes are the blueprint for our bodies.
- A genetic mutation means that a gene contains a change – like a 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.
- Although most related parents will have healthy children, they are more likely than unrelated parents to have children with health problems or genetic disorders.

Parents pass on traits or distinguishing characteristics such as eye and hair colour to their children through their genes. Many health conditions and diseases are also genetic. Genes may also influence some behavioural characteristics, such as intelligence and natural talents.

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.

A DNA strand looks like a twisted ladder. The genes are like a series of letters strung along each rung. 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).

The genes are copied ‘letter for letter’ to a similar substance called ribonucleic acid (RNA). The working parts of the cell read the RNA to create the protein or hormone according to the instructions. Each gene codes the instruction for a single protein only, but one protein may have many different roles in the human body. Also, one characteristic, such as eye colour, may be influenced by many genes.

Sometimes, a gene contains a variation – like a spelling mistake – that disrupts the gene’s coded message. A variation can occur spontaneously (no known cause) 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 about 23,000 genes. The 46 chromosomes in the human cell are made up of 22 paired chromosomes. These are numbered from 1 to 22 according to size, with chromosome number 1 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.

The 23,000 genes come in pairs. One gene in each pair is inherited from the person’s mother and the other from their father. A sperm and an egg each contain one copy of every gene needed to make up a person (one set of 23 chromosomes each). When the sperm fertilises the egg, two copies of each gene are present (46 chromosomes), and so a new life can begin.

The chromosomes that decide the sex of the baby are called sex chromosomes. The mother’s egg always contributes an X, while the father’s sperm provides either an X or a Y. An XX pairing means a girl, while an XY

pairing means a boy. As well as determining sex, these 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.

How we inherit characteristics

We can inherit characteristics in many different ways. One characteristic can have many different forms – for example, blood type can be A, B, AB or O. Variations in the gene for that characteristic cause these different forms. Each variation of a gene is called an allele (pronounced 'AL-eel'). We can inherit different alleles of the gene pair (one from each parent) in different ways.

Dominant and recessive genes

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, a number of different genes together determine eye colour and so blue-eyed parents can have a child with brown eyes.

Dominant and recessive blood-group inheritance

Dominant inheritance is when one allele of a gene is dominant within the pair. For blood groups, the A allele is dominant over the O allele, so a person with one A allele and one O allele has the blood group AO. Another way of saying this is that the O group is recessive – a person needs two O alleles to have the blood group O.

So a child may have blood group A because the blood group A gene inherited from their mother is dominant over the blood group O gene inherited from their father.

If the mother has an A allele and an O allele (AO), her blood group will be A because the A is dominant. The father has two O alleles (OO), so he has the blood group O. 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.

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. So someone with one copy of A and one copy of B has the blood group AB.

Genotype and phenotype

'Genotype' and 'phenotype' are terms commonly used in genetics. Continuing the example of blood groups, a person with the alleles AO will have the blood group A. The observable trait – blood group – is known as the phenotype.

The genotype is the genes that produce the observable trait. So the person with blood group A and AO alleles has the blood group A phenotype but the AO genotype.

Chemical communication

Although every cell has two copies of the 23,000 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) and the wrong chemical message is sent to the cell. This spontaneous genetic mutation can cause problems in the way the person's body functions.

Genetic mutations are permanent. Some of the causes of 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 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 a person's germ cells, that 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 although they themselves will not have the condition caused by the faulty gene – 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 colourblind. 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 caused directly or indirectly by changes in the genes. Around half of all miscarriages are caused by 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.

The three ways in which genetic conditions can happen are:

- 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.

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 of 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 such triggers means significantly reducing the risks.

Genes and genetics – related parents

Many cultures approve of marriage between relatives such as first cousins. The aims of intermarrying are often to bolster family unity and keep wealth within the family. A relationship between related people is called consanguinity – meaning ‘shared blood’ in Latin.

Consanguinity is often associated with factors such as:

- cultural and religious practices
- isolated groups (such as migrants) who prefer to marry within their own culture
- low socioeconomic status
- illiteracy
- living in rural areas.

Related parents are more likely than unrelated parents to have children with health problems or genetic disorders. 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 altered (mutated) gene, their children are more likely to have a genetic disorder.

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

Autosomal recessive genetic disorders

If two parents have a copy of the same altered gene, they may both pass their copy of this altered gene on to a child, so the child receives both altered copies. As the child then does not have a normal, functioning copy of the gene, the child will develop the disorder. This is called autosomal recessive inheritance. The parents are ‘carriers’ of the genetic condition but are unaffected themselves. Autosomal recessive genetic disorders are more likely if two parents are related, although they are still quite rare.

Examples of autosomal recessive genetic disorders include cystic fibrosis and phenylketonuria (PKU). When both parents are carriers of the same altered gene, there is a one in four (25 per cent) chance that each pregnancy will be affected. Other children of the same parents may also be affected or may be carriers, having only one copy of the altered gene.

A child with only one copy of the altered gene will not be affected, as that child also has a normal copy of that gene – the same as the healthy parents.

Degrees of relationship

Relatives are described by the closeness of their blood relationship. For example:

- First-degree relatives share half their genetic information. First-degree relatives include a person’s siblings, non-identical twin, parents and children.
- Second-degree relatives share one-quarter of their genetic information. Second-degree relatives include a person’s half-siblings, uncles and aunts, nephews and nieces, and grandparents.
- Third-degree relatives share one-eighth of their genetic material and include a person’s first cousins, half-uncles, half-aunts, half-nephews and half-nieces.
- The closer the genetic relationship between the parents, the greater the risk of birth defects for their children.

Incidence of birth defects in children of related parents

A child of unrelated parents has a risk of around two to three per cent of being born with a serious birth defect or genetic disorder. This risk is approximately doubled (to between four and six per cent) for children of first cousins without a family history of genetic disorders. The risk of birth defects or death for children of first-degree relatives – for example, parent and child or brother and sister – rises to about 30 per cent.

Australian law and consanguinity

Australian law allows marriage between cousins, an uncle and niece, or an aunt and nephew. Unions that are illegal in Australia include those between closely related family members such as siblings, half-siblings, or grandparents and grandchildren. This law also includes adoptees, despite the fact they don't have blood relations within their adopted families.

Genetic counselling and testing

Genetic services in Victoria provide information and counselling for couples considering prenatal diagnosis or following diagnosis of fetal abnormalities, and referral to community resources including support groups if needed.

A couple who suspect they may be related can seek genetic counselling. If the family has a history of a known autosomal recessive genetic disorder, genetic testing may be possible to see whether the couple are both carriers of the condition.

Where to get help

- Your doctor
- Paediatrician
- Genetic counselling services – available at most large public maternity hospitals
- Victorian Clinical Genetics Services (VCGS) Tel. (03) 8341 6201
- The Murdoch Children's Research Institute Tel. (03) 8341 6200

Things to remember

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