

Some Genetic Terms You May Hear or Read

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ACGT: These four letters stand for what are known as chemical 'bases': Adenine (A), Cytosine (C), Guanine (G), and Thymine (T) that make up our DNA. These bases form pairs that create each 'rung of the ladder of DNA. On these rungs, Adenine always pairs with Thymine and Cytosine with Guanine.

Base pair: The binding of these pairs of chemical bases or 'base pairs' form the structure of DNA (see illustration). Bases Adenine (A) and Thymine (T) pair together, and Cytosine (C) and Guanine (G) pair together.

Cell: this is the smallest unit of life. Genetic diagnostic tests examine cells from the baby that contain chromosomes.

Chromosome: humans have 23 pairs of chromosomes—22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, X and Y (XY for males and XX for females). When they have a baby, each parent contributes one chromosome to each pair so a baby gets half of their chromosomes from their mother and half from their father.

Clinical geneticist: doctor who specialises in genetic conditions and their diagnosis and management.

Copy number variant (CNV): is used to describe a variation in the number of copies of a gene or genes between different people. Sometimes this can involve extra copies or missing copies. We all have CNVs but research has shown that some may cause health conditions.

De novo: if a genetic change is described as 'de novo' it means it has appeared in a pregnancy for the first time and is not a change that has been inherited from the parents.

Deletion: refers to a loss of genetic material. it can be a small loss, perhaps just one base pair is missing, or a much larger loss involving a part of a chromosome. Small losses (too small to be seen under a normal microscope) are sometimes known as **microdeletions**.

Duplication: refers to extra genetic material. It can be a small gain or occasionally can involve whole chromosomes. Small gains (too small to be seen under a conventional microscope are sometimes known as **microduplications**).

DNA (deoxyribonucleic acid): This is what genes are made of. It is a long molecule, made up of a sequence of chemical bases (AGCT), that contains all the instructions for life. DNA is passed on to children by their parents.

Exome: this is the part of the **genome** that makes proteins and proteins have a major part to play in a baby's development. We know that most of the genetic changes that cause health conditions or disability are in the exome. It makes up about 2% of the **genome**.

Gene: genes are specific sections of **DNA** made up of a number of base pairs (the number can range from a few hundred to over two million). It is thought that humans have up to 25,000 genes in total. Genes are arranged on **chromosomes** and carry the instructions that determine our physical characteristics, growth and development.

Genetics: the study of genes, genetic variations and how genes pass through families.

Genome: our genome is our complete set of **DNA** that contains the genetic instructions needed to for us to develop and live. It can be helpful to think of the genome as a huge manual or book of instructions, each **chromosome** is

a chapter in the book, each gene is a paragraph and each sentence is made of a series of letters A, C, G, and T. All the letters have to be present and in a particular order. If there are extra or missing chapters, paragraphs or letters, or if some are in the wrong order the instructions won't work and this can affect how we develop. There are more than three billion letters in the human genome.

Genetic counsellor: professionals trained to work with individuals and families who have or carry genetic conditions or are interested in genetic tests for themselves, their children or in pregnancy. They aim to help families understand genetic conditions and genetic tests and support them in decisions they might face.

Genomics: the study of the whole of a living creature's **DNA** or genetic code, i.e. the **genome**.

Molecule: a tiny particle made of atoms bonded together by chemicals. For instance, a water molecule is made of two hydrogen atoms and one oxygen atom. You will sometimes hear **molecular testing** used to describe techniques that look at biological markers in an individual's genes to try to diagnose genetic changes.

Mutation: a change in the DNA sequence that may occur in a pregnancy or because of environmental factors.

Sequencing: refers to testing technologies that can 'read' the sequences of **DNA base pairs**.

Single gene disorder (also called monogenic disorder): these are genetic conditions that are caused by a change in one gene. There are many different single gene disorders, two examples are cystic fibrosis and sickle cell disease. They can be inherited or passed down in families or can occur for the first time in a particular pregnancy (**de novo**).

Translocation: when part of one **chromosome** is broken off and attached to another. If this does not involve a loss or gain in genetic material it is called a **balanced translocation**. If there is missing or extra material it is known as an **unbalanced translocation**.

Variant: refers to an alteration in a **DNA** sequence. All humans have variants in their DNA which make each of us unique.

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info@arc-uk.org to arrange an evening call.

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