

Genetics 101 - Glossary of Terms

GENE: Unit of DNA that codes for proteins – instructions to make molecules called proteins. They are passed down to us from our parents.

PROTEIN: A large molecule made of amino acids – proteins do most of the work in cells and are required for the cell functioning correctly. In this way they regulate most of the organs and tissues in the body. Examples of proteins include antibodies and enzymes.

DNA: Short for deoxyribonucleic acid - is the material our genetic code is built from. In humans it is almost exclusively stored in the nucleus of the cell and consists of four chemical bases repeated over and over 3 billion times and packaged very tightly in 46 chromosomes.

CHROMOSOMES: Large units of genetic material containing both genes and non-coding genetic material. Humans usually have 46 chromosomes. 22 pairs that are the same whether you are male or female and two sex chromosomes. Women usually have two X chromosomes and men usually have one X and one Y chromosome.

- ❖ *Book Analogy:* If we imagine that *DNA* is the alphabet that writes the instructions for our body and *proteins* are the workers being instructed, then *genes* are like individual books with instructions and *Chromosomes* are the book shelves that contain many of the instruction books. Humans have 46 book shelves and about 20,000 books.

GENE TESTING: Reading through the DNA of that gene to see if there are any errors – basically, the DNA is read and checked for “spelling mistakes”.

PANEL TESTING: A test designed to test several genes at the same time. These tests can help your medical team determine which gene has a mutation that causes your condition in a more time effective way.

SINGLE SITE TESTING: When a mutation is known in a family, members of the same family can have this faster test that looks only at the area of the gene where the familial mutation has been described.

- ❖ *Book Analogy:* A panel test looks at several books at the same time checking for spelling mistakes, a single gene test looks only at one book and a single site test looks only at the page that contained the spelling mistake in a family member.

MUTATION: A change in the DNA sequence – a “spelling mistake” in the sequence. This can cause the protein to be unchanged, it can cause the protein to be changed, or it can cause the protein to be shortened. A changed or shortened protein is less likely to be able to function correctly.

PATHOGENIC MUTATION: A change in the DNA sequence that causes a condition. The protein is changed or shortened rendering it unable to function correctly.

NON-PATHOGENIC MUTATION: A change in the DNA sequence that does not cause a condition. Usually this means that the protein is not changed or not changed significantly enough to be impaired in its functioning.

VUS or “Variant of Unknown Significance”: A change in the DNA sequence that may or may not cause a condition. These mutations are further explored and may be reclassified as pathogenic or non-pathogenic later in time.

TRUNCATION MUTATION: A truncating mutation means that the change in the sequence makes the protein end prematurely.

- ❖ *Book Analogy:* If the original sentence is **The bad cat saw the big dog and ran and bit it**, a *mutation* would be a change to any part of this sentence.

A *non-pathogenic mutation* would be for instance **The bad cat saw one big dog and ran and bit it** - while the sentence has changed slightly, the meaning did not – the protein is unchanged.

A *pathogenic mutation* would be **The bad cat saw the big kid and ran and bit it** – the sentence now has changed and the meaning is different. The protein would not be able to do the job it was supposed to do.

A *VUS* would be **The bad cat saw the big dog and ran and bit her** – this sentence could be correct if the dog is female, but we don't know that yet. More research is needed to find that out. If the dog is indeed female, then the sentence is still fine and the mutation would be downgraded to non-pathogenic. If the dog is male, then the sentence is now wrong and it might be upgraded to pathogenic.

If the mutation added a full stop and the sentence was now **The bad cat saw**, the sentence is now extremely shortened and the meaning is completely lost – this is a *truncating mutation*.