

THE BASICS OF GENETICS



WHAT IS GENETICS?

Genetics is the study of genes and inheritance. Genetics influences how we look and why certain traits and diseases run in families. The study of genetics allows us to understand how our genes are involved in the development of disease. To understand genetics, you need to know a bit about DNA, genes and chromosomes.

DNA (deoxyribonucleic acid) is a long molecule with a twisting ladder-like structure, called a double helix, that carries the instructions for making the human body. DNA is made of a chain of molecules called bases. There are four types of bases represented by the letters A, T, C and G and the order in which they occur is called the DNA sequence.

Genes are sections of DNA with a unique code (specific base sequence) and most genes contain the instructions to produce proteins in our bodies which carry out specific functions in the cell.



Genes are packaged inside **chromosomes**, which are found inside the nucleus of our cells.



HUMAN CELLS CONTAIN 46 CHROMOSOMES CONSISTING OF TWO SETS OF 23

We inherit one copy of all genes from each parent, **22 chromosomes** called autosomes that are the same in males and females and a **single sex chromosome, X or Y**. Following fertilization, we end up with **46 chromosomes** with two copies of each gene found on autosomes and two sex chromosomes. Females are XX and males are XY.



Sex chromosome X inherited from his mother



Sex chromosome X inherited from her mother

Sex chromosome Y inherited from his father



Sex chromosome X inherited from her father



Inheritance is the passing down of genetic material (DNA) from parent to child and is responsible for some of the traits that make us who we are, like hair color or eye color.

HUMAN GENOME

All the genetic material within our cells makes up the **human genome**. Each chromosome contains many genes and the human genome is estimated to contain **20,000 to 25,000 genes**.



GENETIC VARIATION

Overall, our DNA sequences are very similar. However, there are small differences between the DNA sequences of individual genomes and this is called **genetic variation**. These variations make us **unique**.



Genetic variations can be caused by differences in a single base (**single nucleotide polymorphisms**), a common cause of natural variations in the DNA sequence

OR

by **mutations** which alter the sequence of larger sections of DNA. Mutations within genes are also known as **variants**. Variants can lead to changes in genes that affect the production or function of proteins. In turn this can have a negative impact on your body and cause disease.



Basic Concepts In Medical Genetics

Gregory M. Pastores



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