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**AN INTRODUCTION TO DNA, GENES AND CHROMOSOMES**

DNA contains the instructions for growth and development in humans, and all living things. Our DNA is packaged into chromosomes that contain all of our genes.

In humans genetic information, also known as our genome, can be described as the ‘Book of Life’. This book can be thought of as being made up of two volumes, each volume of the book is given to a person by one of their parents.

Reading a person’s genetic book of life:

* One volume of the book is inherited from the mother and the other from the father.
* Both volumes contain 23 chapters each, equivalent to the 23 pairs of chromosomes in human body cells that contain genetic information.
* The 23 chapters (chromosomes) are made up of a number of recipe pages (DNA or genes).
* Some of the chapters contain many pages while others only have a few. Some chromosomes are large and contain many thousands of genes and non-coding DNA while others are much smaller.
* Genes are sections of DNA that code for the proteins our body needs to function.
* In-between (non-coding) sections of DNA have various jobs, not all of which we understand.
* Careful examination of the words within genes shows that all the words are made up of three letters such as AGT, GGT, ACT, CAA etc.
* There are four letters used in the genetic book. They are A, T, C & G.

Just as reading the words on the page of a book allows an understanding of the author’s message, the body is able to read the three letters in genes to make the protein needed for our cells to work.

Our cells don’t need all the instructions all the time. Pages of our genetic book can be closed and then reopened when needed. Each type of cell can have different parts of the genetic book opened or shut because different cells do different jobs in our body. Which genes are turned on or off can be influenced by our diet, chemical exposure, exercise, aging and messages from other genes in the body.

**DNA, GENES AND CHROMOSOMES IN THE BODY**

Our bodies are made up of millions of cells. Each cell contains a complete copy of a person's genetic book of life.

Chromosomes can be thought of as being made up of strings of genes. The chromosomes, including the genes, are made up of a chemical substance called DNA. The chromosomes are very long strands of DNA, coiled up like a ball of string.

Chromosomes are found in the nucleus of all body cells except for red blood cells which have no nucleus and therefore do not contain chromosomes.

**Chromosomes**

There are 46 chromosomes contained in the **nucleus** of body cells**.** Of these, 23 came from the mother's egg and 23 came from the father's sperm. At conception, the baby’s body cells now have 46 chromosomes, made up of 23 pairs, just like the parents. As we age and grow, our cells are continually dividing to form new cells. During this division process, each of the long thin chromosomes coils up tightly, so that each of the 46 individual chromosomes in the nucleus become rod-shaped structures and can be seen when using a microscope.

The DNA making up each chromosome is usually coiled up tightly. If we imagine it stretched out, it would look like beads on a string:

* Each of these beads is called a ***gene.***
* Each gene is an instruction for a specific protein.
* Thousands of genes make up each chromosome.
* Between the genes are segments of non-coding DNA.

Since the chromosomes come in pairs, there are also two copies of each of the genes. The exception to this rule applies to the genes carried on the sex chromosomes, X and Y. Since men have only one copy of the X chromosome, they have only one copy of all the genes carried on the X chromosome. Women have two copies of the X chromosome in their cells and so they have two copies of all the genes carried on the X chromosome. There are very few genes on the Y chromosome and their role is mainly to make a person male, so they are not needed in female cells.

**DNA**

There are over 20,000 genes found in the DNA of each person. Each gene has its own specific location on a chromosome and the genes (coding DNA) plus the non-coding DNA make up that person’s **genome**.

* The DNA code is made up of very long chains of four basic building blocks (*nucleotide bases*) called **A**denine (A), **G**uanine (G), **T**hymine (T) and **C**ytosine (C).
* A chromosome consists of two of these DNA chains running in opposite directions. The bases pair up to form the rungs of a ladder twisted to form a double helix.
* Pairing of the bases follows a pattern where base A can only pair with base T and base G can only pair with base C. Roughly three billion of these base pairs of DNA make up the human genome.
* Our DNA code is made up of a combination of three of these four chemical ‘letters’.
* Each three-letter word tells the cell to produce a particular amino acid, the building blocks of proteins.
* The sequence of three-letter words in the gene enables the cells to assemble the amino acids in the correct order to make up a protein.
* Only about 2% of the entire DNA in the human cell is made up of genes that contain the information codes for making proteins.
* The remaining 98% of DNA does not contain the information for proteins and used to be called **junk** DNA. This non-coding DNA separates genes from each other along the chromosomes and there is increasing evidence that it has a role in turning genes **on** and **off**. This non-coding DNA therefore has a control function within the genome.

**DNA VARIATIONS**

We all have small variations in our genetic code. That is why we are all unique. Even identical twins have some variations in their DNA by the time they are born. Because we inherit our genes from our parents, members of the same family share their DNA including its variations.

There may be changes in the sequence of letters in the gene message; nucleotide base/s (A, G, T or C) can be missing (called a deletion) or base/s can be added (called an insertion) and these can be of one or many DNA bases. Variations in the code can occur during our life for a variety of reasons including exposure to radiation, certain chemicals or by chance. Aging is a common cause of genetic variation. Throughout our lives, our cells are continually being replaced. Some variations in the genetic information do not seem to make any difference to the function of our cells. These types of DNA variations are quite common. Other DNA variations can be associated with an increased chance of a health condition, for example diabetes or cancer. Some DNA variations can mean the gene instruction is incorrect. This can cause a **mutation.** A DNA mutation can cause a problem for one cell type but not another, since not all cells use all of the possible proteins.

When a DNA change causes a faulty protein in cells that need that protein, it usually results in disease symptoms that can sometimes be recognized as a genetic disorder. Since we have two copies of each gene, if one copy has a mutation and the other copy is working, then we may not develop any problems. We are all born with DNA mutations and sometimes these can be beneficial or cause no problem.

When a gene variation is present in egg or sperm cells, it can be passed on to children (inherited). In other cases, a new gene variation can arise in an egg or sperm cell. The person arising from that egg or sperm cell will be the first in the family to have the DNA change which may then be passed down to his or her children and future generations.