Fact sheet **01**

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 SUMMARY

- DNA stands for (**D**eoxyribo**N**ucleic **A**cid), which is made up of very long chains of chemical 'letters': Adenine (A), Guanine (G), Thymine (T) and Cytosine (C)
- DNA contains the instructions for our genes
- Genes are the instructions for making proteins. Proteins do the work within our cells and body
- In humans, most genes are arranged on chromosomes that are found in the nucleus of cells.

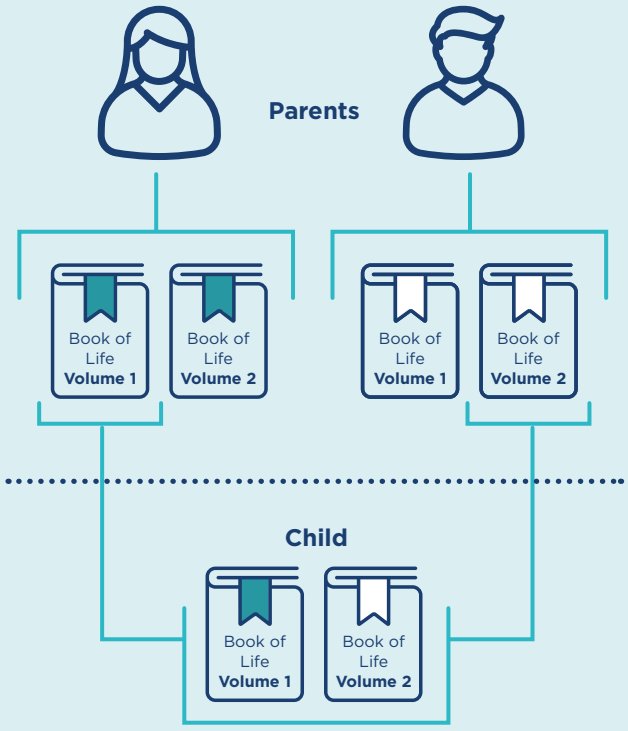
THE GENETIC RECIPE BOOK

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, and each volume of the book is given to a person by one of their parents (*Figure 1.1*).

Reading a person's genetic 'Book of Life' (*Figure 1.2*):

- One volume of the book is inherited from the mother and the other from the father
- Both volumes contain 23 chapters each, just as there are 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 (coding DNA or genes) and in-between (non-coding) pages of DNA
- 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 that our body needs to work properly
- In-between (non-coding) sections of DNA have various jobs. We understand only some of these
- A closer look at the words within genes shows that all the words are made up of three letters (triplets) such as AGT, GGT, ACT, CAA etc.
- There are four letters used in the genetic book. They are A, T, C & G.

Figure 1.1:
The human genome, sometimes called the 'Book of Life'

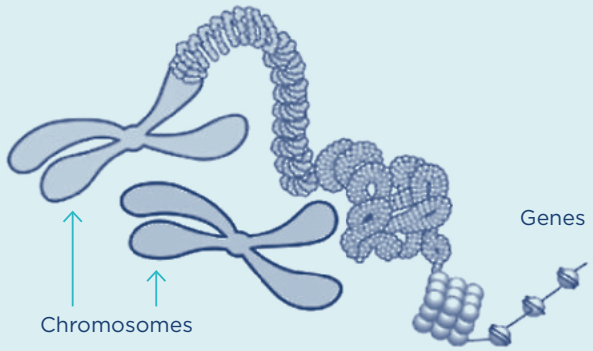


Content updated July 2020

Figure 1.2:
Genetic terminology and the 'Book of Life'

The Book of Life	Genetic terminology
Volume 1 & 2	Genome
Chapters	Chromosomes
Pages	Genes
Words	Triplets
Letters	A, T, C, G
Spelling change	Genetic variant

Figure 1.3:
Chromosomes are like strings of beads (genes)



Just as reading the words on the page of a book helps us understand the author's message, the body is able to read the triplets in genes to make the protein needed for our cells to work.

Our cells do not 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 sometimes be influenced by our diet, chemical exposure, exercise, ageing and messages from other genes in the body.

CHROMOSOMES, GENES AND DNA IN THE BODY

Our bodies are made up of billions of cells. Each cell contains a complete copy of a person's genetic 'Book of Life'.

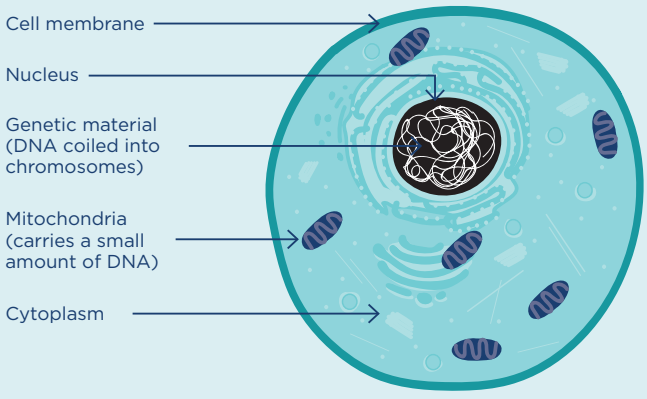
Chromosomes can be thought of as being made up of strings of genes (DNA that codes for proteins) with non-coding DNA between them. The chromosomes, including the genes, are made up of a chemical substance called DNA (**D**eoxyribo**N**ucleic **A**cid).

The chromosomes are very long strands of DNA, coiled up like a ball of string as shown in *Figure 1.3*.

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

Another place in the cell where DNA is found is in very small compartments called **mitochondria** (the energy centres of the cell) that are found scattered outside the nucleus (*Figure 1.4*). The DNA in mitochondria is much smaller and has mostly coding DNA.

Figure 1.4:
Diagram of a human cell

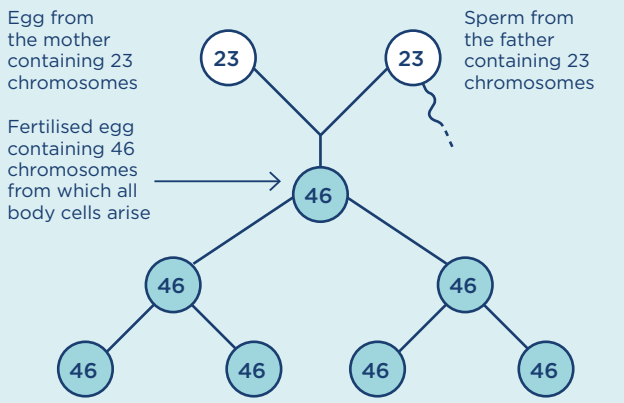


Chromosomes

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
- When the egg and the sperm join together at the time of conception, the first cell of the baby is formed. This cell is copied to make all of the cells of the baby
- The baby's body cells now have 46 chromosomes, made up of 23 pairs, just like the parents (*Figure 1.5*).

Figure 1.5:
At conception the sperm and egg combine



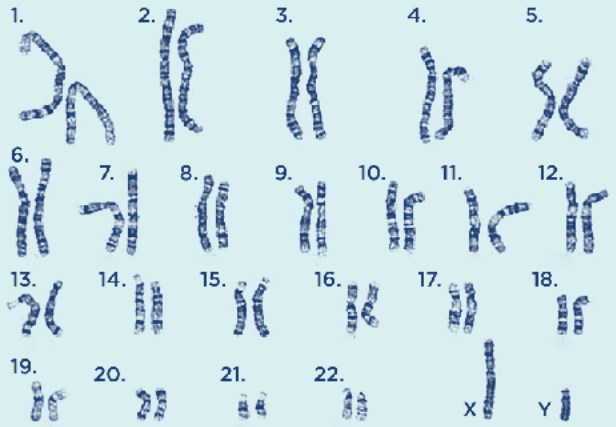
As we age and grow, our cells are continually dividing to make new cells.

During this division process, each of the long chromosomes coils up tightly, so that each of the 46 chromosomes in the nucleus become rod-shaped structures and can be seen when using a microscope.

In a genetic testing laboratory the chromosomes may be coloured (stained) with special dyes to produce distinctive banding patterns and lined up in size order. This produces what we call a **karyotype**. These patterns allow the laboratory to check the size and structure of each chromosome.

Figure 1.6 shows a banded chromosome karyotype where each chromosome has been numbered from chromosome number 1-22 and arranged in pairs, roughly in order of size. These numbered chromosomes are called **autosomes**.

Figure 1.6:
Chromosome picture (karyotype) from a male (46,XY).



There are two chromosomes that have been given the labels X and Y. These are the **sex chromosomes**. It is these sex chromosomes that determine whether the chromosomes have come from a male or a female.

In females, typically cells in the body have 46 chromosomes (44 autosomes plus two copies of the X chromosome). They are said to have a **46,XX karyotype**. Eggs (female reproductive cells) are different as they only contain half of the chromosomes (23, made up of 22 numbered chromosomes and an X chromosome).

In males, typically cells in the body have 46 chromosomes (44 autosomes plus an X and a Y chromosome). They are said to have a **46,XY karyotype**. Sperm (male reproductive cells) are different as they only contain half of the chromosomes (23, made up of 22 numbered chromosomes and an X chromosome or a Y chromosome).

Genes

The DNA making up each chromosome is usually coiled up tightly. If we imagine it stretched out, it might look like beads on a string (*Figure 1.3*):

- 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 sections 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 males typically have only one copy of the X chromosome, they have only one copy of all the genes carried on the X chromosome. Females typically 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.

To adjust for the fact that females have two X chromosomes with lots of genes while males have only one, one of the female's X chromosomes is **switched off** or inactivated in each of their cells.

There are very few genes on the Y chromosome and their role is mainly to make a person have male features, so they are not needed in female cells.

Figure 1.7:
The DNA bases pair up to make genes

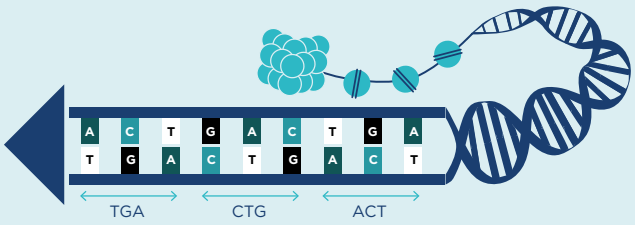
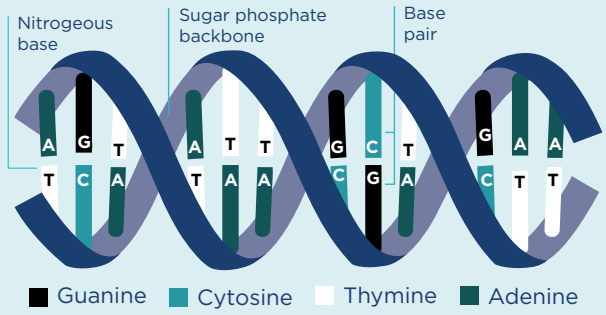


Figure 1.8:
The DNA helix



DNA

There are over 20,000 genes found in the DNA of each person. Each gene has its own specific location on a chromosome or on the mitochondrial DNA 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 that twists to form a double helix (*Figures 1.7 & 1.8*)
- 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' called a triplet.
  - Each three-letter word (**triplet**) tells the cell to produce a particular amino acid, the building blocks of proteins
  - The sequence of three-letter words in the gene allows cells to place the amino acids in the correct order to make up a protein
  - Only about 1-2% of the entire DNA in the human cell is made up of genes that contain the information codes for making proteins

- The remaining 98-99% of DNA does not contain the information for proteins and used to be called **junk** DNA. Non-coding DNA sometimes separates genes from each other along the chromosomes and there is increasing evidence that it has a role in turning genes on and off. Non-coding DNA may therefore control how the genome works.

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 variations 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 include one or many DNA base pairs.

Variations in the DNA code can occur during our life for a variety of reasons including exposure to radiation, certain chemicals, by chance or other unknown factors. Ageing is one of the most common causes of genetic variation. Throughout our life, our cells are continually being replaced.

The genes in our DNA provide the instructions for proteins, which are the building blocks of the cells that make up our body. Although we all have variation in our genes, sometimes this can affect how our bodies grow and develop. Generally, DNA variations that have no impact on our health are called benign variants or polymorphisms. These variants tend to be more common in people. Less commonly, variations can change the gene so that it sends a different message. These changes may mean that the gene does not work properly or works in a different way that is harmful. A variation in a gene that causes a health or developmental condition is called a pathogenic variant or **mutation**.

A pathogenic variant can cause a problem for one cell type but not another, since not all cells use all of the possible proteins.

Since we have two copies of each gene, if one copy has a variant that makes it not work properly and the other copy is working, then we may not develop any problems with health or development.

We are all born with gene variations 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).

Sometimes, a new gene variant can arise in an egg or sperm cell. This is called a **de novo** change.

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.

Genes contain recipes for the body to make proteins - the 'Book of Life' is like a recipe book for our bodies