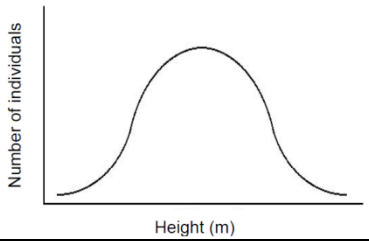
AS90948 Science - Demonstrate understanding of biological ideas relating to genetic variation

This achievement standard involves demonstrating understanding of biological ideas relating to genetic variation.

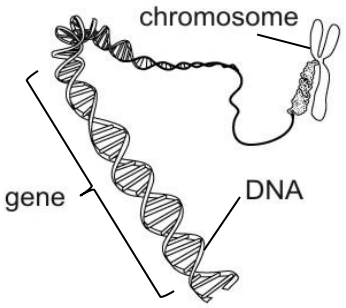
Achievement	Achievement with merit	Achievement with excellence
Demonstrate understanding of biological ideas relating to genetic variation.	Demonstrate in-depth understanding of biological ideas relating to genetic variation.	Demonstrate comprehensive understanding of biological ideas relating to genetic variation.

Part 1 – DNA Structure & Cell Division

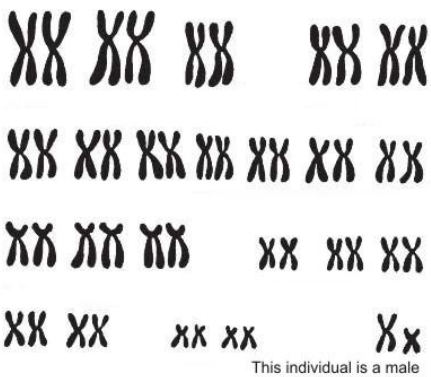
Variation Variation means differences between individuals. Variation arises from inheritance, environmental factors and mutations. There are two types of variation:

Continuous	Discontinuous variation
There is a smooth range of variation between individuals, with all intermediates possible between the extremes. It makes a bell shaped curve when graphed. Examples - skin colour, intelligence, height, and mass. It involves complicated genetics involving many genes and the environmental also has a significant effect. Examples - exposure to sun, nutrition, exercise.	Variation is of two types (either/or) or more than two types with no intermediates and makes a histogram when graphed. Caused by simple genetics with very little or no environmental effect. Examples - blood group (O, A, B, AB), sex (m/f), the ability to roll tongue* (can/can't).
	 *There is evidence that the ability to roll a tongue or not may be much more complicated than the simple genetics we see in many text books & examination questions. It is possible to have identical twins where one can roll & the other not; Non-rolling parents can have a child who can tongue roll.

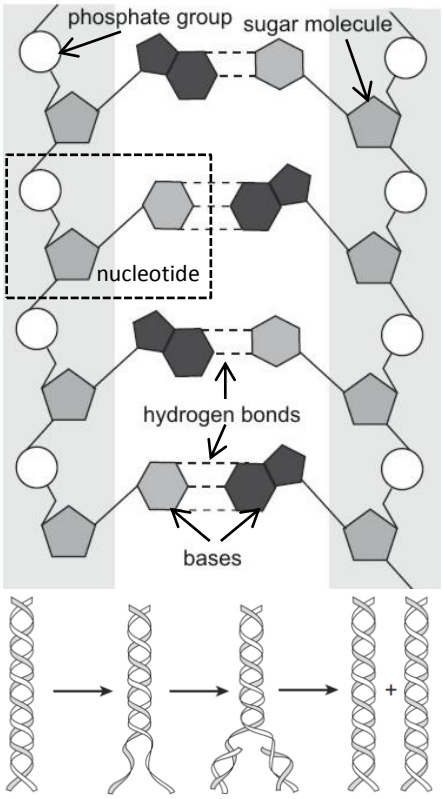
Chromosomes Chromosomes are long coiled lengths of DNA (deoxyribonucleic acid) found in the nucleus of a cell. Each chromosome is made up of many genes. Different species of animals and plants contain different numbers of chromosomes. Humans have 46 chromosomes (23 pairs). A pair of chromosomes is called a homologous pair.



Genes - are sections of DNA that code for a particular characteristic such as eye colour, hair colour, ear shape.
Alleles - the name given to one of the forms that a gene can have. E.g. the gene for tongue rolling has 2 alleles – a dominant form (tongue roller) and a recessive form (non tongue roller).
DNA (deoxyribonucleic acid) – a molecule that contains the instructions to make a new organism. It is found in the nucleus of the cell. It is usually super twisted to form chromosomes.



Chromosomes exist as pairs so that individuals inherit two copies of each gene.



The backbone of the DNA molecule consists of 2 strands of alternating sugar molecules and phosphate groups, the 2 strands twisted to form a double helix. Each sugar molecule is attached to one of 4 bases called Adenine, Guanine, Cytosine or Thymine (A, G, C & T for short). The bases are paired up on opposite strands A always pairs with T and G with C. This is called complementary base pairing –the order of bases in one strand determining the order of bases in the other. A nucleotide is a basic building block for DNA made up of phosphate, sugar and base. The 2 strands are held together by weak hydrogen bonds. A gene consists of hundreds or thousands of bases. A gene codes for a particular protein by its particular base sequence.

DNA must replicate or copy itself before any cell division can take place. The **DNA double helix** is perfectly suited for replication because each strand can serve as a **template** (pattern) to produce a strand opposite to itself.
First it is "unzipped" – the 2 strands are separated. New nucleotides in the cell line up alongside the unpaired bases, A pairing with a T and G with a C. The new nucleotides are joined together using an enzyme called DNA polymerase.

DNA replication is semi conservative i.e. each strand in the original DNA molecule is used as a template to make a new strand of DNA. Each new DNA molecule contains an original strand and a newly made strand.

Cell division

Cell division occurs through **mitosis & meiosis**. Biological ideas relating to mitosis and meiosis are limited to:

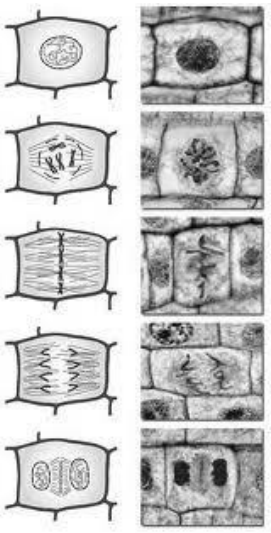
- purpose
- where they occur
- sequence of events (the names of stages are not required)
- reasons for maintenance or change of chromosome number
- significance of the number of cells produced

Cells grow old and die. They need to be constantly replaced. This is done by a type of cell division called mitosis. **Mitosis** takes place in all cells except the sex organs (testes and ovaries). It is used for growth and replacement of cells. If you are going to grow you have to make more cells! Daughter cells have the same number of chromosomes as the parent cell. **Meiosis** – also called reduction division – is the cell division that produces gametes. Meiosis in animals takes place in the testes and ovaries. It results in the production of sex cells (sperm and eggs). In plants it results in pollen grains and eggs. Sex cells have half the number of chromosomes as the parent cell. When the sperm and egg cells unite at fertilisation, each contributes 23 chromosomes so the resulting embryo will have the usual 46. Meiosis also allows genetic variation through a process of DNA shuffling while the cells are dividing. The 2 types of cell division are compared below:

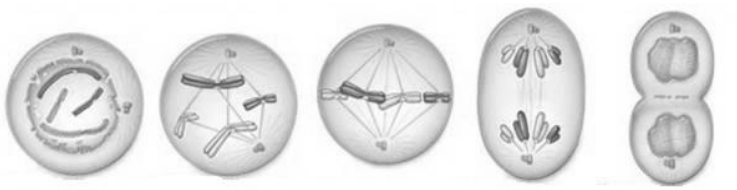
Mitosis	**Meiosis**
In somatic cells (general body cells)	In cells producing the gametes (testes and ovaries)
One cell division, resulting in 2 daughter cells	Two cell divisions, resulting in 4 cells
Chromosome number remains same (diploid) 2n	Chromosome number halved (haploid) n
No pairing of homologous chromosomes	Homologous chromosomes pair before first division
No "crossing over"	"Crossing over" of homologous chromosomes when they pair (exchanging genetic information)
Conservative process: Daughter cells' genotypes identical to parental cell's genotype	Process produces variation: The genetic makeup includes new combinations of genes not found in parental cell
Occurs in many different cell types - important for the animals to grow, develop, and repair damage	Only occurs in specific cells in the body that will become egg cells or sperm cells (gametes)

To understand how the differences between mitosis and meiosis occur, you must understand the sequence of events that takes place inside the cell for each type of cell division.
Remember that the point of mitosis is to produce identical copies of cells for rapid growth and repair.
The point of meiosis is to produce sex cells that contain half of the original number of chromosomes. Also, during meiosis genetic information (bits of chromosomes) is swapped between homologous chromosomes when they line up prior to separation. Mendel formulated what is now known as Mendel's law of independent assortment. This law states that allele pairs separate independently during the formation of gametes. Therefore, traits (characteristics) are transmitted to offspring independently of one another. It is a random process.

Mitosis

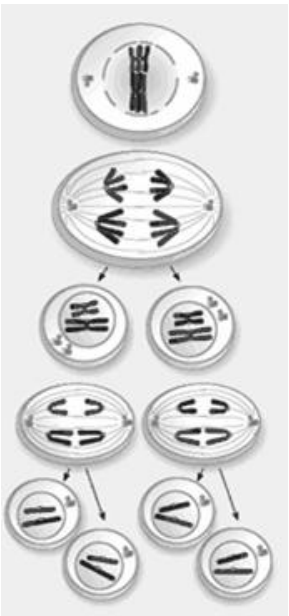


1. Before cell division the chromosomes can't be seen. As division starts, the chromosomes become fatter and more visible. The plant cell (opposite) and animal cell (below) both only shows 4 chromosomes.
2. The DNA making each chromosome is copied. Each chromosome has been copied. The copies are still attached at the centromere. Each copy is called a chromatid.
3. The chromosomes line up SINGULARLY in the middle of the cell (equator).
4. One <u>chromatid</u> of each chromosome goes to opposite ends of the cell (poles), pulled by the spindle apparatus. The parent cell starts to split into 2.
5. Two daughter cells are made. The number of chromosomes is the same in each daughter cell as the original parent cell.



Meiosis

1. Chromosomes become fatter and more visible. This cell has 4 chromosomes (2 homologous pairs).
2. The DNA making each chromosome is copied. Each chromosome has been copied. The copies are still attached at the centromere. Each copy is called a **chromatid.** The **homologous pairs of chromosomes line up** alongside each other in the middle of the cell. Now some **crossing over** of bits of chromosomes can take place between homologous pairs. This swaps genes from one chromosome to another and leads to variation between offspring.
3. One of each homologous pair goes to opposite ends of the cell. The parent cell starts to split into two, making two daughter cells.
4. The chromosomes in each cell line up in the middle and this time the chromatids separate. We now have **4 daughter cells; each one has half the number of chromosomes** as the original parent cell. Also, these chromosomes are not identical to the parent because **crossing over** took place and **independent assortment** (also called segregation) occurred (the two chromosomes of each pair (maternal and fraternal) are separated during meiosis and randomly distributed to the daughter cells).



Mutations – when things go wrong with DNA

It is important that DNA remains unchanged from generation to generation. The complementary base pairing that occurs during DNA replication means this generally occurs. However random, spontaneous mistakes do happen.

A mutation is a change in the <u>type</u> or the <u>amount</u> of DNA in a cell.

Type of DNA: A mistake made in copying the DNA can produce a slightly different allele of a gene. Certain chemicals, ultra-violet light, X-rays, or radiation can increase the chances of changes occurring to DNA even while chromosomes are not being copied.

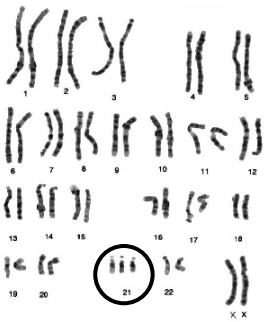
Changes to the base sequence are called *point or gene mutations* and can be (a) insertion (addition of a base) (b) deletion (loss of a base) (c) inversion (2 bases change position) (d) substitution (one base replaces another). If the mutation happens in the gametes, then this new allele can be passed on to offspring. Some diseases caused by mutations can be passed on to offspring – these diseases are called inherited diseases eg Cystic fibrosis.

Mutations can be:

- harmful (causing death or disease): To function correctly cells depend on many proteins. Gene mutations may stop one or more proteins from working properly. By changing a gene's instructions for a protein, the mutation can cause the protein to malfunction or to be missing entirely. If the protein plays a critical role in the body, it can disrupt development or cause a medical condition. Some mutations can cause illnesses such as cancer.
- beneficial (give some advantage or benefit to an organism) e.g. antibiotic resistance in bacteria is beneficial to the bacteria
- silent / neutral – appear to not affect the organism (does not affect phenotype). They may not be evident if the mutation is in a recessive allele. A change to a DNA triplet might not change which amino acid is introduced, or it might change the amino acid to a chemically similar amino acid that works just as well. Silent / neutral mutations do however create the possibility of a future mutation having an effect.

Amount of DNA: Sometimes when cells divide, the wrong number of chromosomes ends up in a gamete and it may have a particular chromosome missing, or two copies of a chromosome.

Down syndrome occurs in about 1 in 900 births. The risk of having a Down syndrome child increases as a woman gets older. Down syndrome is caused each cell in the body having three copies of chromosome 21. Down's children often have learning difficulties and poor muscle tone, and problems with their sight or hearing, and/or have heart problems.



Karyotype of a girl with Down syndrome