3.4 Genetic information, variation and relationships between organisms

3.4.1 DNA, genes and chromosomes

Content

- In prokaryotic cells, DNA molecules are short, circular and not associated with proteins.
- In the nucleus of eukaryotic cells, DNA molecules are very long, linear and associated with proteins, called histones. Together a DNA molecule and its associated proteins form a chromosome.
- The mitochondria and chloroplasts of eukaryotic cells also contain DNA which, like the DNA of prokaryotes, is short, circular and not associated with proteins.
- A gene is a base sequence of DNA that codes for:
    - the amino acid sequence of a polypeptide
    - a functional RNA (including ribosomal RNA and tRNAs).
- A gene occupies a fixed position, called a locus, on a particular DNA molecule.
- A sequence of three DNA bases, called a triplet, codes for a specific amino acid. The genetic code is universal, non-overlapping and degenerate.
- In eukaryotes, much of the nuclear DNA does not code for polypeptides. There are, for example, non-coding multiple repeats of base sequences between genes. Even within a gene only some sequences, called exons, code for amino acid sequences. Within the gene, these exons are separated by one or more non-coding sequences, called introns.

In **prokaryotic** cells, DNA molecules are short, circular and not associated with proteins or a nucleus. On the contrary, in the nucleus of **eukaryotic** cells, DNA molecules are very long, linear and associated with proteins, called histones. Together a DNA molecule and its associated proteins form a chromosome. Mitochondria and chloroplasts of eukaryotic cells also contain DNA which, like the DNA of prokaryotes, is short, circular and not associated with proteins.

DNA is made up of genes, ie short sections of DNA that code for specific polypeptides and functional RNA. The coded information is in the form of a specific sequence of bases along the DNA molecule. Polypeptides make up proteins and so genes determine the proteins of an organism. Enzymes are proteins which control chemical reactions, and are responsible for an organism development and activities. Therefore, genes determine (alongside environmental factors), the nature and development of all organisms. A gene is a section of DNA located at a particular position, this particular position is called the ‘locus’. The gene is a base sequence of DNA that codes for the amino acid sequence of a polypeptide, or a functional RNA for the process of polypeptide synthesis. It codes for ribosomal RNA as well as tRNAs.

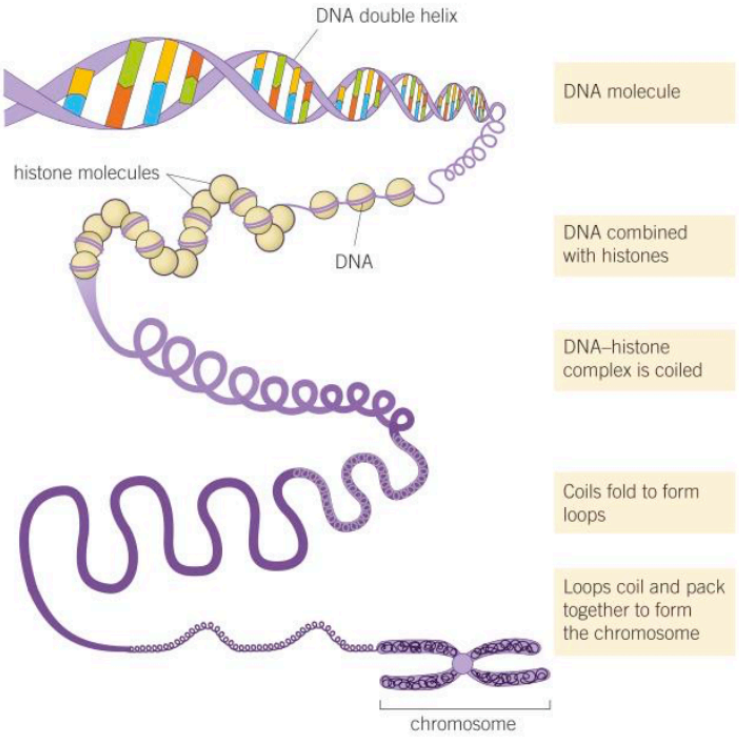
In trying to discover how DNA bases coded for amino acids, scientists suggested that there must be a minimum of three bases that coded for each amino acid. This is called a triplet, although there are 64 different triplets that can be coded for and only 20 regularly occurring amino acids, thus some triplets code for the same amino acids. Another thing worth

mentioning is that if there were pairs of bases, only 16 possible codes would be possible, less than the amount of naturally occurring amino acids found.

The genetic code is universal, non-overlapping and degenerate. It is **degenerate** because most amino acids are coded for by more than one triplet. These triplets are read in only one direction, and the start of a DNA sequence that codes for a polypeptide is always the same triplet (start codon). This start codon always codes for methionine in eukaryotes, and if this first molecule does not form part of the final polypeptide, it is removed. There are three different triplets that encode stop codons, whose role is to designate the termination of translation. The code is **non-overlapping** in that successive triplets are read in order. Each nucleotide is part of only one triplet codon. The code is also **universal,** with minor exceptions ie protozoan species, in that each triplet codes for the same amino acid in all organisms.

Although a large proportion of the DNA in eukaryotes does not code for polypeptides. Only certain parts of sequence code for amino acids, called exons. The non-coding sequences are called introns. There are also non-coding multiple repeats of base sequences between genes. The other genes code for ribosomal RNA and transfer RNAs.

Chromosome structure – chromosomes are only visible as distinct structures when a cell is dividing. For the remainder of time, they are widely dispersed throughout the nucleus. When they first become visible at the start of cell division, chromosomes appear as two threads, joined as a single point. Each thread is called a chromatid because DNA has already replicated to give two identical DNA molecules. The DNA in chromosomes is held together by histones.



DNA is a double helix, wound around histones to fix it in position. The DNA-histone complex is then coiled, and this coil, in turn, is looped and further coiled before being packed into the chromosome. In this way, a lot of DNA is condensed into a single chromosome. It is clear from the diagram above that a chromosome contains just a single DNA molecule. This single DNA molecule has many genes along its length, each gene occupying a specific locus along the DNA molecule.

Homologous chromosomes – these are a pair of chromosomes, one maternal and one paternal, that have the same gene loci. Sexually produced organisms are as a result of the fusion of a sperm and an egg, each of which contributes one complete set of chromosomes to the offspring. Therefore, one of each pair is derived from the chromosomes provided by the mother in the egg, and the other from the paternal DNA. These are known as homologous pairs and the total number of these chromosomes is referred to as the **diploid** number. A homologous pair is always two chromosomes that carry the same genes but not necessarily the same alleles of the gene.

During meiosis, the halving of the number of chromosomes is done in a manner which ensures that each daughter cell receives one chromosome from each homologous pair. In this way each cell receives one gene for each characteristic of an organism, and when these haploid cells combine, the diploid state with paired homologous chromosomes is restored.

An **allele** is one of a number of alternative forms of a gene. We have seen that genes are sections of DNA that contain coded information in the form of specific sequences of bases. Each gene exists in two, occasionally more, different forms. Each individual inherits one allele from each of its parents, these two alleles may be the same but they may be different. Any change in the base sequence of a gene produces a new allele of that gene (a mutation), resulting in a different sequence of amino acids being coded for. This results in a different primary structure that gives rise to a different tertiary structure, hence a different protein. The consequences of this lie with the inability for enzyme substrate complexes to form if the substrate is no longer a complementary fit to the active site of the enzyme.

In eukaryotic cells, the DNA is largely confined to the nucleus. Although, the synthesis of proteins takes place in the cytoplasm as sections of the code of the DNA are transcribed onto a single-stranded molecule called RNA. There are a number of types of RNA, mRNA, tRNA and rRNA. mRNA is messenger RNA, as it takes the role of transferring the DNA code from the nucleus to the cytoplasm. The mRNA is small enough to leave the nucleus through nuclear pores and enter the cytoplasm, where its coded information is used to determine the sequence of amino acids in the proteins which are synthesised there. The structure and function of these different types of RNAs are given in the next section.

3.4.2 DNA and protein synthesis

Content

- The concept of the genome as the complete set of genes in a cell and of the proteome as the full range of proteins that a cell is able to produce.
- The structure of molecules of messenger RNA (mRNA) and of transfer RNA (tRNA).
- Transcription as the production of mRNA from DNA. The role of RNA polymerase in joining mRNA nucleotides.
    - In prokaryotes, transcription results directly in the production of mRNA from DNA.
    - In eukaryotes, transcription results in the production of pre-mRNA; this is then spliced to form mRNA.
- Translation as the production of polypeptides from the sequence of codons carried by mRNA. The roles of ribosomes, tRNA and ATP.
- **Students should be able to:**
- Relate the base sequence of nucleic acids to the amino acid sequence of polypeptides, when provided with suitable data about the genetic code
- Interpret data from experimental work investigating the role of nucleic acids.
- Students will **not** be required to recall in written papers specific codons and the amino acids for which they code.

The **genome** is the complete set of genes in a cell and the **proteome** is the full range of proteins that a cell is able to produce. The words **complete proteome** refers to the the proteins produced by a given type of cell under a set of **conditions**.

RNA has two forms, one being messenger RNA and the other transfer RNA. mRNA is small enough to leave the nucleus through nuclear pores to enter the cytoplasm, where the coded information it contains is used to determine the sequence of amino acids in the proteins which are synthesised here.

The term **codon** means the sequence of three bases on mRNA that codes for a single amino acid.

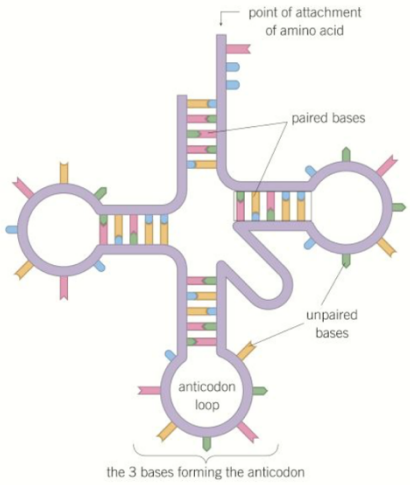
RNA is a polymer made up of repeating mononucleotide sub-units. It forms a single strand where each nucleotide is made up of: a pentose sugar, A G C or U and a phosphate group.

Messenger RNA (mRNA) consists of thousands of mononucleotides. It is a relatively long strand arranged in a single helix. The base sequence on the mRNA is determined by the sequences of bases on a length of DNA in a process called transcription. mRNA then leaves the nucleus via nuclear pores in the nuclear envelope entering the cytoplasm, where it associates with ribosomes. It then stays there and acts as a template for protein synthesis, as tRNA molecules bind.

Its structure is suited to its function because it possesses information in the form of codons (three bases that are complementary to a triplet in DNA). The sequence of codons ultimately determines the amino acid sequence of a specific polypeptide that will be made.

Transfer RNA (tRNA) is a relatively small molecule that is made up of around 80 nucleotides. It is a single-stranded chain folded into a clover-leaf shape, where one end of the chain extends slightly above the other. This is the side of the tRNA that the amino acid can attach easily too, and you get different types of tRNA that bind to different specific amino acids. At the opposite end you find an anticodon, made up of three other organic bases. Each tRNA is specific to one amino acid, with an anticodon specific to one amino acid. Although you do find as many different tRNA molecules as there are coding triplets due to the degenerate code of DNA.

Its structure is suited to it function by having an end for attaching to amino acids, as well as its anticodon for complementary base pairing with the codon of the mRNA. Also it is suited for lining up amino acids on the mRNA template during protein synthesis.



In RNA you find the base uracil instead of thymine, and so the different complementary base pairings you find are:

- Guanine and cytosine
- Adenine and uracil (RNA) or Thymine (DNA).

DNA	Messenger RNA	Transfer RNA
double polynucleotide chain	single polynucleotide chain	single polynucleotide chain
largest molecule of the three	molecule is smaller than DNA but larger than tRNA	smallest molecule of the three
double-helix molecule	single-helix molecule (except in a few viruses)	clover-shaped molecule
pentose sugar is deoxyribose	pentose sugar is ribose	pentose sugar is ribose
organic bases are adenine, guanine, cytosine and thymine	organic bases are adenine, guanine, cytosine and uracil	organic bases are adenine, guanine, cytosine and uracil
found mostly in the nucleus	manufactured in the nucleus but found throughout the cell	manufactured in the nucleus but found throughout the cell
quantity is constant for all cells of a species (except gametes)	quantity varies from cell to cell and with level of metabolic activity	quantity varies from cell to cell and with level of metabolic activity
chemically very stable	Less stable than DNA or tRNA, individual molecules are usually broken down in cells within a few days.	chemically more stable than mRNA but less stable than DNA

Proteins are polypeptides, and essential to our life, particularly enzymes. However each organism needs to make its own unique proteins, and the biochemical chemistry in the cytoplasm of each cell has the capacity to make every protein from just 20 amino acids. Exactly which protein is made is determined by the DNA.

The process of polypeptide synthesis is as follows:

- DNA provides the instructions in the form of a long sequence of bases
- A complementary section of part of this sequence is made in the form of a molecule called pre-mRNA – a process called **transcription**
- Pre-mRNA is splices to remove introns and form mRNA
- The mRNA is used to act as a template to which complementary tRNA molecules attach and the amino acids they carry are linked to form a polypeptide – a process called **translation**

Transcription involves making pre-mRNA using DNA as a template.

- First of all, an enzyme acts on a specific region of DNA causing the two strands to separate and expose the nucleotide bases of that region.
- The nucleotide bases on the two DNA strands act as template strands, where complementary RNA nucleotides pair with these exposed bases. The enzyme RNA polymerase then moves along the strand and joins the nucleotides together to form a pre-mRNA molecule.
- However, the base pairs are now different, with adenine and uracil as opposed to adenine and thymine in DNA.
- As the RNA polymerase adds the nucleotides one at a time to build a strand of pre-mRNA, the DNA strand rejoins behind it. As a result, only about 12 base pairs on the DNA are exposes at any one time.
- When the RNA polymerase reaches a particular sequence of bases on the DNA that it recognises as a 'stop' triplet code, it detaches and then the production of pre-mRNA is complete.

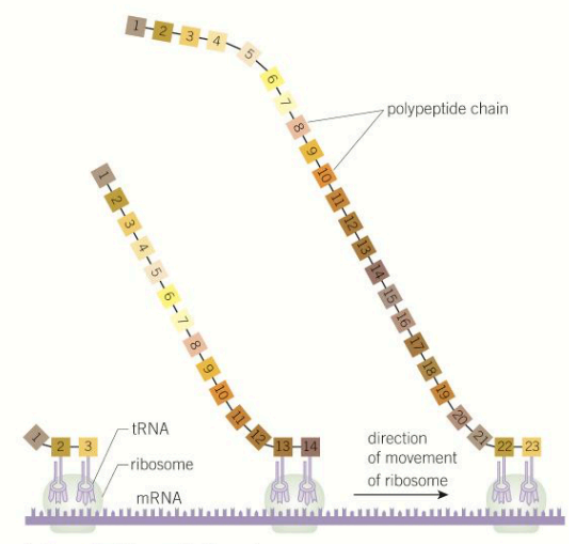
Splicing of pre-mRNA does not occur in prokaryotic cells, as transcription results directly in the production of mRNA from DNA. However, in eukaryotic cells the DNA is made up of introns and exons, which are non coding sections and coding sections. These introns would prevent the synthesis of a polypeptide.

The mRNA molecules formed are too large to diffuse out of the nucleus so leave via the nuclear pore. Once outside the nucleus, the mRNA moved towards the ribosome and attaches to it, ready for the next stage in the process called **translation**. This stage involves translating the codons on the mRNA into a sequence of amino acids. The process is given below:

- A ribosome becomes attached to the starting codon at one end of the RNA molecule
- The tRNA molecule with the complementary anticodon sequence moves to the ribosome and pairs up with the codon on the mRNA. The tRNA carries a specific amino acid
- Another tRNA molecule with a complementary anticodon moves and pairs with the next codon on the mRNA. This tRNA molecule carries another amino acid

- The ribosome moves along the mRNA, bringing together two tRNA molecules at any one time, each pairing up with the corresponding two codons on the mRNA
- The amino acids on the tRNA then join by a peptide bond, using an enzyme and ATP which is hydrolysed to provide the required energy
- The ribosome moves on to the third codon in the sequence and links the next amino acids
- As this is happening, the first tRNA is released from its amino acid and is free to collect another amino acid from the amino acid pool in the cell
- The process continues in this way until a polypeptide chain is built up
- The synthesis of a polypeptide continues until the ribosome reaches a stop codon, where the ribosome, mRNA and the last tRNA all separate and the polypeptide is made.

In this process, up to 50 ribosomes are able to pass on the mRNA so the same polypeptide can be built up many times.



3.4.3 Genetic diversity can arise as a result of mutation or during meiosis

Content

- Gene mutations involve a change in the base sequence of chromosomes. They can arise spontaneously during DNA replication and include base deletion and base substitution. Due to the degenerate nature of the genetic code, not all base substitutions cause a change in the sequence of encoded amino acids. Mutagenic agents can increase the rate of gene mutation.
- Mutations in the number of chromosomes can arise spontaneously by chromosome non-disjunction during meiosis.
- Meiosis produces daughter cells that are genetically different from each other.
- The process of meiosis only in sufficient detail to show how:
    - Two nuclear divisions result usually in the formation of four haploid daughter cells from a single diploid parent cell
    - Genetically different daughter cells result from the independent segregation of homologous chromosomes
    - Crossing over between homologous chromosomes results in further genetic variation among daughter cells.
- **Students should be able to:** Complete diagrams showing the chromosome content of cells after the first and second meiotic division, when given the chromosome content of the parent cell
- Explain the different outcome of mitosis and meiosis
- Recognise where meiosis occurs when given information about an unfamiliar life cycle
- Explain how random fertilisation of haploid gametes further increases genetic variation within a species.

Opportunities for Skills Development

- Students could examine meiosis in prepared slides of suitable plant or animal tissue.
- Students could:
    - Use the expression to calculate the possible number of different combinations of chromosomes following meiosis, without crossing over
    - Derive a formula from this to calculate the possible number of different combinations of chromosomes following random fertilisation of two gametes,
    - Where n is the number of homologous chromosomes pairs.

Gene mutations involve a change in the base sequence of chromosomes. They can arise spontaneously during DNA replication and include base deletion and base substitution. Due to the degenerate nature of the genetic code, not all base substitutions cause a change in the sequence of encoded amino acids. Also, you can get mutagenic agents which increase the rate of mutation.

Two examples of mutations are substitutions and deletions.

Substitution: It is where a nucleotide in a DNA molecule is replaced by another nucleotide that has a different base. If the change in base sequence encodes a different amino acid, then it could be significant depending on the original role of the amino acid. For example if it were important in forming bonds that determine the final tertiary structure of the protein, then the replacement amino acid may not form the same bonds. The protein may then be a different shape and therefore not function properly, so if the protein were an enzyme it could cause a non-functional enzyme.

Deletion: A gene mutation by deletion arises when a nucleotide is lost from the normal DNA sequence. This causes a frame shift to the left, so each triplet code becomes different and the overall structure of the polypeptide becomes different.

Chromosome mutations occur when there are changes in the structure or number of whole chromosomes. They can arise spontaneously and come in two forms:

- **Changes in whole sets of chromosomes** occur when organisms have three or more sets of chromosomes rather than the usual two. This condition is called **polyploidy** and occurs mainly in plants.
- **Changes in the number of individual chromosomes** happens because sometimes individual homologous pairs of chromosomes fail to separate during meiosis. This is called **non-disjunction** and usually results in a gamete having one more or one fewer chromosome. Down's syndrome is as a result of an additional chromosome, an example of non-disjunction.

Meiosis produces daughter cells that are genetically different to each other, unlike mitosis. The process of meiosis must only be known in such detail to show that

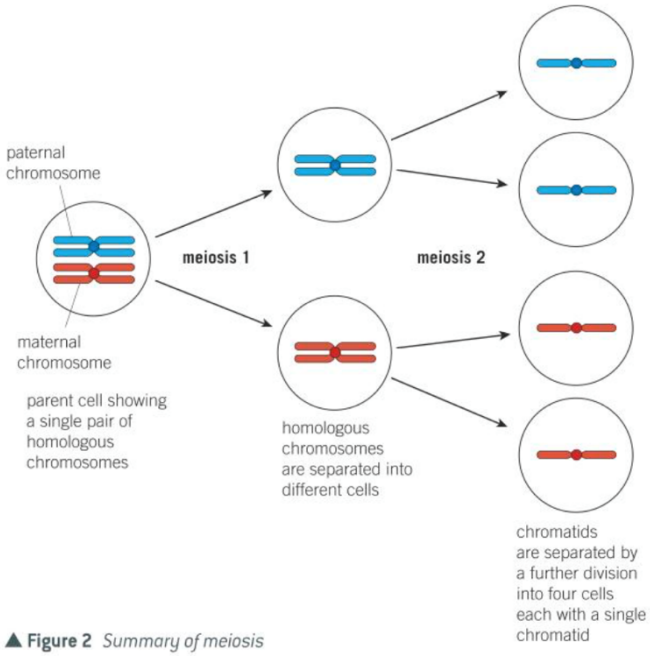
- Two nuclear divisions result usually in the formation of four haploid daughter cells from a single diploid parent cell
- Genetically different daughter cells result from the independent segregation of homologous chromosomes
- Crossing over between homologous chromosomes results in further genetic variation among daughter cells.

The two nuclear divisions usually occur immediately after each other. In the first division (meiosis 1), homologous chromosomes pair and their chromatids wrap around each other. Equivalent portions of these chromatids may be exchanged in a process called crossing over, this process involved exchange of genetic information. By the end of this division the homologous pairs have separated, with one chromosome from each pair going into one of the two daughter cells. In the second division (meiosis 2), the chromatids move apart. Thus four cells are formed, each containing 23 chromosomes in humans.

In addition to halving the number of chromosomes, meiosis also produces genetic variation among the offspring, which may lead to adaptations that could improve survival chances. Meiosis brings about this genetic variation in the following two ways:

- Independent segregation of homologous chromosomes

- New combinations of maternal and paternal alleles by crossing over (formation of chiasmata).



▲ **Figure 2** *Summary of meiosis*

The law of independent assortment – during meiosis 1, each chromosome lines up alongside its homologous partner. In humans, this means 23 homologous pairs will be lying side by side. However, they arrange themselves at random in the line, and only one of each pair will pass to each daughter cell. Since the pairs line up at random, the combination of chromosomes of maternal and paternal origin entering the daughter cell at meiosis 1 is also a matter of chance, called independent segregation. The independent assortment of these chromosomes therefore produces new genetic combinations.

The random fusion of gamete also ensures genetic diversity.

Crossing over contributes to genetic recombination in that genetic information is transferred between the chromosomes. The chromatids of each pair become twisted around one another, and during this twisting process portions of the chromatids break off. These broken portions may rejoin with the chromatids of its homologous partner, and usually it is the equivalent portions of homologous chromosomes that are exchanged. The broken off portions of chromatid recombine with another chromatid, so this process is called recombination.

AQA June 2014 Unit 2 Q1bi

Question:

'Explain the role of independent segregation in meiosis'

Answer:

- (To provide) genetic variation;
- (Allows) different combinations of maternal and paternal chromosomes/ alleles;
- (To produce) haploid cells/half the chromosome number;
- (Allows) homologous chromosomes/ homologous pairs to arrange randomly (at equator/middle of cell)/separate;

3.4.4 Genetic diversity and adaptation

Content

- Genetic diversity as the number of different alleles of genes in a population.
- Genetic diversity is a factor enabling natural selection to occur.
- The principles of natural selection in the evolution of populations.
    - Random mutation can result in new alleles of a gene.
    - Many mutations are harmful but, in certain environments, the new allele of a gene might benefit its possessor, leading to increased reproductive success.
    - The advantageous allele is inherited by members of the next generation.
    - As a result, over many generations, the new allele increases in frequency in the population.
- Directional selection, exemplified by antibiotic resistance in bacteria, and stabilising selection, exemplified by human birth weights.
- Natural selection results in species that are better adapted to their environment. These adaptations may be anatomical, physiological or behavioral.
    - **Students should be able to:**
- Use unfamiliar information to explain how selection produces changes within a population of a species
- Interpret data relating to the effect of selection in producing change within populations
- Show understanding that adaptation and selection are major factors in evolution and contribute to the diversity of living organisms.

Genetic diversity is the number of different alleles of genes in a population. It is the factor that enables natural selection to occur. Differences in DNA are the reason behind the vast genetic diversity found on Earth. Furthermore, a population is a group of individuals of the same species that live in the same place and can interbreed. A species consists of one, or more, populations. The greater range of alleles, the higher chance that some individuals in a population will survive environmental change.

Natural selection is part of the evolution of populations, as not all alleles of a population are equally likely to be passed on to the next generation. This is because only certain individuals are reproductively successful and so pass on their alleles.

Differences between reproductive success of individuals affects allele frequency in populations. This process works as follows:

- Within any population of a species there will be a gene pool containing a wide variety of alleles
- Random mutations of alleles within this gene pool may result in a new allele of a gene in which most cases will be harmful
- However, in certain environments, the new allele of a gene might give its possessor an advantage over other individuals in the population
- These individuals will be better adapted and therefore more likely to survive in their competition with others

- These individuals are more likely to obtain the available resources and so grow more rapidly and live longer. As a result, they will have a better chance of breeding successfully and producing more offspring
- Only those individuals that reproduce successfully will pass on their alleles to the next generation
- Therefore it is the new allele that gave the parents an advantage in the competition for survival that is most likely to be passed on to the next generation
- As these new individuals also have the new, 'advantageous' allele, they in turn are more likely to survive, so reproduce successfully
- Over many generations, the number of individuals with the new 'advantageous' allele will increase at the expense of the individuals with the 'less advantageous' alleles.
- Over time, the frequency of the new, 'advantageous' allele in the population increases whilst that of the 'non-advantageous' one decreases.

The different types of selection are as follows: directional selection and stabilising selection.

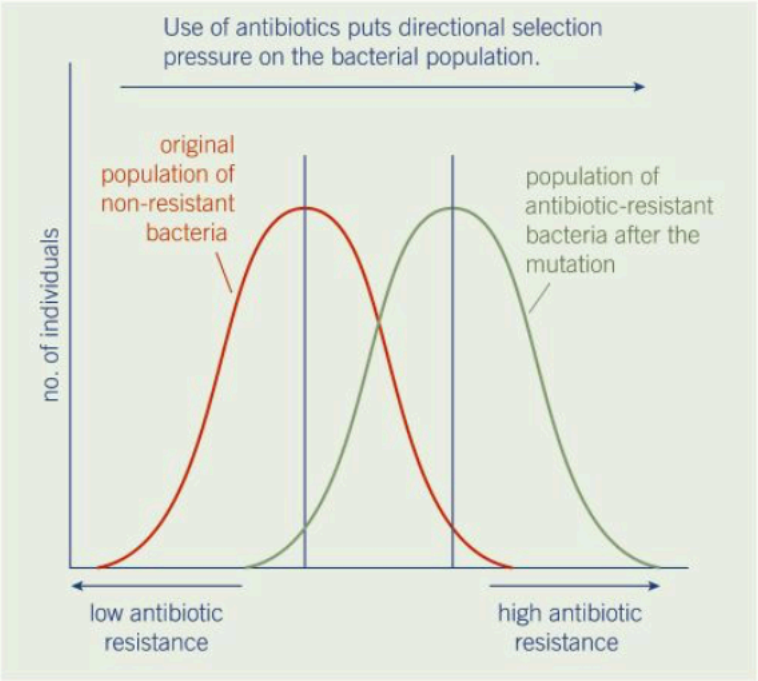
Directional selection is where selection may favour individuals that vary in one direction from the mean of the population. This changes the characteristics of a population, hence directional.

The phenotypes (observable physical and biochemical characteristics of an organism) that are best suited to the new conditions are most likely to survive. Some individuals, which fall to either the left or right of the mean, will posses a phenotype more suited to the new conditions. These individuals are more likely to survive and breed, so will contribute more to the offspring and the alleles of the offspring. So over time the mean phenotype will move in the direction of these individuals.

An example is antibiotic resistance in bacteria because shortly after the discovery of antibiotics it became apparent that the effectiveness of some antibiotics at killing bacteria was reduced. The bacteria had developed resistance, due to mutations within the bacteria. The case of resistance to penicillin is shown below:

- A spontaneous mutation occurred in the new allele of a gene in a bacterium that enabled it to make a new protein. The new protein was an enzyme that broke down the antibiotic penicillin before it was able to kill the bacterium. The enzyme was called penicillinase.
- The bacterium happened, by chance, to be in a situation where penicillin was being used to treat an individual. In these circumstances, the mutation gave the bacterium an advantage of being able to use penicillinase to break down the antibiotic and so survive while the rest of the population of bacteria were killed by it
- The bacterium that survived was able to divide by binary fission and build up a population of resistant bacteria
- Members of this population were more able to survive, and so multiply in the presence of penicillin
- This population increases at the expense of the other population, so the frequency of the allele that enabled production of penicillinase increased in the population
- The population's normal distribution curve shifted in the direction of a population having greater resistance to penicillin.

The graph below shows directional selection.



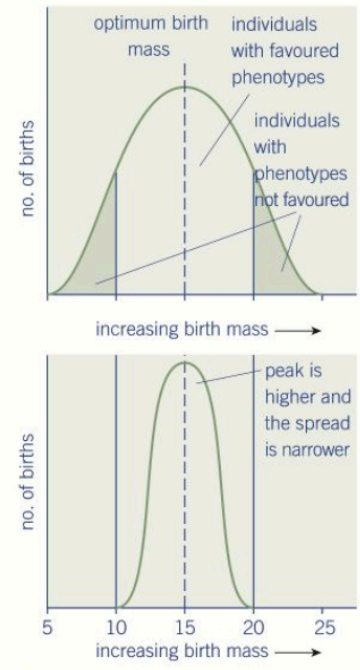
With continued use of antibiotics, there is a greater chance that the mutant population will out-compete, and replace, the original population. Directional selection therefore results in phenotypes at one extreme of the population being selected for and those at the other extreme selected against.

Stabilising selection occurs where the environmental conditions remain stable, so the individuals with phenotypes closest to the mean are favoured. These individuals are more likely to pass their alleles on to the next generation, and those at the extremes are less likely. Stabilising selection tends to eliminate those at the extremes.

An example of stabilising selection is human **birth weights**.

The mortality rate is higher at the extremes of birth weights, as it seems the optimum masses are between 2.5-4kg. The infants with higher and lower masses are more likely to die, thus are being selected against.

Stabilising selection therefore results in phenotypes around the mean of the population being selected for and those at both extremes being selected against.



Natural selection results in species better adapted for the environment that they are living in, these adaptations are:

- **Anatomical** – such as shorter ears and thicker fur in arctic foxes compared to foxes in warmer climates
- **Physiological** – for example oxidising of fat rather than carbohydrate in kangaroo rats to produce additional water in a dry desert environment
- **Behavioural** – such as the autumn migration of swallows from the UK to Africa to avoid food shortages in the UK winter.

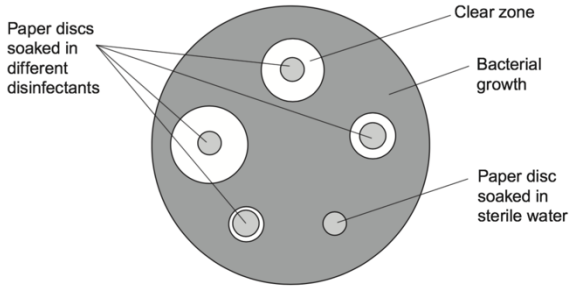
AQA June 2013 Unit 2 Q5d

Pseudomonas aeruginosa is a bacterium that can cause infections in hospital patients suffering from burns. Disinfectants are substances used to kill bacteria on non-living objects, such as medical equipment. Doctors in one hospital investigated how effective four disinfectants were at killing *P. aeruginosa*.

The doctors:

- Took samples from many patients in the hospital
- Isolated *p. Aeruginosa* from those samples
- Suspended the *p. Aeruginosa* in a solution
- Spread many samples of this solution on nutrient jelly in many petri dishes.

The doctors then placed five small paper discs on the jelly in each dish. Each disc had been soaked in a different disinfectant or sterile water. The doctors left the plates for 24 hours to allow bacteria to grow and divide. The diagram shows a typical Petri dish after 24 hours.



Question:

Doctors in a different hospital repeated this investigation. They found that hypochlorite had little effect on samples of *P. aeruginosa* they obtained. Suggest how this different result may have arisen.

Answer:

1. Mutation (in bacterium);
2. Gene/allele for resistance;

AQA Jan 2013 Unit 2 Q3ci)ii)

Question:

All modern cheetahs are thought to have descended from a single female. This female was part of a small population that survived an ice age a long time ago that killed almost all cheetahs. After the ice age, the number of cheetahs increased.

Use this information to explain what is meant by a genetic bottleneck

Answer:

1. Drop in population / many killed / only single female left;
2. Idea of reduced/low genetic variation/diversity / reduction in (variety of) alleles / smaller gene pool;

Question:

The fertility of cheetahs is low. The proportion of abnormal sperm cells produced is higher in cheetahs than in other members of the family Felidae. Suggest an explanation for this.

Answer:

1. Mutation affecting sperm cell or production (in small population);
2. Errors during meiosis;
3. Inbreeding / closely related cheetahs breed;
4. High chance of inheriting allele / high frequency of allele (in the population)

AQA A Level Specimen (set 2) Q10.2

Question:

Penicillin has been the antibiotic of choice for the treatment of bacterial meningitis. Since the year 2000, strains of *Neisseria meningitidis* that are resistant to penicillin, sulfonamides and rifampin have been discovered in the UK.

Describe how a population of *Neisseria meningitidis* (Nm) can become resistant to these antibiotics.

Answer:

1. Mutation;
2. Results in Nm cell with allele for resistance to one antibiotic/to named antibiotic;
3. (This) cell survives and passes the allele for resistance to offspring;
4. Process repeated with different genes conferring resistance to each of the other (two) antibiotics

3.4.5 Species and taxonomy

Content

- Two organisms belong to the same species if they are able to produce fertile offspring. Courtship behaviour as a necessary precursor to successful mating. The role of courtship in species recognition.
- A phylogenetic classification system attempts to arrange species into groups based on their evolutionary origins and relationships. It uses a hierarchy in which smaller groups are placed within larger groups, with no overlap between groups. Each group is called a taxon (plural taxa).
- One hierarchy comprises the taxa: domain, kingdom, phylum, class, order, family, genus and species.
- Each species is universally identified by a binomial consisting of the name of its genus and species, eg, *Homo sapiens*.
- Recall of different taxonomic systems, such as the three domain or five kingdom systems, will **not** be required.
- **Students should be able to** appreciate that advances in immunology and genome sequencing help to clarify evolutionary relationships between organisms.

It can be said that two organisms belong to the same **species** if they are able to breed to produce fertile offspring. Essentially, when a species reproduces sexually, any of the genes of its individuals can, in theory, be combined with any other.

Before breeding, courtship behaviour is necessary for successful mating as it plays a role in species recognition. Members of the same species will resemble each other physically and biochemically, helping them to distinguish members of their own species from others. The same is true of behaviour, and the behaviour of members of the same species is more alike than that of members of different species.

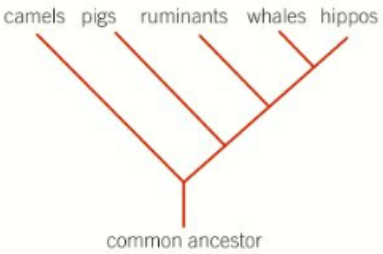
The ability to display behaviour is genetically determined, so individuals can therefore recognise members of their own species by the way they act. Reproduction is the means by which a species can survive over time, so each individual has adaptations to help ensure their DNA is passed on. The females of most species only produce eggs at specific times, sometimes as little as once a year. It is therefore important to ensure mating is successful and that the offspring have the maximum chance of survival.

Courtship behaviour helps to achieve mating is successful by:

- Recognising members of their own species: to ensure mating only takes place between members of the same species because only members of the same species can produce fertile offspring
- Identify a mate that is capable of breeding because both partners need to be sexually mature, fertile and receptive to mating
- Form a pair bond that will lead to successful mating and raising of offspring
- Synchronise mating so it takes place when there is maximum probability of the sperm and egg meeting
- Become able to breed by bringing a member of the opposite sex into a physiological state that allows breeding to occur.

A **phylogenetic classification** system attempts to arrange species into groups based on their evolutionary origins and relationships. It uses a **hierarchy** in which smaller groups are placed within larger groups, with no overlap between groups. Each group is called a taxon (plural taxa). There is also another type of classification called **artificial classification** which divides organisms according to differences useful at the time ie colour, size, number of legs etc.

The hierarchical order of taxonomic ranks is based upon the supposed evolutionary line of descent of the group members. The evolutionary relationship between organisms is known as phylogeny, and the term derives from 'phylum', which, in classification is a group of related or similar organisms. The phylogeny of an organism reflects the evolutionary branch that led up to it. In a phylogenetic tree-like diagram, the oldest species is at the base of the tree, and most recent at the ends of the branches. An example is shown below.



The closer the branches, the closer the evolutionary relationship. Hippos and whales are more closely related than hippos and ruminants.

Taxonomy is the theory and practice of biological classification. Each group within the phylogenetic biological classification is called a taxon (plural taxa). Taxonomy is the study of these groups in a hierarchical order, called taxonomic ranks.

One **hierarchy** comprises the taxa: domain, kingdom, phylum, class, order, family, genus and species. The highest rank is domain and it comprises of bacteria, archaea and eukarya.

Each species is universally identified by a **binomial** consisting of the name of its genus and species, eg, *Homo sapiens*. It is a universal system based upon Latin or Greek names. The first name, called the **generic** name, denotes the genus which an organism belongs to ie the surname of a human. The second name called the **specific** name, denotes the species to which an organism belongs, equivalent to the first name of a human. However, these specific names are never shared between other species in the genus.

There are rules when using the binomial system, including:

- The names are printed in italics or, if handwritten they are underlined to indicate scientific names
- The first name of the genetic name is in upper case but the specific name is all lower case
- If the specific name is not known is can be written as '*sp*'

AQA Jan 2012 Unit 2 Q3cii

Question:

The variety of colours displayed by catfish is important in courtship. Give **two** ways in which courtship increases the probability of successful mating

Answer:

1. Attracts/recognises same species;
2. Attracts/recognises mate/opposite sex;
3. Indication of sexual maturity/ fertility / synchronises mating;
4. Stimulates release of gametes;
5. Form pair bond;

3.4.6 Biodiversity within a community

Content

- Biodiversity can relate to a range of habitats, from a small local habitat to the Earth.
- Species richness is a measure of the number of different species in a community.
- An index of diversity describes the relationship between the number of species in a community and the number of individuals in each species.
- Calculation of an index of diversity (d) from the formula $d = \frac{N(N-1)}{\Sigma n(n-1)}$
- Where N = total number of organisms of all species and n = total number of organisms of each species.
- Farming techniques reduce biodiversity. The balance between conservation and farming.

Opportunities for Skills Development

- Students could be given data from which to calculate an index of diversity and interpret the significance of the calculated value of the index.

Biodiversity can relate to a range of habitats, from a small local habitat to the Earth. It can come in three forms:

- Species diversity: refers to the number of different species and the number of individuals of each species within any one community
- Genetic diversity: refers to the variety of genes possessed by the individuals that make up a population of a species
- Ecosystem diversity: refers to the range of different habitats, from a small local habitat to the whole of the Earth.

One measure of species diversity is **species richness**, which is the measure of the number of different species in a community in a particular area at a given time. Two communities may have the same number of species by the proportions of the community made up of each species may differ markedly.

Measuring the index of diversity is done by using the equation $d = \frac{N(N-1)}{\Sigma n(n-1)}$

An example of how this equation is used is given below

Species	Numbers (n) found in habitat X	$n(n-1)$	Numbers (n) found in habitat Y	$n(n-1)$
A	10	$10(9) = 90$	3	$3(2) = 6$
B	10	$10(9) = 90$	5	$5(4) = 20$
C	10	$10(9) = 90$	2	$2(1) = 2$
D	10	$10(9) = 90$	36	$36(35) = 1260$
E	10	$10(9) = 90$	4	$4(3) = 12$
	$\sum n(n-1)$	450	$\sum n(n-1)$	1300

You can now calculate the species diversity index for each habitat.

Habitat X: $d = \frac{50(49)}{450} = \frac{2450}{450} = 5.44$

Habitat Y: $d = \frac{50(49)}{1300} = \frac{2450}{1300} = 1.88$

The higher the value d, the greater is the species diversity. So, in this case, although the total number of species and the total number of individuals are the same in both habitats, the species diversity of habitat X is much greater.

Farming techniques reduce biodiversity, thus there is difficulty in finding a balance between conservation and farming.

Impact of **agriculture** on biodiversity: As natural ecosystems develop over time they become more complex communities with many individuals of a large number of different species, thus a high index of diversity. Agricultural ecosystems are controlled by humans so very different, as farmers often select species for particular qualities that make them more productive. As a result, the number of species, and the genetic variety of alleles they possess, is reduced to the few that exhibit the desired features.

Since any particular area can only support a certain amount of biomass, if a farmer has taken most of the area up by one species they see as desirable, it leaves less space for other species. These many other species have to compete for the remaining resources and space, but most will not survive this competition. Even if species evolved to adapt to the changes, the population would be drastically reduced. In addition, pesticides are used to exclude these species because they compete for the light, mineral ions, water and food required for farmed species. The overall effect is reduced species diversity.

The balance between conservation and farming is difficult to get as food is essential to life, so with an ever-expanding human population there is pressure to produce it more and more intensively. The food production in the UK has been boosted by things like improved genetic varieties of plant and animal species, greater use of chemical fertilisers and pesticides, greater use of biotechnology and changes in farm practices, leading to larger farms and the conversion of land supporting natural communities into farmland. These changes have had many ecological impacts, but the overriding effect of intensive food production has been to reduce the variety of habitats within ecosystems and consequently reduce species diversity.

Certain practices directly remove habitats and reduce species diversity:

- Removal of hedgerows and grubbing out woodland
- Creating monocultures, for example replacing natural meadows with cereal crops or grass for silage
- Filling in ponds and draining marsh and other wetland
- Over-grazing of land, for example upland areas by sheep, thereby preventing regeneration of woodland

Other practices have had a more indirect effect:

- Use of pesticides and inorganic fertilisers
- Escape of effluent from silage stores and slurry tanks into water courses
- Absence of crop rotation and lack of intercropping or undersowing

However, there are management techniques that can be applied to increase species and habitat diversity, without unduly raising food costs or lowering yields. Examples of these include:

- Maintain existing hedgerows at the most beneficial height and shape, an A-shape provides better habitats than rectangular ones.
- Plant hedges as opposed to erecting fences at field boundaries
- Maintain existing ponds and where possible create new ones
- Leave wet corners of fields rather than draining them
- Plant native trees on lands with a low species diversity rather than in species-rich areas
- Use organic, rather than inorganic, fertilisers
- Use crop rotation that includes nitrogen-fixing crop, rather than fertilisers, to improve soil fertility
- Use intercropping rather than herbicides to control weeds and other pests
- Create natural meadows and use hay rather than grasses for silage
- Leave the cutting of verges and field edges until after flowering and when seeds have dispersed
- Introduce conservation headlands – areas at the edges of fields where pesticides are used restrictively so that wild flowers and insects can breed

These practices will make food production slightly more expensive, so financial incentives are provided to farmers ie from DEFRA or the Department for Environment, or the EU. If biodiversity is reduced the global living system will become more unstable and we all rely on the global system for food and other resources.

AQA June 2014 Unit 2 Q7a

Question:

What two measurements are needed to calculate an index of diversity

Answer:

1. Number of (individuals of) each species;
2. Total number of individuals / number of species;

AQA June 2013 Unit 2 Q2bii

Question:

'The forest was cleared to make more land available for agriculture

After the forest was cleared the species diversity of the insects in the area decreased. Explain why.'

Answer:

1. Decrease in variety of plants / fewer plant species;
2. Fewer habitats/niches;
3. Decrease in variety of food / fewer food sources;
4. Aspect of clearing forest (killing insects) eg machinery, pesticides;

3.4.7 Investigating diversity

Content

- Genetic diversity within, or between species, can be made by comparing:
  - The frequency of measurable or observable characteristics
  - The base sequence of DNA
  - The base sequence of mRNA
  - The amino acid sequence of the proteins encoded by DNA and mRNA.
- **Students should be able to:** interpret data relating to similarities and differences in the base sequences of DNA and in the amino acid sequences of proteins to suggest relationships between different organisms within a species and between species
- Appreciate that gene technology has caused a change in the methods of investigating genetic diversity; inferring DNA differences from measurable or observable characteristics has been replaced by direct investigation of DNA sequences.
- Knowledge of gene technologies will **not** be tested.
- Quantitative investigations of variation within a species involve:
  - Collecting data from random samples
  - Calculating a mean value of the collected data and the standard deviation of that mean
  - Interpreting mean values and their standard deviations.
- Students will **not** be required to calculate standard deviations in written papers.

Opportunities for Skills Development

- Students could:
  - Design appropriate methods to ensure random sampling
  - Carry out random sampling within a single population
  - Use random samples to investigate the effect of position on the growth of leaves.
- Students could use standard scientific calculators to calculate the mean values of data they have collected or have been given.
- Students could calculate, and interpret the values of, the standard deviations of their mean values.

Genetic diversity within, or between species, can be made by comparing:

1. The frequency of measurable or observable characteristics
2. The base sequence of DNA
3. The base sequence of mRNA
4. The amino acid sequence of the proteins encoded by DNA and mRNA.

An explanation of each factor is given below

1. This was the traditional method of assessing genetic diversity. The method is based on the fact that each observable characteristic is determined by a gene or genes (with environmental influences), so the variety within a characteristic depends on the number and variety of alleles of that gene (plus environmental influences). Although

this has limitations as a large number of observable characteristics are coded for by more than one gene (polygenic). Also differences may be as a result of environmental conditions as opposed to allelic differences. Human height for example, is determined by a number of genes but environmental factors like diet can affect the actual height reached. Therefore, inferring DNA sequences has been replaced by directly observing DNA sequences

2. Comparing DNA base sequences has been enabled by gene technology. DNA sequencing is now done by computer systems, and in these systems each nucleotide base can be tagged with a different coloured fluorescent dye to produce a series of bands, each of which represents one of the four nucleotide bases. We can measure the genetic diversity of a species by comparing the appearance of coloured bands between different species, or between species to show diversity within species. These techniques can also be used to determine the evolutionary relationships between species. When one species gives rise to another species during evolution, the DNA of the new species will initially be very similar to that of the species that gave rise to it. However, as time progresses, and mutations occurs, the sequences of nucleotide bases will become more and more different. As a result, species more closely related should show more similarity in their DNA base sequences than species less closely related.
3. The base sequence of mRNA is determined by the sequence of DNA, as they are complementary to that of the DNA strand that has made it. By comparing sequences of mRNA you are able to see genetic diversity.
4. Comparing amino acid sequences in proteins is useful as the amino acid sequence is determined by mRNA, which is in turn determined by DNA. The degree of similarity will also reflect the degree of similarity between different species.

Quantitative investigations of variation

Random sampling involves taking measurements of individuals, selected from the population of organisms which is being investigated. In theory, if these individuals are representative of the whole population then the measurements can be relied upon. However, they are not always representative, for reasons including:

- Sampling bias. The selection process may be biased, and the investigators may be making unrepresentative choices, either deliberately or unwittingly.
- Chance. Even if sampling bias is avoided, the individuals chosen may, by pure chance, not be representative.

The best way to prevent sampling bias is to eliminate, as far as possible, any human involvement in choosing the samples. This can be achieved by carrying out random sampling:

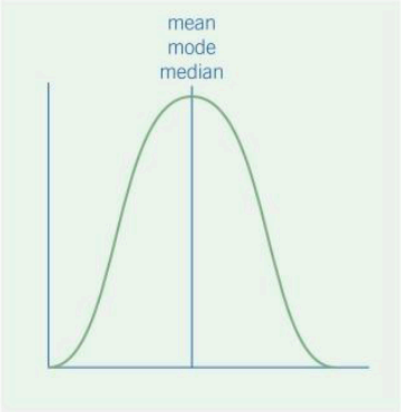
- Divide the study area into a grid of numbered lines, for example by stretching two long tape measures at right angles to each other
- Using random number generated from a table or by a computer to obtain a series of coordinates
- Take samples at the intersection of each pair of coordinates

We can only minimise the effect of chance from the sampling process by:

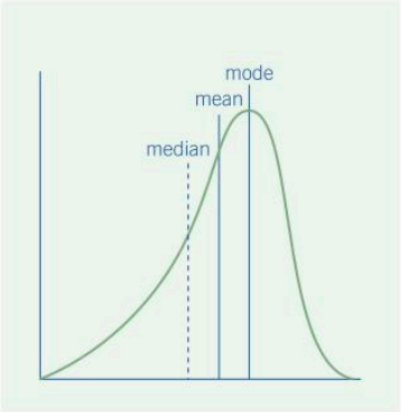
- Using a large sample size. The more individuals that are selected the smaller is the probability that chance will influence the result, also the less influence anomalies will have.
- Analysis of the data collected – this can be done using statistical test to determine the extent to which chance may have influenced the test (chi-squared test). These test allow us to decide whether any variation observed is the result of chance or is more likely to have some other cause.

The normal distribution curve usually shows a bell-shape for a continuous variation (ie height in humans). The graph is symmetrical about a central value. Occasionally the curve is shifted slightly to one side, called a skewed distribution. There are three terms related with normal distribution curves:

- The mean (arithmetic mean)
- The mode
- The median



▲ **Figure 2** *A normal distribution curve where the mean, mode and median have the same value*



▲ **Figure 3** *A skewed distribution where the mean, mode and median have different values*

The mean does not give useful information when comparing one sample with another. The standard deviation (s) is a measure of the width of the curve. It gives an indication of the range of values either side of the mean. To calculate standard deviation you use the equation below:

$$\text{standard deviation} = \sqrt{\frac{\sum (x - \bar{x})^2}{n - 1}}$$

Where:

$\sum$ = **the sum of**

x = **measured value (from the sample)**

$\bar{x}$ = **mean value**

n = **total number of values in the sample.**

References:

A Level Biology Textbook Kerboodle
https://micro.magnet.fsu.edu/cells/nucleus/images/nucleusfigure1.jpg
http://www.yourgenome.org/facts/what-is-mitosis
http://www.anatomy-diagram.info/digestive-tract-diagram/
http://academic.brooklyn.cuny.edu/biology/eckhardt/chap4.html
http://www.cancerresearchuk.org/about-cancer/cancers-in-general/treatment/biological/types/about-monoclonal-antibodies#M2CrhAJYAxwSBp1b.99
http://www.tokresource.org/tok_classes/biobiobio/biomenu/membranes/c8.7x17.transport.jpg