AQA Biology A-level

Topic 4: Genetic information, variation and relationships between organisms

Notes

DNA, genes and chromosomes

Both DNA and RNA carry information, for instance DNA holds genetic information whereas RNA then transfers this genetic information from DNA to ribosomes made of rRNA and proteins. Both deoxyribonucleic acids and ribonucleic acid are polymers of nucleotides. Nucleotides consist of pentose, a 5 carbon sugar, a nitrogen containing organic base and a phosphate group:

- The components of a DNA nucleotides are deoxyribose, a phosphate group and one of the organic bases adenine, cytosine, guanine or thymine. Adenine and guanine both have double ring structures and are classified as purine bases.
- The components of an RNA nucleotides are ribose, a phosphate group and one of the organic bases adenine, cytosine, guanine or uracil. Thymine, uracil and cytosine all have single ring structure and are classified as pyrimidines.
- Nucleotides join together by phosphodiester bonds formed in condensation reactions.

A DNA molecule is a double helix composed of two polynucleotides joined together by a hydrogen bonds between complementary bases whereas RNA is a relatively short polynucleotide chain.

Genetic code

The order of bases on DNA is called the genetic code which consists of triplets of bases, with each triplet of bases coding for a particular amino acids known as a codon. The amino acids are then joined together by peptide bonds and form a polypeptide chain. Therefore a gene is a sequence of bases on a DNA molecule coding for a sequence of amino acids in a polypeptide chain. The location of a gene is called the locus. However, not all the genome codes for proteins – the non-coding sections of DNA are called introns and the coding regions are called exons.

There are four nucleotide bases which code for 20 different amino acids. Scientists were able to conclude that each amnio acid is coded for by one or more combination of triplets. There are therefore 64 possible triplets (4^3) meaning that each amino acid is represented by more than one triplet.

Features of the genetic code:

- The genetic code is non-overlapping meaning that each triplet is only read once and triplets don't share any bases.
- Genes are separated by non-coding repeats of bases.
- Genetic code is degenerate meaning that more than one triplet codes for the same amino acid, this reduces the number of mutations which are mistakes in the base sequence such as base deletion, insertion or substitution. A change in the base sequence of DNA alters the amino acid sequence and the protein therefore it can have various effects. Some mutations are harmful such as the mutation which leads to production of sticky mucus and causes cystic fibrosis or sickle cell anaemia in which a mutated form of haemoglobin distorts the shape of red blood cells.
- The genetic codes contains start and stop codons.

Different types of DNA are found in eukaryotic and prokaryotic cells. In eukaryotic cells the DNA molecules are found in the nucleus and are long and linear. They are associated with proteins

called histones to form structures called chromosomes. Chromosomes are visible at the start of cell division and is the result of the DNA being tightly coiled around the histones. Finally in eukaryotic cells the mitochondria and chloroplasts contains DNA. In prokaryotic cells the DNA is short and circular and not associated with proteins.

Humans have 46 chromosomes in every cell in the body that are arranged into homologous pairs. A homologous pair consists of two chromosomes that carry the same genes. They are not identical as they can carry different alleles of the same gene. The 23rd chromosome determines the sex of the individual with female having two X chromosomes but males having and X and Y chromosome. An allele is described as an alternative form of the same gene, with every gene existing in two or more possible forms. Every individual inherits two allele of every gene from their parents.

DNA and protein synthesis

There are two stages of protein synthesis. Transcription which occurs in the nucleus and involves DNA and mRNA and translation which involves mRNA, tRNA and ribosomes. During transcription, a section of a DNA strand is transcribed into mRNA which is then translated into a polypeptide chain formed of amino acids.

Structure of mRNA and tRNA

Messenger RNA (mRNA) - this is a long single strand created during transcription in which the base sequence is complementary to DNA. Each set of three bases, called the codon, match a triplet on the DNA as well as the tRNA.

Transfer RNA (tRNA) - Transfer RNA is a small molecule made up of around 80 nucleotides. It is a single strand that is folded into a clover leaf shape in which one end extends from the others. Here an amino acids attaches. At the opposite end is an anti-codon which is specific to the amino acid that the tRNA attaches to.

Transcription:

During transcription, a molecule of mRNA is made in the nucleus:

1. The hydrogen bonds between the complementary bases break due to the action of an enzyme and the DNA uncoils thus separating the two strands exposing the organic bases.

2. One of the DNA strands is used as a template to make the mRNA molecule, the template is called the antisense strand.

3. Free nucleotides line up by complementary base pairing and adjacent nucleotides are joined by phosphodiester bonds thus forming a molecule of mRNA. The enzyme RNA polymerase catalyses the formation of phosphodiester bonds. When a stop codon is reached this ceases. As the RNA polymerase moves away the DNA rejoins, with only 12 bases being exposed at a time to reduce the chance of damage to the DNA.

4. In eukaryotic cells the pre-mRNA is then spliced to remove the introns leaving just a strand of exons. The mRNA then moves out of the nucleus through a pore and attaches to a ribosome in the cytoplasm which is the site of next stage of protein synthesis called translation.

Translation:

During translation amino acids join together to form a polypeptide chain:

1. mRNA attaches to a ribosome and transfer RNA collects amino acids from the cytoplasm and carries them to the ribosome. tRNA is a single stranded molecule with a binding site at one end thus it can only carry one type of amino acid, and a triplet of bases (anti-codon) at the other.

2. tRNA attaches itself to mRNA by complementary base pairing – two molecules attach to mRNA at a time.

3. The amino acids attached to two tRNA molecules join by a peptide bond and then tRNA molecules detach themselves from the amino acids, leaving them behind.

4. This process is repeated thus leading to the formation of a polypeptide chain until a stop codon is reached on mRNA and ends the process of protein synthesis.

A ribosomes can join up to 15 amino acids per second until the stop codon is reached. Furthermore up to 50 ribosomes can move along the same strand of mRNA behind one another so that several proteins can be assembled simultaneously.

Genetic Mutation

Changes in DNA can arise spontaneously during replication and any change in the base sequence or quantity of DNA is called a mutation. A change in the base sequence of a gene can change the sequence of amino acids. Sometimes this can result in a mutation that is harmful, however due to the genetic code being degenerate the amino acid sequence may not always be changed. There are two keys types of mutation, these are:

1. *Substitution* - this is when one nucleotide in the DNA sequence is replaced by another. The effect of the change in an amino acid depends on the role of the original amino acids in the overall shape and function of the protein. A substitution may not always be harmful as the substituted nucleotide may code in that triplet for the same amino acid.

2. *Deletion* - a deletion event is when a nucleotide in the DNA sequence is lost. The loss of a single nucleotide can have a significant impact as it leads to a frame shift, resulting in completely different amino acids being coded for.

Mutations may also occur in chromosomes for which there are two forms:

1. *Polyploidy* - changes can occur in the whole set of chromosomes so that an individual has three of more sets of chromosomes instead of two. This is common in plants, with many modern wheats arising to be polyploidy.

2. *Non-disjunction* - non-disjunction occurs when chromosomes fail to separate correctly in meiosis. As a result the gametes and any zygotes formed will have one more or one less chromosome than they should. Down's syndrome is the result of non-disjunction where individuals have an extra chromosome 21.

Meiosis and Genetic Variation

Meiosis is a form of cell division that gives rise to four daughter cells that are all **genetically different** and have **half the number of chromosomes** found in the parent cell. The main role of meiosis is the **production of haploid gametes.** as cells produced by meiosis have half the number of chromosomes. This is necessary to maintain a stable number of chromosomes. Meiosis produces genetically different cells, genetic variation is achieved through:

- **Independent assortment of chromosomes** – there are various combinations of chromosome arrangement. During meiosis 1 homologous chromosomes line up in pairs, the arrangement of these pairs is random, meaning that the division into the daughter cells is also random.
- **Crossing over of chromatids** - When pairs of chromosomes line up they can exchange some of their genetic material. Crossing over occurs when one chromosome may swap places with the same part of its homologous pair leading to a different combination of alleles on the gene.

Stages of Meiosis

The stages of meiosis can be split into two nuclear divisions, they are summarised below:

Meiosis 1 - homologous chromosomes pair up whereby crossing over at the **chiasmata** may take place. The cell then divides whereby each daughter cell contains one chromosome from each homologous pair.

Meiosis 2 - the chromatids of each chromosome are separated producing 4 haploid daughter cells.

Genetic diversity and adaptations

Genetic diversity is the **total number of different alleles in the population**. A **population** is defined as a group of individuals of the same species that live in the same place and are able to breed with one another. A species consists of more than one population. The greater the number of alleles in a population the greater the genetic diversity, and therefore the greater the chance that a population would survive a change in their habitat.

The **niche** of a species is **its role within the environment.** Species which share the same niche compete with each other. The idea that better adapted species survive is the basis of **natural selection**. **Natural selection** is the process in which **fitter individuals** who are betted adapted to the environment **survive and pass on the advantageous genes to future generations.** Evolution is the process by which the **frequency of alleles in a gene pool changes over time as a result of natural selection.**

Evolution via natural selection:

- There's a **variety of different phenotypes** within a population.
- An **environmental change occurs** and as a result of that the **selection pressure changes.**
- Some individuals possess **advantageous alleles which give them a selective advantage and allow them to survive and reproduce.**

- The advantageous alleles are passed on to their offspring.
- Over time, the frequency of alleles in a population changes and this leads to evolution.

Types of Selection

Selection is the process by which individuals that are better adapted to their environment are more likely to survive and breed. This means that they can pass on their advantageous alleles. Every living organism is subject to selection determined by the conditions which they are living in. There are two key types of selection, these are:

1. ***Directional Selection*** - directional selection occurs when the environmental conditions change and the phenotypes best suited to the new conditions are more likely to survive. As a result these individuals will breed and produce offspring. Overtime the mean of the population will move in the direction of these individuals. An example of this is bacteria being resistant to antibiotics. A single bacteria will have had a mutation that meant it was not killed by penicillin as it could produce the enzyme penicillinase. As a result it was able to grow and populate, and the frequency of the allele that enabled penicillinase production increased in the population. Therefore the population moved to have greater penicillin resistance.

2. ***Stabilising Selection*** - In stabilising selection the phenotypes with successful characteristics are preserved and those of greater diversity are reduced. This selection doesn't occur due to changes in the environment. If the environment stays the same then the individuals closest to the mean are favoured because they have the alleles that have given them the survival advantage. The furthest from the mean are selected against. A example is new born babies weights. Those that have a birth weight of around 3kg are more likely to survive than those at the extremes.

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Organisms are adapted to their environment in various ways:

- Anatomical adaptations are physical adaptations, either external or internal e.g. length of the Loop of Henlé - in desert mammals this is very long to produce concentrated urine and minimise water loss.
- Behavioural adaptations are changes in behaviour which improve the organism's chance of survival e.g. mating calls.
- Physiological adaptations are processes inside an organism's body that increase its chance of survival e.g. regulation of blood flow through the skin.

Species and taxonomy

Species are capable of breeding to produce living, offspring. We name species using the **binomial naming system**. Every species is given a name with two parts, based on Latin and Greek.

- The ***generic name*** is the genus to which the organism belongs. If a species is closely related it will share the same name
- The ***specific name*** is the species to which the organism belongs.

When hand writing these the names must be **underlined** and the **first letter of the generic name must be capitalised**, but not the first letter of the specific name.

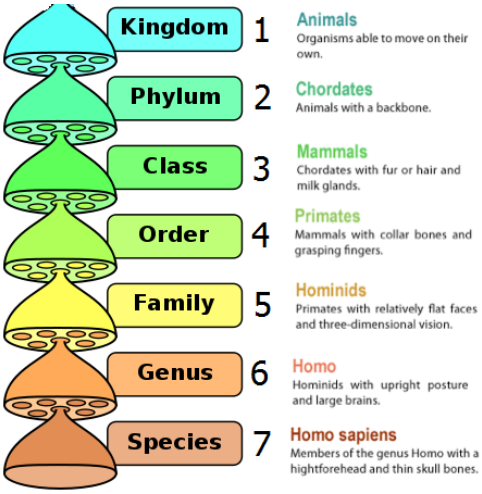
Courtship Behaviour

In order for members of the same species to recognise one another during mating **courtship behaviour** is often used. Members of the same species share similar physical, biochemical and behavioural characteristics. Courtship behaviour takes place before mating and enables individuals to:

- Recognise members of their own species
- Synchronise mating
- Form a pair bond
- Become able to breed

Classification

Classification is the process of **naming and organising organisms into groups based on their characteristics.** Organisms can be grouped into one of the five kingdoms: **animals, plants, fungi, prokaryotes and protoctists.** They can then be grouped further into **phylum, class, order, family, genus and species.**



The analysis of molecular differences in different organisms to determine the extent of their evolutionary relatedness is known as **molecular phylogeny**. The data obtained by molecular phylogeny has been accepted by scientists and this gave rise to new taxonomic groupings – all organisms can be separated into one of the **three domains: Bacteria, Archaea and Eukaryota.**

The scientific community evaluates the data in the following ways:

- The findings are published **in scientific journals** and presented **scientific conferences.**
- Scientists then study the evidence in a process called **peer review**
- Scientists start **collecting evidence** to either **support or reject the suggestion**

$$D = \frac{N(N-1)}{\sum n(n-1)}$$

Biodiversity within a community

Biodiversity is the **variety of living organisms.** Over time the variety of life on Earth has become more extensive but now it is being **threatened by human activity** such as **deforestation**. Biodiversity can be measured in terms of:

- **Species diversity** is the **number of different species and the number of different individuals in a community**. It can be measured by simply counting the number of species present via methods such as **random sampling.**
- **Genetic diversity** is a measure of the **genetic variation** found in a particular species, in other words it is **the number of alleles in a gene pool.**
- **Ecosystem diversity** is the range of different habitats.

Biodiversity can also be measured using the **index of diversity (D)** which can be calculated as following:

D = Diversity index
N = total number of organisms
n = total number of organisms of each species
Σ = the sum of

Impacts of Agriculture

Agricultural ecosystems reduce the biodiversity and the number of species present because **humans select for particular characteristics**. This therefore **reduces the number of alleles** and therefore the genetic diversity of the population. Farmland is typically used for only a single species and therefore means that fewer individuals of other species can survive there.

In order to maintain biodiversity a number of techniques have been used, some of these include:

- Using **hedgerows** instead of fences.
- Growing different crops in the same area, known as **intercropping.**
- Reducing the use of **herbicides and pesticides.**
- **Preserving wetlands** instead of draining these for farming use

Comparing Genetic Diversity

The **genetic diversity** within or between species can be determined by comparing the **frequency of measurable or observable characteristics**, the **base sequence of DNA**, the base sequence of **mRNA** and the **amino acid sequence** of the proteins encoded by DNA and mRNA.

Comparing observable characteristics like the **shape or size** used to be one way in which scientists classified species. However this has many limitations such as:

- The characteristics could be **coded for by more than one gene.**
- The **same characteristics could have arisen separately.**
- The characteristics could be **influenced by the environment** and not the genes.

Given the fact that this is a poor way of classifying species scientist now use **gene technology** to read the base sequences of organisms. The genetic diversity of a species can be measured by sampling DNA or mRNA from different individuals.

The **sequence of amino acids** can also be studied as this can be taken back to the mRNA sequence and thus the DNA sequence. The differences between species are called **interspecific variation**. There are also differences between individuals of the same species which is known as **intraspecific variation**.

In order to measure characteristics in species scientist cannot measure each individual, therefore **sampling** is used. Sampling is the random selection of individuals to measure, however it may not be representative of the population due to **chance** and **sampling bias**.

To eliminate sampling bias a **quadrat** or **transect** can be used by placing these on randomly generated coordinates in the sample area. To increase the reliability of the results a **larger sample size** is needed.