WJEC (England) Biology GCSE

Topic 7: Inheritance, Variation and Evolution

Notes

('Higher Tier only' in **bold**)

7.1 The genome and gene expression

Key terms

- Genome - the entire genetic material of an organism.
- Chromosomes - The structures that the long strands of DNA are compacted into.
- Gene - a length of DNA that codes for a particular protein.
- Allele - one of a number of different versions of a gene.
- Genotype - the genetic makeup of an organism, consisting of all the alleles present.

DNA structure

DNA stands for deoxyribonucleic acid. DNA is a polymer made up of nucleotide monomers bound together in a chain. Nucleotides form two long strands, which wind around each other and bind together to form a double helix. There are four types of nucleotide. Each one is made from a common sugar, a phosphate group and one of four different bases.

The four different bases are: adenine (A), thymine (T), cytosine (C) and guanine (G). When the two complementary strands of DNA bind together, A always binds to T, and C always binds to G. The order of these bases in DNA is very important for protein synthesis.

Protein synthesis

Proteins are extremely important in cells as they maintain structure, as well as carry out reactions and transport molecules. Protein synthesis is the process of making proteins from amino acids.

The main steps of protein synthesis are:

1. **A messenger RNA (mRNA) strand is made from a section of DNA which holds the code to a particular gene. mRNA is similar to DNA and is also made of nucleotides (although different in chemical structure). This occurs in the nucleus. A mRNA copy is necessary as the DNA is too large to leave the nucleus.**
2. **The mRNA exits the nucleus and travels through the cytoplasm to a ribosome, which is the site of protein synthesis.**
3. **The ribosome uses the mRNA strand as instructions to join amino acids together according to their specific order, which produces a particular protein. Each amino acid is coded by three nucleotides, thus it is referred to as a triplet code.**

The order of nucleotides in the original DNA strand will affect the structure of the mRNA strand, which will affect the sequence of amino acids. These changes therefore will affect the final structure of the protein. Different sections of DNA (genes) are used to code for different proteins which have a range of functions.

Genes can occur in different forms (alleles) which each have a slightly different DNA base sequence. When protein synthesis occurs, different proteins can be made due to this which can affect the characteristics of the organism, such as eye colour or blood group.

Non-coding DNA

Not all DNA is used to code for proteins: a large amount of DNA is not found in genes and hence is not used in protein synthesis. This DNA is therefore known as non-coding DNA. The role of these sections of DNA is to regulate when protein synthesis occurs by controlling which genes are 'switched on' and 'switched off'. When a gene is switched on, it becomes active and can be transcribed and then translated into a protein, whereas when it is switched off it is inactive. This is important as not every protein is needed by every cell at all times.

Genetic profiling

Genetic profiling is the process of analysing an organism's DNA to see the DNA base sequence. This can be compared to other DNA profiles to see the evolutionary closeness between two organisms, as although every organism's DNA sequence is unique, two organisms closely related will have more similarities in their DNA than two organisms distantly related. This can therefore be used in classifying organisms and seeing evolutionary relationships.

Producing a genetic profile:

1. A sample is taken from the organism.
2. DNA is isolated from other organic material and is cut into short pieces using an enzyme.
3. These DNA pieces are separated into bands using electrophoresis. This is done by placing the DNA into a gel and passing an electric current through the sample, causing the DNA fragments to move into different bands based on size and charge.
4. The pattern of bands is compared to other organisms to show similarities.

This method is commonly used in forensic investigations and in paternity tests, as well as to study evolutionary relationships between organisms.

Genetic profiling and the increased knowledge about the genome play an important role in the development of new medicines and the detection of disease:

- Knowledge of how DNA and proteins function allow earlier diagnosis of diseases as it is more obvious when they are not working correctly.
- Mutations in the genome and their effects can be identified.
- Knowing how diseases and mutations interact with the genome can allow medicines which counteract this to be developed more effectively.
- The genomes and proteins of pathogens can also be studied in detail to find potential medicines that target them specifically.

7.2 Inheritance

Key Terms:

- Inheritance - the transmission of genetic information from parents to offspring.
- Chromosome -The structures that the long strands of DNA are compacted into.
- Gene - a length of DNA that codes for a particular protein.
- Allele - one of a number of different versions of a gene.
- Genotype - the genetic makeup of an organism, consisting of all the alleles present.
- Phenotype - the observable features of an organism as a result of the expression of particular alleles of the gene.
- Homozygous - an organism containing two identical alleles of a particular gene.
- Heterozygous - an organism containing two different alleles of a particular gene.
- Gamete - a reproductive cell which carries one half of each chromosome pair. During fertilisation, two gametes fuse to create a full set of chromosomes. Egg and sperm cells are examples of gametes.

Genes and alleles

Offspring inherit characteristics from both their mother and father and two sets of genes are inherited, one from each. If the mother and father both pass down the same allele for a particular trait, e.g. they both pass down the allele for blue eyes, the offspring will have two identical alleles for this trait, which is referred to as homozygous. If two separate alleles are passed down, e.g. the mother has blue eyes and the father has brown eyes, the offspring will have two different alleles for the gene, which is called heterozygous.

Alleles are different versions of the same gene, common examples being eye colour and blood group. Alleles can be either dominant or recessive. A dominant allele is always expressed if present, whereas the recessive allele is only expressed in the absence of the dominant allele. For example, the allele for brown eyes is dominant and the allele for blue eyes is recessive. This means that if two parent organisms with blue and brown eyes were bred, the offspring would have brown eyes.

Sex determination

There are 23 pairs of chromosomes in the nucleus of all diploid human cells. One of these pairs determines gender. These chromosomes are therefore called the sex chromosomes. For females these chromosomes are XX and for males the chromosomes are XY. Gametes are haploid cells which only contain one half of each chromosome pair, meaning that each egg or sperm cell will only have half of the sex chromosomes: all female egg cells contain only an X chromosome, whereas male sperm cells may contain an X or a Y. Thus, the gender of the baby depends on which sperm cell fertilises the egg cell.

Monohybrid crosses

Most characteristics of an organism are determined by multiple genes interacting, however some are determined by a single gene. This is called single gene, or monohybrid, inheritance. Monohybrid crosses show this and are used to predict the ratios of inherited characteristics in a population. There are always four outcomes. The dominant allele is written as a capital letter and the recessive as the lowercase of that letter.

Parent one → Parent two ↓	**G**	**g**
G	GG	Gg
g	Gg	gg

E.g.1) Crossing two heterozygous green and yellow pea plants (both parents contain the alleles for both traits):

G= green (dominant allele) g= yellow (recessive allele)

Genotypes: The outcomes are GG, Gg, Gg and gg.

Phenotypes: As G is dominant, there is a 75% chance that the offspring will display this allele in the phenotype and be green. There is a 25% chance that the offspring will be yellow therefore the ratio is 3:1.

E.g.

2) Crossing a homozygous recessive (yellow) pea plant with a heterozygous pea plant:

Parent one (homozygous) → Parent two (heterozygous) ↓	**g**	**g**
G	Gg	Gg
g	gg	gg

G= green (dominant allele) g= yellow (recessive allele)

Genotypes: The outcomes are Gg, Gg, gg and gg.

Phenotypes: There is a 50% chance of the offspring being green or yellow, therefore the ratio is 1:1.

The development of genetics

Genetic inheritance was first explored by Gregor Mendel in 1866, who discovered the basic principle of genetics through experimenting with pea plants. He crossed different types of pea plants which had different characteristics, such as flower colour and seed shape, and found a correlation in characteristics between the parents and offspring. This allowed him to create the first genetic crosses and coin the terms ‘dominant’ and ‘recessive’. He did not, however, understand what caused these results. Consequently, when he first published his findings, his work was not seen as significant and was even criticised by other scientists. It was not until the 20th century, when other scientists verified his results, that his work became recognised. Now, better microscopes and technology have proved that Mendel was correct.

7.3 Variation and evolution

Sexual reproduction

Sexual reproduction requires two parents. During sexual reproduction, the nuclei of two gametes, one from each parent, fuse together during fertilisation to form a genetically unique zygote.

In humans, a sperm cell and an egg cell fuse their nuclei together to form a zygote. Sexual reproduction in plants takes place in the flowers using pollen. Cross-pollination occurs when grains of pollen are transferred from the anthers of one plant to the stigma of another plant of the same species. Some flowers can produce male and female gametes, and thus can self-pollinate by transferring pollen from their own anther to their own stigma.

Advantages of sexual reproduction:

- Wide diversity - the offspring are likely to have a wider range of characteristics.
- Promotes survival - if the population is diverse in terms of characteristics then it is likely that some individuals will be able to survive if the environment changes or if the population is hit by a disease.
- Organisms can adapt - as each offspring is born with different genes, those with a genetic advantage are more likely to survive and pass their positive traits on to their offspring, whilst those with a genetic disadvantage are more likely to die without producing offspring. This allows the species to evolve through natural selection.

Disadvantages of sexual reproduction:

- Two parents are required - it may be difficult for some species to find mates, especially when there is an imbalance of males and females in an area or if the species is endangered.
- Fewer offspring produced - it takes longer and requires more energy to produce offspring, therefore it is less efficient than asexual reproduction.

Asexual reproduction

Asexual reproduction is a process which results in the production of genetically identical offspring, known as clones. This only requires one parent, unlike sexual reproduction. Asexual reproduction occurs predominantly in plants, although some animals such as starfish also reproduce in this way.

Advantages of asexual reproduction:

- Only one parent is required - this is helpful for organisms which live in desolate environments where finding a mate is difficult.
- Can reproduce quickly - large quantities of offspring can be produced quickly to rapidly populate an area. This helps to dominate a habitat and prevent competition from other species.
- It takes less energy to reproduce asexually.

Disadvantages of asexual reproduction:

- Lack of diversity - all offspring are genetically identical.
- Prone to extinction - as each organism produced is genetically identical, a disease which harms one will be dangerous to all of them, thus is easy for the whole population to be destroyed by one pathogen.
- Cannot adapt - organisms are adapted to one environment and cannot adapt to changes. If the environment changes, e.g. the temperature rises, they are likely to be killed.
- Overpopulation - too many offspring may be produced, which causes overcrowding in a habitat.

Variation

Variation refers to the differences between each organism in a species. There is usually an extensive genetic variation in each species, which is beneficial as it allows natural selection to occur and reduces the risk of extinction from a change in the environment or disease. There are two types of variation, genetic variation and phenotypic variation:

- Genetic variation - each organism in a species has a unique set of DNA, which is due to genetic variation. Each gamete has a different set of alleles, which means that when the two gametes fuse an entirely new combination of genes are produced.

- Phenotypic variation - The phenotype of an organism refers to its observable characteristics, such as height or hair colour. Phenotypical variation can be caused by both genetic and environmental factors. For example, the potential height and weight of an organism is determined by its genes which come from the parents, although some organisms will never reach this height and weight as they do not receive enough nutrients from their environment.

Gene mutation

Mutations are genetic changes which result in a change in the sequence of DNA bases. These changes can be caused by a variety of factors, including exposure to some chemicals and ionising radiation. If the mutation occurs at a particular allele, this allele may be altered, changing how it functions. This is how new alleles are formed. New alleles lead to genetic variation.

Evolution and natural selection

Natural selection is where organisms with **favourable alleles** and **advantageous inherited** characteristics have a higher probability of surviving and reproducing. This is due to competition within a population for resources and mates. As there is variation in the alleles of each species, each organism within a species has different inherited characteristics, some positive and some negative. Those with more positive traits can adapt to the environment more effectively and are thus more likely to survive and produce many offspring, which inherit these advantageous alleles. Over time, negative characteristics are lost from the species as organisms with those characteristics are less likely to reproduce to pass on their genes. Evolution is the process that

allows a population to change over time and can result in new species forming. Sometimes, environmental conditions change too rapidly for natural selection to take place, leading to extinction.

Evidence for evolution

- A mutation in rats has made them resistant to warfarin, a common rat poison. As these rats are no longer killed by the poison, they have a higher chance of surviving and reproducing to pass on the mutated gene. Consequently, the amount of resistance to warfarin is increasing. This shows that evolution is ongoing.

- Some bacterial strains have become resistant to antibiotics as a result of natural selection. As bacteria reproduce much more quickly than animals, evolution and natural selection in bacterial strains can be observed and monitored more easily.

- Fossils - the presence of fossils supports the theory of evolution since fossils found in the oldest rocks contain the simplest organisms, whereas later fossils are made up of more complex organisms, proving that organisms have evolved to become more complex. The differences between the organisms present in fossils and living organisms also show that organisms have changed and evolved over time.

Classification of organisms

The classification of organisms is known as taxonomy. Organisms are classified into similar categories to make them easier to study and to compare similar characteristics. The classification system splits organisms into a hierarchy of different levels called taxa:

Domain --> Kingdom --> Phylum --> Class --> Order --> Family --> Genus --> Species

Each level becomes more specific, with a large number of organisms in the first taxa and a small amount in the last taxa.

This system was first created in the 18th century, with the exception of the three domains (archaea, bacteria and eukaryotes), which were added in 1977. When humans first began to classify organisms, technology was very limited which meant that animals were not originally classified using DNA. Consequently, organisms were placed into categories based on physical or behavioural similarities. This method was flawed, however, as many animals that looked similar were very different genetically. For example, although hares and rabbits look alike, they cannot interbreed and have a different number of chromosomes. Organisms that live in the same habitat often adapt via natural selection to appear alike as it is beneficial to survival in that environment, leading to errors when classifying these organisms.

Today, the same classification hierarchy is still used, although many changes have been made due to the advancement of technology and the improvement of immunology and genome sequencing, which has allowed genetic similarities between organisms to be found. For example, the 'domain' level of classification depends on differences in RNA between organisms. This has helped to clarify evolutionary relationships between different organisms and reduces the errors from classifying organisms using physical characteristics alone, since organisms in the same group will have a very

similar genome. Genome sequencing also shows if two different groups of organisms have a common ancestor, and how recently speciation occurred.

The development of evolution

The theory of evolution by natural selection was first developed in the mid 19th century by a number of scientists: Most famously Charles Darwin but also Alfred Russel Wallace and Jean-Baptiste Lamarck. Darwin's evolution theory was based on a number of observations from studying fossils, selective breeding and similarities in anatomy between closely related species. With modern technology, even more evidence has been found to support the theory of evolution, such as bacterial antibiotic resistance.

7.4 Selective breeding and gene technology

Selective breeding

Selective breeding is where humans select animals or plants with desirable features and breed them together to make offspring which possess these desirable features. This process is repeated over many generations to promote beneficial characteristics and remove negative characteristics. As this breeding is controlled by humans, it is known as artificial selection.

An example of selective breeding in animals is the German Shepherd. These dogs were originally bred as working dogs to herd sheep as they are known for their intelligence and agility. Humans selectively breed these dogs to exaggerate desirable qualities, such as their sloping backs and large ears. This involves crossing dogs which show these traits so that the alleles are passed on to their offspring. These dogs now look very different to how they did 100 years ago.

Farmers also selectively breed crops. For example, bananas are selectively bred for their size, shape and easiness to peel. This means that plants which express these characteristics are chosen and bred to produce more offspring with these characteristics.

Genetic engineering and gene technology

Genetic engineering is the process of artificially altering genes in a cell to change the way it works. This could be to make the cell perform a desired function, such as making a specific protein, or to make the cell resistant to different factors. Being able to manipulate genes means that organisms can be given desirable characteristics immediately without the need for selective breeding, which can take many years to produce results.

The main steps in genetic engineering are:

1. **The desired gene is located in the DNA of an organism.**
2. **This gene is isolated and removed from the organism using enzymes.**
3. **The gene is replicated and placed into a vector, e.g a plasmid.**
4. **The gene is inserted into a bacterium cell using the vector.**
5. **Successfully modified cells are selected.**

If the gene codes for a specific protein, such as insulin, this can be harvested from the bacteria. The gene may also be inserted into another organism, such as a crop plant, so that it displays the desired characteristics.

Benefits of genetic engineering and gene technology:

- Crops can be modified to improve yield by becoming resistant to frost, diseases and herbicides. E.g. Some strawberries have been genetically engineered to become frost-resistant by inserting an anti-freeze gene found in arctic fish.

- Crops can be modified to provide scarce nutrients. E.g. Golden rice is a genetically modified variety of rice that produces beta-carotene, which is converted into vitamin A in the body. Areas which lack dietary vitamin A grow this rice to prevent vitamin A deficiency,

which can lead to blindness and death.

- Genetic testing can be used to inform individuals about family planning and healthcare since genetic screening can show any diseases they could have and determine whether the individual is a carrier of a disease which may be inherited by their children.

Risks of genetic engineering and gene technology:

- Genetically modified crops can have a number of negative impacts on the environment, including loss of biodiversity, contamination of wild species through crossbreeding, and leading to herbicide-resistant weeds. Genetically modified crops are also more expensive.
- Long term health impacts of consuming genetically modified food products are not known.
- Genetic testing means that embryos can be screened to find out their characteristics or if they have any inherited diseases or disabilities. This could lead to 'designer babies' and is a highly contentious topic.