OCR (B) Biology GCSE

Topic B1: You and your genes

Notes

(Paragraphs in **bold** are higher tier only)

What is the genome and what does it do?

Key Terms:

- **Genome** - the entire genetic material of an organism.
- **Chromosome** - a thread-like structure of DNA that carries genetic information in the form of genes.
- **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 and interaction with the environment.

Eukaryotes and Prokaryotes

Cells can be viewed using a **light microscope** to study their structure. Plant and animal cells are known as **eukaryotic** cells as they contain a nucleus and membrane bound **organelles** such as ribosomes and mitochondria. Bacteria are known as **prokaryotes** as they do not contain these. Genetic material in prokaryotes is stored in **plasmids.** Plasmids are **circular loops** of DNA that are found in the **cytoplasm** of the cell. In eukaryotes, DNA is stored in the **nucleus**.

Characteristics

Most characteristics of an organism, such as eye colour and blood group, are determined by the **genome**. **Alleles** are different versions of a gene which produce different characteristics. Some alleles are **dominant**, meaning that they will be **expressed** if present in the genome. Characteristics can also be influenced by **external factors** as the organism interacts with the environment. For example, skin colour is influenced by the intensity of sunlight.

DNA

DNA stands for **deoxyribonucleic acid**. It acts like instructions, allowing the cell to carry out a variety of key processes. For example, DNA allows **protein synthesis**, where **amino acids** are bonded together to make proteins. Proteins are extremely important in cells as they maintain **structure**, as well as carry out **reactions** and **transport** molecules.

DNA is a **polymer** made up of many **nucleotide** monomers bound together in a chain. Nucleotides form **two** long strands, which wind around each other to form a **double helix**.

DNA (Biology only)

There are four types of nucleotides. Each are made from a **common sugar**, a **phosphate group** and one of four different **bases.**

Protein Synthesis (Higher/Biology only)

Protein synthesis is the process of making proteins from amino acids. Each amino acid is coded for by three nucleotides on a DNA strand.

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. This occurs in the nucleus. A mRNA copy is necessary as 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 in a specific order, which produces a particular protein.**

Mutations (Biology only)
Mutations are genetic changes which result in a change in the sequence of DNA bases (nucleotides). These changes can occur due to a variety of factors, including exposure to some chemicals and ionising radiation. If the mutation occurs at a particular allele in coding DNA, this allele may be altered, changing how it functions. This is how new alleles are formed, resulting in genetic variation, as altered alleles produce different proteins which influence the phenotype. Non-coding DNA can also be mutated and can alter the phenotype by altering how genes are expressed.

How is genetic information inherited?

Key Terms:

- **Inheritance** - the transmission of genetic information from parents to offspring.
- **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.
- **Homozygous** - an organism containing two identical alleles of a particular gene.
- **Heterozygous** - an organism containing two different alleles of a particular gene.
- **Dominant allele** – a dominant allele, if present in an organism, is always expressed in the phenotype and will mask recessive alleles.
- **Recessive allele** – recessive alleles are only expressed in the phenotype in the absence of a dominant allele.

Single gene inheritance
Offspring **inherit characteristics** from both their mother and father. **Two sets of genes are inherited**, one from each. If the mother and father 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**. If two **identical homozygous** individuals are bred together, the offspring will have the **same characteristics as the parents**.

Alleles can be **dominant** and **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 parents with blue and brown eyes were bred, the offspring would have brown eyes.

Most characteristics, however, are determined by **multiple genes**, meaning that most characteristics are not passed on through single gene inheritance.

Single gene crosses

Single gene crosses 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.

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)

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

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

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

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

The outcomes are Gg, Gg, gg and gg. There is a 50% chance of the offspring being green or yellow, therefore the ratio is **1:1**.

Sex determination

All humans have **23 pairs of chromosomes** in their DNA. One of these pairs is used to determine gender and they are therefore called the **sex chromosomes**. The sex chromosomes in **males are called XY**, whereas the two in **females are called XX**.

When **gametes** (egg and sperm cells) are produced, they carry one half of each chromosome pair. **Egg cells from the mother always carry X chromosomes**, however **sperm cells may carry a X or a Y chromosome**. This means that there is a 50% chance of producing either male or female offspring as it is random which sperm cell will fertilise the egg cell.

Sex inheritance can be displayed on a genetic cross, which shows that there is a 50% chance of the offspring being male or female:

Mother → Father ↓	**X**	**X**
X	XX	XX
Y	XY	XY

Development of genetics (Biology only)

Genetic inheritance was first explored by **Gregor Mendel** in 1866, who discovered the basic principal 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 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.

Since then more research has been carried out, and due to the advancement of technology and research from the Human Genome Project, these genes have now been mapped, allowing researchers to see how they function. This has a range of benefits, including identification of mutations and their effects, improvement of treatments for a range of diseases, and allowing advancement in a number of fields, including biofuels, agriculture and molecular medicine.

How can and should gene technology be used?

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**.

Genetic engineering

1. **The desired gene is located in the DNA of an organism. This gene is isolated and removed from the organism using enzymes.**
2. **The gene is replicated and placed into a vector, e.g a plasmid.**
3. **The gene is inserted into a bacterium cell using the vector.**
4. **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 to genetic engineering:

- 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 artic 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.

- Knowledge of the human genome can help **improve medicines and vaccines** as specific genes and alleles associated with a particular disease can be targeted.

- **Genetic testing** can be used to inform individuals **about family planning and healthcare** as 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:

- 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.