Edexcel Biology GCSE

Topic 3: Genetics

Notes

(Content in **bold** is for higher tier only)

CONTENTS

3.1B and 3.2B - Sexual and Asexual Reproduction

1. **Sexual reproduction** involves the joining of male and female **gametes**, each containing genetic information from the mother or father.
    - Sperm and egg cells in animals
    - Pollen and egg cells in flowering plants

Gametes are formed by meiosis, as they are non identical.
A normal cell has **46 chromosomes**. There are two sets of chromosomes (i.e. 23 pairs). In each pair, one chromosome is from the father and the second set are from the mother.
Each gamete has 23 chromosomes and they fuse in **fertilisation**.
The genetic information from each parent is mixed, producing variation in the offspring.

2. **Asexual reproduction** involves one parent with no gametes joining.
   It happens using the process of mitosis, where two identical cells are formed from one cell.
   There is no mixing of genetic information.
   It leads to clones, which are genetically identical to each other and the parent.
   Examples of organisms that reproduce this way are bacteria, some plants and some animals.

Advantages of sexual reproduction	Advantages of asexual reproduction
Produces **variation** in offspring. • This means that if the environment changes it is likely that an organism in the species will have a characteristic that allows them to survive (called a **survival advantage**). • Although some individuals may die, variation decreases the chance of the whole species becoming **extinct**.	Only one parent is needed.
It allows us to use **selective breeding**. • This type of reproduction mixes the genetic information from two organisms • Organisms with different **desirable characteristics** can be bred to produce offspring with even more desirable characteristics.	Uses less energy and is faster as organisms do not need to find a mate.

• This speeds up **natural selection**. • An example is to increase food production by breeding two animals with lots of meat.	
	In favorable conditions lots of identical offspring can be produced.

3.3 - The Importance of Meiosis

While mitosis is used for the division of ordinary body cells to produce diploid (2n) daughter cells, **meiosis** is used to produce haploid gametes (sperm and egg cells). Gametes only have one copy of each chromosome.

Meiosis is the formation of four non-identical cells from one cell.

- The cell **makes copies** of its chromosomes, so it has double the amount of genetic information.
- The cell divides into two cells, each with half the amount of chromosomes **(46)**.
- The cell divides again producing four cells, each with a quarter the amount of chromosomes **(23)**.
- These cells are called gametes and they are all **genetically different** from each other because the chromosomes are shuffled during the process, resulting in random chromosomes ending up in each of the four cells.

These gametes with 23 chromosomes join at fertilisation to produce a cell with 46 chromosomes, the normal number.

- This cell divides by mitosis to produce many copies.
- More and more cells are produced, and an **embryo** forms.
- The cells begin to take on different roles after this stage (**differentiation**).

3.4, 3.5, 3.6 - The Structure of DNA, The Genome and Extracting DNA

DNA, found in the nucleus, is a chemical that contains genetic material.

DNA stands for **deoxyribonucleic acid**, and this is a polymer that contains instructions for the body.

- It is made up of many small parts called **nucleotides**.
- Each nucleotide is made up of **one sugar molecule**, **one phosphate molecule** (which form the backbone) and **one of the four types of organic bases**.
- The four types of organic bases are **A, C, G, T**.

- Each DNA molecule is made up of **two DNA strands** which are twisted together. Each base is connected to another base in the other strand.
- A bases only connect to T bases, and C bases only connect to G bases. This is called **complementary base pairing**.
- The order of the different bases forms a **genetic code** - e.g. A, G, T, T, C, A, A etc.

DNA is a **polymer** (long molecule) made up of two strands which are wound around each other to form a structure called a **double helix**.

A **gene** is a short section of DNA. Each gene codes for many **amino acids**, which are joined together to make a specific **protein**.

- There are 20 types of amino acid.

The word **genome** describes all the genetic information (DNA) of a single organism. The human genome has been studied, which has improved our understanding of the genes linked to different types of disease, the treatment of inherited disorders and has helped in tracing human migration patterns from the past.

Extracting DNA from Fruit

It is possible to extract DNA from fruit using household ingredients:

- Gently mix together 50ml cold water, half a teaspoon of salt and 10ml washing up liquid. Gently heat this mixture at 50C for 5-10 minutes.
- Peel the skins of a kiwi and chop into small pieces. Pulverise the kiwis.
- Add the solution from Step 1 to the kiwi.
- Filter the solution using a few sheets of kitchen paper and a sieve. Pour the filtrate into a test tube.
- Add 10ml of pineapple juice to the filtrate and allow to rest for a few minutes.
- Add 2 teaspoons of cold ethanol to the solution and wait 10 minutes.

What should we see?

A white mass should precipitate at the top of the tube after 10 minutes; this is the DNA from the kiwi.

Why do we add pineapple juice?

Pineapple juice contains an enzyme called **bromelain** which breaks down proteins attached to the DNA. This helps us see the DNA more clearly.

Why do we add ethanol?

Ethanol causes the DNA to precipitate out of the solution - making it visible at the top of the container.

3.7B and 3.8B **Biology and Higher Only** - The Stages of Protein Synthesis

- Each group of three bases (e.g ACT, AGG, GAC) codes for an amino acid.
- The amino acids are joined together and fold to make a protein. It is the different types and order of amino acids that determine which type of protein it is. Often these proteins are enzymes, which need to have a very specific shape.
- Therefore it is the order of bases in DNA that determine which proteins are produced.

There are also non-coding parts of DNA that do not code for proteins. Some of them are responsible for switching genes on or off, i.e. controlling whether the gene is used to form a protein or not.

Protein synthesis: the process of producing a protein from DNA

If a gene is coded to make a protein, it has been expressed.

1. DNA contains the genetic code for making a protein, but it cannot move out of the nucleus as it is too big.
2. The mRNA nucleotides themselves are then joined together, creating a new strand called the mRNA strand. This is a template of the original DNA.
3. An enzyme called RNA polymerase binds to non-coding DNA located in front of a gene on the DNA strand.
4. The two strands of DNA pull apart from each other, and RNA polymerase allows mRNA nucleotides (messenger RNA: a different type of nucleotide) to match to their complementary base on the strand.
5. The mRNA then moves out of the nucleus to the cytoplasm and onto structures called ribosomes.
6. At the ribosomes, the bases on the mRNA are read in threes (triplets) to code for an amino acid (the first three bases code for one amino acid, the second three bases code for another etc).
7. The corresponding amino acids are brought to the ribosomes by carrier molecules called tRNAs - transport RNAs.
8. These amino acids connect together to form a polypeptide (amino acids linked by peptide bonds).
9. When the chain is complete the protein folds to form a unique 3D structure, which is the final protein.

3.9B and 3.10B **Biology and Higher Only** - Genetic Variants and their Effects

Genetic variants are small changes in the order of bases that make up a strand of DNA. They can affect the structure of proteins in different ways, depending on whether they occur in coding DNA or non-coding DNA.

Genotype refers to the genes present in the DNA of an individual, whereas phenotype refers to the visible effects of those genes (e.g the proteins that they code for).

Coding DNA: A genetic variant will alter the sequence of bases (e.g ACT -> AGT) and therefore will change the sequence of amino acids (e.g Glycine -> Valine). This alters the final structure of the protein produced.

Non-Coding DNA: A genetic variant in the coding DNA can affect phenotype differently. The enzyme RNA polymerase (see Section 3.8B) binds to non-coding DNA, and a change in the order of bases in this non-coding DNA can affect the amount of RNA polymerase that can bind to it. If less RNA polymerase is able to bind, less mRNA can be formed and the structure of the final protein is affected.

Mutations change the sequences of bases in DNA. Either:

1. **A base is inserted into the code**
    - **As they are read in threes, this changes the way it is read.**
    - **It may change all the amino acids coded for after this insertion.**
2. **A base is deleted from the code**
    - **Like insertions they change the way it is read.**
    - **It may change all the amino acids coded for after this deletion.**
3. **A base is substituted**
    - **This will only change one amino acid in the sequence or it may not change the amino acid (as the new sequence can sometimes still code for the same amino acid)**

A change in the type/sequence of amino acids will affect the way it folds and therefore the structure.

Most mutations do not alter the protein or only do so slightly.

Some can have a serious effect and can change the shape

- **The substrate will not fit into the active site so it cannot act as a protein.**
- **A structural protein may lose its shape.**

There can also be mutations in the non-coding parts of DNA that control whether the genes are expressed.

3.11B **Biology Only** - Mendelian Genetics

Gregor Mendel

- Trained in mathematics and natural history in Vienna
- Worked in the monastery gardens and observed the characteristics passed on to the next generation in plants
- He carried out breeding experiments on pea plants.
- He used smooth peas, wrinkled peas, green peas and yellow peas and observed the offspring to see which characteristics they had inherited
- Through keeping a record of everything he did and eventually publishing his work in 1866, he came to these conclusions:
    - Offspring have some characteristics that their parents have because they inherit '**hereditary units**' from each.
    - One unit is received from each parent.
    - Units can be dominant or recessive, and cannot be mixed together.

Mendel was not recognised till after his death as genes and chromosomes were not yet discovered, so people could not understand.

- In the late 19th century chromosomes as a part of cell division were observed
- In the 20th century, it was understood that chromosomes and units had similar behaviours. It was decided that units (now known as genes) were on the chromosomes.
- The structure of DNA was determined in 1953, which meant we were able to understand how genes worked.

3.12 and 3.12 - Alleles and Basic Definitions

You need to know the definitions for a number of terms:

Gamete	An organism's reproductive cell (egg in female and sperm in males), which has half the number of chromosomes (23).
Chromosome	A structure found in the nucleus which is made up of a long strand of DNA.
Gene	A short section of DNA that codes for a protein, and therefore contribute to a characteristic. Some characteristics are controlled by a single gene, such as fur colour in mice and red-green colour blindness in humans. However, most characteristics are the result of many different genes interacting.
Alleles	The different forms of the gene - humans have two alleles for each gene as they inherit one from each parent.
Dominant allele	Only one (out of the two alleles) is needed for it to be expressed and

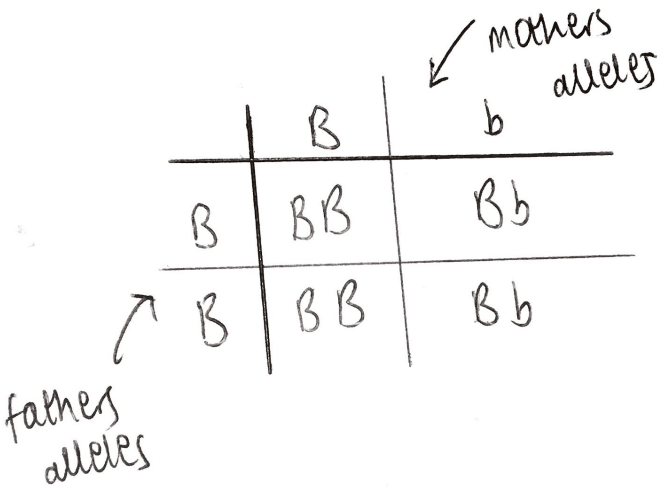
	for the corresponding phenotype to be observed.
Recessive allele	Two copies are needed for it to be expressed and for the corresponding the phenotype to be observed.
Homozygous	When both inherited alleles are the same (i.e. two dominant alleles or two recessive alleles).
Heterozygous	When one of the inherited alleles is dominant and the other is recessive.
Genotype	The combination of alleles an individual has, e.g. Aa
Phenotype	The physical characteristics that are observed in the individual, e.g. eye colour
Zygote	The stage of development immediately after fertilisation - a diploid (2n) cell formed from the fusion of two haploid gametes

Alleles (different forms of the same gene) lead to **differences in inherited characteristics**. This is because different alleles code for different forms of the same protein - an allele that codes for a **damaged form of a protein** can cause illness. For example, in a condition called Huntington's Disease, an allele of the gene that codes for a particular protein is different. This leads to the protein becoming **folded incorrectly** and causing the condition.

3.14 - Monohybrid Inheritance and Genetic Diagrams

Family trees show the inheritance of different phenotypes over generations in the same family.

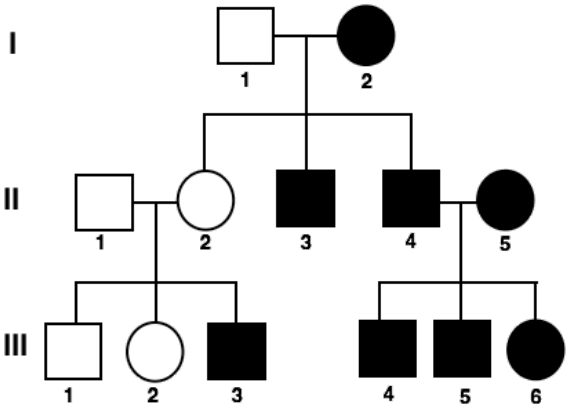
A **monohybrid (single gene) cross** looks at the probability of the offspring of two parents having certain genotypes and phenotypes.This is done using the alleles the two parents have for a gene and a **Punnett square diagram**. You should be able to draw and use a Punnett square diagram.



Uppercase letters are used to represent **dominant characteristics**. Lowercase letters represent recessive characteristics. You can choose any letter but usually either A or B is used for simplicity. Notice that combining the alleles shown above results in **¼ chance** of having an offspring who is **homozygous dominant (BB, or has two dominant alleles),**

and there is no chance of having a homozygous recessive offspring (as both parents have the allele). Whether or not the Bb (heterozygous recessive) offspring show symptoms depends on whether the condition itself is **recessive** or **dominant.**

Family pedigrees are used to show how a condition (or more specifically, the allele which causes it) are passed down through different generations. We can use them to **better visualise certain patterns** - for example, the way that recessive alleles normally 'skip a generation':



We usually use **squares to represent males**, and **circles to represent females** in the lineage.
Black shapes represent an affected individual, and **white shapes** represent an unaffected individual.
A **line through the shape** means that the individual is deceased.

A line passing directly between two shapes means that the two are **partners**, and a line overhanging a group of individuals means that they are **siblings.**

3.15 - Sex of Offspring

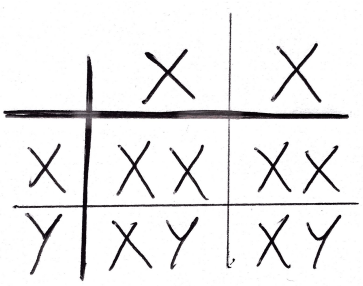
Human body cells have 23 pairs of chromosomes.

- 22 control characteristics, and the chromosomes in each pair look very similar
- The 23rd pair carries **sex determining genes**, and the two chromosomes can look different to each other (Y chromosomes are much smaller than X chromosomes)

The two possible chromosomes in the 23rd pair are **X chromosomes** and **Y chromosomes**. When cells undergo meiosis to form a gamete, one sex chromosome goes into each gamete.

- Females have two X chromosomes, so therefore only pass on X chromosomes in their eggs.

- Males have one X chromosome and one Y chromosome, so therefore can pass on X or Y chromosomes in their sperm.



Punnett squares can be used to show sex inheritance. As shown in this diagram, there is a 50% chance of the child being male, and a 50% chance of the child being female.

3.16 - Outcomes and Pedigree Analysis

We can use monohybrid crosses (Punnett squares) and pedigree analysis to analyse the probabilities of particular outcomes:

If the cystic fibrosis gene is autosomal recessive, and we use C/c for the normal and defective allele respectively:

	C	c
C	CC	Cc
c	Cc	cc

We can see that the risk of having both defective alleles (CC) is one out of four, or ¼ (25%).

3.17B **Biology Only** - ABO Blood Group Inheritance

There are often important patterns to be seen in inheritance of particular genes. For example, sometimes 2 dominant alleles can be expressed **together** in the same individual. This is called **codominance.** When three or more alleles can be present at the same loci (but not necessarily expressed at once), we say that **multiple alleles** are present at the same position, or **locus**.

An example of **codominance** and **multiple alleles** is the **ABO blood group system**, where there are alleles for A, B and O that can all be expressed at the same locus (position). Only two of them are expressed at once, however, with the following pattern (note that the O allele is recessive, and the A and B alleles are dominant):

A and O alleles: becomes type A

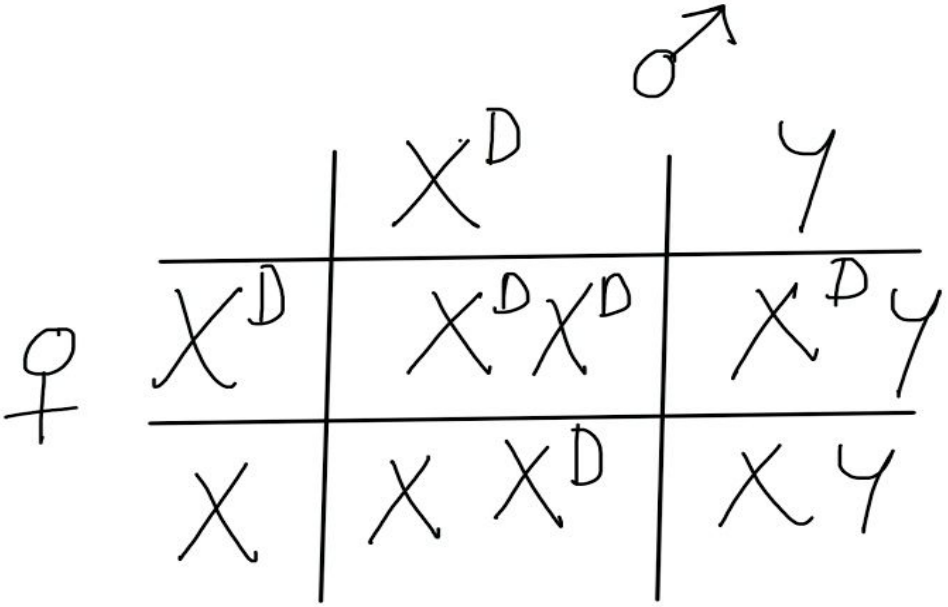
A and B alleles: becomes type AB (codominantly expressed)

A and A alleles: becomes type A
B and B alleles: becomes type B
O and O alleles: becomes type O

It should be clear from the above that Type O blood is significantly more rare than types involving A or B alleles.

3.18B **Biology and Higher Only** - Sex-linked Inheritance

Some alleles are not found on chromosome pairs 1-22: instead, they are found on the sex chromosomes X and Y. Remember that in humans, males carry an X and a Y chromosome (XY), whereas females carry two X chromosomes (XX). If these alleles cause a genetic disorder, it is known as a sex-linked genetic disorder. The majority of sex-linked conditions are found on the X chromosome.



When using Punnett squares to demonstrate sex-linked inheritance, we can use X^D to represent an X chromosome with the affected gene, and X to represent one that is unaffected, for example.

X-linked conditions can be recessive or dominant.

Usually, X-linked recessive syndromes are more common in males - because in females, there is an extra X chromosome to mask the effect of the recessive allele. However, in males, there is only one X chromosome and if it contains the defective allele, the individual will have the condition.

3.19 and 3.20 - Multiple-gene Inheritance and Causes of Variation

Most **phenotypic features** are the result of **multiple genes** acting together, and not single genes. For example, as many as **16 genes** are thought to be responsible for human eye colour.

Variation between individuals is an important factor in allowing natural selection to happen, and it originates from one of two main areas:

a) **Genetic variation -** different characteristics can arise as a result of both random **mutation** and **sexual reproduction**. Random mutation occurs in gametes to produce offspring with 'brand new' phenotypic characteristics, whereas sexual reproduction causes the offspring to have a new combination of characteristics from both its mother, and its father.

b) Environmental variation - characteristics can also be caused by an organism's environment, but these changes are generally not **heritable** (there is no change in the DNA of the organism). For example, a child who does not receive adequate nutrition will not grow to their full height, however this has no effect on their potential height as determined by their genetics.

3.21, 3.22 and 3.23 - Human Genome Project, Genetic Variation and Mutation affecting Phenotype

Remember - the word **genome** describes all the genetic information of that organism. The human genome has been studied, or 'mapped' as part of the **Human Genome Project**, which has:

- improved our understanding of the **genes linked to different types of disease**
- helped in the **treatment of inherited disorders**
- helped in **tracing human migration patterns** from the past.

By mapping the entire human genome, we are better able to understand **which genes cause inherited disorders** and which genes are linked with each other.

There is usually extensive genetic variation within a population of a species - this arises through **random mutation**.

Genetic mutation can have varying effects on the phenotype. For example, the majority of mutations have no effect on the phenotype as they occur in DNA which **does not code for proteins (non-coding DNA).** Some mutations can have a small effect on the phenotype, and other mutations, rarely, can significantly affect the phenotype - for example, changing one base can change one amino acid in a protein. If the protein was an enzyme, this has the potential to **change the shape of the active site** so that substrates can no longer bind to it.