# OCR (A) Biology GCSE

## Topic 5: Genes, Inheritance and Selection

Notes

(Content in bold is for higher tier only)

# Inheritance

## Definitions (5.1a)

| | |
|---|---|
| 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.<br>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. |
| Allele/variant | 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 |

## The genome (5.1b-d)

The genome describes all the genetic information of that organism.
The genetic information and its interaction with the environment influence how genetic traits are presented, i.e. the phenotype. This can be with continuous variation, such as height, or in discontinuous variation, such as with eye colour.
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.

**Genetic variance**

All variants (alleles) are caused by mutation. Most of these mutations have no effect on the phenotype, as most of DNA is non-coding and therefore does not cause a change in any proteins. However, some do have a small influence on phenotype and very few can completely change the phenotype if they are in coding regions.

## Coding and non-coding DNA
## *Biology only*

**If the mutation occurs in a coding region of DNA then it can alter the activity of the protein that the altered area of DNA is meant to code for. This can occur by changing the protein structure, such as by changing the shape of the active sites of enzymes so substrates no longer fit into them.**

**If the mutation is in a non-coding region of DNA it can still affect how genes are expressed by stopping transcription of mRNA in the protein synthesis process (detailed below).**

**Protein synthesis**

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 two strands pull apart from each other, and mRNA nucleotides (messenger RNA: a different type of nucleotide) match to their complementary base on the strand.**
3. **The mRNA nucleotides themselves are then joined together, creating a new strand called the mRNA strand. This is a template of the original DNA.**
4. **The mRNA then moves out of the nucleus to the cytoplasm and onto structures called ribosomes.**
5. **At the ribosomes, the bases on the mRNA are read in threes to code for an amino acid (the first three bases code for one amino acid, the second three bases code for another etc).**
6. **The corresponding amino acids are brought to the ribosomes by carrier molecules.**
7. **These amino acids connect together to form a protein.**
8. **When the chain is complete the protein folds to form a unique 3D structure.**

## Asexual and sexual reproduction (5.1f)
## *Biology only*

The advantages of sexual reproduction are the disadvantages of asexual reproduction and vice versa.

| Advantages of sexual reproduction | Advantages of asexual reproduction |
|---|---|
| Produces variation in offspring.<br>• 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).<br>• 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. | Uses less energy and is faster as |

| | |
|---|---|
| • This type of reproduction mixes the genetic information from two organisms<br>• Organisms with different desirable characteristics can be bred to produce offspring with even more desirable characteristics.<br>• This speeds up natural selection.<br>• An example is to increase food production by breeding two animals with lots of meat. | organisms do not need to find a mate. |
| | In favorable conditions lots of identical offspring can be produced. |

## Meiosis (5.1g and h)

Meiosis is the formation of four non-identical cells from one cell. Cells in the reproductive organs divide by meiosis to form gametes. Gametes only have one copy of each chromosome.

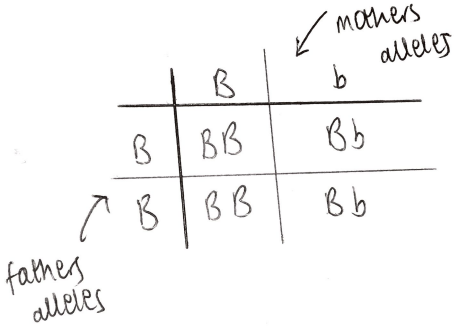
- The cell makes copies of its chromosomes, so it has double the amount of genetic information and so are called diploid.
- The cell divides into two cells, each with half the amount of chromosomes, giving the normal amount of 46 chromosomes.
- Each cell divides into two again to produce four cells, each with 23 chromosomes. As they have half of the normal amount of chromosomes they are called haploid.
- 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.

## Inheritance (5.1i-l)

**Punnett square diagrams**

A 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.
However, it is important to remember that most phenotypic features are the result of multiple different genes interacting rather than a single gene inheritance.

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.



**Sex determination**

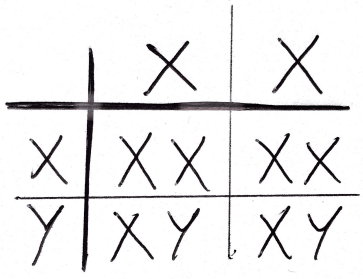
Human body cells have 23 pairs of chromosomes.

- 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.

In a similar way as above, we can show that there is a 50% chance of babies being born as either a boy or a girl using a Punnett square.




## Development of genetics by Mendel
## *Biology only*

Gregor Mendel worked in the monastery gardens and observed the characteristics passed on to the next generations in plants. He carried out breeding experiments on pea plants, using smooth peas, wrinkled peas, green peas and yellow peas and observed the offspring to see which characteristics they had inherited.

He came to the conclusions that:

- 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 his work.

- 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.

# Natural selection and evolution

## Evolution (5.2a, c and d)

Evolution is a change in the inherited characteristics of a population over time through a process of natural selection which may result in the formation of a new species.

Theory of Evolution (Darwin and Wallace): All species have evolved from simple life forms that first developed more than three billion years ago.

Evolution occurs because of natural selection.

- Mutations occur which provide variation between organisms.
- If a mutation provides a survival advantage the organism is more likely to survive to breeding age.
- The mutation will then be passed onto offspring.
- Over many generations, the frequency of the mutation will increase within the population.

Within a population there is usually a large amount of genetic variation between individuals of the same species. This natural variation occurs through small mutations that have occurred throughout time. For example, we can see that within a particular breed of dog there are slightly different coat colours and patternings due to random mutations.

This may cause one population of a species to become so different that they can no longer interbreed to produce fertile offspring. This means they have become a new species. This is called speciation.

## Classification systems (5.2b)

**Artificial classification**

- Artificial classification is when classification is based purely on observations
- Carl Linnaeus invented the The Linnaean system. Living things were divided into kingdoms, phylum, class, order, family, genus, species.
- Linnaeus' system was based on human judgement

**Natural classification**

- The current classification system has been developed based on some key advances in biology:
    - Improvements in microscopes
    - Studies of biochemistry
    - DNA evidence
- Phylogenetics is the study of how closely related two organisms are, allowing us to see which species originated from what
- Natural classification is where molecular techniques are used to see similarities between species, such as DNA sequencing to compare protein structures.

- Carl Woese added three large groups called domains above kingdoms by using molecular techniques:
    - Archaea: primitive bacteria which live in extreme environments such as hot springs
    - Bacteria: true bacteria (despite having similar features to archaea)
    - Eukaryota: organisms who have a nucleus enclosed in membranes, includes the kingdoms protists, fungi, plants and animals
- The binomial system gives each organism a name which is used worldwide (overcomes language barriers). The first part is their genus and the second part is their species.
    - E.g. the ladybug, which has the name *Harmonia* (genus) *axyridis* (species).

## Evidence for Evolution (5.2e)

Evidence for evolution is seen in fossils and in antibiotic resistance in bacteria.

### Fossils

The remains of organisms from many years ago, which are found in rocks. They are formed by:

1. Parts of organisms that have not decayed because oxygen or moisture were not present, meaning that the microbes that cause decay cannot survive.
2. Parts of the organism such as teeth, shells and bones are replaced by minerals as they decay, forming a rock structure of the original part.
3. Preserved traces such as footprints, burrows and rootlet traces (the plants roots) remain due to the ground hardening around them and forming a cast.

Fossils are used to show how the anatomy of organisms has changed over time. They can be used to compare how closely related two organisms are, through looking at the number of similarities they have. This information is used to create evolutionary trees.

### Antibiotic resistance in bacteria

Bacteria are organisms that reproduce at a very fast rate and therefore advantageous genes, such as those for antibiotic resistance, can become prominent within a population very quickly.

Exposure to antibiotics creates a selection pressure, as those with antibiotic resistant genes survive and those without die. As a result those with antibiotic resistance can reproduce and pass on the advantageous gene to their offspring and so the population of antibiotic resistant bacteria increases.

An example is the MRSA 'superbug' that is resistant to many different types of antibiotics. It is found in hospitals as it spreads when doctors and nurses move between different patients.

## The Theory of Evolution: Darwin and Wallace (5.2f)
## *Biology only*

### Darwin

Darwin's work was published in his book 'On the Origin of Species':

- Variation exists within species as a result of mutations in DNA
- Organisms with characteristics most suited to the environment are more likely to survive to reproductive age and breed successfully – called survival of the fittest.

- The beneficial characteristics are then passed on to the next generation
- Over many generations the frequency of alleles for this advantageous characteristic increase within the population

There was lots of controversy surrounding his ideas for many reasons:

1. It contradicted the idea that God was the creator of all species on Earth.
2. There was not enough evidence at the time as few studies had been done on how organisms change over time.
3. The mechanism of inheritance and variation were not known at the time.

**Wallace**

Wallace worked alongside Darwin to help prove evolution, but his work focused more on the theory of speciation.

The process of speciation:

1. Variation exists within a population as a result of genetic mutations.
2. Alleles which provide a survival advantage are selected for through natural selection.
3. Populations of a species can become isolated, for example through physical barriers such as a rock fall preventing them from breeding together.
4. Different alleles may be advantageous in the new environment, leading to them being selected for.
5. Over time the selection of different alleles will increase the genetic variation between the two populations.
6. When they are no longer able to breed together to produce fertile offspring, a new species has formed.