- Triple Science Content only in purple
- Triple Science and Higher Content Only in blue

GCSE Biology AQA Topic 6

Inheritance, Variation, and Evolution

Reproduction

Sexual vs Asexual Reproduction

- **Sexual reproduction**:
  - two parents needed.
  - gametes (sperm & egg in animals; pollen & ovule in plants) fuse together.
  - Offspring receives a mix of genes from both parents, generating **genetic diversity**.

- **Asexual reproduction**:
  - Single parent required
  - No gamete fusion.
  - Offspring are **genetically identical clones**.

- **Advantages / Disadvantages**:
   • Sexual reproduction: variation helps for survival if environment changes, but needs two parents and takes more energy
   • Asexual: fast, needs no mate, but risk if environment changes as they are susceptible to diseases (all are vulnerable)

Examples you should know of organisms that reproduce sexually and asexually (exam-board approved):

- **Malarial parasites (Plasmodium):**
  • Asexual reproduction in the human host (liver/blood cells)
  • Sexual reproduction in the mosquito vector

- **Fungi:**
  • Asexually by spores (producing genetically identical offspring)

• Sexually to introduce variation when conditions change

● **Plants (e.g. strawberries, daffodils):**
    • Asexually by runners (stolons) or bulb division
    • Sexual by producing seeds using pollen (pollination)

Meiosis-

● Purpose: produce gametes with **half the chromosome number** (haploid cells).

● Process in brief:

    1. DNA replicates in parent cell

    2. First division: homologous chromosomes separate → two cells (both have 42 chromosomes in each of the 2 cells)

    3. Second division: sister chromatids separate → total of four gametes (all 4 cells has 23 chromosomes)

● Outcome: 4 genetically different haploid gametes (in humans, each with 23 chromosomes)
● **Haploid** - Has half the number of chromosomes (23)
● **Diploid** - Has full number of chromosomes (46, double 23)

● Fertilisation restores full (diploid) chromosome number

DNA, Genes & Genome

● **Meiosis** is a type of cell division that produces **gametes** with **half the number of chromosomes**.
    ○ The process ensures that when fertilisation occurs, the offspring has the correct number of chromosomes.

Stages of Meiosis:

1. The parent cell duplicates its DNA.
2. The cell divides **twice**, producing four genetically different cells with **half the number of chromosomes** (23 in humans).

DNA and the Genome

DNA as a Polymer

- DNA is a **large polymer**, which means it's made up of many smaller, repeating units joined together.

- These smaller units are called **nucleotides**.

Each nucleotide consists of **three parts**:

1. A **phosphate group**

2. A **sugar molecule** (deoxyribose)

3. A **nitrogenous base** — one of four types:

   - **A** = Adenine

   - **T** = Thymine

   - **C** = Cytosine

   - **G** = Guanine

DNA Structure

- **DNA** (deoxyribonucleic acid) is the molecule that carries all the genetic information in living organisms.
  - It is shaped like a **double helix**, which looks like a twisted ladder.

Structure of DNA:

- DNA is made up of two **strands** forming the double helix.
- These strands are made of units called **nucleotides**.
  - Each nucleotide has:
    1. A **sugar** molecule.
    2. A **phosphate** group.
    3. One of four **bases: Adenine (A), Thymine (T), Cytosine (C)**, or **Guanine (G)**.
- The bases pair up in a specific way called **complementary base pairing**:

- ○ **Adenine (A)** always pairs with **Thymine (T) with a hydrogen bond to pair them**.
- ○ **Cytosine (C)** always pairs with **Guanine (G) with a hydrogen bond to pair them**.
  - 3 bases (e.g ATC) code for an amino acid, which join together with other amino acids to make proteins in protein synthesis.

What is a Gene?

- A gene is a short section of DNA that codes for a particular sequence of amino acids, which then forms a specific protein.

- Each gene occupies a fixed position on a DNA molecule (called a locus).

- Proteins are responsible for nearly all functions in the body — structural (e.g. collagen), catalytic (enzymes), hormonal (insulin), or protective (antibodies).

What is the Genome?

- The **genome** is the **entire set of genetic material** in an organism.

- It includes **all the DNA** found in the **nucleus** (in chromosomes) and, in some cells, in the **mitochondria** or **chloroplasts**.

- The human genome contains roughly **3 billion base pairs** across **46 chromosomes** (23 pairs).

Term	Description
DNA	Polymer of nucleotides carrying genetic information
Gene	Section of DNA coding for a protein
Genome	Entire genetic material of an organism
Polymer	Large molecule made from repeating units
Base Pairing	A with T, C with G

Protein Synthesis

- **Protein synthesis** is the process by which cells make proteins, following instructions coded in the DNA.
    - Proteins are made from long chains of **amino acids**.

Two Main Stages of Protein Synthesis:

1. **Transcription** (in the nucleus):
    - The DNA Molecule splits apart into 2 strands, each a template strand.
    - mRNA nucleotides join onto each DNA strand through complementary base pairing.
    - This forms a single-stranded molecule called **mRNA** (messenger RNA).
    - The mRNA then leaves the nucleus and moves into the **cytoplasm**.
2. **Translation** (at the ribosomes):
    - The mRNA attaches to a **ribosome**.
    - **tRNA** (transfer RNA) molecules bring **amino acids** to the ribosome, matching their **anticodons** to the mRNA's codons (three-base sequences).
    - The ribosome reads the mRNA sequence and assembles the amino acids in the correct order to form a **protein**.

Importance of Proteins:

- **Enzymes**: Proteins that speed up chemical reactions.
- **Hormones**: Regulate body processes and act as chemical messengers (e.g., insulin).
- **Structural proteins**: Give strength and support (e.g., collagen).
- **Antibodies**: Help the immune system fight infections.

What is a Mutation?

A **mutation** is a **change in the sequence of bases** in DNA.

Most mutations occur **spontaneously**

Types of Mutations

Gene mutations affect **one or a few bases** within a single gene. There are three main types you must know:

1. Substitution

- One base is **replaced** with a different base.

- Example:
  Original DNA: **A T G C C A**
  Mutated DNA: **A C G C C A**

- Effect:

  - Sometimes it has **no effect**, because multiple codons can code for the same amino acid (this is called a **silent mutation**).

  - Sometimes it can **change one amino acid**, possibly altering the protein's shape or function.

  - Rarely, it can create a **stop codon**, leading to a shortened, non-functional protein.

2. Insertion

- An **extra base** is added into the DNA sequence.

- Example:
  Original DNA: **A T G C C A**
  Mutated DNA: **A T G T C C A**

- Effect:

  - This causes a **frameshift mutation** — it changes how the sequence of bases is "read" in triplets (codons).

  - Every codon after the insertion may change, often producing a completely different (and non-functional) protein.

  - This usually has a **major effect** on the organism.

3. Deletion

- A **base is removed** from the DNA sequence.

- Example:
  Original DNA: **A T G C C A**
  Mutated DNA: **A T C C A**

- Effect:

  - Like insertion, this causes a **frameshift**, shifting the reading frame and changing all amino acids after the point of deletion.

  - The resulting protein is usually **non-functional** or severely altered.

Effects of Mutations

- **Most mutations have little or no effect**, as they occur in non-coding regions of DNA or result in the same amino acid.

- **Some mutations are beneficial**, leading to **variation** that drives **evolution** by natural selection.

- **Harmful mutations** can disrupt normal biological functions and may cause **genetic disorders** (e.g. cystic fibrosis).

- Mutations can happen **spontaneously** when DNA is copied during cell division, or they can be caused by:
  - **Radiation** (e.g., UV rays, X-rays).
  - **Chemicals** (e.g., carcinogens in tobacco smoke).

Genetic Inheritance

- Characteristics are controlled by **genes**, which come in different versions called **alleles**.

- **Alleles:** different versions of a gene (e.g. allele for blue eyes vs brown eyes).

- **Dominant allele:** expressed if one copy is present (written with capital letter).

- **Recessive allele:** expressed only if both copies are recessive (lowercase).

- **Homozygous:** two identical alleles (e.g. AA or aa)

- **Heterozygous:** two different alleles (e.g. Aa)

Punnett Squares:

- A **Punnett square** can be used to predict the probability of inheriting certain traits based on the combination of alleles from each parent:

	A	A
a	Aa	Aa
A	AA	AA

AA = Father's alleles, Aa = Mothers Alleles

Inherited Disorders

Some disorders are caused by **faulty alleles** (mutations in genes) that can be **passed from parents to offspring** through their gametes.
 These are known as **inherited disorders**.

1. Cystic Fibrosis (CF)

Cause:

- Caused by a **recessive allele** (represented as *f*).

- The gene codes for a membrane protein that controls the movement of salt and water in and out of cells.

- The faulty protein leads to **thick, sticky mucus** building up in the lungs and digestive system.

Effects:

- Breathing difficulties and frequent lung infections.

- Difficulty digesting food due to blocked pancreatic ducts.

- Reduced life expectancy (though treatment has improved).

Treatment (not required in huge depth for GCSE):

- Physiotherapy, enzyme supplements, and medication to clear mucus and prevent infection.

2. Polydactyly

Cause:

- Caused by a **dominant allele** (represented as *P*).

- Only **one copy** of the allele is needed for the condition to appear.

Effects:

- Babies are born with **extra fingers or toes**.

- Usually doesn't affect health and can be corrected surgically.

Inheritance Example (Punnett Square)

For **cystic fibrosis**:

	F	f
F	FF	Ff
f	Ff	ff

→ **25 % (1 in 4)** chance child will have CF if both parents are carriers.

Embryo Screening

Definition:
 Embryo screening is a process used to detect **inherited disorders** in embryos before birth. It is done using **genetic testing**.

Arguments For and Against Embryo Screening

Arguments For	Arguments Against
Can **prevent serious genetic diseases**, improving quality of life.	May lead to **ethical concerns** — embryos with "undesirable" traits are destroyed.
Saves **health-care costs** long-term by reducing need for treatment.	Could encourage ideas of **"designer babies"** (choosing traits like intelligence or eye colour).
Parents can make **informed decisions** about pregnancy.	**Expensive**, may only be accessible to wealthy families — raises fairness issues.
Reduces **emotional suffering** for families affected by genetic disorders.	Small **risk of miscarriage** with some testing methods (e.g. amniocentesis).

Sex Determination

Human sex is determined by the **23rd pair of chromosomes** (the sex chromosomes).**How It Works:**

- During fertilisation:

  - If an **X-carrying sperm** fertilises the egg → **female (XX)**

  - If a **Y-carrying sperm** fertilises the egg → **male (XY)**

Probability:

- There is a **50 % chance** of having a boy and a **50 % chance** of having a girl.

Variation and Evolution

Types of Variation

- **Variation** refers to the differences between individuals in a population.

2 Types of Variation:

1- Genetic Variation (Genotype): Caused by **differences in genes (DNA)** inherited from parents.
- Each individual has a unique combination of alleles.
- Main causes:
Mutations (random changes in DNA)
Meiosis (mixing of genetic material during gamete formation)
Fertilisation (random fusion of gametes)

2- Environmental Variation: Caused by external factors like climate, diet, or lifestyle. This type of variation does not change an individual's genes.

Natural Selection and Evolution

Natural selection is the process through which organisms better adapted to their environment tend to survive and reproduce.

Evolution is the **gradual change in the inherited characteristics** of a population over time.

Proposed by **Charles Darwin**

Steps in Natural Selection

1. **Variation exists** — individuals in a species show differences (from mutations and sexual reproduction).
2. **Competition occurs** — for resources like food, space, and mates.
3. **Survival of the fittest** — individuals with **advantageous traits** survive better.
4. **Reproduction** — those survivors pass on their beneficial alleles to offspring.
5. **Gradual change** — over many generations, these alleles become more common, leading to evolution.

Example: Antibiotic Resistance in Bacteria

- Random mutations make some bacteria resistant to antibiotics.

- When antibiotics are used, non-resistant bacteria die, but resistant ones survive and reproduce.

- Soon, most of the population becomes resistant — evolution in action.

Selective Breeding

- **Selective breeding** (artificial selection) is when humans breed plants or animals for specific traits.

Steps in Selective Breeding:

1. Choose individuals with desirable traits.
2. Breed them together.
3. Select the offspring with the desired traits and repeat the process.

Impacts of Selective Breeding:

- Can improve crop yield or livestock productivity.

Problems of Selective Breeding

❌ Reduced Genetic Diversity

- Breeding from a small gene pool means many individuals share the same alleles.

- Less variation = less ability to adapt to environmental changes.

❌ Inbreeding

- Closely related individuals breed together → increased chance of inheriting harmful recessive alleles.

- Can cause genetic disorders, weaker immune systems, and fertility problems.

🧠 *Example:* Purebred dogs often suffer from inherited health issues.

Genetic Engineering

- **Genetic engineering** involves modifying the DNA of an organism to introduce new traits or characteristics.

The Process of Genetic Engineering:

1. **Identifying the Gene:**
   - Scientists first identify the **gene of interest**

     Isolating the Gene:

   - The gene is then **isolated** from the organism's DNA using **restriction enzymes**.
   - The gene is often left with **sticky ends** (short, single-stranded sections of DNA) to make it easier to insert into a new DNA sequence.
2. **Inserting the Gene into a Vector:**
   - The isolated gene is inserted into a **vector**. A vector is a carrier that can transfer the gene into another organism.
   - Common vectors include **plasmids** (small circular DNA found in bacteria) or **viruses**.
   - The **sticky ends** of the gene and the vector's DNA are joined together using an enzyme called **ligase**, creating **recombinant DNA**.
3. **Transferring the Gene into the Target Organism:**
   - The recombinant DNA (with the new gene) is introduced into the target organism's cells. This can be done in several ways:
     - **Microinjection:** Injecting the DNA directly into the cells.
     - **Gene gun:** Shooting tiny particles coated with DNA into the cells.
     - **Bacterial infection:** Using bacteria (like **Agrobacterium** in plants) to naturally transfer the gene.
4. **Cloning and Growing the Modified Cells:**
   - Once the gene is inside the target cells, the organism is allowed to grow and divide.
   - As the cells divide, they create copies of themselves with the **new gene** included in their DNA.
   - This creates **genetically modified organisms (GMOs)** that express the new trait, such as faster growth, disease resistance, or higher yield.

Applications of Genetic Engineering:

1. **Agriculture:**
   - **Genetically modified crops** can be created to:
     - Resist pests (e.g., Bt crops that produce a toxin harmful to insects).
     - Tolerate herbicides, making it easier to control weeds.

■ Grow in harsh conditions (e.g., drought-resistant crops).
2. **Medicine**:
   ○ **Insulin production**: Bacteria have been genetically modified to produce **human insulin**. This insulin can then be harvested and used to treat diabetes.
   ○ **Gene therapy**: Genetic engineering can be used to insert healthy genes into patients with genetic disorders, such as cystic fibrosis, to treat the condition.
3. **Research**:
   ○ Scientists use genetically modified organisms to study the function of specific genes and to develop treatments for diseases.

Benefits of Genetic Engineering:

- **Increased crop yields**: GM crops can produce more food in the same amount of land, helping to combat hunger.
- **Resistance to pests and diseases**: Reduces the need for chemical pesticides, which can harm the environment.
- **Improved medical treatments**: Genetic engineering has revolutionised medicine, making it possible to produce life-saving drugs like insulin more efficiently.

Concerns and Ethical Issues:

- **Environmental Impact**: There are concerns that GM crops could crossbreed with wild plants, leading to unpredictable effects on ecosystems.
- **Food Safety**: Some people worry about the long-term health effects of eating genetically modified foods, although research has found them safe for consumption.
- **Ethics**: The idea of "playing God" by altering the genes of organisms raises ethical questions, particularly when it comes to genetic engineering in humans.

Cloning

- **Cloning** is the process of creating genetically identical copies of an organism or cell. There are several methods of cloning used in both plants and animals.

1. Cloning in Plants

Tissue Culture:

- Small groups of **cells** from a plant are taken and grown in a sterile culture medium, containing nutrients and hormones to encourage growth.
- These cells develop into **plantlets**, which can be transferred to soil to grow into **identical plants**.

- Tissue culture is used to **preserve rare plant species** or to rapidly produce large numbers of plants with desired traits (e.g., disease resistance).

Cuttings:

- A **simple method** where part of a plant, usually a stem or leaf, is cut and placed in water or soil.
- This part will develop roots and grow into a **new plant**, which is genetically identical to the original plant.
- It is widely used by gardeners to clone plants quickly and cheaply.

2. Cloning in Animals

Embryo Transplants:

- A developing **embryo** from an animal is split into several smaller embryos before they become specialised.
- Each smaller embryo is placed into the uterus of a surrogate mother, where they continue to develop.
- The offspring are genetically identical to each other, though not to the original parents, as they are produced from **sex cells** (egg and sperm).

Adult Cell Cloning (Somatic Cell Nuclear Transfer):

1. A **nucleus** is removed from an unfertilized egg cell.
2. The nucleus from an **adult body cell** (somatic cell) is inserted into the egg cell.
3. The egg cell is then stimulated (often with an electric shock) to start dividing like a normal embryo.
4. The developing embryo is implanted into a surrogate mother, and the clone is born genetically identical to the adult donor of the body cell.
- This method was used to create **Dolly the Sheep**, the first mammal cloned from an adult somatic cell.

Advantages of Cloning:

- **Preservation of endangered species**: Cloning can help conserve species that are at risk of extinction by creating more individuals.
- **Replicating animals with desired traits**: Useful in agriculture to produce livestock with valuable characteristics such as high milk yield or disease resistance.
- **Medical research**: Cloned animals can be used for testing treatments for diseases or studying genetic disorders.

Disadvantages and Ethical Concerns:

- **Reduced genetic diversity**: Since clones are genetically identical, they are more susceptible to diseases or changes in the environment that could wipe out the entire population.
- **Ethical issues**: Many people raise concerns about the morality of cloning, especially when it involves animals or the potential for human cloning. The long-term effects of cloning on health and lifespan are also not fully understood.
- **High failure rate**: Cloning is not always successful, and many cloned embryos may not survive, which raises concerns about animal welfare.

The Genetics and Evolution

The Theory of Evolution
Charles Darwin (1809–1882) developed the **theory of evolution by natural selection**.

Darwin's Observations:

While studying animals (especially finches) on the **Galápagos Islands**, Darwin noticed:

- Individuals of the same species show **variation** in their characteristics.

- More offspring are produced than can survive — there is **competition** for limited resources.

- Those with characteristics best suited to their environment are **more likely to survive and reproduce**.

  Theory Of Natural Selection:

1. **Variation** exists within a species due to mutations and genetic differences.
2. **Competition** occurs for food, mates, and shelter.
3. **Survival of the fittest:** Individuals with advantageous characteristics are more likely to survive.
4. **Reproduction:** These survivors pass their beneficial alleles to their offspring.
5. **Over time:** The proportion of individuals with advantageous traits increases → the species evolves.

Why Was Darwin's Theory Controversial?

- It **conflicted with religious beliefs** about creation.

- He **lacked scientific evidence** at the time (genes and DNA were not yet understood).

- The **mechanism of inheritance** (how traits were passed on) was unknown.

Alfred Russel Wallace independently developed a similar theory of evolution around the same time as Darwin.

- Wallace focused particularly on **speciation** and **geographical isolation** — how new species arise when populations are separated by physical barriers.

- His work and correspondence with Darwin encouraged Darwin to publish his own ideas.

- Wallace is therefore credited as a **co-discoverer of natural selection**.

Speciation — How New Species Form

Speciation occurs when **populations of the same species become so different that they can no longer interbreed** to produce fertile offspring.

Process of Speciation (AQA)

1 **Isolation:**

- A population becomes **separated by a physical barrier** (e.g., mountain, river, ocean).
- This is called **geographical isolation**.

2 **Genetic Variation:**

- Each isolated group already shows genetic variation due to mutations and sexual reproduction.

3 **Natural Selection:**

- Each group experiences **different environmental conditions** (e.g., climate, predators, food).
- Different alleles become advantageous in each environment.
- Individuals with beneficial traits survive and reproduce.

4️⃣ **Evolution of Differences:**

> Over many generations, genetic differences accumulate.

- Eventually, individuals from the two groups become **so different genetically** that they can no longer interbreed.

➡️ Result: **Two new species** are formed.

Gregor Mendel's Discoveries

- Traits are **determined by "factors"** (now called genes) that are passed unchanged from parents to offspring.

- Each individual has **two versions** of a gene (we now call them alleles).

- Some alleles are **dominant** and mask the effect of **recessive** ones.

- Traits are inherited in **predictable patterns**.

Why Mendel's Work Was Ignored

- He had **no knowledge of chromosomes or DNA**.

- His work was published in 1866 but not widely read until after his death.

- Scientists rediscovered his findings in the early 1900s, which helped explain **how Darwin's theory of evolution worked genetically**.

Fossils and Evidence for Evolution

Fossils are the **preserved remains or traces** of ancient organisms, usually found in sedimentary rocks.

How Fossils Form

1. From hard parts (bones, shells, teeth) that don't decay easily.
2. When organisms are buried rapidly by sediment, which prevents decay.
3. When minerals gradually replace bone or shell material, forming a rock-like copy.

What Fossils Show

- How species have **changed over time**.

- Transitional forms showing **links between species** (e.g., Archaeopteryx showing both reptile and bird features).

- Evidence that **simple organisms evolved before complex ones**.

Limitations of Fossil Evidence

- The **fossil record is incomplete**.

- Many organisms had **soft bodies** that decayed completely.

- Geological activity (erosion, earthquakes) may have destroyed older fossils.

Extinction

Extinction occurs when **no individuals of a species remain alive**.

Factors Contributing to Extinction

Cause	Explanation
Environmental changes	Climate change, sea level rise, or habitat destruction.
New predators	Species introduced into new ecosystems (e.g., humans introducing cats to islands).
New diseases	Pathogens can wipe out populations with no immunity.
Competition	A better-adapted species outcompetes another for resources.
Catastrophic events	Asteroid impacts or massive volcanic eruptions (e.g., extinction of dinosaurs 65 million years ago).

Example:
Dinosaurs became extinct likely due to a massive asteroid impact causing climate change.

Resistant Bacteria — Evolution in Action

Bacteria reproduce very rapidly, and random **mutations** can make some individuals resistant to antibiotics.

Development of Antibiotic Resistance

1. A mutation occurs, producing a resistant strain.
2. When antibiotics are used, **non-resistant bacteria are killed**, but resistant ones survive.
3. These resistant bacteria **reproduce**, passing on the resistance gene.
4. Soon, the population becomes resistant.

Example: MRSA (Methicillin-Resistant *Staphylococcus aureus*)

- MRSA is resistant to several antibiotics.

- It can cause severe infections in hospitals and is difficult to treat.

How to Slow the Spread of Resistance

✅ Avoid unnecessary use of antibiotics (only use when truly needed).
✅ Complete the **full course** prescribed — don't stop early.
✅ Reduce the use of antibiotics in **farming**.
✅ Develop **new antibiotics**, though this is costly and slow.

Classification of Living Organisms

Classification is the process of **organising organisms into groups** based on similarities and evolutionary relationships.

Traditional Classification — Linnaean System

Developed by **Carl Linnaeus (18th century)**.
 Organisms are grouped into hierarchical levels based on their features.

Hierarchy (largest → smallest):
Kingdom → Phylum → Class → Order → Family → Genus → Species

Example:
Homo sapiens

- Kingdom: Animalia

- Phylum: Chordata

- Class: Mammalia

- Order: Primates

- Family: Hominidae

- Genus: Homo

- Species: sapiens

Binomial Nomenclature

Each species has a **two-part Latin name**:

- **Genus** (capitalised)

- **Species** (lowercase)

 Example: *Homo sapiens*, *Panthera leo*

This system is used worldwide to avoid confusion between languages.

The Three-Domain System — Modern Classification

Proposed by **Carl Woese (1990)** after DNA and molecular analysis revealed new insights.

He found that some bacteria were very different from others genetically, leading to a new top-level grouping:

Domain	Description
Archaea	Primitive bacteria living in extreme conditions (e.g., hot springs, salt lakes).
Bacteria	True bacteria with typical cell walls and structures.
Eukaryota	All organisms with complex cells — animals, plants, fungi, and protists.