

Topic: Genetics

Duration:
Combined 9 lessons

Composite:
Unit test

Key vocabulary:

Sexual reproduction
Asexual reproduction
Gamete
Chromosome
Fertilisation
Variation
Meiosis
Mitosis
DNA
Nucleotide
Complementary bases
Polymer
Double helix
Gene
Amino acid
Protein
Genome
Codon
Ribosomes
Genotype
Phenotype
Allele
Dominant
Recessive
Homozygous
Heterozygous
Zygote
Monohybrid
Genetic variation
Environmental variation
Inheritance
Mutation

Core knowledge Components

Powerful knowledge components crucial to commit to long term memory (IN RED BOX)

Sexual Reproduction and Meiosis

Meiosis leads to non-identical cells being formed with half number of chromosomes, while **mitosis** leads to identical cells being formed.

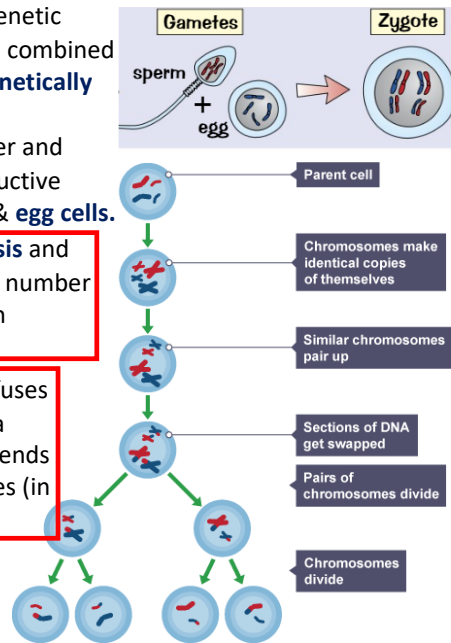
1) **Sexual reproduction** is where genetic information from two organisms is combined to produce offspring which are **genetically different** to their parent.

2) In sexual reproduction, the father and mother produce **gametes** (reproductive cells). In animals these are **sperm & egg cells**.

3) Gametes are produced by **meiosis** and are **haploid** – they contain half the number of **chromosomes** of normal cells, in humans this is 23 chromosomes..

4) At **fertilisation**, a male gamete fuses with a female gamete to produce a fertilised egg (**zygote**). The zygote ends up with the full set of chromosomes (in humans, **46**), so it is **diploid**.

5) The zygote then undergoes cell division (by **mitosis**) and develops into an embryo.



Variation



Variation: Organisms of the same species have differences, this can be genetic or environmental variation or both.

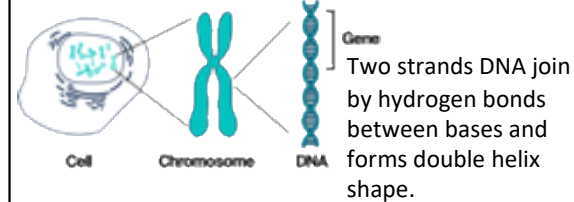
Genetic variation (inheritance): Within a species is caused by organisms having different alleles which can lead to differences in **phenotype** (the characteristics an organism displays). It can be caused through sexual reproduction or by new alleles arising through **mutations**.

Environmental variation: Is also known as **acquired characteristics** and is caused by conditions something lives in, e.g. suntan, diet and exercise, tattoos.

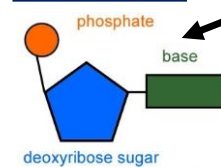
DNA (Genetic Material)

- **Chromosomes** are long strands of **DNA** and are found in the **nucleus** of **eukaryotic cells**.
- Humans have **46** chromosomes in each cell.
- A **gene** is a section of DNA on a chromosome that codes for a particular **protein**.
- All of an organism's DNA makes up its **genome**.

Codon: A sequence of 3 bases is the code for a particular amino acid. The order of bases controls the order in which each amino acids combine and fold to produce a specific shaped protein such as enzymes.



DNA Structure:

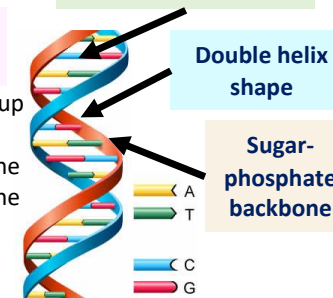


Nucleotide: Basic unit of DNA. Made up of a sugar, phosphate and base. Four possible bases; A, T, C, G.

Hydrogen bonds (joins bases)

Complementary base pairs:

Bases always pair up in a certain way.
Adenine to Thymine
Cytosine to Guanine



(HT only) Not all parts code for proteins. Non-coding parts can switch genes on & off. Mutations may affect how genes are expressed.

Links to previous and future topics

Year 9: Genetics
Year 10: Topic 4
Natural Selection

Genetic Diagrams and Family Pedigree

Monohybrid inheritance:



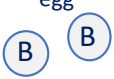
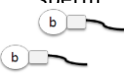
When characteristics are determined by a single gene.

A **genetic diagram** can be used to show how the certain characteristics are inherited and calculate the probability of different outcomes.

The alleles present, or genotype operate at a molecular level to develop characteristics that can be expressed as a phenotype. Some characteristics are controlled by a single gene e.g. fur colour, colour blindness. Most characteristics are as a result of multiple genes interacting.

Dominant and recessive allele combinations	
Dominant	Recessive
Represented by a capital letter e.g. B.	Represented by a lower case letter e.g. b.
3 possible combinations: Homozygous dominant BB Heterozygous dominant Bb Homozygous recessive bb	

One pair of chromosomes carry the genes that determine sex		
Female	Male	
XX	XY	
Gametes	X	Y
X	XX	XY
X	XX	XY
The probability of a male or female child is 50%. The ratio is 1:1. Remember that this ratio is only a probability for each individual. If you had four kids, they could all be boys.		

Using a punnet square (using mouse fur colour as an example)		
Parent phenotype	Black fur 	White fur 
Parent genotype	BB	bb
What gametes are present	In each egg 	In each sperm 
Gametes	b	b
B	Bb	Bb
B	Bb	Bb
The probability of black fur offspring phenotype is 100%. All offspring genotypes are heterozygous (Bb).		

Crossing two heterozygous mice (Bb)			
Gametes	B	b	Bl:Wh
B	BB	Bb	3:1
b	Bb	bb	
The probability of black fur is 75% and white fur 25%.			

The Human Genome Project (HGP)

Thousands of scientists from over the world collaborated on the **Human genome project** to map the human genome.

The **genome** is the entire DNA of an organism.

Prediction and prevention of diseases: Many common diseases (e.g. cancer & heart disease) are caused by interaction of different genes, as well as lifestyle factors. By understanding human genes, can get individually tailored advice to reduce chances of getting these diseases.

Testing and treatment for inherited disorders: Inherited disorders (e.g. cystic fibrosis) are caused by presence of one or more **faulty alleles** in a person's genome. Scientists can now identify these suspected genes and alleles much more quickly and may be able to develop better treatments/cures for these diseases.

New and better medicines: Genome research has highlighted some common genetic variations between people. Some variations affect how an individual will react to certain diseases and treatments. Scientists can use this knowledge to make more targeted drugs for individuals that would have fewer side-effects and be more effective.

Issues surrounding this: Increased stress (people more aware of potential diseases earlier on). Gene-ism (people with genetic problems could be pressured not to have children. Discrimination by employers and insurers (if you have greater chance of disease, they would be less likely to hire you/give you life insurance).

Extraction of DNA

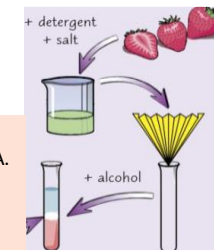
Method:



Aim: The aim of this experiment is to extract DNA from a fruit e.g. kiwi, strawberry.

1. Peel the skin from half a kiwi fruit and mash it up.
2. Mix a teaspoon of salt and small volume of washing up liquid into the fruit.
3. Gently heat this mixture at about 60°C for five minutes.
4. Filter the mixture and retain only the filtrate (the filtered liquid).
5. Cool using an ice bath and gently pour chilled ethanol onto the top of the filtrate.

Results: You will see strands of DNA with bubbles in them at the boundary between the filtrate and the chilled ethanol.



Extraction of DNA

Detergent breaks down cell membrane to release DNA.

Salt makes DNA stick together.

Filtration to remove insoluble part.

Alcohol releases DNA (precipitates).

Family Pedigree and Carriers

A family pedigree is a diagram that shows how a characteristic (or disorder) is inherited in a group of related people.

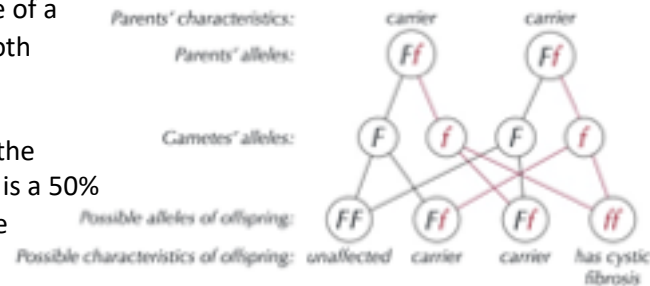
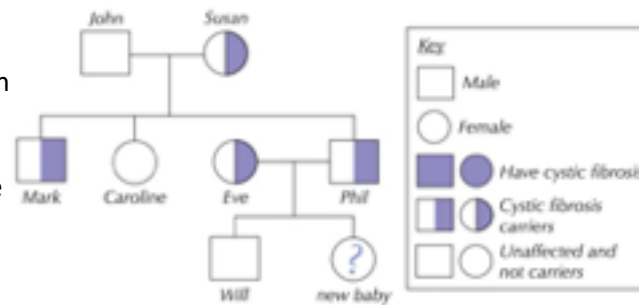
Cystic fibrosis (CF) is an inherited disorder of the cell membranes. The allele which causes cystic fibrosis is a recessive allele, carried by about 1 person in 30. People with one copy of the allele won't have the disorder, because it is recessive. They are known as **carriers**.

For a child to inherit the disorder, both parents must either be carriers or have the disorder themselves.

There is a 1 in 4 (25%) chance of a child having the disorder if both parents are carriers.

If one parent is a carrier and the other has the disorder, there is a 50% chance of the child having the disorder.

Carrier: Someone that has the faulty allele, but don't have any symptoms.



KEYWORDS

Gamete – Sex cells produced in meiosis.

Zygote – Single cell that results from fusion of egg and sperm cell.

Chromosome – A long chain of DNA found in the nucleus.

Gene – Small section of DNA that codes for a particular protein.

Alleles - Different versions of same gene. Can be due to mutations.

Homozygous – Two alleles for a particular gene that are the same.

Heterozygous – If two alleles for a particular gene are different.

Dominant allele – shown by a capital letter and this is the characteristic that is expressed as it overrules the recessive allele.

Recessive allele – shown by a small letter and only expressed when paired with another recessive.

Genetic diagrams can show the inheritance of a single characteristic (**monohybrid inheritance**).

Genotype – Alleles that are present for a particular feature, e.g. Bb or bb.

Phenotype – Physical expression of an allele combination, e.g. black fur.

Mutation – Changes to the base sequence of DNA. Can lead to changes in phenotype, but this isn't always the case.

Topic: Natural Selection and Genetic Modification

Duration:
Combined 6 lessons

Composite:
Unit test

Key vocabulary:

Alleles
Mutations
Selection pressures
Antibiotics
Hominids
Ardi
Natural selection
Classification
Evolution
Fossils
Stone tools
Gene
Genome
Genetic modification
Stratigraphy
Domain
Archae
Eukarya
Restriction enzyme
Ligase
Sticky ends
Vector
Recombinant DNA
Population
Kingdom
Selective breeding
Selection pressure
Inheritance
Binomial name
Common ancestor

Core knowledge Components

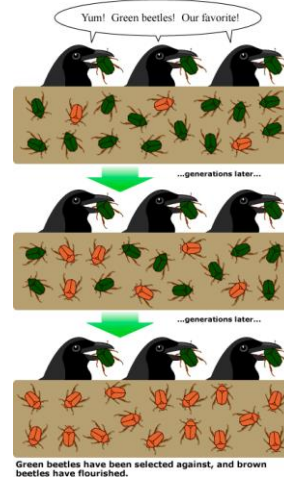
Powerful knowledge components crucial to commit to long term memory (IN RED BOX)

Natural Selection

Natural selection is all about “survival of the fittest” and occurs through a series of stages. The Theory of Evolution by Natural Selection was by Charles Darwin.

- 1) Genetic Variation** - Individuals in a population show genetic variation because of differences in their alleles. New alleles arise through mutations.
- 2) Selection pressure** – Things like predation and competition for resources (food, water, mates etc.) and disease act as selection pressures. This means they affect an organism’s chance of surviving & reproducing.
- 3) Natural Selection** – Those individuals with characteristics that make them better adapted to the selection pressures in their environment have a better chance of survival to reproduce.
- 4) Inheritance** – The alleles that are responsible for the useful characteristics are passed on to the next generation. Those that do not have the characteristic are more likely to die.
- 5) Evolution** – The beneficial allele increases in the population over time.

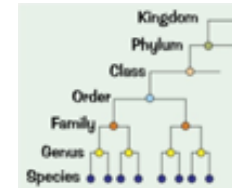
Evolution: is the slow and continuous change of organisms’ inherited characteristics from one generation to the next.



Classification

Five Kingdom Classification: Was originally used to organise organisms due to their similarities and differences in their characteristics. Organisms were classified into five kingdoms and then subdivided into smaller and smaller groups that have common features.

The five kingdoms are; **animals, plants, fungi, prokaryotes & protists.**



Classification is organising living things into groups.

Three Domain Classification: Genetic analysis of DNA led to changes in our grouping of organisms and a three domain system was proposed. The three domains are:

- 1) ARCHAEA:** Look similar to bacteria, but found to be genetically different.
- 2) BACTERIA:** Contains true bacteria.
- 3) EUKARYA:** Includes fungi, plants, animals and protists.

Binomial name is Genus and species, e.g. humans is ***Homo sapiens***.

Evidence for Evolution

Bacteria and Antibiotic Resistance:

- Bacteria develop random mutations in their DNA that can create new alleles. Some mutations can make the bacteria resistant to antibiotics (drug that kills bacteria).
- This means the antibiotic resistant bacteria have an advantage against the selection pressure (antibiotic) and so are able to live for longer and reproduce more.

Fossil Evidence:

- A fossil is any trace of an animal or plant that lived a long time ago. They are most commonly found in rocks and the deeper the rock, the older the fossil.
- By arranging fossils in chronological order, gradual changes in organisms can be observed. This provides evidence for evolution as it shows how species have changed over billions of years.



Fossil Evidence of Human Evolution

Evidence from **fossils** suggests that **humans** and **chimpanzees** evolved from a **common ancestor** that existed around 6 million years ago.

Hominids: Term used for human beings and their ancestors.

“Ardi” (*Ardipithecus ramidus*): Found in Ethiopia and is approx. **4.4 million years old**. Features include:
1) Structure of feet suggest tree climbing and an ape-like big toe to grasp branches.
2) Long arms and short legs, brain size similar to a chimp.
3) Leg structure suggests she walked upright like a human.

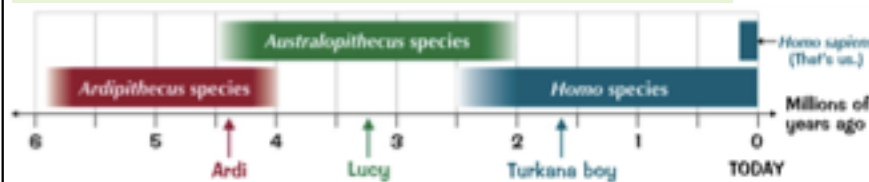


“Lucy” (*Australopithecus afarensis*): Found in Ethiopia and is approx. **3.2 million years old**. Features include:
1) Arched feet more adapted for walking than climbing.
2) Arms and legs between that of a humans and apes.
3) Brain slightly larger than Ardi’s but still similar to a chimps.
4) Leg structure implies more efficient upright walking than Ardi.



A Turkana Boy (*Homo erectus*): Found by scientist Richard Leakey along with other hominoid fossils of other species in Kenya, 1984:

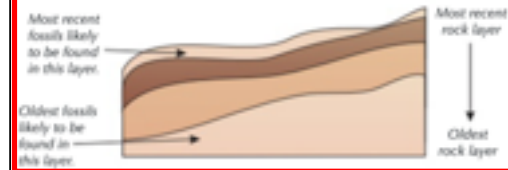
- 1) Fossil skeleton was **1.6 million years old** species and had a mixture of human and ape-like features, but more human like than Lucy.
- 2) Short arms and long legs more like a human than an ape. Brain size was much larger than Lucy’s (similar to human).
- 3) Leg structure suggests he was even better adapted to walking upright than Lucy.



Stone Tool Evidence of Human Evolution

When an ancient stone tool or hominid fossil is found, scientists can determine how old the object is by:

- 1) **Structural features** – Older tools are less complex in design. Earliest tools date back to 3.3 million years ago.
- 2) **Using stratigraphy** – location of object in rock.



- 3) **Carbon-14 dating** – Carbon 14 is slightly radioactive and decays slowly over time. This can be used to date the material.

Tool development over time

Homo habilis (2.5 - 1.5 million years ago)

Simple stone tools called pebble tools made by hitting rocks together to make sharp flakes. Used to scrape meat from bones or crack bones open.



Homo erectus (2 – 0.3 million years ago)

Sculpted rocks into more complex tools e.g. simple hand-axes. Used to hunt, dig, chop and scrape meat from bones.



Homo neanderthalensis

(300,000 – 25,000 years ago)

More complex tools. Evidence of flint tools, pointed tools and wooden spears.

Homo sapiens (200,000 years ago - present)

Flint tools widely used. Pointed tools including arrowheads, fish hooks and needles appeared around 50,000 years ago.



Selective Breeding

Selective breeding: When humans artificially select the plants or animals that are going to breed so that genes for particular characteristics remain in the population.

Organisms are selectively bred to develop features that are

useful or **attractive** such as:

- 1) Animals that produce more meat or milk.
- 2) Crops with disease resistance.
- 3) Dogs with good, gentle temperament.
- 4) Plants that produce bigger fruit.



The process of selective breeding is as follows:

- 1) From your existing stock, select the ones that have the desired characteristics.
- 2) Breed them with each other.
- 3) Select the best of the offspring and breed them together.
- 4) Continue this process over several generations.



Disadvantages of selective breeding:

- 1) It **reduces the gene pool** – number of different alleles in a population.
- 2) Inbreeding can cause **health problems** as greater chance of organisms inheriting harmful genetic defects when gene pool is limited.
- 3) Can be serious problems if a **new disease** appears. Not much **variation** in the population so **less chance of resistant alleles** being present.

Selective Breeding

Risks: alleles that may be useful in future may be bred out.

Populations with low variation can be vulnerable to genetic diseases.

Benefits: Increased growth and yield of plants and animals for food.

Genetic Engineering

Genetic engineering –

Involves modifying an organisms genome to introduce desirable characteristics.

Genetically modified organism (GMO) –

The organism into which the gene has been transferred.

This involves the use of **enzymes** or **vectors** (carriers).

Vector – Is something that is used to transfer DNA into a cell. There are two types (plasmids and viruses). Plasmids are small, circular molecules of DNA that can be transferred between bacteria. Viruses insert DNA into the organisms they infect.

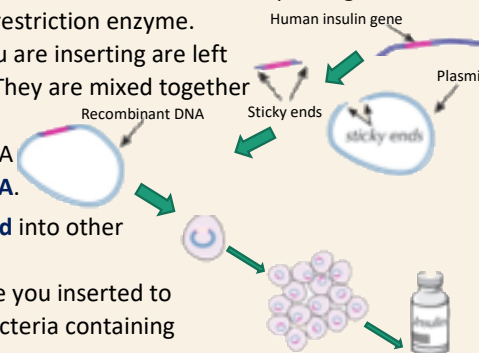
Recombinant DNA – Term used for two different bits of DNA stuck together.

Advantages of GM include: production of herbicide or insect attack resistant crops, high yield of insulin, high crop yield.

Concerns include: unknown side effects of modifying organisms DNA, could adversely affect the food chain, weeds could become herbicide resistant.

Genetic engineering process (HT only)

1. **Restriction enzymes** are used to isolate and cut out the required gene. The **vector DNA** is also cut using same restriction enzyme.
2. The vector DNA and the DNA you are inserting are left with **complementary sticky ends**. They are mixed together with **ligase enzymes**.
3. The **ligases** join the pieces of DNA together to make **recombinant DNA**.
4. The **recombinant DNA is inserted** into other cells, e.g. bacteria.
5. These cells can now use the gene you inserted to make the protein you want. E.g. bacteria containing gene for human insulin.



Risks: Seeds from GM plants can be very expensive. Some people think eating GM plants is bad for health although there is no evidence to support this view.

Benefits: decreased use of herbicide with increase in yield from food crops. Medicines tailored for individuals.