

AQA BIOLOGY - UNIT 6: GENETICS, VARIATION AND INHERITANCE

1. Variation There is usually extensive variation within a population of a species. There are 2 main causes of variation:

- Genetic** - inherited e.g. eye colour, hair colour
- Environmental** - scars, tattoos, piercings
- Both** - skin colour (tan), hair style (naturally curly but straightened) **MOST CHARACTERISTICS ARE AFFECTED BY BOTH**

Chromosome - long strands of DNA coiled up (23 pairs in normal cells, 23 in sex cells (gametes))
DNA - double helix, all info to make an organism

2. Types of reproduction

Sexual Reproduction

- Two parent cells join
- Fertilization involved
- Gametes made by meiosis
- Genetic variation found in offspring caused by mixing of genetic information.

Asexual Reproduction

- Only one parent cell required
- No fertilization involved
- No gametes involved
- Identical clones are made
- E.g. runners in plants
- Mitosis only involved

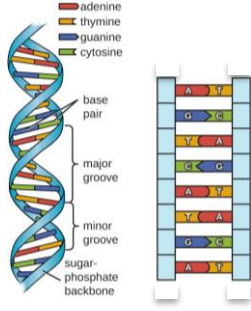
3. Gender Determination - 23rd pair of chromosomes (sex chromosomes)

XX = female XY = male

All ova (egg cells) contain one X chromosome. 50% of the sperm contain an X and 50% contain a Y chromosome. The **punnet square** shows there is a 50% chance of having a boy or a girl every time.

		Dad (XY)	
		X	Y
Mum (XX)	X	XX	XY
	X	XX	XY

4. DNA - Instructions to make an organism.



- Is a double helix **polymer**, which is wound into chromosomes.
- Sections of coding parts of DNA are called **genes**.
- Each gene codes for a particular sequence of amino acids, to make a specific protein.
- A mutation is a change (mistake) in the DNA code.

SEPARATES ONLY

- Sugar phosphate backbone
- Nucleotides made up of 4 bases that pair up A-T and G-C.
- Every 3 base pairs is the instructions given to a ribosome to make an **amino acid**. These are combined to make **proteins**.

SEPARATES HIGHER ONLY

Non-coding sections of DNA can turn on and off different genes to make different proteins. Mutations in coding = change to characteristic. Mutations in non-coding = bigger changes to the organism.



5. Genome - Entire genetic material of an organism

Human Genome Project (HGP) - the sequence of the entire genetic material of a human.

Advantages of HGP	Issues/concerns with HGP
<ul style="list-style-type: none"> Inherited cancer diagnosis Forensics (used in solving crimes) Evidence for evolution/human migration patterns Understanding of inherited diseases resulting in better choice of drugs or developing new treatments. 	<ul style="list-style-type: none"> Genetic discrimination for individuals who are found to have genetically linked conditions Abuse of knowledge - potential to re-engineer human species Very expensive

6. Genetic Crosses

e.g. A heterozygous brown eyed dog mates with a homozygous blue eyed dog. Brown eyes is dominant.

RULES

- Write the genotype of the parents
- Draw the punnet square, write parents genotype on top and side and fill in the boxes to show possible allele pair combinations. (FOUNDATION - complete a punnet square, HIGHER - construct one)

Bb x bb

	Mum (bb)	
	b	b
Dad (Bb)	B	Bb
	b	bb

- Write out the possible phenotypes of the offspring
- 50% chance heterozygous brown eyed
 50% chance homozygous blue eyed
 or a 1:1 chance of brown : blue
 Most characteristics are a result of **MULTIPLE** genes interacting.

Gene - a section of DNA that codes for 1 characteristic

Allele - different forms of a gene

Genotype - Symbols used to show genes for 1 characteristic e.g. Bb

Phenotype - Description of genes e.g. Brown eyes

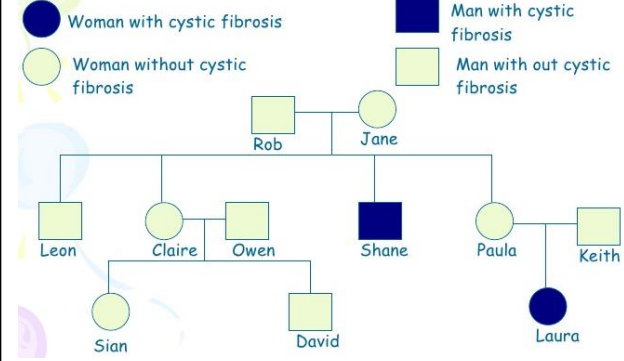
Homozygous - Both genes are the same i.e. BB or bb

Heterozygous - Both genes are different i.e. Bb

7. Genetic Diseases

- Polydactyly - caused by a **dominant** allele - extra finger or toe
- Cystic Fibrosis (CF) - caused by a **recessive** allele - thick, sticky mucus on body linings e.g. digestive, respiratory and reproductive tracts.

Genetic family tree



Rob and Jane must be Ff (where f means has a cystic fibrosis causing allele)

Ff x Ff

	F	f
F	FF	Ff
f	Ff	ff

25% chance of offspring inheriting an f allele from both parents and having the symptoms of CF.

8. Genetic Engineering - adding wanted characteristics to organisms by "cutting out" genes from one organism and transferring them to another.

HIGHER ONLY

- e.g. Making Insulin
- Remove wanted insulin gene from human genome using restriction enzymes.
 - Take a plasmid from a bacteria (vector)
 - Cut open plasmid with restriction enzyme and insert insulin gene with DNA ligase
 - Put plasmid back in bacteria
 - Incubate to allow bacteria to grow, multiply by mitosis.
 - As all the bacteria are clones they all have the insulin gene and so all make insulin - a large amount is produced quickly.

9. GM Crops

- + Resistant to insects, viruses, fungi
- + Grow bigger, taste better, more nutritious
- + Crops can be grown all over the World
- + Increased crop yield
- Worries over long term effects
- Reduced biodiversity
- Could develop allergies to the food
- Herbicide resistant gene could spread to weeds making superweeds!

10. Genetic screening (amniocentesis → gene/genome sequencing)

- Whilst screening an embryo for specific genes e.g. inherited diseases might alleviate suffering, there are concerns about it including:
- Potential for prejudice against people with inherited diseases,
 - Prompt termination of embryo,
 - Financial costs for NHS,
 - Ethical concerns related to the fact that embryos cannot give consent,
 - Procedure may damage the embryo/miscarry,
 - Procedure may harm the mother etc

11. Natural Selection

- Variation occurs naturally within a species due to mutations
- Some organisms have adaptations increasing their chances of survival - this is known as survival of the fittest.
- These organisms are more likely to reproduce.
- The genes responsible for the adaptation are passed on to their offspring.

Reasons why people didn't believe Darwin at first:

- Theory was against religious beliefs
- They didn't know about genes or mutations at the time so Darwin couldn't explain why some organisms had more useful characteristics
- Not enough evidence

12. Selective Breeding

Humans breed animals/plants to gain desirable characteristics in offspring (takes many generations).
 e.g. disease resistance, increased milk production, behaviour, scented flowers etc.

Downsides - Reduces variation limiting success of survival if conditions change, new diseases might wipe out every member of the same species, inbreeding in animals leads to defects.

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13. Evidence for evolution - fossils

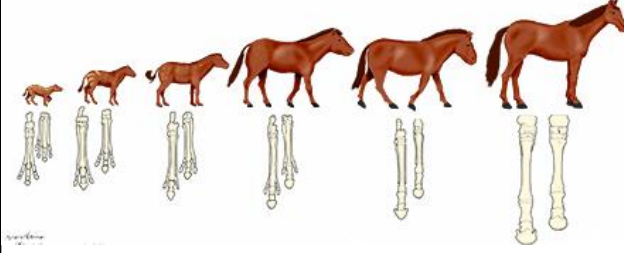
Fossils are made from parts of organisms or imprints they have made whilst alive. Usually one or more conditions required for decay are absent and so parts are preserved.

- Bones and teeth
- Minerals that have replaced bone and tissue in rock
- Organisms trapped in amber, ice, peat or tar where decay cannot occur due to temperature, lack of oxygen and/or acidity.
- Burrows, tracks, where organisms have laid

Softer body parts such as tissue, muscle etc. **decay** if conditions are suitable. As many early life forms were soft bodied, we have little evidence for how life began on Earth.

Fossil record

- Collection of fossils that show evolution of an organism over many years.
- Usually incomplete as most organisms don't become fossils, fossils melt underground due to Earth movement, not been found yet.
- Usually need to comment on changes over time e.g. shape, length or number of bones.



15. Extinction - Living things become extinct because:

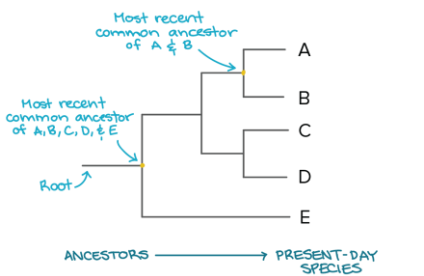
- Habitat changes - not adapted to survive
- New predator - not adapted to get away or hide
- Disease - lack of immunity
- New, more successful competitor - better adapted species will get food, space, water etc.

16. Classification

Carl Linnaeus	Carl Woese
<ul style="list-style-type: none"> • Grouped according to characteristics and structures that make up organisms. • Kingdom, Phylum, Class, Order, Family, Genus, Species 	<ul style="list-style-type: none"> • Three-domain system (added above the kingdom classification) • Based on a better understanding of internal structures (due to better microscopes) and new chemical analysis techniques that prove some species aren't as closely related as once thought. <ul style="list-style-type: none"> • Archaea - primitive bacteria • Bacteria - true bacteria • Eukaryota - fungi, animals, plants, protists

Organisms are named using **binomial system** (genus and species name in Latin).

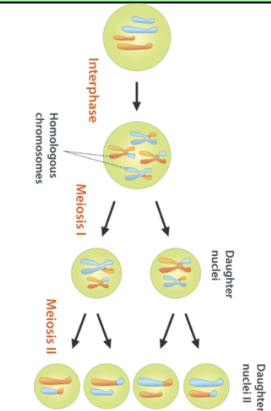
Evolutionary trees show common ancestors. The more recent the common ancestor, the more closely related they are.



17. Meiosis

Meiosis is a type of cell division used to produce gametes. In the process of meiosis:

1. A copy of the genetic information is made (46 → 92 chromatids)
2. The cell divides twice (92 → 46 → 23 in each)
3. Four gametes are produced, all of which have half the genetic information/half the chromosomes of the original cell.
4. ALL four gametes produced are NOT identical to each other or to the parent cell.



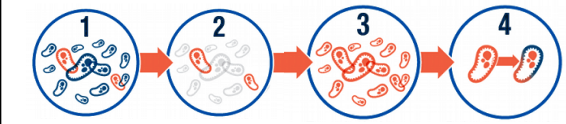
The gametes then go to find gametes of the opposite type/sex in sexual reproduction and when they fuse. This results in **fertilisation**, which **restores the full number of chromosomes** e.g. 23 chromosomes in sperm + 23 chromosomes in egg = 46 chromosomes in fertilised egg.

This **produces variation** because there is **mixing of the genetic information** from two different organisms.

The fertilised egg divides then by **mitosis** (one division, all cells identical) to grow into an embryo. Once big enough the cells in the embryo start to differentiate into different types of specialised cells.

14. Evidence for evolution - antibiotic resistant bacteria

Bacteria evolve rapidly because they reproduce at a faster rate. Some bacteria have evolved to become resistant to antibiotics:



1. High number of bacteria. A few of them are resistant to antibiotics due to a mutation in their DNA.
2. Antibiotics kill bacteria causing the illness, as well as good bacteria protecting the body from infection.
3. The resistant bacteria now have preferred conditions to grow and take over because they are no longer in competition with the other bacteria.
4. They survive and reproduce (survival of the fittest).
5. All the bacteria present are antibiotic resistant and they can even transfer their drug-resistance to other bacteria.

To reduce the rate of development of resistant strains:

- Doctors must not prescribe antibiotics for non-serious or non-bacterial infections.
- Patients must complete the full course of antibiotics.
- Agricultural use of antibiotic (e.g. in chickens) must be restricted.

The development of new antibiotics is costly and slow and is unlikely to keep up with the emergence of new resistant strains of bacteria.

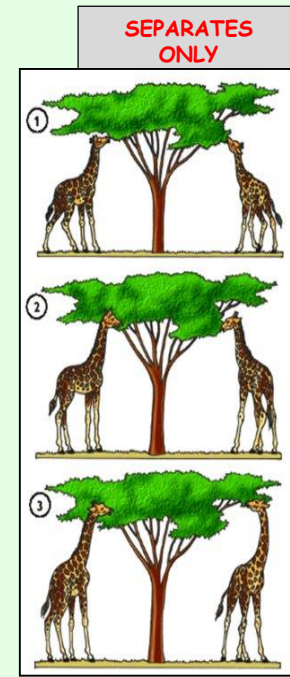
18. Evolution Theories

Lamarck - the more a characteristic is used the more developed it becomes and is then passed on to offspring. (which is nonsense!)
E.g. Giraffes stretched their necks to reach higher food and passed on the characteristic to their offspring.

Darwin proposed that a mutation made some giraffes have longer necks so they would be more likely to eat, survive and reproduce.

Darwin published his ideas in "Origin of the species" (1859) to much controversy. His ideas were not initially accepted because:

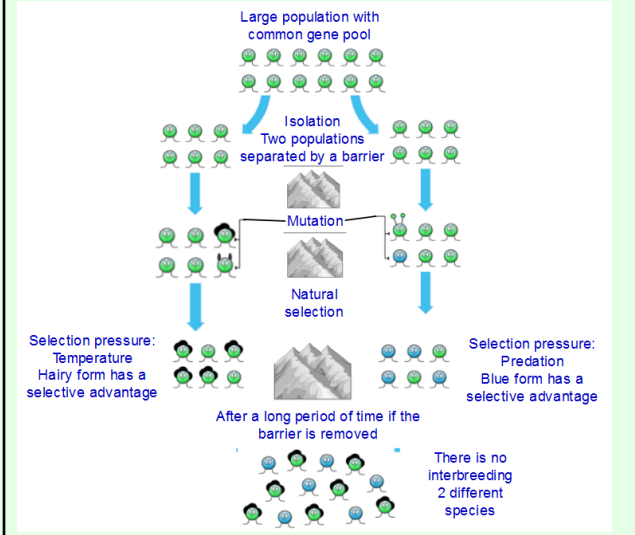
- It challenged existence of God
- There was insufficient evidence at the time (from fossils)
- Mechanisms of inheritance and variation was unknown (until 50 years later).



19. Speciation - making a new species

A new species is made by:

SEPARATES ONLY



Speciation is the process by which a population is divided by a barrier and different selective pressures exist on the two separate populations so that when mutation and natural selection occur different phenotypes survive in the two populations. These differences increase over time until the two populations are genetically dissimilar enough that they are now two separate species that can no longer interbreed.

20. Animal Cloning

SEPARATES ONLY

Adult Cell Cloning - makes copy of adult

1. Take nucleus out of an egg cell
2. Take nucleus from an adult cell
3. Put adult nucleus into empty egg cell
4. Electric shock to fuse nucleus to empty egg cell
5. When it becomes an embryo, insert into uterus

Embryo Transplant - makes cloned offspring

1. Sperm and egg mixed in petri dish
2. Grow into an embryo
3. Split the embryo into separate cells
4. Each cell develops into an identical embryo
5. Insert into the uterus of many host organisms.

21. Plant Cloning

Cuttings - Cut a bit off the stem, add auxins to stimulate root growth and plant it.

- + Cheap and quick
- Only a few clones created

Tissue Culture - Meristem cells put in growth medium with hormones forming tiny plantlets.

- + Grown all year
- + Can make lots
- More expensive.

Know your definitions:

1. Gamete - sex cells of an organism that only contain half the genetic information of normal cells e.g. sperm/egg in humans or pollen/egg in plants.
2. Chromosome - a structure made up of very long pieces of DNA wound up into coils.
3. Gene- a small section of DNA on a chromosome that codes for a particular sequence of amino acids to make a protein.
4. Allele - the form of the gene present
5. Dominant - a dominant allele is always expressed, even if only one copy is present.
6. Recessive - a recessive allele is only expressed when two copies are present (or when no dominant alleles are present)
7. Homozygous - when the two alleles present for a gene are the same
8. Heterozygous - when the two alleles present for a gene are not the same.

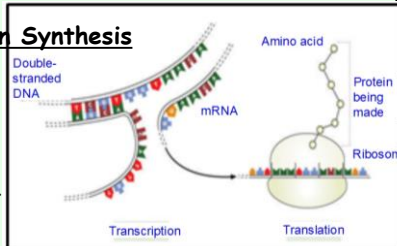
22. History of Genetics

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- Mendel studied pea plants and discovered that characteristics are controlled by 2 'units' that can be dominant or recessive.
- In the late 19th century behaviour of chromosomes during cell division was observed.
- In the early 20th century it was observed that chromosomes and Mendel's factors behaved in similar ways, leading to the idea that the factors (genes) were located on chromosomes.
- In the mid-20th century the structure of DNA was determined and the mechanism of gene function worked out.

23. HIGHER Protein Synthesis

1. DNA strands unwind.
2. A corresponding template of ATGC is made called mRNA.
3. This leaves the nucleus and binds to a ribosome.
4. With the help of tRNA, amino acids are made.
5. The protein is then released from the ribosome.
6. Protein folds into unique shape and can then do its job (hormone, enzyme, structural)



A change in the DNA base pair code can change the amino acid used in the protein chain - many do not, but if they do the protein may no longer fold up into the correct shape.

Know your definitions:

9. Genotype - the combination of two alleles present for a gene
10. Phenotype - the expressed characteristic as a consequence of the genotype
11. Genome - the entire genetic material of an organism
12. Mutation - an accidental change in the genetic code, which might be caused by ionising radiation or chemicals
13. Natural selection - a process by which variants of a species that are best adapted to live and survive in their environment are the ones that successfully reproduce and pass on their characteristics/phenotypes that helped them survive to their offspring.
14. Selective breeding - the choosing of parents with a desired characteristic from a mixed population, which are bred together and the offspring of which who show the desired characteristic are bred again, over many generations until all the offspring show that desired characteristic.
15. Genetic engineering - a process of modifying the genome of an organism by introducing a gene from another organism to give a desired characteristic
16. Extinction - when there are no remaining individuals of a species still alive.

24. Sexual vs asexual reproduction

SEPARATES ONLY

Pros of sexual reproduction

- Variation in offspring
- Survival advantage when a change in the environment occurs.
- Humans can selectively breed the species to increase food production

Pros of asexual reproduction

- Only one parent needed
- More efficient as do not need to find a mate/other type of gamete
- Faster form of reproduction
- Many offspring can be produced in favourable conditions.

Some organisms use both sexual and asexual reproduction to get the best of both:

Malarial protists

- Asexual reproduction in human host when conditions are favourable
- Sexual reproduction in mosquito host

Fungi

- Asexual reproduction to make spores
- Sexual reproduction by joining of gametes to make spores

Some plants

- Asexual reproduction through runners or bulbs
- Sexual reproduction by seeds

Know your definitions:

SEPARATES ONLY

17. Tissue culture - using a small group of cells from part of a plant to grow identical new plants.

18. Embryo transplant - splitting apart cells from a developing animal embryo before they become specialised, then transplanting the identical embryos into host mothers.

19. Speciation - the process by which a population is divided by a barrier and different selective pressures exist on the two separate populations so that when mutation and natural selection occur different phenotypes survive in the two populations. These differences increase over time until the two populations are genetically dissimilar enough that they are now two separate species that can no longer interbreed.