

 **SL IB Biology**
Your notes

Diversity of Organisms

Contents

- * Biological Species Concept
- * Chromosome Number
- * Karyograms: Skills
- * Genomes
- * Comparing Genome Sizes: Skills
- * Uses of Genome Sequencing



Your notes

Biological Species Concept

Variation Between Organisms

What is variation?

- **Differences** exist between organisms; these differences are known as **variation**
- There are multiple aspects of an organism that can vary, e.g.
 - Visual appearance, such as fur colour or body length
 - Behaviour, such as mating rituals and level of aggression
 - Biochemistry, such as antibiotic resistance or metabolic products

Causes of variation

- Variation is the result of a **combination of genetic and environmental factors**
 - The genes determine **which proteins** an organism is capable of producing, so influencing an organism's characteristics
 - Genetic variation is generated when mutation occurs and when alleles are combined in different ways during sexual reproduction
 - The environment may determine whether or not an organism has the resources needed to produce a particular protein, so **may affect gene expression**
 - Factors in the environment that may generate variation include
 - Environmental temperatures
 - Nutrient availability
 - Oxygen concentration

Variation between and within species

- Variation exists **between organisms of different species**
 - This variation can be used to **classify organisms** into different groups, e.g. morphological differences between species have historically been the main way of classifying organisms, and can still aid classification today
- Variation exists **between members of the same species**
 - While members of a species will have a similar genetic makeup, different individuals have different combinations of alleles
- **No two individuals are identical** when all characteristics are compared
 - Even identical twins, which have the same combination of alleles, will differ due to subtle environmental differences

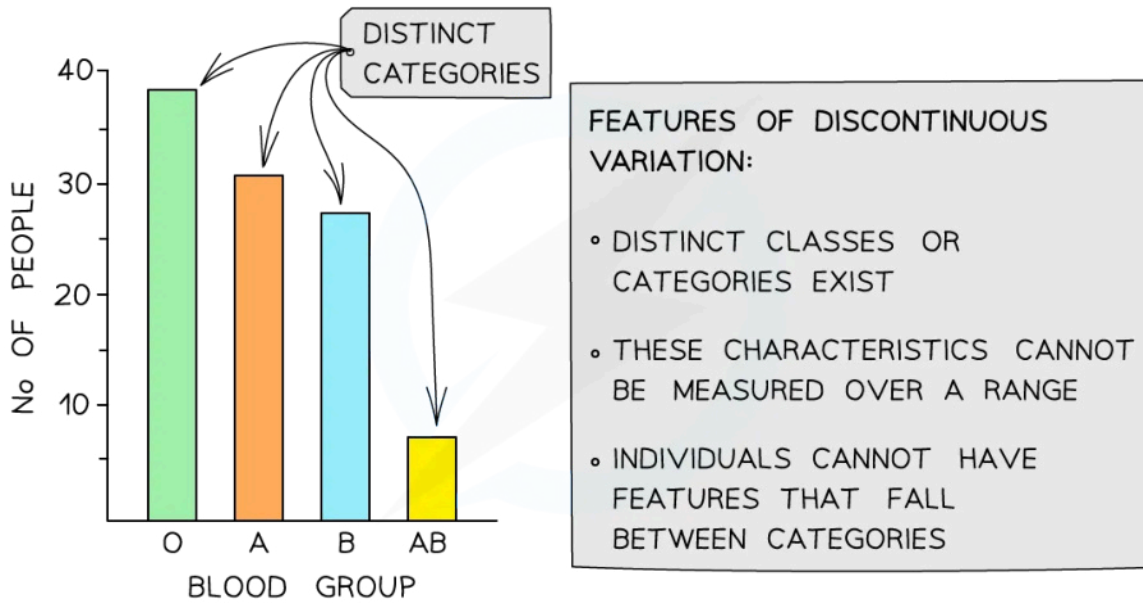
Types of variation

- Variation can be **discontinuous**, meaning that characteristics fall into **distinct categories**, e.g. black fur vs brown fur, or human blood type
- Variation can be **continuous**, meaning that characteristics can be **measured incrementally** on a scale, e.g. height

Discontinuous and continuous variation graphs

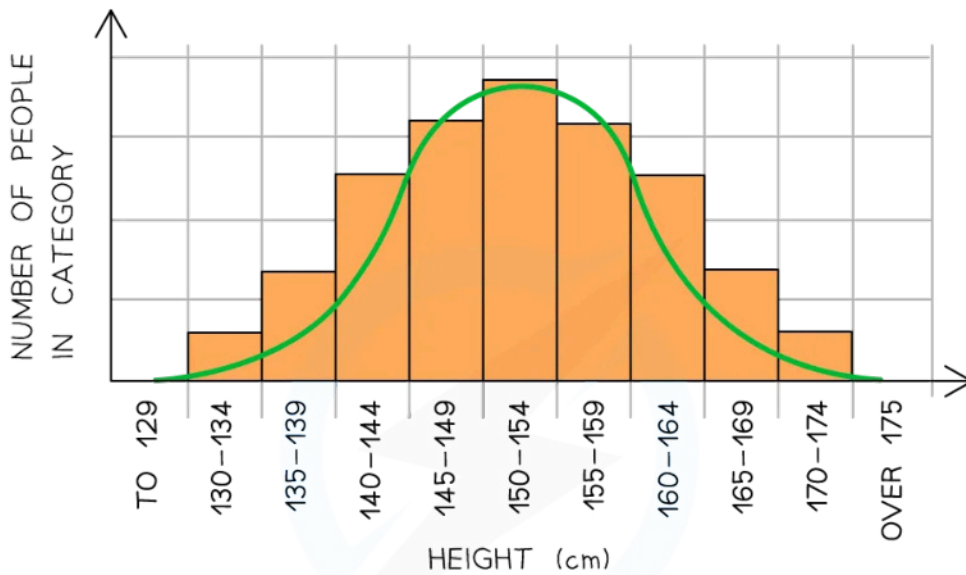


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FEATURES OF CONTINUOUS VARIATION:

- NO DISTINCT CLASSES OR CATEGORIES EXIST
- CHARACTERISTICS CAN BE MEASURED AND FALL WITHIN A RANGE BETWEEN TWO EXTREMES

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Discontinuous and continuous variation have different features



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Species: Linnaeus

Species classification using morphology

- For biologists to make sense of the huge array of species on Earth, organising them into **logical groups** is essential
 - This process of putting organisms into groups is known as **classification**
 - The science of classification is known as **taxonomy**, and scientists working in the field of taxonomy are **taxonomists**
- Classifying an organism involves deciding which biological group, or **taxon** (plural taxa), it fits into best, and then **naming** it according to its taxon
 - The smallest taxonomic group is the **species**
- Historically an organism's species was determined on the basis of its **observable characteristics**; this is **morphological classification**
- The morphological species concept states that:
 - A species is a group of organisms that are morphologically unique**
- While morphology can still be a useful guide for taxonomists, **classification is no longer carried out using the morphological species concept** alone

Linnaeus: the father of taxonomy

- Carl Linnaeus** was an 18th century Swedish botanist, famous today for his work on taxonomy
- Linnaeus' developed the **method of naming species** that is used by scientists all over the world today
 - He is sometimes referred to as the 'father of taxonomy' for this reason
- Linnaeus noticed that traditional methods of naming species were **long and descriptive**, for example the tomato plant was named *Solanum caule inermi herbaceo, foliis pinnatis incisae, racemis simplicibus*, meaning 'solanum with the smooth stem which is herbaceous and has incised pinnate leaves'
- Under Linnaeus' new system, species were given **two-part Latin names** which would be **the same everywhere in the world**, e.g. the tomato became *Solanum lycopersicum*
 - Many species still have the same two-part Latin names that Linnaeus gave them
- It is worth noting that while Linnaeus' work on taxonomic naming shaped modern taxonomy, he **didn't always get his classification correct**; many species named by him have since been reclassified and given new two-part Latin names
 - Linnaeus used the morphological species concept; **classification based on morphology alone often leads to mistakes**; species with a similar appearance are not always closely related



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TRADITIONAL NAME

LINNAEUS' BINOMIAL LATIN NAME



*Solanum caule inermi herbaceo,
folii pinnatis incisae, racemis
simplicibus*

Solanum lycopersicum

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Linnaeus' binomial naming system allowed species to be given simplified names that scientists all over the world would recognise



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Binomial System

The binomial naming system

- The biological system of naming, used to name species according to their taxa, is known as the **binomial system of nomenclature**
 - This system is **universal**, ensuring that scientists around the world all use the same method of naming species
- The system involves giving a species a **two-part name**, hence **binomial**
- Both parts of the name are in **Latin**, or a **latinised version** of a non-Latin word
 - e.g. *Eriovixia gryffindori* is a species of orb spider named after a famous school house
- The first part of the name is an organism's **genus**, and the second is its **species** name
 - E.g. the binomial name of a wolf is *Canis lupus*; wolves belong to the genus *Canis*, and the species *lupus*
- Species that are grouped into the same genus will have **similar characteristics**
 - E.g. the genus *Canis* includes the wolf (*Canis lupus*), the coyote (*Canis latrans*), and the domestic dog (*Canis familiaris*)

Using binomial names

- There are several **conventions**, or rules, that should be used when writing binomial names
 - The **genus should begin with a capital letter**, and the **species with a lower-case letter**, e.g. the honey bee is *Apis mellifera*
 - When typed, binomial names should appear in italics, and when written by hand, they should be underlined e.g. a limpet is *Patella vulgata* when typed, or Patella vulgata by hand
 - The first time a binomial name is used in a text it should appear in full, e.g. wheat is *Triticum aestivum*, but the genus name can from then on be abbreviated so that the name is given as *T. aestivum*



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Biological Species Concept

Biological species concept

- The morphological species concept relies on the **observable features** of a species, and often leads to mistakes in classification
 - Species with similar traits may not be closely related
- Biologists now rely on other definitions of a species, one of which is the **biological species concept**
- The biological species concept states that a species is:
A group of organisms that can interbreed to produce fertile offspring

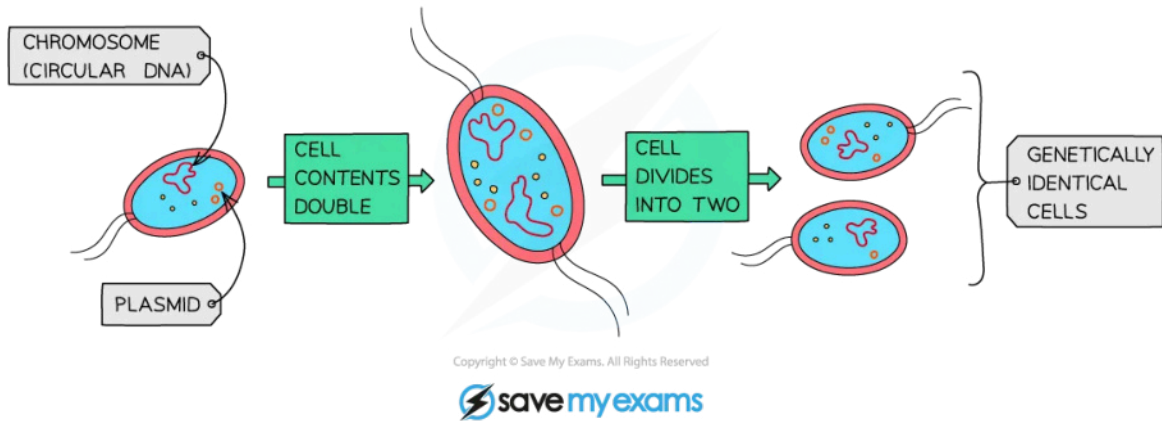
Limitations of the biological species concept

- The biological species concept can be very useful to biologists, but there are some situations to which it can be difficult to apply:
 - **Asexual reproduction**
 - Organisms that reproduce by asexual reproduction cannot be classified using this method
 - E.g. **bacteria reproduce asexually**, so the question of whether or not they can breed together is irrelevant
 - **Fertile hybrids**
 - On rare occasions, animals of different species breed together and produce **fertile offspring**
 - E.g. the so-called 'wholphin' is the fertile offspring from a cross between a melon-headed whale and a common bottlenose dolphin
 - According to the biological species concept the wholphin would be a new species, but while scientists do believe that hybridisation can lead to new species, it needs to be a **frequent event** for this to occur, and wholphins are rare
 - Note that the melon-headed whale is actually a species of dolphin, so the name 'wholphin' is a bit inaccurate!
 - **Extinction**
 - Some species are extinct, so it is not possible to breed them together with members of an existing species to determine the fertility of their offspring
 - E.g. the woolly mammoth is quite similar in morphology to modern elephants, though it is classified as a different species; there is no way of checking this classification using the biological species concept

Asexual reproduction in bacteria diagram



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The biological species concept cannot be applied to bacteria because they reproduce asexually

- The imperfect nature of the biological species concept means that **other characteristics must sometimes be used** to determine species
 - **Morphology:** organisms of the same species share similar morphology
 - **DNA:** sequences can be compared, with a certain level of similarity indicating that organisms are the same species
 - **Biochemistry:** species may produce different molecules as products of their metabolism
 - E.g. some bacteria produce carbon dioxide, while others may produce methane
 - **Ecology:** the precise ecological niche of a species is likely to be distinctive from other, similar species
 - **Evolutionary lineage:** fossilised remains of extinct species can be compared with morphologically similar existing species and classified within their evolutionary lineage
- The characteristics used to aid classification will **differ depending on the organism**, e.g:
 - Bacteria may have very similar morphology, so may need to be classified on the basis of their biochemistry or their ecology
 - We might know very little about the biochemistry and ecology of a long-extinct species, but we can classify it according to its evolutionary lineage or morphology



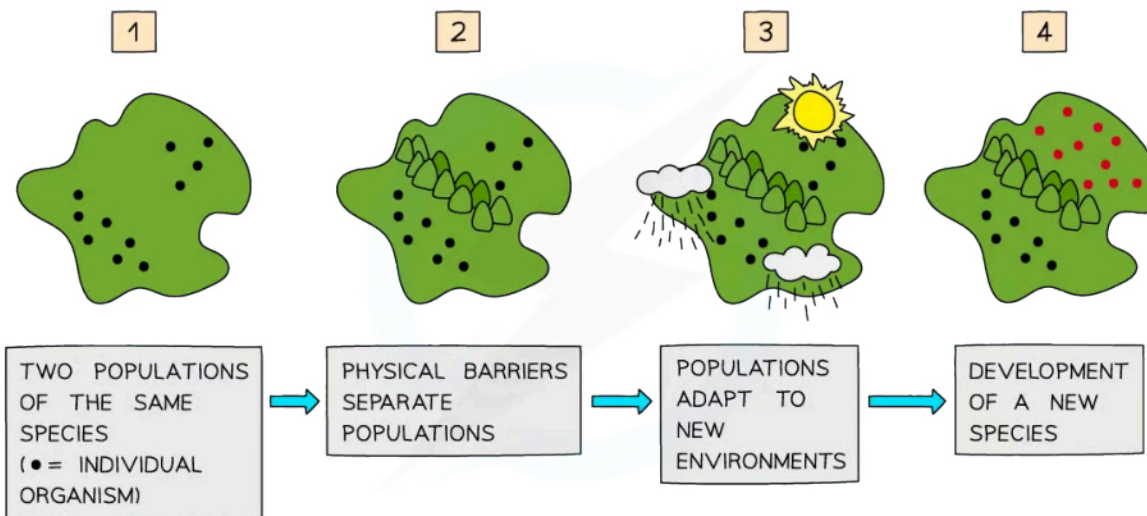
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Distinguishing Between Populations & Species

Speciation

- Species **do not stay the same over time**; the species that we see around us today have developed over millions of years
 - This process of species change is known as **evolution**
- The process by which one species gives rise to two or more new species is **speciation**
 - Speciation can occur when a population becomes isolated from other populations of the same species due to living in a different area
 - This isolation means that members of the separate populations cannot breed together and **gene flow cannot take place** between them
 - If the environmental conditions affecting each population are different, then natural selection could act differently on each population and eventually lead to speciation
 - Genetic drift can also lead to speciation
 - Once speciation has taken place, the two species can **no longer breed to produce fertile offspring**; they are **reproductively isolated** and are said to be **separate species**
- Note that speciation is covered in more detail [later in the course](#)

Speciation diagram



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Speciation can occur when gene flow does not occur between two populations of the same species

Distinguishing between populations and species

- The process of speciation occurs over very long time periods, and the differences between isolated populations accumulate incrementally
- In most cases it is likely that **the ability of two populations to interbreed successfully declines gradually**, rather than a sudden cut-off point occurring, meaning that it is difficult to pinpoint the stage

at which two separate populations have become two new species

- The **decision as to when to assign separate species status to two populations can therefore seem arbitrary**, and is often down to the opinions of scientists, i.e. it is **subjective**
 - E.g. killer whales (*Orcinus orca*) show significant variation between populations, and are currently said to consist of several 'ecotypes', but some scientists believe that there could in fact be more than one species of orca



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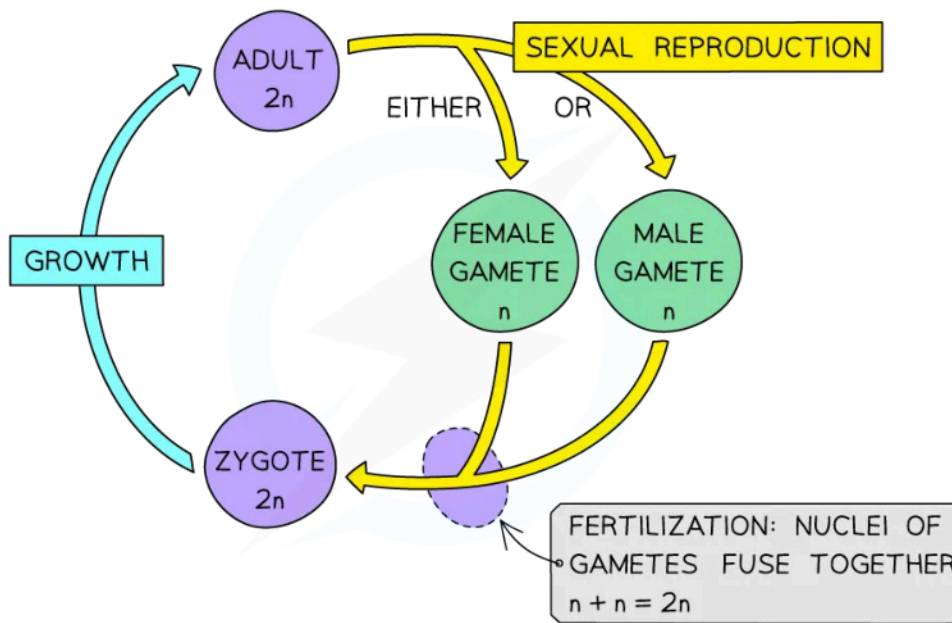
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Chromosome Number

Chromosome Number

Diploid & haploid

- A **diploid** cell is a cell that contains **two complete sets** of chromosomes ($2n$)
 - Adult body cells are usually diploid cells
- **Haploid** cells contain **one complete set** of chromosomes (n), meaning that they have **half** the number of chromosomes compared to normal body cells
 - In most cases these haploid cells are called **gametes** and they are involved in **sexual reproduction**
 - In animals, they are the female egg cell and the male sperm cell
 - There are some unusual species which have haploid cells for other parts of their life cycle
- During **fertilisation** the nuclei of haploid gametes **fuse together** to form the nucleus of a diploid zygote
- Both gametes must contain the **same number of chromosomes** in order for the zygote to be viable.
 - For a diploid zygote this means that the gametes must be haploid
- **Every body cell** that arises from the zygote will contain **the same number of chromosomes**
 - Exceptions to this include red blood cells, which have no nucleus and so contain no chromosomes



Chromosome number is halved in gametes, and restored to diploid after fertilisation

Chromosome number

- The number of chromosomes possessed by different species **varies** and is dependent upon changes that have occurred during that species' evolution



- Each individual in a species always has the **same number of chromosomes**
 - There are a few rare instances where a chromosome mutation has occurred, giving a different chromosome number
- Differences in chromosome number is one reason why organisms from different species are **unable to breed together** successfully
- When stating the chromosome number of an organism, it needs to be clear whether you are giving
 - The number of chromosomes found in a **diploid cell**
 - The **number of pairs of chromosomes** found in a diploid cell
 - This will be the same as the haploid chromosome number
 - The number of chromosomes found in a **haploid cell**

Species chromosome number table

Name of species	Diploid chromosome number (2n)	Haploid chromosome number
Human (<i>Homo sapiens</i>)	46	23
Chimpanzee (<i>Pan troglodytes</i>)	48	24
Rice (<i>Oryza sativa</i>)	24	12
Horse threadworm (<i>Parascaris equorum</i>)	4	2

Different species have different numbers of chromosomes

- The diploid number must always be an **even number**
 - This is because the diploid number (2n) must **always be divisible by two** to produce a whole haploid number (n)
- Note that the number of chromosomes a species possesses is not linked to how 'advanced' a species is in evolutionary terms

Examiner Tip

Note that you need to know the diploid chromosome numbers of **humans (46)** and **chimpanzees (48)**, but that you do not need to learn any other specific chromosome numbers



Your notes

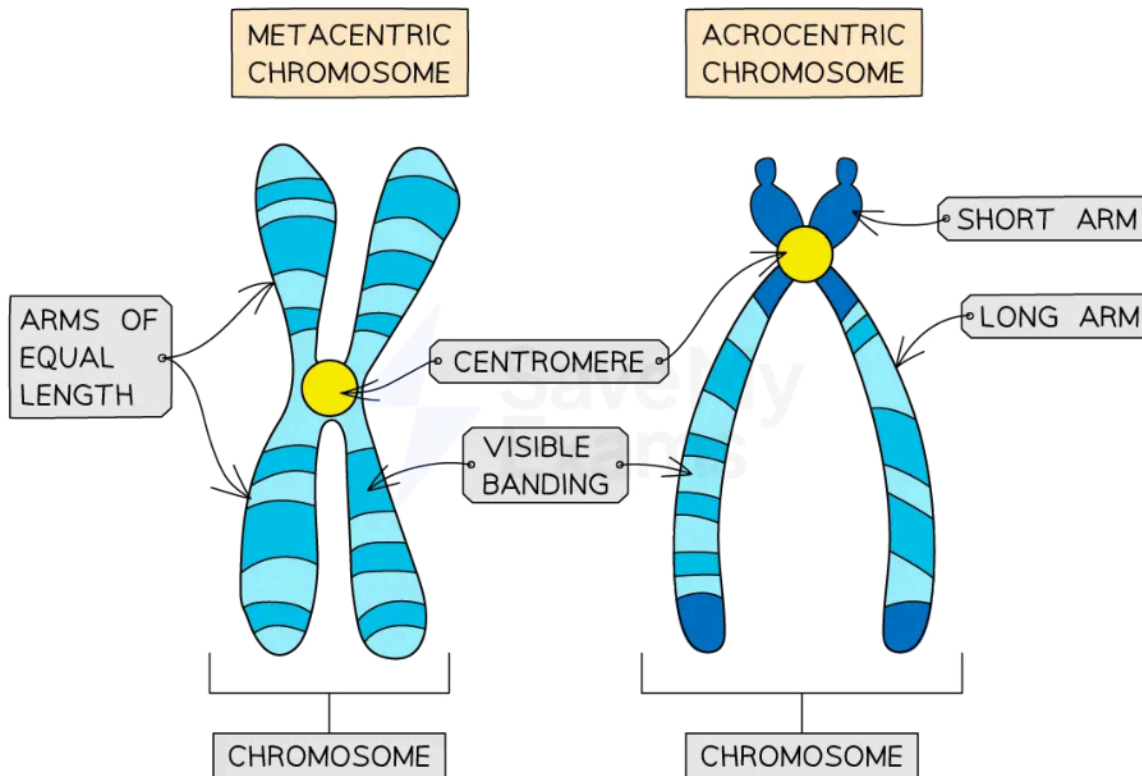
Karyograms: Skills

Karyograms

Chromosomes

- Chromosomes are **strands of DNA**, along which are sections known as genes
 - A gene is a section of DNA that codes for one polypeptide
- Chromosomes that have undergone DNA replication have the appearance of an 'X' shape, where the 'legs' of the X are made up of two strands of DNA attached at a region known as the **centromere**
 - Chromosomes with a centromere located roughly in the middle are known as **metacentric** chromosomes
 - Chromosomes with the centromere near the end are **acrocentric**
- Chromosomes that have been stained with a dye have a **banded** appearance

Metacentric and acrocentric chromosomes diagram



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Chromosomes with a central centromere and arms of equal length are metacentric, and chromosomes with a near-terminal centromere and arms of unequal length are acrocentric

Karyotypes



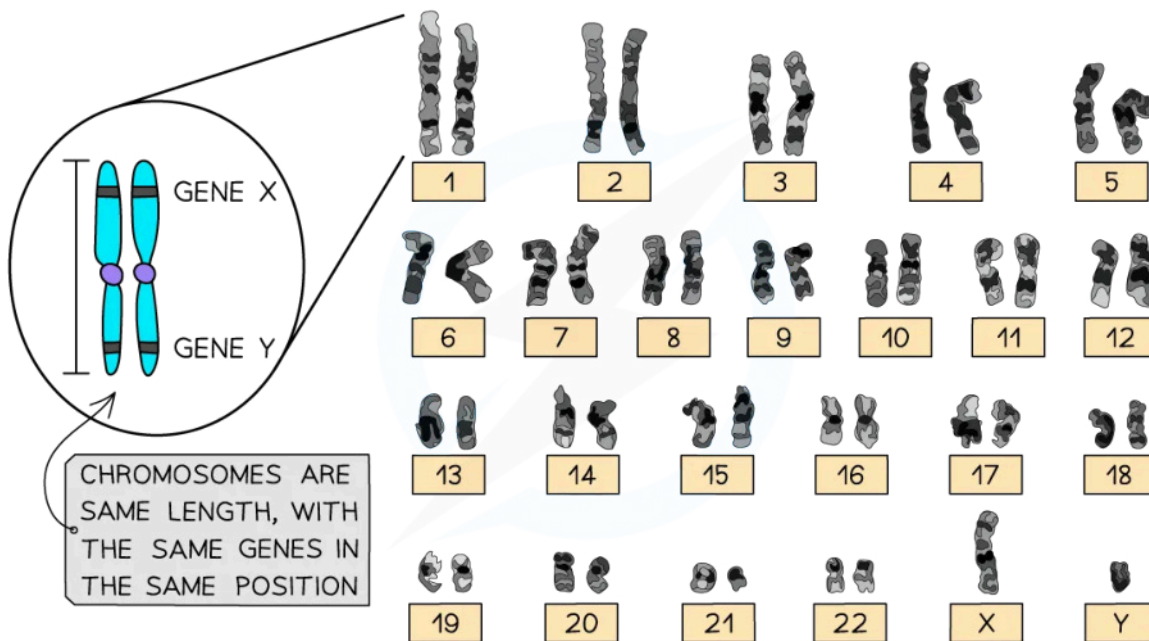
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- A **karyogram** is an image that shows **all of the chromosomes in a cell**, arranged by size, shape, and banding pattern, and placed with their homologous pairs
- A karyogram shows the **karyotype** of an individual, which can be defined as

The appearance of a complete set of an individual's chromosomes, including their number, size, shape, and banding

- Note that chromosome pair 23 often does not fit the size-order pattern, as pair 23 contains the **sex chromosomes** and the X chromosome is very large

Human karyogram diagram



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A karyogram contains an individual's chromosomes arranged in homologous pairs. It shows a karyotype; the appearance of a complete set of chromosomes arranged by size, shape, and banding pattern

Making a karyogram

- A karyogram can be produced as follows
 - Cells are **stained** and viewed under a **light microscope**
 - Photographs are taken of the contents of the nucleus during metaphase of cell division
 - The photographs of the chromosomes are cut up and **arranged by size, shape, and banding pattern**
 - This can be done with paper and scissors or on a computer

Evaluating chromosome evidence relating to human evolution



Your notes

- As you will know from the section on **chromosome number**, **humans have 46 chromosomes**, while **chimpanzees have 48**
 - Gorillas and bonobos also have 48 chromosomes
- Given that evidence shows that **humans share a recent common ancestor** with these species, this raises the question of **how these extra chromosomes were lost** during evolution
 - A whole pair of chromosomes must be removed for 48 chromosomes (24 pairs) to become 46 (23 pairs)
- The possible mechanisms by which this loss could have occurred when humans split from chimpanzees and gorillas include:
 - A pair of chromosomes **disappeared** from the genome
 - A pair of chromosomes **fused with another pair** to form a single pair
- The loss of an entire pair of chromosomes would have had a significant effect on the characteristics of human ancestors, and may have put their survival at risk, so the first mechanism is unlikely to be correct, but scientists have been able to use karyograms to test the following hypothesis:

Chromosomes in pairs 12 and 13 in a common ancestor fused to form the chromosomes in human pair 2

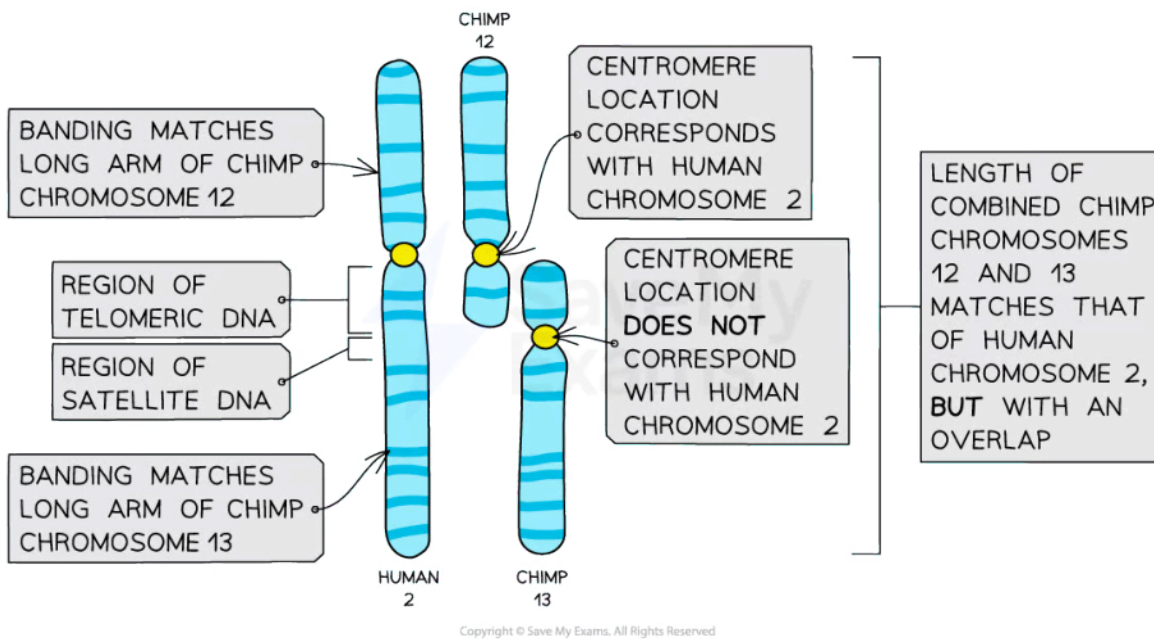
(Note that from this point onwards the notes will only refer to one chromosome from each pair, but the same will also be true for the other member of the pair)

- Evidence that **supports this hypothesis** includes:
 - Chimpanzee chromosomes 12 and 13, when placed end-to-end, **match the length** of human chromosome 2
 - The **location of the centromere** of chimpanzee chromosome 12 matches that of human chromosome 2
 - Human chromosome 2 contains a region of non-coding DNA known as satellite DNA that corresponds to the location of the centromere in chimpanzee chromosome 13; this could be a remnant of a centromere
 - The **banding** of the long arms of acrocentric chimpanzee chromosomes 12 and 13 corresponds to the banding of metacentric human chromosome 2
 - Human chromosome 2 contains telomeric DNA in the middle of the chromosome
- Evidence that **does not support this hypothesis** includes:
 - The length of chimpanzee chromosomes 12 and 13 combined is **not a perfect match** for human chromosome 2; there is a slight overlap
 - The location of the centromere of chimpanzee chromosome 13 **does not match** that of human chromosome 2
- Note that chimp chromosomes 12 and 13 are sometimes referred to as **chromosomes 2A and 2B**, in acknowledgement of the fusion event discussed above

Chromosome fusion evidence diagram



Your notes



Comparing the structures of chimp chromosomes 12 and 13 with human chromosome 2 provides evidence both for and against the chromosome fusion hypothesis

NOS: Distinguishing between testable hypotheses and non-testable statements

- The hypothesis above, relating to the origin of chromosome 2, is an example of a **testable hypothesis**
- For a hypothesis to be **testable**, it must have the following characteristics:
 - There needs to be **access to evidence** that supports it or refutes it
 - The hypothesis above about the fusion of ancestral chromosomes 12 and 13 can be **tested by examining chromosome evidence** from humans and modern chimps
 - A hypothesis such as '*the common ancestor of humans and chimps enjoyed singing*' would not be testable, as there is **no available evidence** to support or refute it
 - The hypothesis needs to be a **testable statement**:
 - The hypothesis written above is '*chromosomes in pairs 12 and 13 in a common ancestor fused to form the chromosomes in human pair 2*'; this statement can be **accepted or rejected**
 - Another example might be '*organisms with a large surface area to volume ratio lose heat more quickly*'; an investigation can be carried out to determine whether to accept or reject this statement
 - A hypothesis **should not contain vague statements** that use terms like '*may*' or '*could*'; this makes it difficult to entirely accept or reject a statement, e.g.
 - '*The fusion of ancestral chromosomes may have reduced the chromosome number*'
 - '*Surface area to volume ratio could affect the rate of heat loss*'
 - A hypothesis **should not make predictions**, e.g.
 - '*Surface area to volume ratio will affect the rate of heat loss*'
 - A hypothesis **should not draw causal conclusions**, e.g.

- 'A larger surface area to volume ratio causes increased heat loss'

Examiner Tip

Be careful to avoid statements that imply that humans have 'evolved from chimps', that is not what the chromosome evidence suggests. Instead, we should say that this evidence provides information about how humans and chimps may have **diverged from their common ancestor**.

This section about human and chimp chromosomes is about the skill of **evaluating evidence**; you don't need to learn all of the details given here, but you should know how to consider the evidence for and against a hypothesis.



Your notes



Your notes

Genomes

Genomes

Genomes

- The term genome can be defined as follows:
 - **All of the genetic information in an organism**
- This refers to the DNA present within **every cell** of an organism
- This includes **genes** that code for proteins as well as **non-coding DNA** sequences
- Mitochondrial DNA and chloroplast DNA are included in the genome of eukaryotic cells
- In a prokaryote cell, plasmid DNA is included in the genome

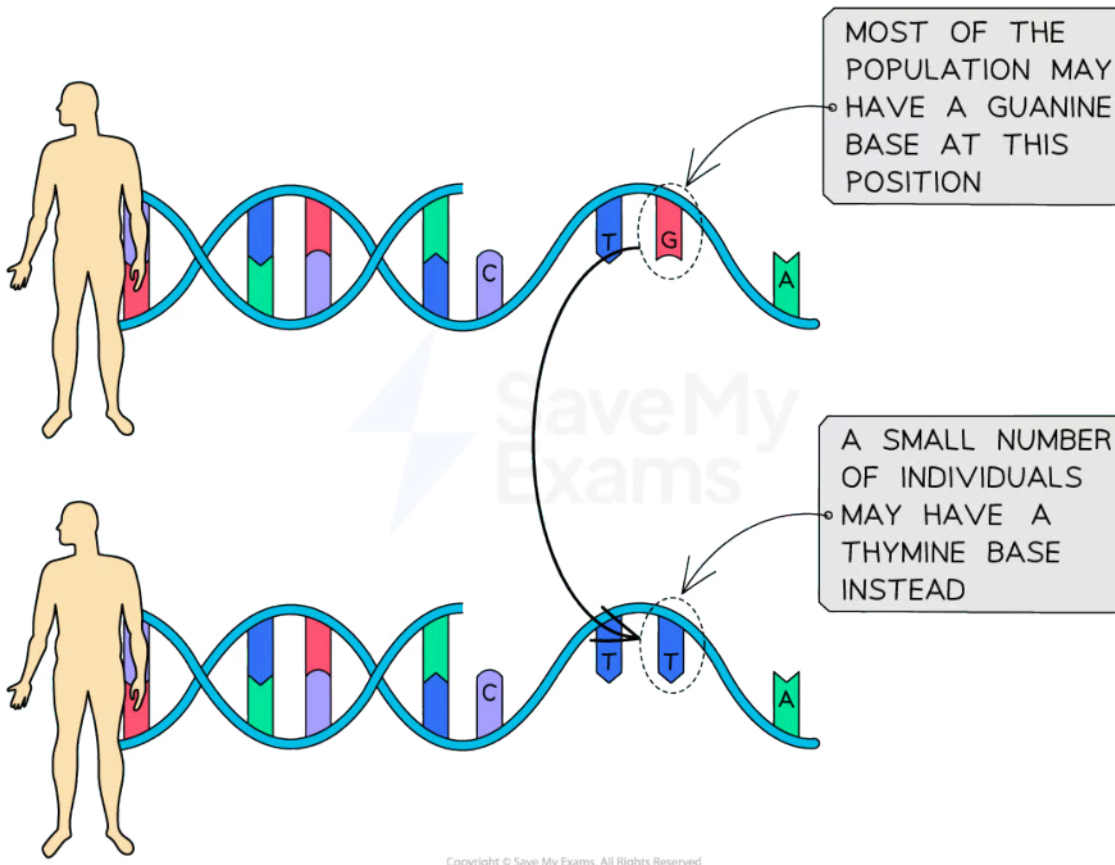
Unity and diversity of genomes

- Advances in technology have allowed scientists to **determine the entire base sequence** of the genes within an organism's genome
 - Determining the base sequence of DNA is referred to as **DNA sequencing**
- Genome-wide comparisons can now be made between individuals and between species
- Comparisons show that there is a **high level of genome similarity** within, and even between, species
 - Humans share around 99.9 % of their DNA with other humans
 - Humans share around 99 % of their DNA with chimpanzees
- Humans in fact share **all of their coding genes** with other humans; the differences between individual humans are due to possessing different alleles of genes
 - Different alleles arise due to mutations
- Differences between DNA sequences that involve a single base change are known as **single nucleotide polymorphisms (SNPs)**
 - Because SNPs are areas of difference between individuals who share 99.9 % of their DNA, scientists will often use SNPs to determine ancestry or disease risk

Single nucleotide polymorphism diagram



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A difference in a single nucleotide is known as a single nucleotide polymorphism, or SNP. Most SNPs have no effect on the protein produced, but a few may lead to production of an altered protein.

Eukaryote Genome Diversity

Comparing eukaryotic genomes

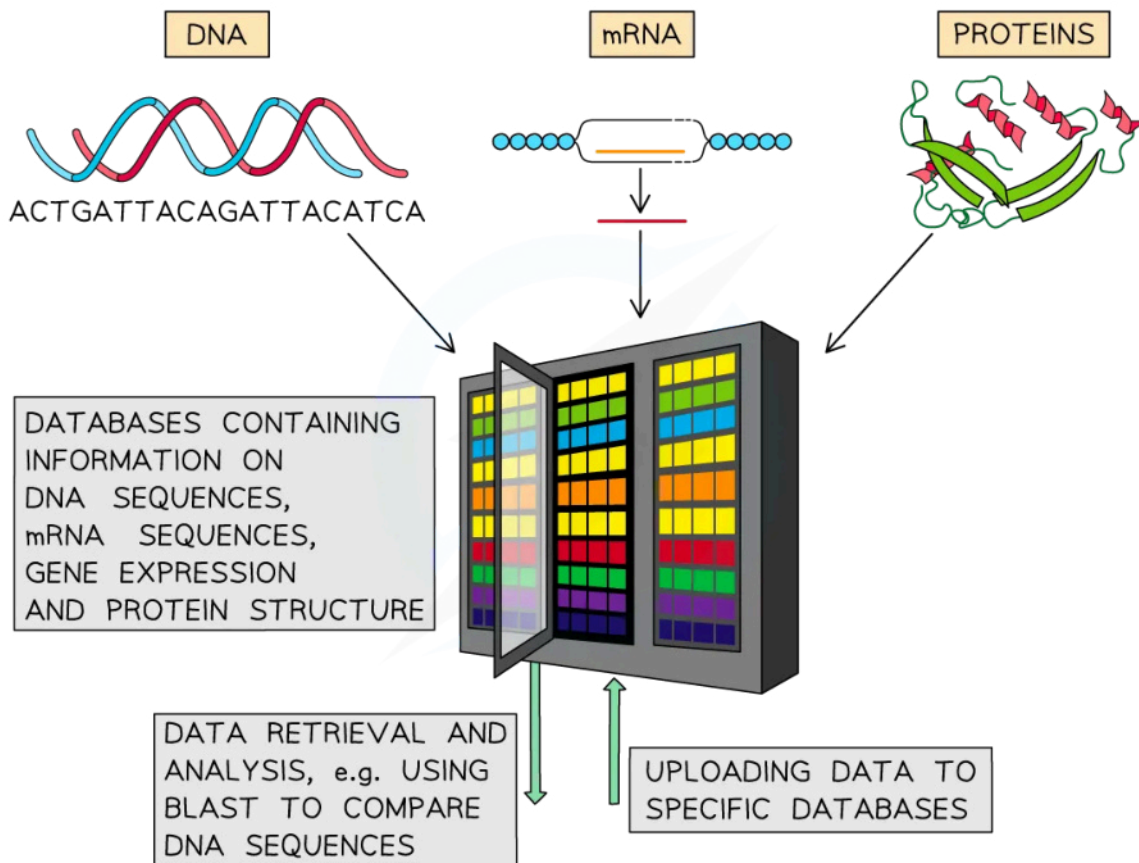
- Eukaryotic genomes can **vary in size**; this is determined by the mass of DNA present in a nucleus
 - Size variation means that some organisms will have genes that others lack
 - E.g. plants need genes that code for enzymes involved in photosynthesis, while humans do not need these genes
- Eukaryotic genomes can also **vary in base sequence**
 - DNA sequence data can be compared using information from **online databases**
 - Scientists all around the world enter information into such databases, allowing anyone to retrieve the information for analysis
 - Examples of such databases include GenBank and NCBI
 - Databases allow you to:
 - Select a specific gene to compare
 - Select species to compare
 - Choose whether to compare amino acid sequences or DNA base sequences
- Comparison of eukaryotic species sequence data shows that **closely related individuals have more similar genomes** than distantly related individuals
 - E.g. individuals of the same species have more similar genomes than individuals of different species



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Databases contain information on DNA, RNA and protein sequences, as well as protein structure. This information can be retrieved for analysis of variation between genomes



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Comparing Genome Sizes: Skills

Comparing Genome Sizes

Comparing genome sizes

- Advances in technology have allowed scientists to **sequence genomes** of many species
- Genome-wide comparisons can now be made **between individuals** and **between species**
- Genome sizes can differ in different organisms:
 - Viruses and bacteria tend to have **very small genomes**
 - Prokaryotes tend to have smaller genomes than eukaryotes
 - The size of **plant genomes can vary widely**

Comparing the genome size of different organisms table

Organism	Common name / description	Genome size (million base pairs)
Enterobacteria phage T2	Virus that infects <i>E. coli</i>	0.17
<i>Escherichia coli</i>	<i>E. coli</i> bacteria	5
<i>Drosophila melanogaster</i>	Fruit fly	140
<i>Homo sapiens</i>	Human	3000
<i>Paris japonica</i>	Japanese canopy plant	150 000

Using a genome size database

- It is possible for anyone to look up the genome size of a wide range of organisms using a **genome size database**, e.g. the Animal Genome Size Database or the Plant DNA C-Values Database
- Databases present genome sizes using a measure known as a C-value
 - The C-value is the haploid nuclear DNA content of an organism
 - C-value units can be given in mass: picograms (pg) where $1 \text{ pg} = 10^{-12} \text{ g}$
 - C-value units can be in number of bases: megabases (Mb) where $1 \text{ Mb} = 10^6 \text{ bases}$
 - $1 \text{ pg} = 978 \text{ Mb}$

Genome size and organism complexity

- We might expect there to be a clear relationship between genome size and organism complexity, but there are **plenty of examples that do not fit with this pattern**, e.g.

- Humans = 3 100 Mb
- Hagfish = 4 200 Mb
- Common wheat = 17 000 Mb
- In some cases **similar species may have very different genome sizes**
 - E.g. common wheat above has a genome size of around 17 000 Mb, while red wild einkorn wheat has a genome size of around 5 000 Mb
- There are several factors that should be considered when thinking about the reason for these unexpected numbers:
 - The genome contains **all of an organism's DNA**, not just the DNA that codes for proteins
 - Plants can have **polyploidy**, meaning that their cells can contain many sets of chromosomes, giving them very large genomes
 - **Our view of 'complexity' may not be correct**; we generally associate complexity with brain function, but there are other ways in which an organism can be complex



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Uses of Genome Sequencing

Uses of Genome Sequencing

Genome sequencing

- DNA sequencing allows for the **base sequence** of an organism's genome to be identified and recorded
- Sequencing methods are continuously advancing to become **faster and cheaper**
 - Advances in technology have allowed scientists to **rapidly sequence** the genomes of organisms
 - Most sequencing methods used are now **automated**
- Newer methods of genome sequencing are known as **next-generation sequencing (NGS)** techniques
- The data obtained from sequencing can be **entered into computers** with specialised programmes that can **analyse the information** for purposes such as:
 - Determining evolutionary relationships
 - Personalised medicine

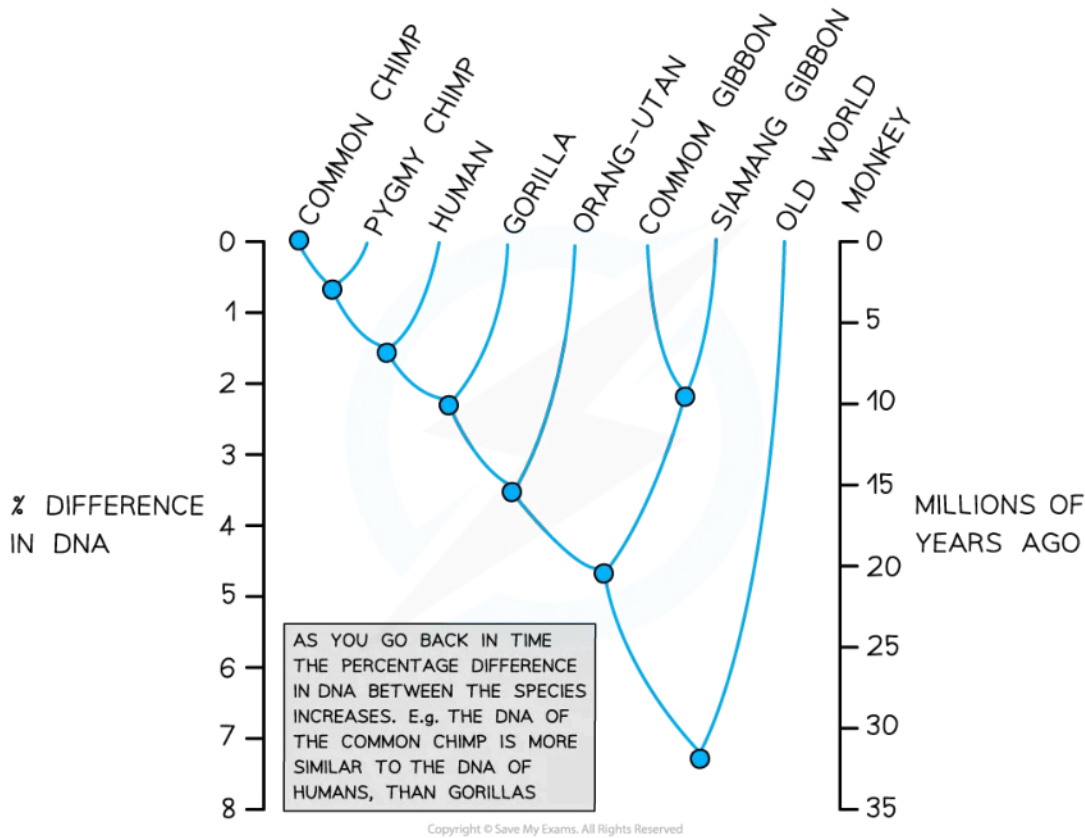
Genome sequencing & evolutionary relationships

- **Phylogenetics** is the classification of species based on their **evolutionary origins and relationships**
 - This means that organisms are grouped together on the basis of shared **common ancestry** rather than, e.g. similarities in appearance
- Advances in **sequencing technology** have allowed scientists to understand the true phylogeny of taxa
 - Note that this technology is especially useful for comparison with an **extinct species** (using ancient DNA) or when distinguishing between species that are **very physically similar**
- Three types of sequence data are used to investigate evolutionary relationships
 - DNA
 - mRNA
 - Amino acids (of a protein)
- For all types of sequence data it can be said that **the more similar the sequences, the more closely related the species are**
 - Two groups of organisms with **very similar sequences will have separated into separate species more recently** than two groups with less similarity in their sequences
 - Species that have been **separated for longer have had a greater amount of time to accumulate mutations** and changes to their DNA, mRNA and amino acid sequences
- Scientists will choose specific proteins or sections of the genome for comparison between organisms
 - Looking at **multiple proteins or multiple regions of the genome** will allow for a **more accurate estimate** of evolutionary relatedness
 - Note the protein used **needs to be present in a wide range of organisms** and **show sufficient variation** between species
 - Cytochrome c is often used as it is an integral protein in aerobic respiration, which occurs in many organisms
- Sequence analysis and comparison can be used to create **phylogenetic trees** that show the evolutionary relationships between species

Primate phylogenetic tree diagram



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Genome sequence data can be used to produce phylogenetic trees that accurately show the relationships between species

Genome sequencing & personalised medicine

- Information gathered from genome sequencing projects like the Human Genome Project can be used to develop **genomic medicine**, which uses **information about the genes to design medical treatments**
 - The Human Genome Project involved the sequencing of the entire human genome
- The information gained during the Human Genome Project is stored in **databases**, within which genes that code for certain proteins can be found and analysed
- Knowing the sequence and structure of proteins involved in disease allows the development of **drugs that target specific proteins**
 - E.g. if an enzyme is involved with disease, a drug that acts as an enzyme inhibitor can be developed
 - Targeted treatments can mean **fewer unpleasant side-effects** for patients
- By combining information about the genome with other clinical and diagnostic information, patterns can be identified that can help to **determine an individual's risk of developing disease**
 - **Genetic screening** allows individuals with a high chance of developing specific diseases to be identified and means that preventative measures can be taken, e.g.

- Certain genetic mutations are known to increase the risk of an individual developing breast cancer, so those who know that they have such a mutation can have surgery in advance to reduce their risk
- An individual may be able to make certain life choices regarding diet and lifestyle based on knowledge of their genetic risk of cancers and heart disease
- Doctors can also use an individual's genome to work out **how well they might respond to specific treatments**, allowing treatments to be selected on the basis of an individual's genotype



Your notes