

### 9.3 – Blueprint of Life:

#### 1. Evidence of evolution suggests that the mechanisms of inheritance, accompanied by selection, allow change over many generations:

- *Outline the impact on the evolution of plants and animals of:*
  - ◆ *Changes in the physical conditions in the environment:*
  - ◆ *Changes in the chemical condition in the environment:*
  - ◆ *Competition for resources:*
- **Evolutionary theory** states that all organisms have developed from previous organisms and that all living things have a common ancestor in some initial form of primitive life.
- It also states that all organisms are fundamentally similar because their basic chemistry was inherited from this very first organism.
- Changes in the Physical Environment:
  - The Earth has continually changed since life first evolved.
  - Changes in the environment force species to either die out, or survive and diversify.
  - An Example - The Peppered Moth:
    - Prior to the Industrial Revolution of the late 18<sup>th</sup> Century, most Peppered moths were light coloured (there was also a dark form). They survived better as they could camouflage against the white lichen on the trees.
    - During the revolution, the pollution caused the trees to blacken with soot. The trees could no longer hide the white moths. The darker variant of the moth was better able to hide, and so the population of the Peppered moth shifted from mainly white to mainly dark.
    - This is how a change in the environment can affect the evolution of an organism by selecting those more suitable to survive
- Changes in the Chemical Environment:
  - Chemical changes in the environment impact on the evolution of organisms by acting as a selection pressure that decides which organisms are the ‘fittest’.

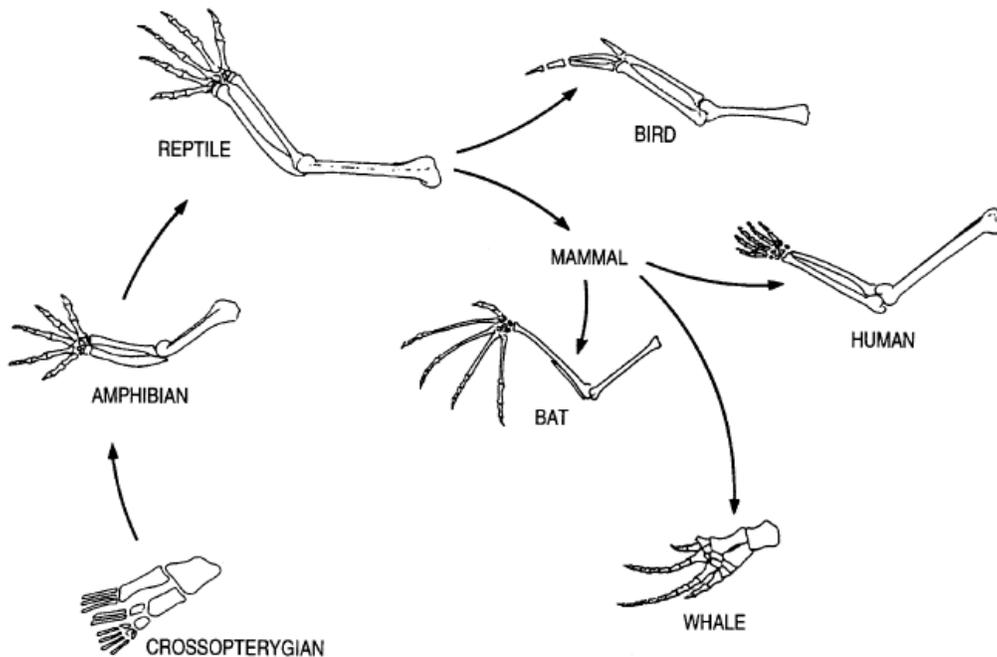
- An Example - Mosquitoes and DDT:
  - When DDT (dichloro-diphenyl-trichloroethane) was first used as an insecticide to kill malarial mosquitoes, low concentrations were effective.
  - In subsequent doses, higher concentrations were needed and the sprayings became less effective.
  - The few DDT-resistant mosquitoes that had survived passed on their genes to their offspring, and now the mosquito population is mainly resistant.
  - Thus the gene pool had changed
- Competition for Resources:
  - Competition for resources affects evolution because the survival of a species relies heavily on its ability to obtain the resources needed for life.
  - For example, when the dinosaurs were the dominant life form on Earth, mammals were very scarce
  - The dinosaurs had access to most of the resources and so mammals were unable to proliferate into different niches.
  - When the mass extinction of the dinosaurs occurred, the mammals that so scarcely populated the planet quickly diversified to take advantage of all the available resources, such as plants, or other organisms.
- Competition for resources, and changes in the physical and chemical environment influence the evolution of plants and animals.
- *Describe, using specific examples, how the theory of evolution is supported by the following areas of study:*
  - ◆ *Palaeontology; including fossils that have been considered to be transitional forms:*
  - ◆ *Biogeography:*
  - ◆ *Comparative embryology:*
  - ◆ *Comparative anatomy:*
  - ◆ *Biochemistry:*

- Because the age of fossils can be determined, the sequence from the very earliest life to the present can be observed
- Fossils show a clear change from simple to very complex organisms.
- This suggests a change over time, which is evidence for evolution
- **Transitional Forms:**
  - Transitional forms are examples of organisms that indicate development from one group of organisms to another
  - They help biologists to understand how evolution may have come about
  - Eg 1: Crossopterygian (lobe-fin) Fish:
    - Fish that could absorb oxygen from air appeared 40 mya
    - It is thought that amphibians developed along this line of descent
    - A special feature is that it had bones in its fins, which suggests it could drag itself on the land.
    - FISH Features: scales, fins, gills
    - AMPHIBIAN Features: lobe-fins, lungs
  - Eg 2: Archaeopteryx:
    - This was a small flying dinosaur with feathers
    - It appeared in the late Jurassic
    - It shared features with both birds and reptiles, suggesting that birds evolved from these reptiles
    - REPTILE Features: long-tail, claws, no keel, solid bones, teeth
    - BIRD Features: wish-bone, feathers
- **Biogeography:**
  - Biogeography is the study of the distribution of living things
  - Basic principle is that each plant and animal species originated only once
  - This is called the centre of origin (or the common ancestor)
  - Regions that have separated from the rest of the world, e.g., Australia and NZ, often have organisms that are distinctive and found nowhere else (endemic)
  - General principles of dispersal and distribution of land animals:

- Closely related organisms in different geographical areas probably had no barrier to dispersal in the past
- The most effective barrier to distribution is the oceans and seas
- By looking at the pattern of distribution today plus its fossil distribution in the past, we are able to reconstruct its evolutionary history
- For Example - Waratahs:
  - 3 genera of waratahs; distribution spans southern pacific ocean
  - The present-day distributions of these closely related species in the eastern parts of Australia, and New Guinea and the Western part of South America suggest that the 3 regions may have been connected in the past
- **Comparative Embryology:**
  - The embryos of different vertebrates are very similar
  - The embryos of many different vertebrates all have gill pouches at some stage of development.
  - This suggests that these vertebrates evolved from a common aquatic ancestor, such as the crossopterygian fish.
- **Comparative Anatomy:**
  - Comparative anatomy is the study of the differences and similarities in structure between different organisms.
  - The structures they have in common are evidence of similar inherited characteristics from a common ancestor.
  - For Example - Pentadactyl Limb:
    - The pentadactyl limb is a 5-digit limb
    - It is a structure of bones that is found in many vertebrates
    - It is believed that this limb was inherited from an aquatic ancestor
  - Xylem is found in almost all plants. This suggests a common ancestor
- **Biochemistry:**
  - All organisms share the same basic biochemistry. They all:
    - Consist primarily of organic compounds
    - Share a common genetic code of DNA or RNA

- Rely on enzymes to control chemical reactions
- Share the same cell membrane structure
- Rely on cellular respiration to make energy for cell processes (except chemosynthetic bacteria)
- Similarities in the base-pairing of DNA strands have been analysed to show evolutionary links between organisms
- The amino-acid sequence of certain proteins found in many organisms (such as haemoglobin and cytochrome-c) has been analysed across a range of organisms, and similarities provides evidence for evolution in general
- *Explain how Darwin/Wallace's theory of evolution by natural selection and isolation accounts for divergent and convergent evolution:*
  - Natural Selection:
    - There are variations within every population of species
    - Organisms that don't reproduce have their genes removed from the population
    - Organisms that survive and reproduce are well suited to their environments
    - Favourable variations are passed onto offspring and become common
  - The Role of Isolation:
    - For a new species to evolve, groups of organisms need to become isolated from each other
    - Usually the organisms become separated by a physical barrier
    - Within each separate population, different mutations occur, and therefore, different variations are produced
    - Natural selection acts differently on each isolated population, as there are different environmental conditions and selection pressures
    - Over time the populations differ so much that they no longer interbreed, and as such, a new species was produced
    - Isolation can be created by a difference in food preference, to the splitting of the continents.
  - Divergent Evolution:
    - Also known as adaptive radiation

- It is the process whereby one species radiates out into different environments and as a result produces organisms that look different from each other, and may have many other differences.
- One of the best known examples are Darwin's finches
- 14 different species were described; all with similar greyish-brown to black feathers and all had similar calls, nests eggs and courtship displays
- However, their habitats, diets, body size and beak sizes differed throughout
- Darwin believed that they had all evolved from a common ancestor
- Convergent Evolution:
  - Natural selection over many generations can result in similar adaptations in species that live in similar environments, even though they may be unrelated
  - This is called evolutionary convergence
  - For example, the seal and the dolphin both live in the ocean
  - They have flippers as limbs, they are strong swimmers, can hold their breath longer than most mammals, and they have a layer of fat under their skin.
  - But they belong to different orders of mammals and are unrelated.
- *Prepare a case study to show how environmental change can lead to changes in a species:*
  - **The Peppered Moth:**
    - The population consisted of 2 variations: the light and dark moth.
    - Originally population was mainly composed of lighter moths
    - They camouflaged on lichen covered trees to hide from birds
    - During Industrial Revolution, trees covered in soot and lichen die off
    - Light moths can no longer camouflage, become easier prey. Darker moths can hide better now, on soot covered trees.
    - Population shifts from mainly light to mainly dark.
- *Gather information from secondary sources to observe analyse and compare the structure of a range of vertebrate forelimbs:*



- They all consist of a forearm bone, connected to a dual lower arm group, connected to wrist bones (carpals in humans) which connect to the digits. Usually 5 in number (pentadactyl). Common ancestor.
- *Use available evidence to analyse, using a named example, how advances in technology have changed scientific thinking about evolutionary relationships:*
  - New technologies, especially in the field of biochemistry, have increased knowledge about the relationships between species.
  - Techniques such as DNA hybridisation, amino acid sequencing and analysis of the antibody-antigen reaction between different species have shown the degree of similarity and evolutionary pathways of organisms.
  - **DNA Hybridisation:**
    - DNA hybridisation is a process by which the DNA of different species can be compared
    - The process uses heat to separate the 2 strands of the double helix, from 2 different species

- The single strands of the different species are then mixed, and cooled
  - On cooling, the hydrogen bonds re-form in varying degrees
  - The greater the number of bonds between the strands, the greater the degree of genetic similarity between the two species
  - E.G. DNA hybridisation technology has shown that humans are more closely related to primates than previously thought – shows we share 97% of DNA with chimpanzees.
- *Analyse information on the historical development of theories of evolution and use available evidence to assess social and political influences on these developments:*
    - Influences Prior To Publishing of Evolutionary Theory:
      - Christianity was a very dominant force during the time of Charles Darwin.
      - Creationism was widely accepted, as a religious and a scientific concept
      - Darwin knew what a huge impact his knowledge would make on the world when he released it, so he withheld his theory for 25 years.
      - It was only when he felt the social and political climate was right, did he publish his information
      - He chose to publish it during a time of great societal change; i.e. the Industrial Revolution, and a time when the power of the Church was waning.
      - Also, Wallace's willingness to propose his own version of evolution prompted Darwin to finally publish his papers
    - Influences of Evolutionary Theory on Society:
      - Darwin's theory caused great furore in the society at the time. Great debates were fought out by evolutionists and creationists (a famous one being between Thomas Huxley and Bishop Samuel Wilberforce).
      - Darwin was also blamed for many catastrophes in history, as people continued to wrongly apply the "Survival of the Fittest" to normal life.
      - Darwin has been blamed for the destruction of religion and the rise of atheism, fascism, communism and even the Second World War, as people like Karl Marx base their philosophies on *The Origin of Species*.

## 2. Gregor Mendel's experiments helped advance our knowledge of the inheritance of characteristics:

- *Outline the experiments carried out by Gregor Mendel:*
  - Genetics is the study of heredity
  - Heredity is the transfer of characteristics from one generation to the next
  - The founder of the modern study of genetics was an Austrian monk, Gregor Mendel, who lived in the 19<sup>th</sup> century
  - He studied the genetics of the garden pea plant (*pisum sativum*)
  - Mendel's Method:
    - Mendel first chose 7 pairs of characteristics that he wanted to study
    - These were:
      - round/wrinkled seed
      - yellow/green seed
      - smooth/constricted seed pods
      - green/yellow pods
      - violet/white flowers
      - tall/short stem
      - terminal (at the top) / auxiliary (off the sides) flowers
    - Before he began his experiment, he selectively bred plants for each characteristic for 2 years to produce ONLY *pure breeding offspring*.
  - Firstly he crossed two pure breeding plants
  - Then crossed their off-spring.
    - Mendel's conclusions about organisms that he made his results are summed up in his Law of Segregation:
      - An organism's characteristics are determined by 'factors' (we call them genes), that occur in pairs.
      - In a sex cell (gamete) only one 'factor' is present.
      - During fertilisation, the factors pair up again; they don't blend, but match up with each other

- *Describe the aspects of the experimental techniques used by Mendel that led to his success:*
  - He studied a large number of CHARACTERISTICS – increased accuracy.
  - He performed a large number of CROSSES; this produced very reliable results
  - He made sure he used PURE BREEDING plants – to produce accurate results.
  - He made exact counts of the characteristics, producing QUANTITATIVE data that could be easily analysed.
  - He studied separate, easily identifiable characteristics, one at a time
  - He chose a plant (the pea plant) that shows easily identifiable, alternative forms.
  - He strictly controlled fertilisation (hand-pollination) to ensure accurate results.
- *Distinguish between the terms allele and gene, using examples:*
  - A chromosome is a long strand of DNA located in the nucleus
  - Chromosomes always come in pairs, one from the mother (maternal) and one from the father (paternal)
  - The pair of chromosomes are called HOMOLOGOUS chromosomes
  - A gene is a section of DNA on a chromosome; it codes for a particular characteristic
  - Alleles are corresponding (matching) pairs of genes, at the same position on homologous chromosomes
  - Eg. The gene for eye colour, and the brown allele or the blue allele.
- *Describe outcomes of monohybrid crosses involving simple dominance using Mendel's explanations:*
  - Each characteristic is coded for by *at least* a pair of genes.
  - A monohybrid cross is where we are examine only one characteristic, like Mendel did with his pea plants.
  - The genotype of an organisms is its genetic make-up
  - The phenotype is the physical characteristics of an organism.
  - Mendel's Monohybrid Crosses:
    - Mendel only studied one pair of characteristics at a time (e.g. stem height)

- This type of breeding experiment is called a *monohybrid cross*
- Mendel first bred one variety of pure-breeding plant (e.g. tall plants) with another variety, also pure-breeding (e.g. short plants).
- But they were ALWAYS different varieties of the same characteristic.
- F<sub>1</sub> is known as the first generation, F<sub>2</sub> the second generation, and so on.
- The parents were cross-pollinated, and all the off-spring was tall.
- **Parents:** tall plants x short plants
- **F<sub>1</sub>:** all tall plants
- Mendel then took these tall offspring and self-pollinated them: He transferred the pollen by hand from the stamens onto the stigmas.
- **F<sub>1</sub>:** tall plants x tall plants
- **F<sub>2</sub>:** approximately - 3 tall plants : 1 short plant
- Mendel repeated this experiment many times, and with different characteristics such as seed colour, but the same ratio kept occurring.
- This ratio 3:1 is called the *monohybrid ratio*
- *Distinguish between homozygous and heterozygous genotypes in monohybrid crosses:*
  - Homozygous genotypes have the same allele for a characteristic. Also called pure-breeding. They are represented by the same letter twice. E.g. TT
  - Heterozygous genotypes have different alleles. E.g. Tt
- *Explain the relationship between dominant and recessive alleles and phenotypes using examples:*
  - Dominant and recessive alleles:
    - For every characteristic, there are 2 alleles.
    - They are ALWAYS present in pairs in body cells
    - If the two genes are the same allele, then the organism is said to be homozygous for that characteristic.
    - If the two alleles are different, then the organism is heterozygous for this characteristic.

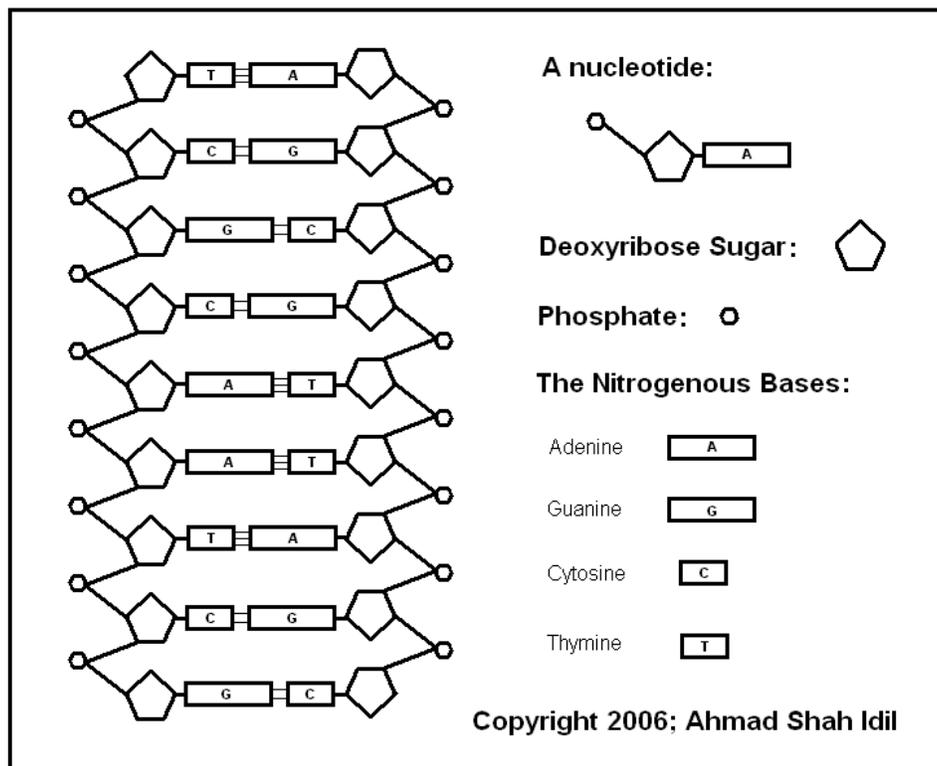
- In simple genetics, one of the alleles is DOMINANT, and one of them is RECESSIVE
- Taking a characteristic, e.g. Pea Plant height. We represent it's genotype with 2 letters, each letter representing a gene. T is the dominant tall allele, t is the recessive, short allele
- A tall pea plant can be either TT or Tt, as the dominant gene is always expressed
- A short plant is always tt
- *Outline the reasons why the importance of Mendel's work was not recognised until some time after it was published:*
  - He only presented his paper to a small group of scientists
  - His work was radically different to previous ideas – possibly not understood
  - Significance was possibly not realised at the time
  - He had no outstanding reputation as a scientist – possible ignored by scientific community.
- *Perform an investigation to construct pedigrees or family trees, trace the inheritance of selected characteristics and discuss their current use:*
  - Pedigrees are family trees. They show the inheritance of a trait over many generations.
  - Patterns to recognise characteristics are inherited:
    - If two non-affected parents have an affected child, then the trait is a recessive one.
    - If two affected parents, have a non-affected child, then the trait is dominant
    - If there is a large bias towards males being affected, and sometimes generations are skipped, than the trait is recessive sex-linked
- *Solve problems involving monohybrid crosses using Punnett squares or other appropriate techniques:*
  - ???

- *Process information from secondary sources to describe an example of hybridisation within a species and explain the purpose of this hybridisation:*
  - In Kenya, hybridisation has been carried out by means of artificial insemination between Jersey cattle and the African Sahiwal cattle. The purpose of the hybridisation is to increase milk yield and quality. The milk yield of the Jersey-Sahiwal crossbred cows is generally higher than that of either the Jerseys or the Sahiwals.

### 3. Chromosomal structure provides the key to inheritance:

- *Outline the roles of Sutton and Boveri in identifying the importance of chromosomes:*
  - Theodor Boveri:
    - A German zoologist
    - **1986** – Boveri studied sea urchins - obtained eggs without nuclei (with no female chromosomes) and fertilized them with sperm (just male chromosomes)
    - The larvae born showed none of the characteristics of the female (mother)
      - This proved the nucleus carried the information for inheritance.
      - Suggested that chromosomes (observed in the nucleus) might be the means of inheritance
      - At this time, he knew nothing of Mendel's factors.
    - Showed that chromosomes were transferred from one generation to the next by the process of cell division
    - Argued that chromosomes could exchange factors with each other during meiosis (**crossing over**)
  - Walter Sutton:
    - An American geneticist
    - Sutton observed grasshopper chromosomes, during meiosis, as well as researching patterns and observations (of previous scientists)
    - 1902 proposed the **Chromosomal Theory of Inheritance**
    - Observations of grasshopper cells:
      - During meiosis, chromosomes line up in pairs, same size and shape
      - Homologous pairs **segregate randomly** so that each gamete receives one chromosome from each pair
      - After fertilisation, the resulting zygote had a full set of chromosomes
    - Suggested Mendel's inheritance "factors" are carried on chromosomes
    - Recognized that chromosomes provided a mechanism for the operation of Mendel's laws

- *Identify that DNA is a double-stranded molecule twisted into a helix with each strand comprised of a sugar-phosphate backbone and attached bases – Adenine (A), thymine (T), cytosine (C) and guanine (G) – connected to a complementary strand by pairing the bases, A-T and G-C:*
  - DNA (deoxyribonucleic acid):
    - A double stranded helix
    - Made up of sub-units called nucleotides
    - Each nucleotide is made up of a phosphate, a sugar and a nitrogenous base
    - The sugar is deoxyribose
    - The four different bases are adenine, thymine, guanine, and cytosine
    - Adenine pairs with thymine (A-T) and guanine with cytosine (G-C)
  - A single DNA strand is made up of a chain of nucleotides (a polynucleotide) where the phosphate and sugar alternate as the backbone of the strand
  - The bases attach to the sugar
  - The other strand of DNA attaches to the strand by complementary pairing of the nitrogenous bases. The other strand is ‘flipped’ (it goes in the opposite direction).



- *Describe the chemical nature of chromosomes and genes:*
  - Each chromosome is made up of about 60% protein and 40% DNA
  - The DNA is coiled tightly around a protein core (histone proteins)
  - A gene is a section of DNA on a chromosome
  - The DNA is made of sugar, phosphate, and nitrogenous bases.
  - It is made up of a particular sequence of bases
  - Different genes are different lengths
- *Explain the relationship between the structure and behaviour of chromosomes during meiosis and the inheritance of genes:*
  - The stages of meiosis that lead to the creation of gametes and the inheritance of genes are:
    - The chromosomes duplicate. The single stranded chromosomes become double stranded, linked at the centre by a centromere
    - In the first meiotic division, the homologous chromosomes separate, but the double-strands of the chromosomes are still joined.
    - In the second division, the chromatids of the chromosomes separate and form 4 gametes altogether.
- *Explain the role of gamete formation and sexual reproduction in variability of offspring:*
  - The events that create variation in sexual reproduction are:
    - **RANDOM SEGREGATION:** During meiosis, genes on different chromosomes sort independently. They can line up in the middle of the cell in many different ways. This produces many gene combinations, which are different from the parents
    - **CROSSING OVER:** Crossing over of genetic material during meiosis results in the exchange of genes between chromosome pairs. The combinations of alleles of the gametes will vary across cells and differ from the parent
    - **RANDOM FERTILISATION:** When the male and the female mate, the two different gametes randomly fuse. Many different combinations are possible, and this causes variation.

- Describe the inheritance of sex-linked genes, and alleles that exhibit codominance and explain why these do not produce simple Mendelian results:

– Co-Dominance:

- In this case, the two alleles are not dominant over each other
- Both alleles are expressed in each others presence
- There is no “blending”, the alleles do not mix, but both can be seen
- An example is roan coloured cattle
- If a type of cattle has the gene for red, and white, it would **not** make a pink cow, but the hairs on the cow would be both red AND white, making an interesting roan colour.

	R	R	A cross between a white and a red cow
W	RW	RW	
W	RW	RW	
	R	W	A cross between two roan cows
R	RR	RW	
W	RW	WW	

Cross: Red x White  
RR x WW

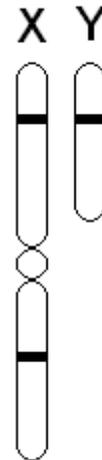
Result: All offspring RW  
All offspring roan

Cross: Roan x Roan  
RW x RW

Result: 1 Red : 2 Roan : 1 White  
1 RR : 2 RW : 1 WW

- Looking at the cross in the form of a Punnett square, we can see that a cross concerning a codominant trait does not give the simple Mendelian ratio of 3:1

- The cross between the two roan cows of the F<sub>1</sub> generation does not give the 3:1 ratio because a **heterozygous** animal does not give the dominant trait, as would happen in simple dominant-recessive cases. A “heterozygous” animal gives the roan colour, which results in the 1:2:1 ratio.
- Incomplete Dominance:
  - The alleles in this case do not show simple dominance either
  - If the both alleles are present (heterozygous) blending of phenotype will occur
  - For example if a snapdragon (a flower) has a red & white gene, it will be pink.
- Sex-linked Characteristics:
  - SEX is a genetically determined characteristic
  - The sex of an individual is determined by a pair of chromosomes called the sex chromosomes; in humans, it is the 23<sup>rd</sup> pair
  - For females, both the sex chromosomes are the same. This combination is called XX. Females have two X chromosomes, and the chromosomes are homozygous
  - For males, the sex chromosomes are different. The combination is XY. The Y chromosome is shorter than the X chromosome.
  - Because the Y chromosome is much shorter than the X chromosome, some characteristics are only coded for by the X chromosome (the X has genes Y does not have)
  - This is a special case of inheritance of characteristics.
  - Most sex-linked characteristics are recessive
    - Take, eg, haemophilia. ‘H’ is the dominant, normal allele; ‘h’ is the recessive, haemophiliac allele.
    - Females:
      - ◆ A normal female’s genotype – X<sup>H</sup>X<sup>H</sup>
      - ◆ A carrier female has the genotype - X<sup>H</sup>X<sup>h</sup>
      - ◆ A haemophiliac female has the genotype – X<sup>h</sup>X<sup>h</sup>
    - Males:
      - ◆ A normal male - X<sup>H</sup>Y (Y chromosome is missing the gene)



- ◆ A haemophiliac male -  $X^hY$ 
  - As you can see for the example, males only have to inherit a single gene to have the characteristic.
  - A single recessive gene has the same phenotypic effect as a single dominant gene
  - This is why some sex-linked characteristics are much more common in males than females.
  - PUNNETT SQUARE??
- *Describe the work of Morgan that led to the understanding of sex linkage:*
  - Morgan studied the breeding of the fruit fly (*drosophila*)
  - As he was breeding the flies, he noticed one white-eyed male fly among the offspring of red-eyed parents
  - This was strange as the normal eye colour was red
  - He further bred this white male with other males
  - His results showed that all the white-eyed flies were male
  - He hypothesised that the characteristic was ‘sex-limited’, and that it was on the X chromosome.
- *Explain the relationship between homozygous and heterozygous genotypes and the resulting phenotypes in examples of codominance:*
  - In simple dominance cases, if an organism is homozygous dominant, the phenotype is obviously that of the dominant allele. If it was homozygous recessive, then the phenotype would be that of the recessive allele.
  - If the organism was heterozygous, then the dominant allele would be the phenotype of the organism, as the dominant allele would preside over the recessive one.
  - HOWEVER, if it was a case of codominance, heterozygous organisms would have both phenotypes expressed at the same time, as no allele is totally dominant over the other. Eg, red and white – roan cattle.
- *Outline the way in which the environment may affect the expression of a gene in an individual:*

- Genes are not the only factor that influence phenotype
- GENES + ENVIRONMENT = PHENOTYPE
- The environment can control to what extent a genotype is expressed
- A lack of sufficient resources can stunt the growth of a person, eg.
- Examples:
  - Phenylketonuria (PKU): Babies born with PKU can not make the important enzyme phe-hydroxylase, and as a result, can not metabolise the amino acid phenylalanine (phe) into tyrosine. This is a genetic disorder. If they eat excessive amounts of phe, the babies will become severely mentally retarded. If phe levels are kept low, the babies will grow up normally.
  - Hydrangeas: This flower's colour is controlled by pigments known as anthocyanins. These are affected by pH. If the hydrangeas grow in acidic environments, the flowers will be bright blue. In alkaline environments, the flowers are pale-pink, or off-white.

**4. The structure of DNA can be changed and such changes may be reflected in the phenotype of the affected organism:**

- *Describe the process of DNA replication, and explain its significance:*
  - The significance of the ability of DNA to replicate itself exactly is that identical copies of genes can be made
  - This allows cells to replicate, as well as allows inheritance of characteristics.
  - DNA replication is made possible because the molecule is a double helix, and because the nitrogenous bases only pair complementarily
  - The steps for DNA replication:
    - The parent DNA molecule unwinds into 2 separate strands, at one end
    - And the two strands become exposed, free nucleotides floating in the nucleus attach to the exposed bases, A with T and C with G. This ensures that the replication is exact
    - The joining of nucleotides together is catalysed by DNA polymerase
- *Outline, using a simple model, the process by which DNA controls the production of polypeptides:*
  - DNA holds the information for creating proteins in cells
  - As we know, a protein is made up of one or more chains of polypeptides, and each polypeptide is made up amino acids and peptide bonds
  - The way DNA codes for proteins:
    - A set of 3 bases is called a triplet code, or a **codon**.
    - Every codon codes for one amino acid
    - There are 20 different amino acids
  - The structures involved in polypeptide synthesis are:
    - DNA: A gene contains a sequence of bases (codons) to code for a protein
    - RNA: RNA is similar to DNA except that instead of deoxyribose as the sugar, it has ribose. It is single stranded, **thymine** is replaced by **uracil**. There are 3 forms of RNA involved in polypeptide synthesis:
      - *mRNA*: Messenger RNA copies the DNA & carries the genetic code outside the nucleus, into the cytoplasm, where it can be read by ribosomes

- *tRNA*: Transfer RNAs carry the loose amino acids to the ribosomes. Here, they link together to form a polypeptide chain. tRNA are shaped like clover leaves; there is a different type for every amino acid. At the bottom of every tRNA molecule is a series of 3 bases (the **anti-codon**) that binds to the complementary codon on the mRNA strand. That is how the amino acid is linked to the codon.
- *Ribosomal RNA*: Ribosomes are made up of protein and RNA
- Ribosomes: The ribosome is the active site for protein synthesis. It is made up of protein and RNA molecules. It can accommodate 2 tRNA at a time.
- Enzymes: The enzyme that controls the formation of mRNA is RNA polymerase. There are, of course, many other enzymes that control the process.
- **STAGE ONE - Transcription:**
  - A double stranded DNA molecule in the nucleus unwinds a section of itself that consists of a single gene.
  - One of the strands coding for the gene exposes itself to the nucleoplasm
  - The enzyme, RNA polymerase moves along the strand, attaching loose RNA nucleotides to the DNA, with A-U and C-G, until the whole gene is copied.
  - This new RNA strand is called messenger RNA (mRNA)
  - A start codon, and a stop codon determine the length of the gene
  - The mRNA strand exits the nucleus and enters the cytoplasm
- **STAGE TWO - Translation:**
  - The mRNA strand binds to a ribosome in the cytoplasm, with the start codon being AUG (always).
  - The ribosome moves along the mRNA strand, to ‘read’ more of its bases.
  - tRNA molecules floating in the cytoplasm, which have anti-codons complementary to the codons of the mRNA enter the ribosome. Eg, if the mRNA had an AAG codon, the UUC tRNA would bind to it
  - The tRNA releases its amino acid to form a chain. The ribosome can only accommodate 2 tRNA.
  - The ribosome moves along the mRNA, and more and more amino acids are attached, with peptide bonds, to the growing polypeptide chain.

- When a 'stop' codon is reached, the polypeptide chain is released into the cytoplasm, for further processing, to become a protein.
- *Explain the relationship between polypeptides and proteins:*
  - A polypeptide is made up of amino acids linked by peptide bonds
  - A protein is made up of one or more polypeptide chains, folded to fit a specific function, often into a globular shape.
- *Explain how mutations in DNA can lead to the generation of new alleles:*
  - A mutation is a change in the DNA information on a chromosome
  - Three things can happen as a result of a mutation:
    - Most mutations are lethal and kill the cell the mutation takes place in
    - In some cases, the mutation is not advantageous or lethal to the organism. It is a neutral mutation
    - VERY RARELY, a mutation will give an organism a phenotypic advantage. These individuals with the new allele will be at a selective advantage, and be better suited to their environment
  - The mutation causes a change in the code of the DNA (base sequence)
  - This means that, possibly, new amino acids will be introduced in polypeptide chains, this will lead to new proteins being produced, and new forms of traits.
  - This leads to the generation of new alleles.
- *Discuss evidence for the mutagenic nature of radiation:*
  - Mutagens are environmental factors that increase the rate of mutation
  - Effect of radiation on DNA strands:
    - E.g. UV light, X-rays, radioactive materials
    - Can cause bases to be deleted, totally removed from strand
    - Can cause thymine bases to link together, affecting protein synthesis.
    - This causes a disruption in the normal functions of DNA
    - High-energy radiation levels can actually break up the whole chromosome
  - Evidence for the mutagenic nature of radiation:
    - UV radiation has been recorded to increase the incidence of skin cancers in humans. Some regard it as the sole cause of skin cancer

- First generation radiotherapists, who did not know the dangers of radiation, often died young. Scientists like Marie Curie would carry uranium around in their pockets, and developed cancers very quickly
- People who live in areas which have been affected by high-level radiation, such as Hiroshima, or Chernobyl, still show high incidences of cancers and other mutations in their offspring.
- *Explain how an understanding of the source of variation in organisms has provided support for Darwin's theory of evolution by natural selection:*
  - We know Darwin's theory requires variation to be present within a population
  - Our knowledge of genetics tells us where this variation comes from:
    - The random segregation of chromosome pairs during meiosis
    - Crossing over of genetic material during meiosis
    - Random fertilisation of sex cells
    - Random mutation of the genetic material
  - The phenotypes that are variable are “chosen” by the environment
  - Over time, some genotypes become more prevalent than others
- *Describe the concept of punctuated equilibrium in evolution and how it differs from the gradual process proposed by Darwin:*
  - Darwin's Gradualism:
    - He proposed that populations change slowly and gradually over time
    - However, the fossil record only shows rare occasions where this happens
  - If an environment remains stable for many years, we would expect there to be no change in the organisms living there
  - It is only when the environment changes that natural selection occurs
  - The fossil record in fact shows periods of stability followed by mass extinctions and rapid change
  - Punctuated Equilibrium:
    - The fossil record suggests that organisms evolve suddenly, and remain stable for millions of years

- In 1972, 2 scientists, Gould and Eldridge, put forward a theory to explain this; they called it punctuated equilibrium.
- Punctuated equilibrium proposes that, instead of gradual change, there have been periods of rapid evolution followed by long periods of stability, or equilibrium
- *Analyse information to outline evidence that lead to Beadle and Tatum's 'one gene – one protein' hypothesis and explain why this was changed to 'one gene – one polypeptide' hypothesis:*
  - Research conducted by Beadle and Tatum
  - They knew that bread mould grows on a base of sugar, salts and vitamin broth
  - This nutrient base was called the “minimal medium”
  - They reasoned that these nutrients must be converted into amino acids, and that enzymes were responsible for this change
  - They then exposed the mould to X-rays, to induce mutations
  - This mutant mould was then grown on the minimal medium
  - If the mould grew, it was discarded
  - If the mould didn't grow, it was grown on a different medium, containing different amino acids.
  - It was found that if the mould was supplemented with other amino acids, it could grow healthily.
  - Beadle and Tatum hypothesised that this mutant mould had lost the ability to make the enzyme to create this amino acid, because the X-rays had mutated the gene.
  - Then they hypothesised that one gene was responsible for one enzyme
  - This was later changed to 'one gene – one polypeptide', because genes code for many proteins that are not enzymes
  - Many proteins are made up of more than one polypeptide, and a gene only codes for one polypeptide.

## 5. Current reproductive technologies and genetic engineering have the potential to alter the path of evolution:

- *Identify how the following current reproductive technologies may alter the genetic composition of a population:*
  - *Artificial insemination:*
  - *Artificial pollination:*
  - *Cloning:*
- Humans have selectively bred plants and animals for centuries
- However, it had always been for the benefit of humans; we breed animals and crops to be bigger, grow-faster, tastier, etc.
- *Selective breeding* is the deliberate crossing or mating of individuals of the same species with the characteristics wanted; over time, these characteristics become dominant.
- However, the overall genetic variation of populations tends to be reduced
- Artificial Insemination:
  - Refers to animals
  - It is the injection of male semen into a female
  - Commonly used with species of large mammals, eg cows, sheep, horses, etc
  - The sperm is collected from a male with desirable characteristics
  - **ADVANTAGES:** Can be used to inseminate many females from one male. Transport of semen is much easier than transporting a whole animal. Semen can be stored for a period of time.
  - **DISADVANTAGES:** Reduced the genetic variations found in populations, making them susceptible to changes in the environment (e.g. new disease)
- Artificial Pollination:
  - Plant breeders carry out artificial pollination to breed plants with specific characteristics (like Mendel did).
  - Pollen from the male anther is collected. It is then dusted onto the female stigma of another plant. The pollinated flower is covered to prevent pollination from other flowers

- **ADVANTAGES:** Particularly useful and easy way of breeding new varieties of plants. No expensive equipment required
- **DISADVANTAGES:** Genetic variation reduced.
- Cloning:
  - Cloning is a method of producing genetically identical organisms
  - A clone is a collection of genetically identical copies
  - **PLANT CLONING:**
    - The most commonly used method, and the oldest, is cutting and grafting. A stem of short section of another plant is cut off, dipped in root-growth hormones, and planted into soil. The plant that grows is a clone of the original plant
    - Tissue culture technology has allowed mass cloning of plants. Firstly, a section of a plant, eg, a root tip, is pulverised using a blender to release the individual plant cells. The cells are grown on a nutrient medium, and incubated under controlled conditions. Genetically identical plants are produced.
  - **ANIMAL CLONING:**
    - Much more difficult than plant cloning
    - First animal to be cloned was Dolly (named after Dolly Parton...LoL)
    - Technique used is called 'nuclear transfer technology' :
      1. Adult sheep tissue cell removed from sheep and cultured in lab
      2. Nucleus removed from one of these cells and placed in an enucleated egg cell (egg cell with genetic info removed)
      3. Gentle electric pulse causes nucleus to fuse with egg cell
      4. A second electric pulse starts cell division and embryo formation
      5. This new cell is implanted into a female sheep where it grows into a new organism
  - **ADVANTAGES:** In agriculture, cloned plants have identical requirements and grow in similar ways to produce similar yields at the same time. In plants and animals identical copies of desirable varieties can be produced

- **DISADVANTAGES:** In crops – all plants susceptible to the same diseases. Cloning is expensive with limited advantages over reproductive techniques. Cloning of animals has raised ethical questions about the cloning of humans. The health/life expectancy of cloned animals is questionable, with the death of Dolly the sheep being earlier than expected.
- *Outline the processes used to produce transgenic species and include examples of this process and reasons for its use:*
  - Transgenic species are organisms which have had genetic material from a different species transferred into their chromosomes
  - That is, genes from one species have been taken and transferred into another
  - The introduced gene instructs the transgenic organism to produce the desired trait or products
  - This trait may be passed onto future generations
  - Processes Used to Produce Transgenic Species:
    - The steps in producing a transgenic species is usually like this
      1. A useful gene, and the chromosome it is on, is identified
      2. The gene is ‘isolated’ or cut-out of its DNA strand
      3. Separate DNA sequences for regulation may have to be added to ensure the gene will work
      4. The gene is inserted into the cell of another organism. Sometimes a vector is used to do this.
    - A vector is a carrier of a substance from one species to another
  - Techniques Used to Produce Transgenic Species:
    - *Isolating Genes:* Once a useful gene is identified, it has to be isolated by ‘cutting’ it out of its DNA strand. Special enzymes, called restriction enzymes are used. More than 800 types are known. They cut DNA by breaking the hydrogen bonds in a triplet – the ends are called “sticky ends”
    - *Making Recombinant DNA:* The DNA strands from 2 organisms are cut using the same enzyme, the sticky ends will match. When they are mixed, the new

gene will match with the DNA strands, and link up. This is called ANNEALING. DNA ligases are added to strengthen the bonds.

- *Making Transgenes:* An isolated gene cannot function if it is transferred alone. It has to be transferred with a promoter sequence attached to ensure it works
- Inserting Genes into Bacteria:
  - Most bacteria contain small, circular pieces of DNA called plasmids
  - Plasmids can be used as vectors or carriers to transfer transgenes into bacteria
- Reasons For Using These Processes:
  - These processes enable scientists to combine the qualities of different organisms
  - Transgenic species are being developed to:
    - Increase the resistance of plants or animals to diseases, pests or extreme environmental conditions
    - For medicines and vaccines and to study human diseases
    - To improve productivity of crops, pastures and animals
    - To improve the quality of food and efficiency of food processing
- Examples of the Use of Transgenic Species:
  - **BT CROPS:** BT is a bacterium that naturally produces chemicals that kills many insects. The chemicals are specific to many pests and do not kill other insects. Genetically modified crops have had the gene of BT pesticide inserted into them. They produce their own BT chemicals, and no longer need to be sprayed
  - **COLD STRAWBERRIES:** A gene from a type of salmon that allows it to survive cold temperatures has been isolated, and inserted into a strain of strawberry. This strawberry can survive and grow in cold temperatures.
  - **BACTERIAL INSULIN:** Diabetics previously obtained their insulin from animals, esp. pigs. The gene for insulin production, taken from the human pancreas, was placed in to the DNA of a bacterium. This now provides mass production of insulin.
- Ethical Issues of Transgenesis:
  - These technologies help treat diseases and increase food production

- Should we be tampering with nature in this way?
  - Is it right to change living organisms for commercial gain?
  - Transgenesis disrupts evolutionary relationships between organisms
  - If a transgenic species was released into the natural environment, it could out-compete the natural organisms
  - Health-risks and side effects with eating GM foods.
- *Discuss the potential impact of the use of reproductive technologies on the genetic diversity of species using a named plant and animal example that has been genetically altered:*
    - The main fear behind the use of genetic and reproductive breeding techniques on organisms is that the natural diversity and variation within populations is decreased
    - E.g. cotton plants. The main crop being grown all over the world is BT cotton.
    - As more and more farmers shift from natural cotton to BT cotton, there are many disadvantages:
      - Many natural varieties of cotton will be lost
      - The species itself becomes vulnerable to extinction. If all cotton grown all over the world is BT, and a disease appears, that kills specifically BT cotton, then there is a risk of cotton becoming an extinct organism
    - In another case, a population of cattle that have all been fathered by the same bull, through artificial insemination techniques, is at risk of environmental changes
    - A lack of variation is a major risk factor in extinction of a species.