IQ1: How does reproduction ensure the continuity of a species?

1.1 explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including but not limited to:

– animals: sexual reproduction, advantages of external and internal fertilisation

- Sexual reproduction involves the union of a male gamete (sperm) and female gamete (ovum) to form a unique individual.
- The two main purposes of sexual reproduction are to form a population with genetic variation in offspring and to facilitate adaptation/promote continuity of species.
- Gametes (haploid, n) are the sex cells in the body e.g. sperm or egg cells.
- Somatic cells (diploid, 2n) are basically every single other non-sex cell in the body e.g. muscle cells or nerve cells.
    - When two gametes are unified and create a zygote (fertilisation), the two haploid cells combine and it becomes the body's first somatic cell, now diploid.

ADVANTAGES AND DISADVANTAGES OF SEXUAL REPRODUCTION

Advantages	Disadvantages
Variation in population	Time consuming/inefficient for rapid reproduction
Unfavourable variation is eliminated more quickly	Mates have to be found and accepted as suitable (risky and energetically costly)
Populations are better able to adapt to and survive changing environmental conditions	Potential for spread of STD's throughout population

INTERNAL AND EXTERNAL FERTILISATION

- As animals moved from protective aquatic environments to exposed terrestrial ones, there was a need to shift from external to internal to prevent dehydration + predation of gametes.

	Internal	External
Definition	Union of a sperm and egg cell inside the body of a parent (sperm must be able to find its way into female)	Union of a sperm and egg cell outside the body of a parent, otherwise known as spawning
Location	Terrestrial environments	Aquatic environments
Examples	Mammals e.g. human reproduction	e.g. frogs: female lays eggs and male drops sperm on top of them

CLASSIFICATIONS FOR INTERNAL FERTILISATION

- Placentals → fetus is nourished in utero and born fully developed e.g. humans
- Marsupials → young are born at a very early stage and continue developing outside the uterus (usually protected by a mother's pouch) e.g. kangaroos
- Monotremes → lay fertilised eggs covered in tough membranes, protecting them until young hatch e.g. platypus

– plants: sexual reproduction

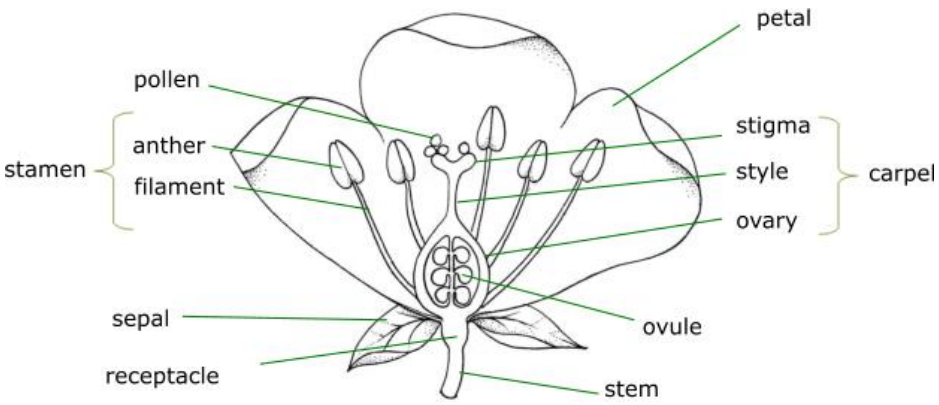
- Most plants have mechanisms that protect against self-pollination as it is disadvantageous.
  - E.g. plants may reject their own pollen, or in flowering plants different parts of the flower ripen at different times.

SEXUAL REPRODUCTION IN NON-FLOWERING PLANTS

- Ferns
  - Grow from spores, which then mature into gametophytes (haploid)
  - Gametophytes release sperm to fertilise eggs - fertilisation occurs on top of gametophyte to form sporophyte
  - Sporophyte grows into new fern plant - produces spores to complete life cycle
- Gymnosperms e.g. conifers
  - Use wind to bring pollen grains from male cones to female cones
  - Female cones at top of tree, male at bottom = makes it hard to self-pollinate since pollen is blown by the wind

SEXUAL REPRODUCTION IN FLOWERING PLANTS (ANGIOSPERMS)

- Pollination is the process of pollen coming into contact with the stigma of a flower.
  - After pollination, male gametes in pollen travel through the pollen tube into the ovary where they then fertilise the female gamete (ova).
  - Fertilisation leads to formation of a zygote -> mitosis forms an embryo -> develops and turns into a seed.
- Flowering plants can be pollinated by insects, mammals, birds or wind.



Feature	Function
Petals	Usually large and colourful to attract birds or insects that help pollinate the flower
Ovary	Central swelling at the base of the flower that contain the ovules (female gametes).
Anther	Small sac attached to the end of a filament that produces pollen.
Stigma	Receptive surface for pollen in order to reproduce sexually.
Pollen	Small, powdery grains that contain male gametes.
Sepal	Encloses and protects flower during the budding stage

– asexual reproduction: bacteria, protists, fungi

- Asexual reproduction is the production of offspring by mitosis from one parent.
  - No genetic recombination since mitosis = cloning, thus offspring are genetically identical to parent.

ADVANTAGES AND DISADVANTAGES

TABLE 2.1.2 Advantages and disadvantages of asexual reproduction

Advantages	Disadvantages
• efficient form of reproduction • amount of time and energy to produce offspring is minimal • population sizes can increase rapidly in optimal environments • there is no need to find a sexual partner • offspring are genetically identical to the parent cell, so they are well suited to a stable environment	• rapid population growth can lead to overcrowding and increased competition for resources • the lack of genetic variation in a population can cause death of the entire population if conditions change (e.g. a disease pathogen arrives or a severe drought) because they are not adaptable to new environmental conditions

METHODS OF ASEXUAL REPRODUCTION

budding	Form of asexual reproduction in which the new organism arises as an outgrowth or bud from the parent.
fission	Form of asexual reproduction of unicellular organisms where the parent cell divides into two approximately equal parts.
fragmentation	Form of asexual reproduction of multicellular organisms in which an organism breaks into two or more parts, each of which regenerates the missing pieces to form a complete new organism.
spore formation	Formation of structures that are resistant to adverse environmental conditions and can give rise to complete organisms when conditions become favourable.
parthenogenesis	Development of an egg in the absence of fertilisation by sperm; a normal part of the life cycle of some insects and crustaceans.

- Plants can reproduce by runners, tubers, or cuttings (vegetative propagation).
- Bacteria e.g. H. Pylori are all unicellular and reproduce by **binary fission**.
  - Numbers increase exponentially by C = 2^n (where C is number of cells and n is the number of cell replication cycles).
  - Bacteria can also exchange genetic material to recombine characteristics (gene transfer).
- Protists e.g. amoeba reproduce by **binary fission** and **budding**.
- Fungi e.g. penicillium mould reproduce both sexually and asexually.
  - Asexual reproduction -> plasmogamy by **budding** or **spores**
    - Sporangia produce spores, hyphae form mycelium threads and develop fruiting body
  - Sexual reproduction -> karyogamy
    - Two separate hyphae fuse and form a zygospore that produces gametes.

1.2 analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals

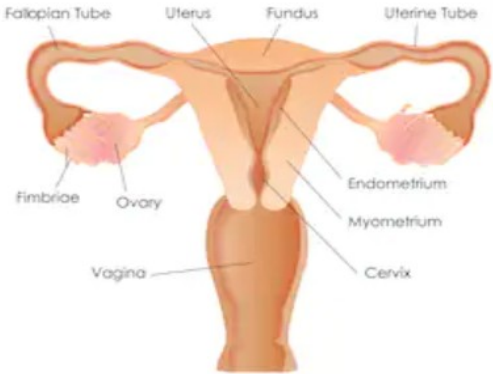
MAMMALIAN REPRODUCTION SYSTEMS

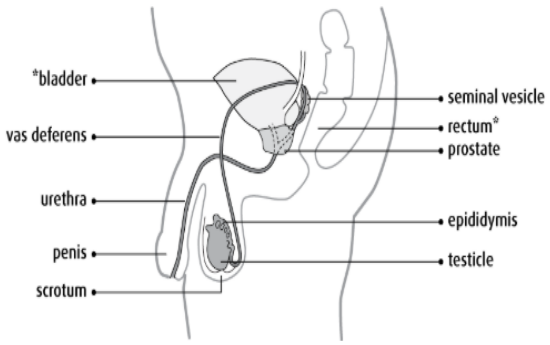
Male:

Structure	**Function**
Vas deferens	Tubes that lead from the testes to the urethra
Penis	Delivers urine and semen to outside of the body (but not simultaneously).
Testis	Produce and store mature sperm

Female:

Structure	**Function**
Vagina	Receives the penis during intercourse, exit canal for menstruation, and exit canal for child during birth
Cervix	Narrow muscular canal that connects the uterus and vagina - dilates during birth
Uterus/uterine cavity	Ovum is implanted in endometrium. Nurtures fertilised egg during development until it is mature enough for birth.
Fallopian tube	Connects the ovary to the uterus
Ovary	Holds egg cells and releases them during ovulation Produces hormones
Fimbria	Surround the ovary to catch eggs when released.





PROCESS OF FERTILISATION

- The male and female have sexual intercourse.
- Muscular contraction (ejaculation) moves semen (sperm) from the male into the vagina.
- Sperm cells travel through the uterus and into a fallopian tube.
- A single sperm cell fertilises each available egg, resulting in one or more zygotes (different species release different numbers of eggs).

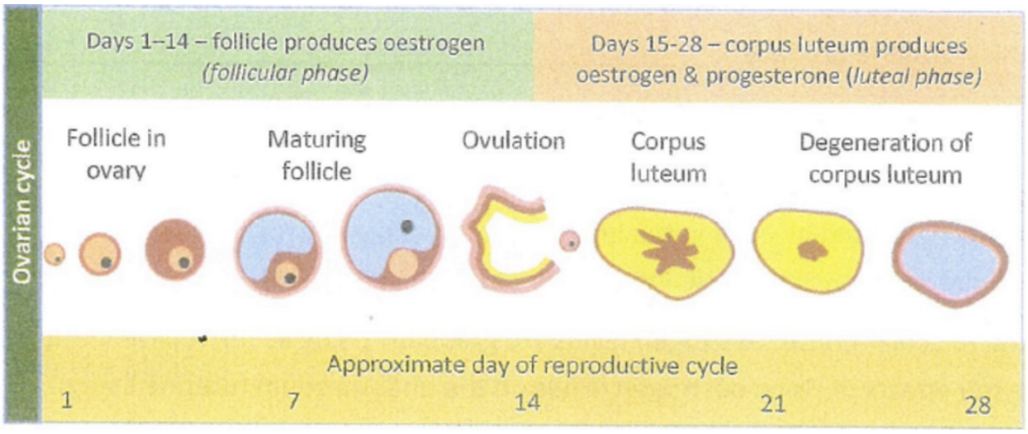
- Zygote grows through mitosis → blastocyst implants into lining → develops into an embryo.

HORMONES AND FUNCTIONS

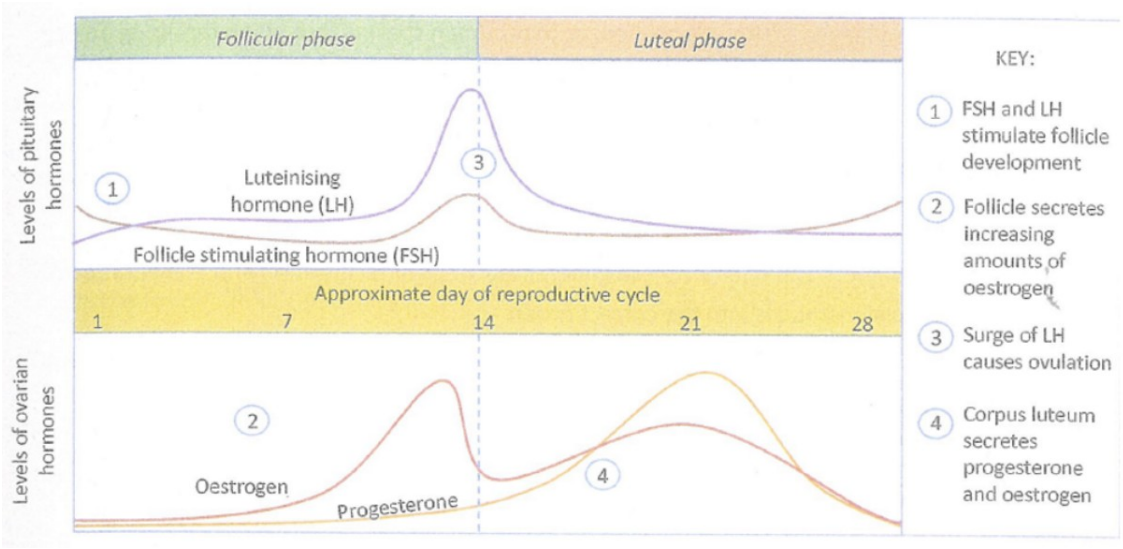
- Hormones are proteins or lipids that control metabolic functions.
- Hormone secretion is controlled by negative feedback (homeostasis).
  - i.e. when a hormone is too high, it will stop being secreted until it is depleted.

Hormone	Secreted by	Action
FSH	Pituitary gland	• Initiates the ripening of follicle and egg in the ovary. • Stimulates secretion of estrogen.
Estrogen	Ovaries/corpus luteum	• Contributes to development/repair of endometrium + maturing of follicle. • Inhibits FSH secretion and stimulates secretion of LH.
LH	Pituitary gland	• Causes follicle to release the egg (ovulation). • Causes empty follicle to grow into corpus luteum. • Inhibits secretion of estrogen.
Progesterone	Corpus luteum	• Causes endometrium to thicken (preparing for pregnancy). • Inhibits release of FSH and LH.
Oxytocin	Pituitary gland	• Stimulates muscle contractions for birth + secretion of milk.
GnRH	Hypothalamus	• Stimulates secretion of FSH and LH.

REPRODUCTIVE CYCLE



- The corpus luteum is a hormone-secreting structure that develops in an ovary after an egg has been released. It degenerates after a few days unless pregnancy has begun.
- Menstruation is caused by a fall in the levels of estrogen and progesterone, which happens when the corpus luteum begins to disintegrate.

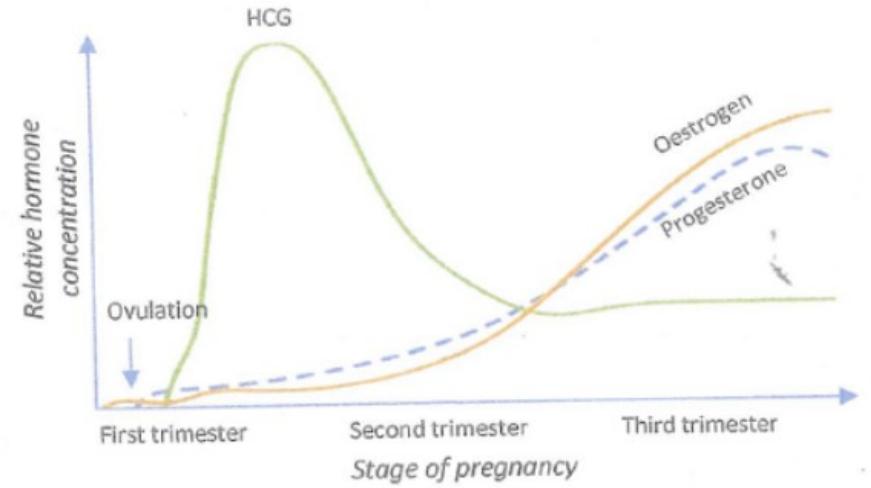


FERTILISATION AND IMPLANTATION

- Fertilisation = when the male gamete penetrates the female gamete and their nuclei meet.
- Cleavage (mitosis occurring) results in a rapid increase in the number of cells, but little to no increase in size.
- Hormones progesterone and estrogen prepare the uterus for pregnancy (allow implantation).
- When the blastocyst brushes against the endometrium, small projections form to attach it to the wall (implantation).
    - After implantation, HCG is released.

PREGNANCY AND BIRTH

- HCG is a hormone that maintains the corpus luteum - it is necessary to maintain estrogen and progesterone levels so the uterine lining can remain thick (receptive to an embryo).
    - Estrogen and progesterone also interact with secretory glands to decrease production of GnRH, FSH and LH which prevents menstruation or ovulation from occurring.
- During the second trimester of pregnancy, HCG declines and the corpus luteum deteriorates - but the placenta takes over the role of producing estrogen and progesterone.
- At the end of the third trimester, both baby and mother produce oxytocin which causes muscular contractions of the uterus for birth.



1.3 evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture

- Speciation = the evolution of a new species.
- Hybrids are the offspring of two plants or animals of different species/varieties, e.g. boysenberry or liger.
- Manipulating reproduction can be negative e.g. pugs bred for aesthetics have breathing problems
  - However, it can also be positive/promote survival – e.g. Brahman cattle are resistant to the effects of cattle ticks + thrive in harsh conditions
- There are four main steps that apply to all forms of selective breeding:
  - Determine the desired trait
  - Interbreed parents who show the desired trait
  - Select offspring with best form of the trait and interbreed those offspring
  - Continue this process until the population reliably reproduces the desired trait.

Knowledge	Technique	Example	Impact & Concerns on Agriculture
• Genes are transferred from parent to offspring • Variation occurs in sexual reproduction	• Selective Breeding (or artificial selection)- deliberate selection of organisms for breeding	• All modern crops and livestock e.g. Holstein Fresian dairy cow (selectively bred for high milk yields)	• Reduced genetic variation - increased susceptibility to diseases or parasites • Gene linkage - other traits close to ideal one on chromosome are also bred e.g. arthritis • Biofortification (improvement to nutritional quality) • Sterility • Monocultures
• DNA of an organism can be altered by the addition of new genes, or removing of genes or modification (genetic engineering)	• Development of transgenic organisms- cutting DNA from one species using enzymes and inserting that DNA into another species = genetic engineering	• BT cotton - added gene into cotton plant to have natural resistance to pesticide • AAT protein in sheep for lung disease	• Can increase fertility, quality and yield of meat, eggs, wool, meat etc • Can provide crop resistance to pests or disease • Monocultures are often grown which are sterile and have reduced genetic variation
• Genetic information is found in the nucleus	• Cloning-artificial production of an organism that is genetically identical to the parent/original-no alteration of DNA	• Cuttings, grafts, tissue culture, embryo splitting and somatic cell nuclear transfer	• Retain desirable traits • Reduces genetic variation

Other main concerns:

- Uncontrollable pest plant species (e.g. if transgenes promote rapid growth)
- Cross-pollination between GM and non-GM crops
- Loss of biodiversity/reduced genetic variation
- Gene linkage (i.e. by selecting for one trait, other linked traits are unavoidably carried with it during cell division, some of which may be undesirable).

IQ2: How important is it for genetic material to be replicated exactly?

2.1 model the processes involved in cell replication

- The cell cycle is the life cycle of a cell, involving growth, DNA replication and division to produce two identical daughter cells. It occurs in three main phases:
  - Interphase (cell spends most of its time in this stage)
  - Mitosis (PMAT)
  - Cytokinesis
- Cell replication allows for growth and development, maintenance and repair, and the restoration of the nucleus-to-cytoplasm ratio.

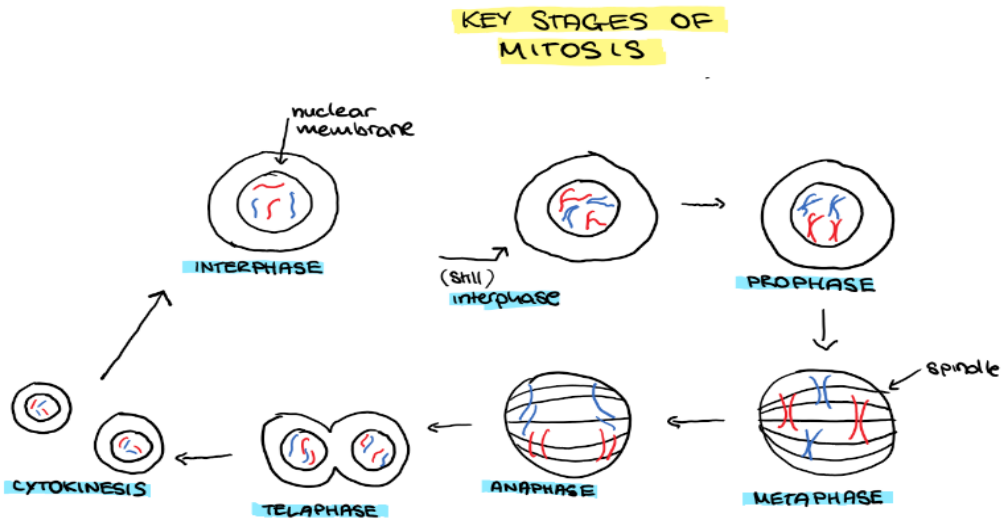
MITOSIS

Interphase	• Resting phase between cell division • Chromosomes are not visible (DNA is uncoiled)

	• DNA replication and protein synthesis occurs
Prophase	• Chromosomes condense and become visible • Centrioles move to opposite ends of the nucleus and form poles • Nuclear membrane breaks down • Centrioles form spindle fibres between the two poles
Metaphase	• Chromosomes align at equator of cell • Spindle fibres attach to centromeres of chromosomes
Anaphase	• Spindle fibres contract, splitting the centromeres and separating sister chromatids • Separated chromosomes are pulled to opposite poles
Telophase	• Nuclear membrane reforms around the two sets of chromosomes • Spindle fibres disappear
Cytokinesis	• Division of cytoplasm occurs • Two whole separate daughter cells are formed (end)

TERMINOLOGY

- Centromeres = parts of a chromosome where the two chromatids join (the bridge in an H).
- Centrioles = parts of cells that control spindles.
- Spindle = filaments involved in moving + segregating chromosomes in cell division
- Chromatids = half of a chromosome



MEIOSIS

- Meiosis (the formation of gametes) creates NEW genetic combinations, increasing genetic diversity and promoting the continuity of species.
- Meiosis I produces TWO haploid cells → HOMOLOGOUS CHROMOSOMES are separated.
- Meiosis II produces FOUR haploid cells → SISTER CHROMATIDS are separated.

MEIOSIS I – REDUCTION DIVISION

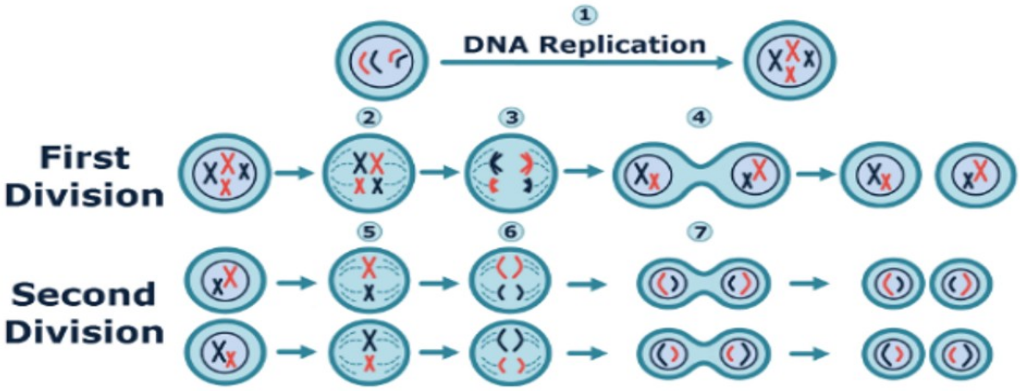
- INTERPHASE
  - Before division, the cell replicates its DNA.
- PROPHASE I
  - DNA condenses into chromosomes
  - Homologous chromosomes pair up

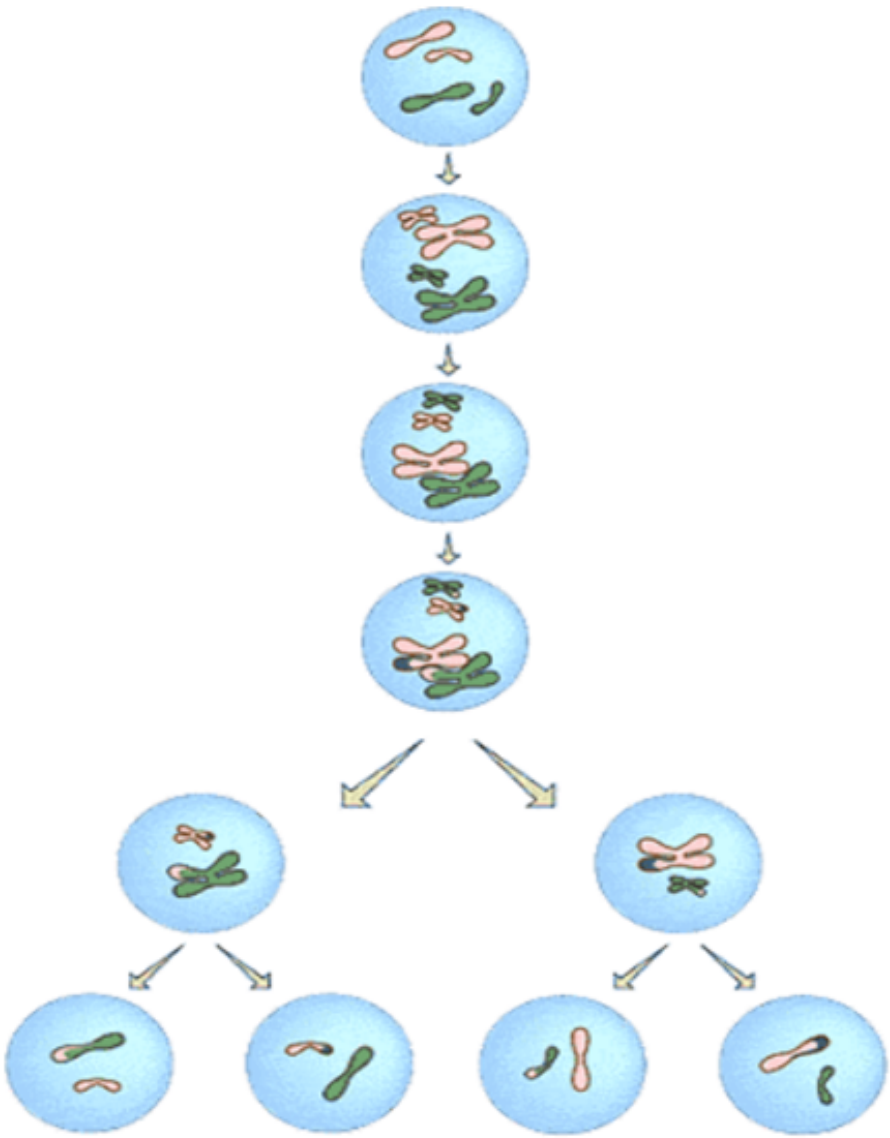
- At the END of PROPHASE I, CROSSING OVER can occur – paired homologous chromosomes may exchange sections of chromosomes at a locus.

- METAPHASE I
  - Parent cell's nuclear membrane breaks down
  - Chromosomes move to the equator/centre of the cell
  - Spindles form
- ANAPHASE I
  - Network of spindle fibres RANDOMLY separate the chromosomes to opposite ends of the cell.
- TELOPHASE I
  - Nuclear membranes form around the separated chromosomes.
- CYTOKINESIS I
  - Cell membrane pinches off to make two daughter cells.

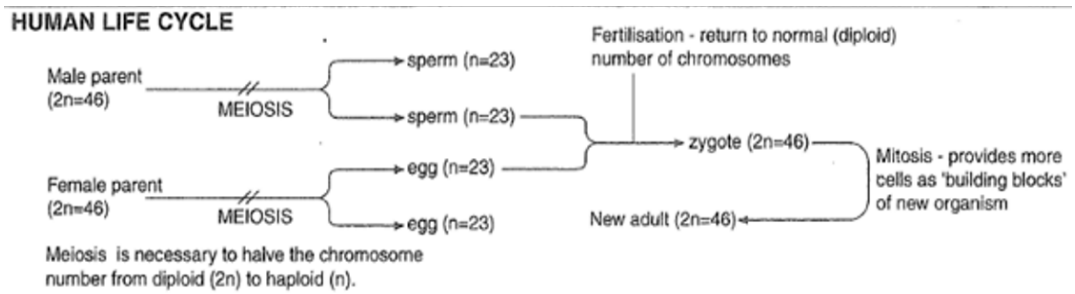
MEIOSIS 2 – SEPARATION DIVISION

- PROPHASE II
  - Nuclear membranes break down.
- METAPHASE II
  - Chromosomes align at the centre of the cell.
- ANAPHASE II
  - Network of spindle fibres separate SISTER CHROMATIDS
  - Separated chromatids move to opposite ends of the cell.
- TELOPHASE II
  - Nuclear membranes form around separated chromatids.
- CYTOKINESIS II
  - Membranes pinch off to make FOUR different haploid daughter cells (gametes).



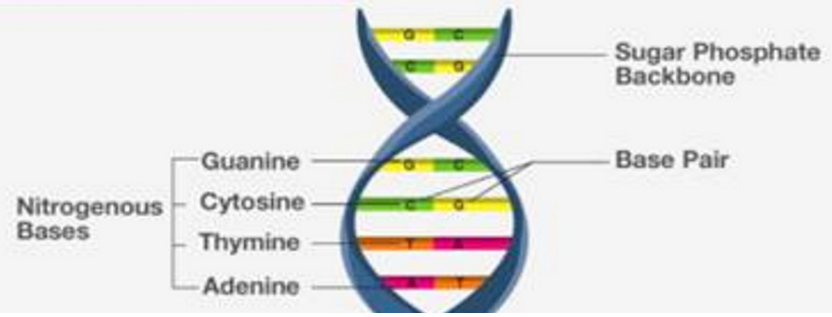


EXAMPLE: HUMAN LIFE CYCLE



DNA STRUCTURE (WATSON & CRICK MODEL)

- DNA is a double-helical structure made up of two polynucleotide strands (deoxyribose sugar attached to a nitrogenous base, plus a phosphate molecule).
  - Nitrogenous bases are adenine, thymine, guanine and cytosine.
- Antiparallel strands = run in opposite directions (5'3' vs. 3'5')
- Strands are joined by hydrogen bonds; AT having a double bond and CG having a triple bond.
- Phosphates are attached to sugars by a covalent bond.
- For the strands to remain equidistant, the bases must bond in a specific way, i.e. purines bond with pyrimidines.
  - Purine (single bond) = A and G
  - Pyrimidine (double bond) = C, T, and U
- Chargaff's rule = in DNA, the ratio of the bases A/T and C/G are always 1:1 (i.e. 5% A = 5% T).

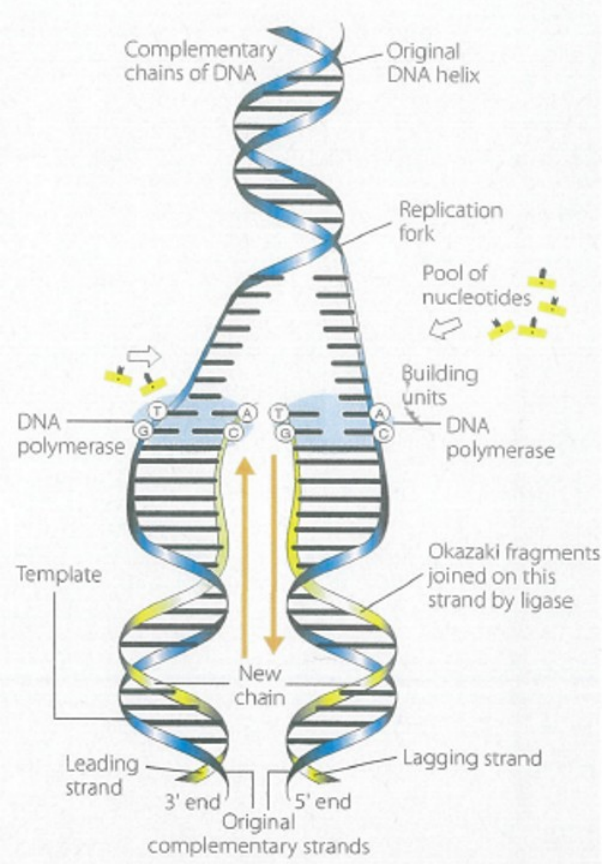


DNA FUNCTION

- DNA is responsible for storing and transferring genetic information.
- RNA directly codes for amino acids and acts as a messenger between DNA and ribosomes to make proteins.
- DNA replicates in interphase prior to mitosis, the process of which is controlled by enzymes.

PROCESS OF DNA REPLICATION

1. Helicase causes the DNA double helix to UNWIND (i.e. go from twisty to straight).
2. Helicase disrupts the weak hydrogen bonds between nucleotide bases, causing the two strands to SEPARATE
3. Nucleotides are ADDED against each single strand.
   - A short strand of RNA (primer) is made by primase and attaches to DNA.
   - DNA polymerase picks up free nucleotide units and inserts them opposite their complementary base partner (AT/CG).
   - This process is antiparallel - that is; each DNA strand has a 3 prime end and 5 prime end on opposite ends. Nucleotides are always added from the THREE prime (leading) end!
4. Replication ERRORS are identified and CORRECTED, and the DNA strand is SEALED.
   - DNA polymerase corrects base pair errors by splicing out the incorrect base and replacing it with the correct base.
   - Ligase seals the two strands together and the final pairing is checked by another DNA polymerase.
     - However, 1 in 10 billion base pairs may still be incorrect despite checking, and this is how mutations occur.



2.2 assess the effect of cell replication processes on the continuity of species

GENETIC CONTINUITY

- Genetic continuity refers to the identical replication of genetic information from a parent cell to two daughter cells, including the continuance of parental traits in offspring.
  - NOT the same as a lack of variation, as sexually reproduced individuals are unique.
- Cell replication plays a role in maintaining genetic continuity as it makes a perfect copy of DNA for new cells.
  - DNA replication occurs prior to both mitosis (production of body cells) and meiosis (production of gametes).
  - Healthy cells replicate in a highly regulated way, controlled with built-in checkpoints – however, the process is prone to errors.
- When there are errors in DNA replication e.g. neoplasms (abnormal/uncontrolled cell division), genetic continuity has been lost because normal cell cycles of replication cannot continue.

GENETIC STABILITY VS. VARIATION

- Genetic stability and variation both play a significant role in ensuring the continuity of species.
- Accurate DNA replication brings about genetic stability (the passing on of consistently accurate genetic information).
  - Genetic stability ensures that new cells/organisms have the working genes they need to survive, promoting the continuity of the species.
  - A lack of genetic stability results in disease, death or extinction.
- Mutation results in genetic variation, which is also vital for evolution.

- Variation better allows organisms to adapt to and survive a changing environment, promoting the continuity of the species.

IQ3: Why is polypeptide synthesis important?

3.1 compare the forms in which DNA exists in eukaryotes and prokaryotes

PROKARYOTES VS. EUKARYOTES

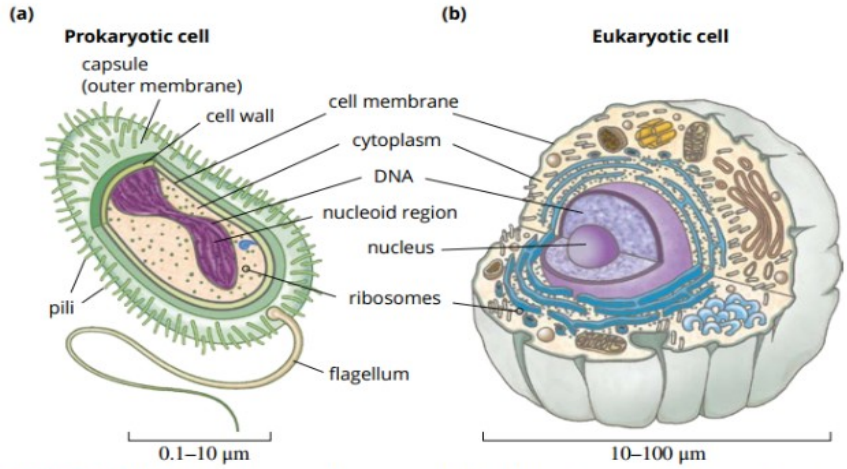
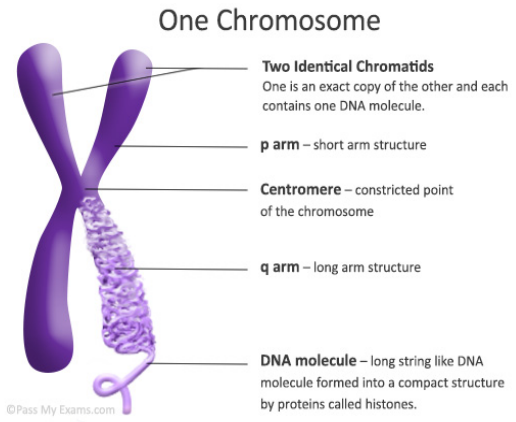


FIGURE 4.1.1 (a) A typical prokaryotic cell compared to (b) a typical eukaryotic cell. Prokaryotic cells and the processes involved in their DNA replication are generally much simpler than eukaryotic cells. Note the different membrane-bound organelles in the eukaryotic cell and the lack of such organelles in the prokaryotic cell.

Prokaryotic cells	Eukaryotic cells
Genetic material consists of one double-stranded, circular DNA chromosome (less).	Genetic material consists of double-stranded, linear DNA chromosomes tightly packaged (more).
DNA located in "nucleoid region", no histones.	DNA located in the nucleus, has histones.
Gene expression is regulated during transcription (first stage)	Gene expression is regulated at ALL stages
DNA replication is significantly faster (40 mins)	DNA replication is much slower (10 hours)
Gene expression consists only of polypeptide synthesis through transcription and translation	Gene expression occurs by transcription, RNA processing and translation

DNA PACKAGING IN EUKARYOTES

- DNA is tightly packed in the nucleus of every cell, wrapped around special proteins called histones - forming loops of DNA called nucleosomes.
  - These nucleosomes coil (supercoiling) and stack to form fibres called chromatids.
  - Chromatids then form larger loops and coil further to form chromosomes.



3.2 model the processes of polypeptide synthesis, including:

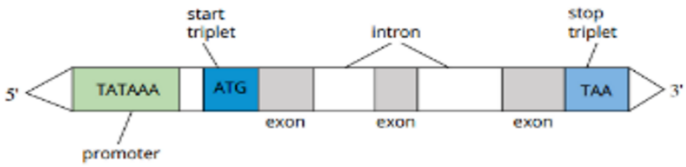
– assessing the importance of DNA and RNA in transcription and translation

SUMMARY

- Transcription → copying DNA onto RNA by initiation, elongation and termination
- RNA processing → making pre-mRNA into mature mRNA
- Translation → chain of amino acids formed (i.e. a polypeptide)

GENE STRUCTURE

- Exons = coding segments of DNA
  - Exons are usually 'expressed' as RNA. They then come together to make mRNA, which is translated into proteins.
  - i.e. gene codes for producing proteins are carried in the exon regions.
- Introns = non-coding regions of DNA
  - Introns are spliced out during RNA processing – i.e. NOT translated in gene expression.
- Stop/start triplet sequences = regions where encoding of DNA ends/begins for a specific gene.
- Promoter = section of a gene found on the DNA before the start triplet, at the 5' end of the site where transcription begins.
  - This is the location where RNA polymerase (enzyme that initiates transcription) attaches to the gene.

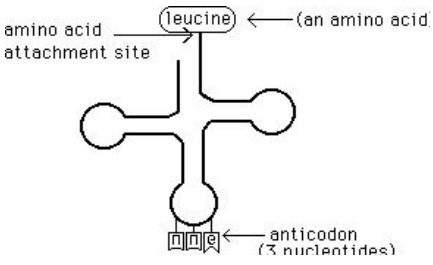


ROLE OF DNA IN POLYPEPTIDE SYNTHESIS

- Gene expression is the process by which information from a gene is used to synthesise a functional gene product.
- The genetic code defines how genetic information is translated into polypeptides.
  - Groups of three nucleotides on DNA are called triplets, and triplets transcribed into mRNA are called codons.
- DNA provides the instructions as a nucleotide sequence, which are translated by RNA into polypeptides.

ROLE OF RNA IN POLYPEPTIDE SYNTHESIS

- RNA expresses the information from DNA, coming in three main forms:
  - Messenger RNA (mRNA) is formed in the nucleus by transcription → carries a copy of DNA nucleotide sequence to the ribosomes, where it is translated into proteins
    - Pre-mRNA is the primary form before it is processed.
  - Ribosomal RNA (rRNA) makes the ribosomes.
    - Ribosomes sit in the cytoplasm on the rough ER.
  - Transfer RNA (tRNA) transfer specific amino acids from cytoplasm to ribosomes → anticodons on tRNA molecules enable AA's to be correctly placed.
    - Once an unrecognizable stop codon is reached, the polypeptide chain is released.



- Diagram of tRNA molecule: →

– analysing the function and importance of polypeptide synthesis

TRANSCRIPTION + RNA PROCESSING

Initiation	• RNA polymerase attaches to the promoter, unwinding and unzipping the DNA by breaking its hydrogen bonds.
Elongation	• RNA moves along the non-coding strand, producing a complementary mRNA strand. • RNA polymerase attaches complementary nucleotides to the template (AUCG).
Termination	• RNA polymerase reaches the termination site (stop codon) and transcription ends. • RNA polymerase detaches, releasing the mRNA and allowing the DNA molecule to reform.

- After transcription, the copy of pre-mRNA contains introns (non-coding sections).
  - RNA processing removes these and splices exons together to create a continuous code.
  - This produces mature mRNA which then moves to the cytoplasm for translation.

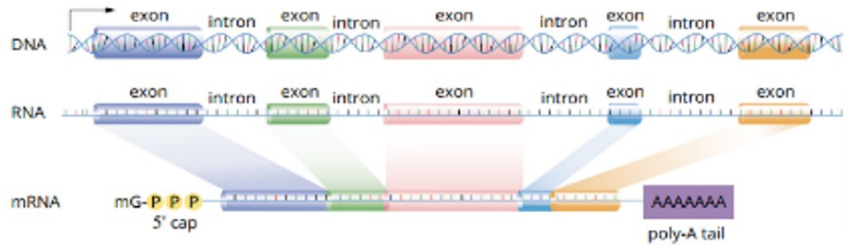


FIGURE 4.2.11 During RNA processing, the introns (non-protein-coding regions) are spliced from the primary pre-mRNA transcript, resulting in mature messenger RNA, which consists of only exons (protein-coding regions).

TRANSLATION

Initiation	• A ribosomal unit attaches to the 5' end of the mRNA strand and moves along until it reaches a start codon. • A tRNA molecule with a matching anticodon brings its amino acid to the mRNA and joins to the codon.
Elongation	• Another tRNA attaches to the next codon and adds its specific amino acid to the polypeptide chain. • Earlier tRNAs are released to be reused as more bind and add amino acids to the chain.
Termination	• The tRNA reaches a stop codon, releasing the polypeptide chain from the ribosome where it synthesises into a functional protein. • The mRNA is broken down into its individual nucleotides and reused.

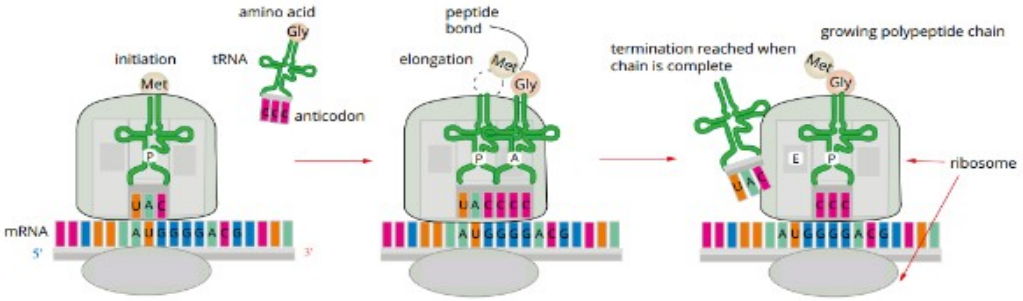


FIGURE 4.2.12 The process of translation on a ribosome. The ribosome moves along the mRNA one codon at a time and tRNA molecules bring their specific amino acids to their complementary mRNA codon. The amino acids join together by peptide bonds to form a polypeptide chain.

– modelling transcription and translation

EQUIPMENT

- A3 piece of paper → represents a simplified double helix DNA
  - 15 base pairs drawn onto paper represent a gene
  - Noncoding strand → right side of the paper
  - Coding strand → left side of the paper
- Central carpeted area of lab → represents the nucleus of a cell
  - Breaks between the desks → represent nuclear pores where the mRNA moves through
- Surrounding lab benches → represent ribosomes
- Staples → represent bonds between nucleotides
- Scissors → represent enzymes

METHOD

1. The piece of paper that represents DNA was cut in half, or 'unwound' by scissors, which represent RNA polymerase (unzipping enzyme).
2. Complementary RNA base pairs (pieces of paper) were matched to the noncoding strand i.e. right half of paper, and the DNA was copied.
3. mRNA bases on the strand were stapled together, representing the formation of an identical nucleotide.
4. The two halves of the A3 piece of paper were put back together, representing the DNA rewinding.
5. The mRNA nucleotide strand was carried out of the central area (nucleus) to a lab bench (ribosome).
6. tRNA codons (paper) were matched with mRNA codons.
7. The amino acids (individual bits of paper) attached to the tRNA were cut off with scissors after more than two had joined to the mRNA.
8. The amino acids were stapled together, representing peptide bonds.
9. When the stop codon was reached, the 'reaction' stopped and the whole polypeptide (chain of paper) was released.

EVALUATION

Benefits	Limitations
• Model was large enough to see a simplified process of polypeptide synthesis, which is usually invisible.	• Did not show the splicing of introns (RNA processing).
• Was able to show that transcription occurs in the nucleus and translation occurs in the ribosomes by	• Polypeptide chain was short.

physically walking the equipment from one area to another.	

– assessing how genes and the environment affect phenotypic expression

GENE EXPRESSION

- The phenotype is a set of characteristics expressed from the genetic information of a cell.
  - Encompasses ALL characteristics of an organism → e.g. behaviour, morphology, development, physiology etc.
- In other words, phenotypic traits are the expression of genes in an observable/measurable way → e.g. hair colour.
  - Genes/alleles which make up the genotype determine the phenotype; however, it is also influenced by the environment → e.g. human skin colour changing due to sunlight exposure.
- The proteome is the complete set of proteins expressed by the genome of an individual cell or organism at any given time.
  - Varies between cell type, developmental stage and environmental conditions.
- Although all cells contain the entire genome, only specific genes will be expressed or 'switched on' at any given time.
  - This ensures a cell produces only the proteins required for the specific function it carries out → e.g. skin cells do not produce insulin.

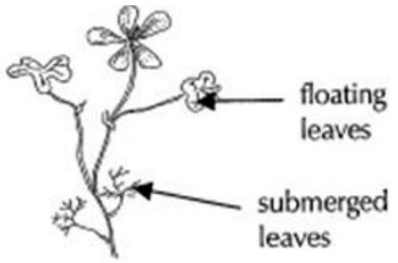
GENES AND THE ENVIRONMENT

- Genotype is non-modifiable, but an organism's phenotype may be modified by a range of environmental factors.
  - e.g. animals → oxygen levels, temperature, presence of mutagens, diet, light, etc.
  - e.g. plants → water, sunlight, temperature, nutrients, etc.
- Studies of identical twins show that even in genetically identical organisms, raising/rearing them in different environments can change their phenotype.

- ANIMAL EXAMPLE: HIMALAYAN RABBITS
  - These animals alter their coat colour/pigmentation with changes in temperature.
  - At low temperatures, the fur on their extremities is black to absorb heat.
  - At high temperatures, their fur becomes white to reflect heat.

- 

- PLANT EXAMPLE: WATER BUTTERCUP
  - This plant grows half-submerged in water.
  - Although its leaves are genetically identical, the leaves underwater are thin and finely divided whereas those above the water are broad and lobed so they can float.



3.3 investigate the structure and function of proteins in living things

PROTEIN FUNCTION

- Nearly every function of a living organism depends on proteins, as they are the building blocks of the body.
  - They speed up chemical reactions (e.g. enzymes), regulate cell function (e.g. storage protein ferritin) and provide structural support (e.g. collagen).
  - Alzheimer's and BSE (mad cow disease) are examples of diseases caused by protein malfunctions in humans.
- Most proteins are required to bind to other molecules to achieve functionality e.g. haemoglobin protein must attach to iron to bind oxygen.
- Classified into TWO main types:
  - Fibrous proteins → typically elongated and insoluble, have structural roles e.g. keratin
  - Globular proteins → compactly folded or coiled into spherical structures, compose most enzymes and hormones e.g. insulin

PROTEIN STRUCTURE

- Amino acids are joined in a condensation polymerisation reaction, which removes water and forms peptide bonds between them.
- The backbone of a polypeptide chain is formed by repeats of carboxyl and amine groups – R groups form the side chains.
- Four different levels of organisation: primary, secondary, tertiary and quaternary.
  - Primary → linear sequence of amino acids in a polypeptide chain that provides information on how the proteins will fold.
  - Secondary → folding or coiling of the polypeptide chain due to hydrogen bonds (alpha helix, beta-pleated sheets or random coil).
  - Tertiary → bonds formed between the R groups creating a more stable folded structure.
  - Quaternary → polypeptide chains joined to create a single functional protein.

Amino acid structure

All amino acids have the same basic structure (Figure 4.3.4):

- an **amine group** (NH_2)
- a **carboxyl group** (COOH)
- an **R group** (or side chain).

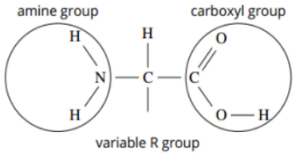


FIGURE 4.3.4 The basic structure of an amino acid, showing an amine, carboxyl and variable R group

CHAPERONINS

BIOFILE CCT

Chaperonins

Crucial to the protein-folding process are **chaperonins** (or chaperone proteins). Chaperonins are protein molecules that assist in the proper folding of other proteins. Chaperonins do not specify the final structure of a polypeptide, but instead provide the polypeptides with an area to fold in without influences from the cytoplasmic environment (e.g. changes in pH) (Figure 4.3.16). Another function of chaperonins is to prevent newly synthesised polypeptide chains and assembled subunits from being denatured and becoming non-functional structures due to high temperatures.

It is now thought that some inherited diseases associated with the lack of function of a particular protein may be due to a fault in chaperonins rather than a mutation in the gene for the protein itself. The sequence of amino acids in the polypeptide may be correct, but the protein may not be correctly folded into its functional structure.

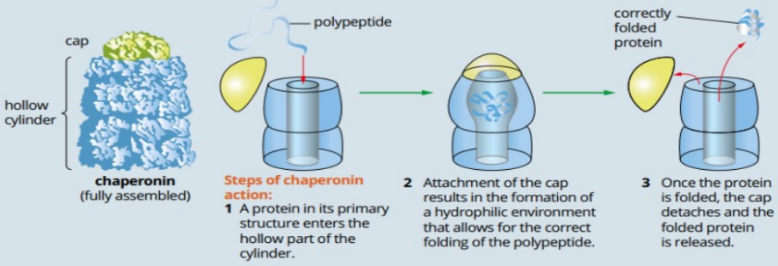
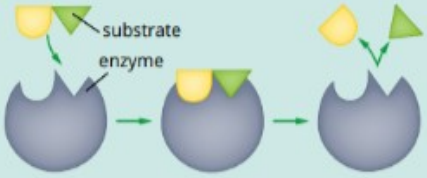


FIGURE 4.3.16 A computer graphic showing a large chaperonin protein complex from the bacteria, *E. coli*. It has an interior space that provides a shelter to allow the correct folding of newly made polypeptides.

PROTEIN STRUCTURE/FUNCTION TABLE

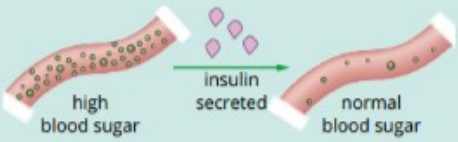
Function: enzymatic proteins

Description: act as catalysts in cellular reactions (enzymes)
Examples: catabolic enzymes, such as lipase and amylase, that catalyse the breakdown of bonds (also known as hydrolysis); anabolic enzymes, such as DNA polymerase, that catalyse the formation of bonds (also known as condensation)



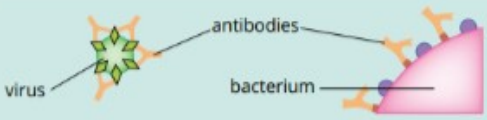
Function: hormonal proteins

Description: coordinate an organism's activities by triggering responses
Examples: insulin, glucagon



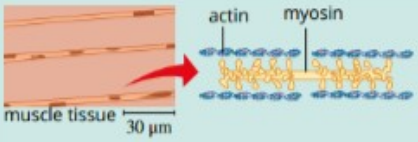
Function: immunological proteins (antibodies)

Description: protect against disease by recognising foreign bodies and microbes; activate immune cells
Examples: immunoglobulins (antibodies), complement, major histocompatibility complex proteins



Function: contractile and motor proteins

Description: contractile proteins aid muscle contraction; motor proteins are responsible for the movement of cilia and flagella
Examples: myosin, actin, kinesin, dynein, spindle fibres used in cell division

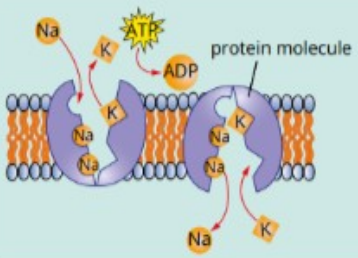


Function: structural proteins

Description: provide support by forming the structural components of cells and organs; assist in contractile functions in tissue such as muscle
Examples: collagen, keratin, actin, cytoskeleton, cell membranes

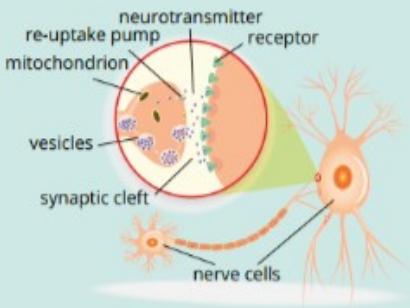
Function: transport proteins

Description: transport of substances by acting as carrier molecules within or between cells; act as membrane channel proteins
Examples: haemoglobin, sodium–potassium pump, calcium channel



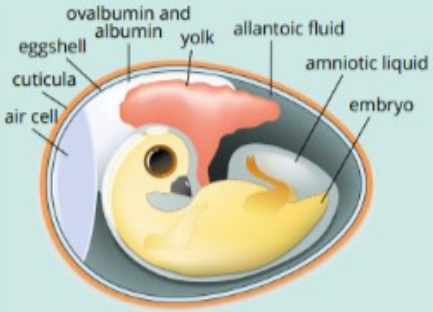
Function: receptor proteins

Description: assist the cell in responding to chemical stimuli
Examples: neurotransmitter receptors, hormone receptors



Function: storage proteins

Description: storage of metal ions and amino acids
Examples: ovalbumin and casein (to store amino acids), and ferritin (to store iron)



IQ4: How can the genetic similarities and differences within and between species be compared?

4.1 examine variations in the genotype of offspring by modelling meiosis – including crossing over, fertilisation and mutations

SPECIES

- Individuals of the same species can (usually) interbreed successfully amongst themselves.
- Biological species concept = "groups of actually or potentially interbreeding natural populations which are reproductively isolated from other such groups."
    - *Note: this cannot be used to classify asexually reproducing organisms.*

SEXUAL REPRODUCTION

- Sexual reproduction is the overarching source of variability in offspring.
    - It differs to asexual reproduction wherein organisms make identical copies of themselves, resulting in little variation.
- Sexual reproduction allows for different combinations of genes to be produced by meiosis, fertilisation, and pre-existing variation in a population's genes (i.e. choosing a mate).
- Variation allows species to better adapt to and survive in changing conditions.

MEIOSIS

- One parent cell undergoes two divisions to create four haploid daughter cells.
- The genes in each haploid cell are a new combination of the parental genes, a result of crossing over and random segregation.
- Crossing over is the exchange of genetic material between homologous chromosomes.
    - This creates new genetic combinations and interactions = major source of variation.
- Random assortment occurs during the second division, wherein genes on sister chromatids may line up and assort into any mixture before dividing.
    - This creates several new combinations of the individual alleles of maternally and paternally derived chromosomes.

FERTILISATION

- A single gamete contains one full copy of each chromosome from the parent, e.g. a human male sperm cell would contain the haploid (n) number 23.
- Fertilisation is the union of a male and female gamete to form a diploid (2n) zygote containing an equal combination of the parental genes.
- It is a completely RANDOM process, making it an important source of variation.
    - i.e. each sperm has an equal chance of penetrating the ovum, so any combination of genes from any sperm could succeed in fertilising a single egg.

MUTATIONS

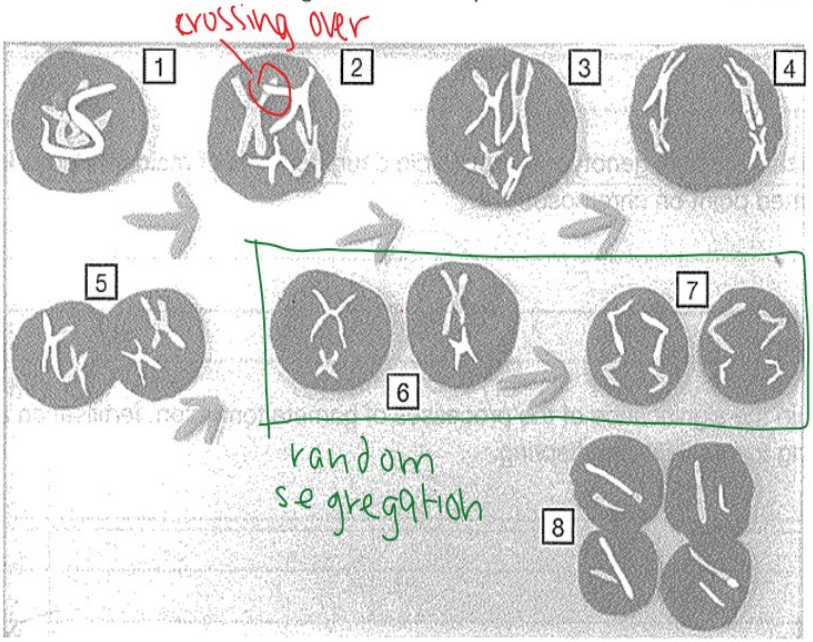
- Mutations are a permanent change in the nucleotide sequence of a gene, e.g. body cell mutations causing cancer
  - Some mutations make genes work better, e.g. single copy of mutated HBB gene giving resistance to malaria, so they are the basis of evolution by natural selection.
  - Other mutations interfere with the actions of a gene in a damaging way e.g. two copies of mutated HBB gene causing sickle cell anaemia
- Somatic cell mutations cannot be passed on, but germline (reproductive) cell mutations can.
  - Missing, extra or malformed chromosomes resulting from defective gametes may have serious consequences for offspring e.g. Down Syndrome.
  - Deletions can cause essential genes to go missing and are usually lethal.
  - Duplications, translocations and inversions of chromosomes can result in changes in physical characteristics.

CALCULATING CHROMOSOME COMBINATIONS IN MEIOSIS

- If you know the haploid (n) number for an organism, you can calculate the number of possible combinations in the gametes.
- The possible combinations are equal to 2^n, where n is the haploid number.
  - e.g. How many combinations of chromosomes would be possible in the gametes formed when a cell with eight chromosomes undergoes meiosis?
    - In this case, the haploid number is 4 (8÷2).
    - $2^4 = 16$, so there could be 16 possible combinations of chromosomes.

PRACTICAL MODEL

A biology student constructed the following model out of plasticine to show meiosis and crossing over.



- *Note: Models need to be labelled and have the chromosomes be different colours.*

4.2 model the formation of new genotype combinations produced during meiosis, including:

– interpreting examples of autosomal, sex linkage, codominance, incomplete dominance and multiple alleles using Punnett squares

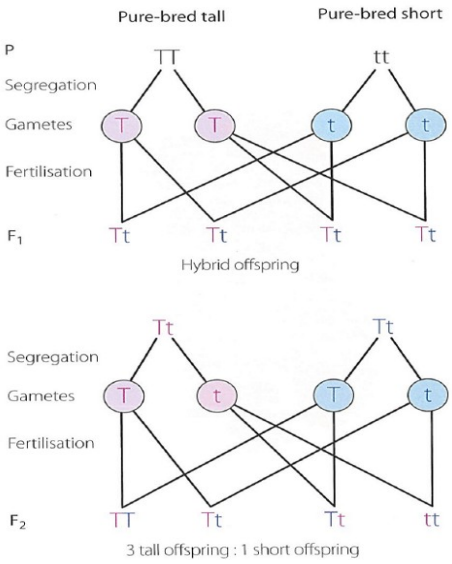
AUTOSOMAL INHERITANCE

- Autosomal inheritance was proposed as a result of Mendel's experiments with pea plants.
  - States that if two alleles of a gene are present in a population, one allele is dominant (seen in the phenotype) and the other is masked or recessive.
  - Assumes that alleles are located on one of the NON-SEX chromosomes (autosomes).

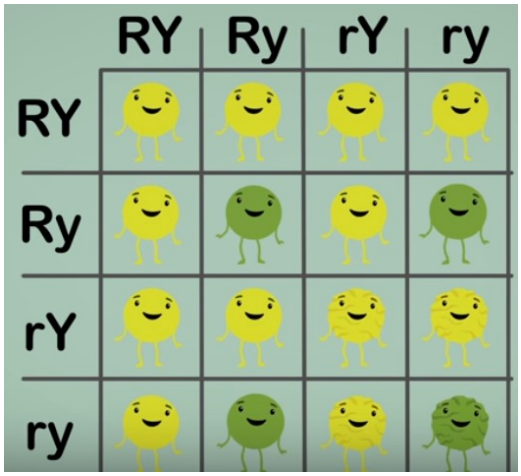
MENDEL'S EXPERIMENTS + LAWS

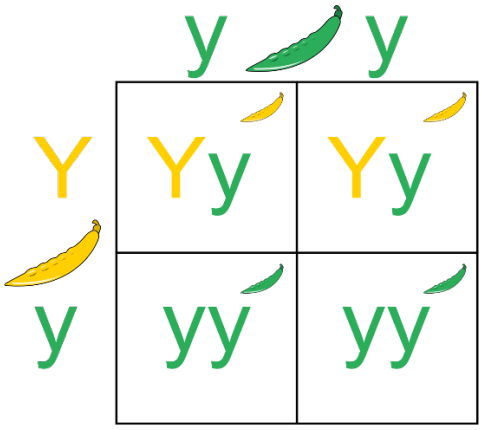
- Mendel carried out a pure-breeding cross followed by a monohybrid cross.

- First law of dominance and segregation → states that the characteristics of an organism are determined by factors that occur in pairs
  - Only one member of a pair of factors can be represented in any gamete (segregation), thus offspring inherit one factor from each parent.
  - When two hybrids breed, they will produce a ratio of three offspring showing the same trait as the parents (dominant trait) to one offspring showing the recessive trait (i.e. 3:1 ratio).



- Second law of independent assortment → assumes genes are located on different chromosomes
  - Studied dihybrid crosses observing two pairs of factors and their separation.
  - Found when individuals with two or more pairs of unrelated, contrasting characteristics are crossed, the different pairs of factors separate out independent of each other.
    - e.g. crossing round yellow peas x wrinkled green peas -> yellow is not always inherited with round nor green with wrinkles





SEX LINKAGE

- Sex determination refers to the combination of X and Y chromosomes to determine sex.
  - In humans, XX = female and XY = male, however in other organisms it can differ e.g. male birds = ZY and female birds = ZW.
- Sex linkage occurs when genes carries on the X and Y chromosomes code for characteristics other than gender, e.g. red-green colour vision, blood clotting
  - Males will express all genes on their X chromosome whether they are dominant or recessive, since there is no equivalent allele on the Y chromosome to mask the gene.
    - This means they are hemizygous – only have one allele for a gene.
    - Hence recessive disorders appear more frequently in males e.g. haemophilia.
    - *Note: the MOTHER passes on the X chromosome to a male child.*
  - If a female inherits a mutant gene on the X chromosome, she will not suffer from the disorder if her other allele is dominant – these people are called carriers.
  - Some genes are Y-linked and appear in males only, but these are less common e.g. chromosome infertility.

SEX LINKAGE IN CATS

- Tortoiseshell cats can only be female, displaying codominance and sex-linked inheritance.
  - X^BX^B = black female
  - X^OX^O = orange female
  - X^OX^B = tortoiseshell
  - X^BY = black male
  - X^OY = orange male
- Punnett square:

Gametes	X^B	Y
X^B	X^BX^B	X^BY
X^O	X^OX^B	X^OY

Results → 1 black male : 1 orange male : 1 black female : 1 tortoiseshell female

INCOMPLETE DOMINANCE

- Incomplete dominance = blending of alleles
  - e.g. crossing red snapdragon flowers with white = pink flowers

- The pink flowers breeding may then produce pink flowers OR pure white or red flowers.

- Special notation is used to represent alleles that show incomplete dominance – a letter is used to represent the gene e.g. C for colour, and alleles are written as superscripts next to the gene
  - e.g. red allele would be C^R and white allele would be C^W

CODOMINANCE

- Codominance = both alleles expressed (creating a new phenotype).
  - e.g. crossing a red bull and white cow = roan calf with mottled appearance (not in patches, but interspersed)
- Like incomplete dominance, it does NOT show a Mendelian pattern.

MULTIPLE ALLELES

- Multiple alleles occur when there are several different alleles for one gene e.g. human blood type, whereas polygenes are a group of genes that control one trait e.g. height in humans.
  - These give populations greater genetic variability and thus greater diversity.
  - *Note: while there may be several different alleles, an INDIVIDUAL can only have two alleles (determined by parental inheritance).*

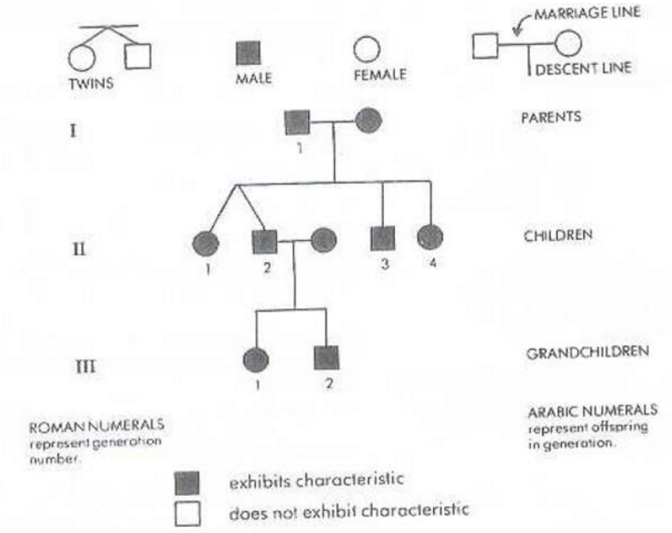
BLOOD TYPE IN HUMANS

- The gene for blood type in humans has three alleles: A, B, and O – represented as I^A, I^B and I^i.
- The A and B alleles are codominant and produce a marker on red blood cells.
- However, the i allele produces no marker and is recessive to A and B.
  - As a result, there are 4 possible phenotypes (A, B, AB or O) and 6 possible genotypes (AA, BB, Ai, Bi, AB, or ii).

– constructing and interpreting information and data from pedigree charts

PEDIGREES

- To determine if a trait is dominant or recessive:
  - Find two parents who have the SAME characteristic who had a child with a DIFFERENT characteristic.
  - The parents will have the dominant trait and the child will have the recessive trait.
- To explain how you know the trait is recessive:
  - For two parents to have a characteristic different to



their child, they must be heterozygous and have both passed on the recessive allele to the child.

- Alternatively, two parents who are recessive can only have recessive children.

PEDIGREE ANALYSIS

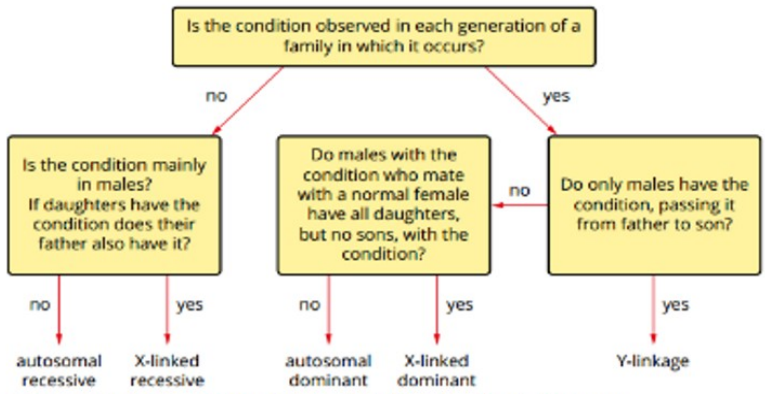


FIGURE 5.3.32 Flow diagram for pedigree analysis of simple modes of inheritance

4.3 collect, record and present data representing frequencies of characteristics in a population to identify trends, patterns, relationships and limitations in data, for example:

– examining frequency data

POPULATION GENETICS

- Population genetics is the study of how the gene pool of a population changes over time, leading to a species evolving.
- The gene pool is the total of all genes and their alleles within a population.
- Genetic diversity is the total of all the genetic characteristics in the genetic makeup of a species.
  - This is dependent on genetic variability, i.e. the tendency of individual genetic traits in a population to vary.
  - Species with greater genetic diversity have greater potential to adapt and survive.
- Population genetics (combination of Mendelian + Darwinian evolution) explains how changes in allele frequencies arise and how these changes can lead to microevolution and macroevolution.

EXAMINING FREQUENCY DATA

- Population geneticists study factors that cause changes in allele frequency.
  - e.g. temperature change (selective pressure) causing natural selection
  - External factors such as gene flow and genetic drift are also studied.
- They use a model based on allele frequencies typical of a stable population with Mendelian inheritance (in equilibrium) and then compare to a real population exposed to selective pressures.

GENETIC VARIATION AND FREQUENCIES OF CHARACTERISTICS

- Microevolution can be studied by examining changes in allele frequency in a population over several generations.
- Genetic variability in a population can be determined by examining the relative proportion (ratio or percentage) of a given phenotype, genotype or allele within that population.
- Allele frequency is a measure of how common an allele is within a population.
  - Can be calculated by counting the number of copies of an allele in a population and then dividing by the total number of copies of all alleles of the gene.
    - e.g. frequency of allele G = copies of G in population / total copies of the gene (G + g) in population

– analysing single nucleotide polymorphisms (SNPs)

SINGLE NUCLEOTIDE POLYMORPHISMS

- SNPs represent a difference in a single DNA nucleotide.
- Occur in 1% of the population – not a mutation, but a variation
- Can use them as biological markers to put individuals into categories called haplotypes
  - e.g. haplotype 1 = c t t g g a c t
  - e.g. haplotype 2 = c t g g g a c t
  - Can help predict an individual's response to certain drugs, susceptibility to environmental factors, and/or risk of developing diseases.
- Most SNPs occur outside of genes and therefore have no effect on genes or proteins → linked SNPs
- When SNPs occur inside of a gene, they can create different versions of an allele e.g. green eyes → causative SNPs
  - SNPs in the coding regions (exons) change the protein product
  - SNPs in the non-coding regions (introns) change how much protein is produced

Use of SNP	Description
Genetic screening and genetic testing	SNPs are used in genetic screening checking for the presence of disease genes, e.g. sickle cell anaemia, beta thalassaemia and cystic fibrosis and the same testing procedures are used in genetic testing which check for a particular condition due to family history or the result of genetic screening. This data can be used to track family inheritance of particular genetic diseases and disorders.
Pharmocogenetics	Pharmacogenetics studies the influence of one or a few genes on the response to a drug. Analysis of particular genomes has shown how particular genotypes respond to certain drugs and therapies. Using SNPs to identify particular alleles in an individual means drugs that cause adverse reactions can be avoided and personalised health care treatments can be implemented for a particular individual's genotype.
Forensics	Forensics and DNA fingerprinting are used to identify individuals and the evidence can be used to solve crimes and in court cases.
Human evolutionary and migratory history	Different human populations have different allele frequencies for different SNPs. Collecting worldwide data and using fossil evidence shows the differences between populations and analyses show trends and patterns in changes suggesting possible human migration routes throughout history and steps in human evolution.

SHORT TANDEM REPEATS

- STRs are sections of non-coding DNA that are in the same position on chromosomes for every individual and are repeated many times.
  - Are unique to individuals because they repeat by different amounts – used in DNA profiling.

IQ5: Can population patterns be predicted with any accuracy?

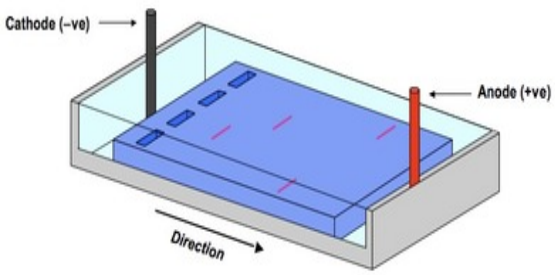
5.1 investigate the use of technologies to determine inheritance patterns in a population, for example:

– DNA sequencing and profiling

DNA SEQUENCING AND PROFILING

Feature	DNA sequencing	DNA profiling
Definition	DNA sequencing involves determining the exact order of nucleotides (e.g. A, T, C, G) in a DNA molecule.	DNA profiling involves determining the identity of an individual using a number of DNA restriction fragments.
What is involved	Determines exact nucleotide sequence in DNA molecule and genome of an organism.	Looks at important variable regions of DNA called microsatellites which are short sequences repeated many times with the number of repeats different from individual to individual.
Technologies used	Polymerase chain reaction which makes many copies of short sections of DNA, sequencing reaction, gel electrophoresis and computer scanning which interprets the sequence from the gel into a sequence of nucleotides.	Polymerase chain reaction and gel electrophoresis is used to separate the markers giving the common banding pattern.
Detail gathered	Very detailed with nucleotide sequence of DNA molecules and genome information.	Does not attempt to determine sequence, providing less information.
Uses	Used in scientific research, e.g. to determine the function of a section of DNA, evolution patterns and relationships between species.	Used in forensics to match samples that can be used in court cases.
Time and cost	Takes longer to gather details and more expensive.	Faster and cheaper.

GEL ELECTROPHORESIS



- Samples of fragmented DNA are placed in the wells of an agarose gel.
- The gel is placed in a buffering solution and an electrical current is passed across the gel.
- DNA, being negatively charged (due to phosphate), moves to the positive anode.
- Smaller fragments move faster through the gel.
- The fragments are then separated according to size.
    - Size is calculated (in kilobases) by comparing against a known industry standard.

5.2 investigate the use of data analysis from a large-scale project to identify trends, patterns and relationships

DATA ANALYSIS

- Qualitative data = characteristic results, subjective e.g. frequent vs. rare
- Quantitative data = numerical results, objective e.g. time
  - Involves calculating averages (outliers may be problematic)
  - Difficult in population genetics
- Valid conclusions can still be drawn with acknowledgement of limitations
- Scatter graphs can show trends/correlations.

– population genetics data in conservation management

POPULATION GENETICS

- Concerns maintaining sufficient genetic diversity within small populations of endangered animals, with the aim to maintain their health and eventually release them back into the wild.
- Genetic diversity is important to identify favourable traits that help them survive.
- Genetic drift = important alleles can be lost in a population by the death of a few individuals.
- Isolated populations are useful for studying disease because they show very little genetic variation (from inbreeding).
  - Certain disorders become more common in these restricted populations.

CONSERVATION STUDY – KOALA

- Collaboration with researchers, consultants, Port Macquarie Koala Hospital, Australia Zoo Wildlife Hospital and Australian Museum Tissue Collection
- Tissue samples were obtained from 662 wild koalas and mitochondrial DNA control region was analysed (sequencing and profiling) to determine base sequence.
  - Identified varying genetic diversity depending on location
  - Positioning on maps, SNPs, and haplotype groups were analysed
  - To increase genetic diversity in some regions, researchers plan to introduce koalas from regions that have very different DNA.

» ▪ The lines connect each haplotype to its most similar relative.

▪ The number of bars on the lines represents the mutational steps between the haplotypes. The more bars, the greater the difference in the sequence of bases between the haplotypes.

A simplified haplotype network is shown in Figure 6.14. This diagram shows three haplotypes in the species being tested, ranging in size from the largest, with 8 individuals sharing a haplotype, to the smallest, with 2 individuals sharing a haplotype. The circle with 8 and the circle with 4 are the most closely related. The bars on the line between 2 and 4 indicate that there are two mutational steps between groups 2 and 4 and, therefore, genetic divergence.

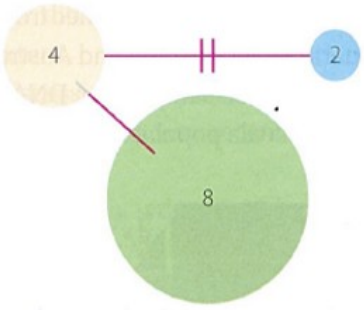


FIGURE 6.14 A simple haplotype network diagram

– population genetics studies to determine the inheritance of a disease or disorder

DISEASE EXAMPLE – BRCA GENES AND CANCER

- BRCA1 and BRCA2 are tumour suppressor genes that help regulate the process of cell division.
- Genes of 46,000 women globally were sequenced to identify the prevalence of BRCA1 and BRCA2 mutations → ancestry also studied to determine any link between ethnicity and risk
  - Found that ethnic groups had higher levels of BRCA1 and 2 than their western counterparts.
  - Found that for BRCA1 carriers, there is an increased risk of developing cancer over the age of 35.
  - Found that for BRCA2 carriers, there is an increased risk of developing cancer over the age of 50.

– population genetics relating to human evolution

HUMAN MIGRATION THEORIES

Human migration theories

There are two main contesting theories regarding human migration out of Africa: the *Multiregional hypothesis* and the *Replacement hypothesis*.

The **Multiregional hypothesis (MRE)** relies mostly on fossil evidence and suggests that all human populations can be traced back to when *Homo erectus* first left Africa, about 2 million years ago. It is suggested that there was gene flow between neighbouring populations and that once they dispersed into other portions of the old world, they slowly evolved into modern humans (Fig. 6.22).

The **Replacement hypothesis**, also called the Out of Africa or Eve hypothesis, suggests that archaic *Homo sapiens* left Africa. It proposes that a second migration out of Africa happened about 100 000 years ago and that modern humans of African origin conquered archaic groups and replaced them by interbreeding with and out-competing them.

EVOLUTION EXAMPLE – OUT OF AFRICA THEORY

- Researchers sequenced over 18,000 whole human mitochondrial DNA genomes globally.
- Mitochondrial DNA chosen because of its maternal inheritance – can link back to "Eve".
  - Discovered that, among modern humans, most of the variation in mtDNA sequences was in African populations → the more diverse the DNA, the longer the population has existed
  - Provides evidence for the replacement (out of Africa) hypothesis.