HSC / Yr 12
BIOLOGY

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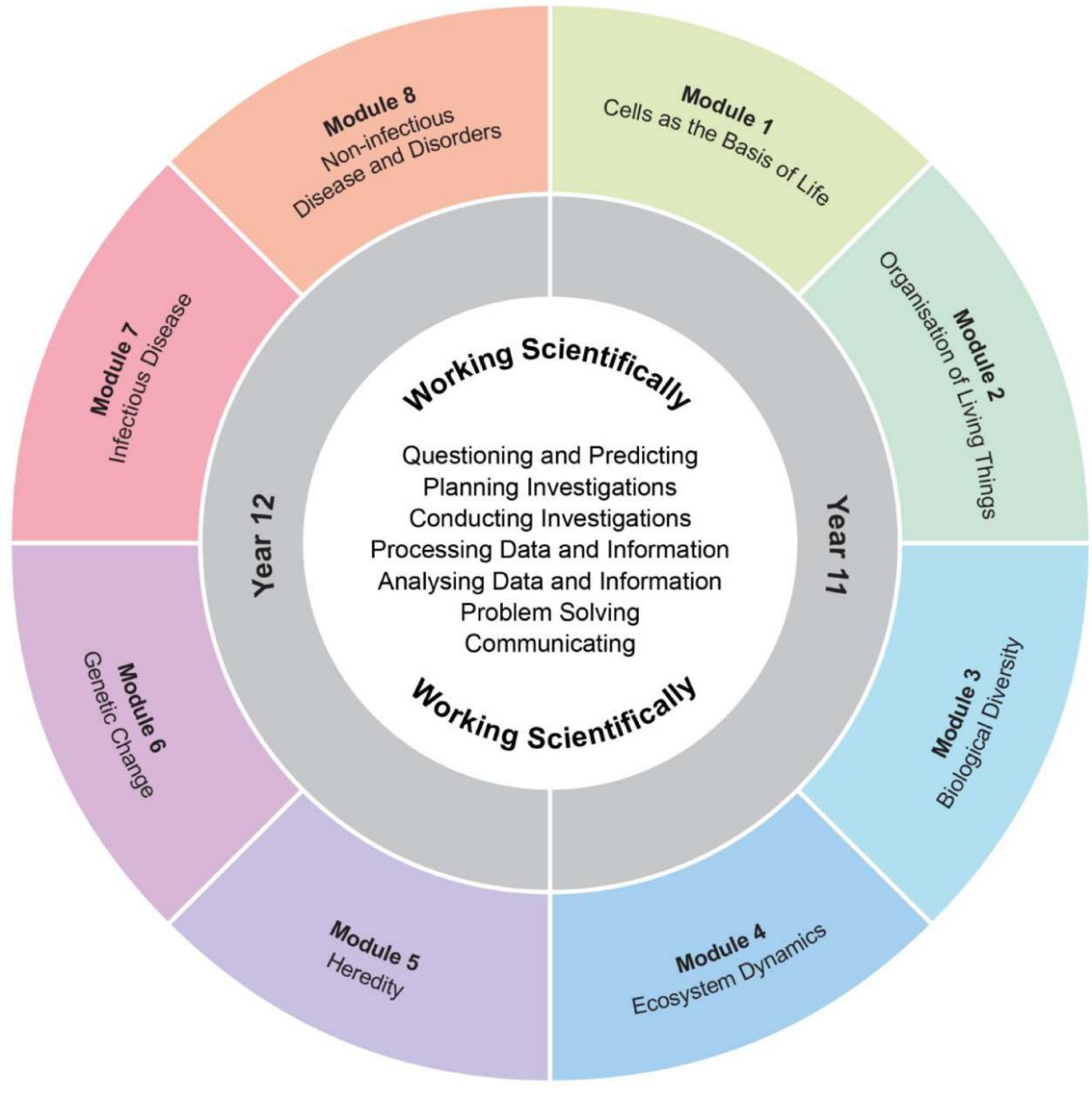
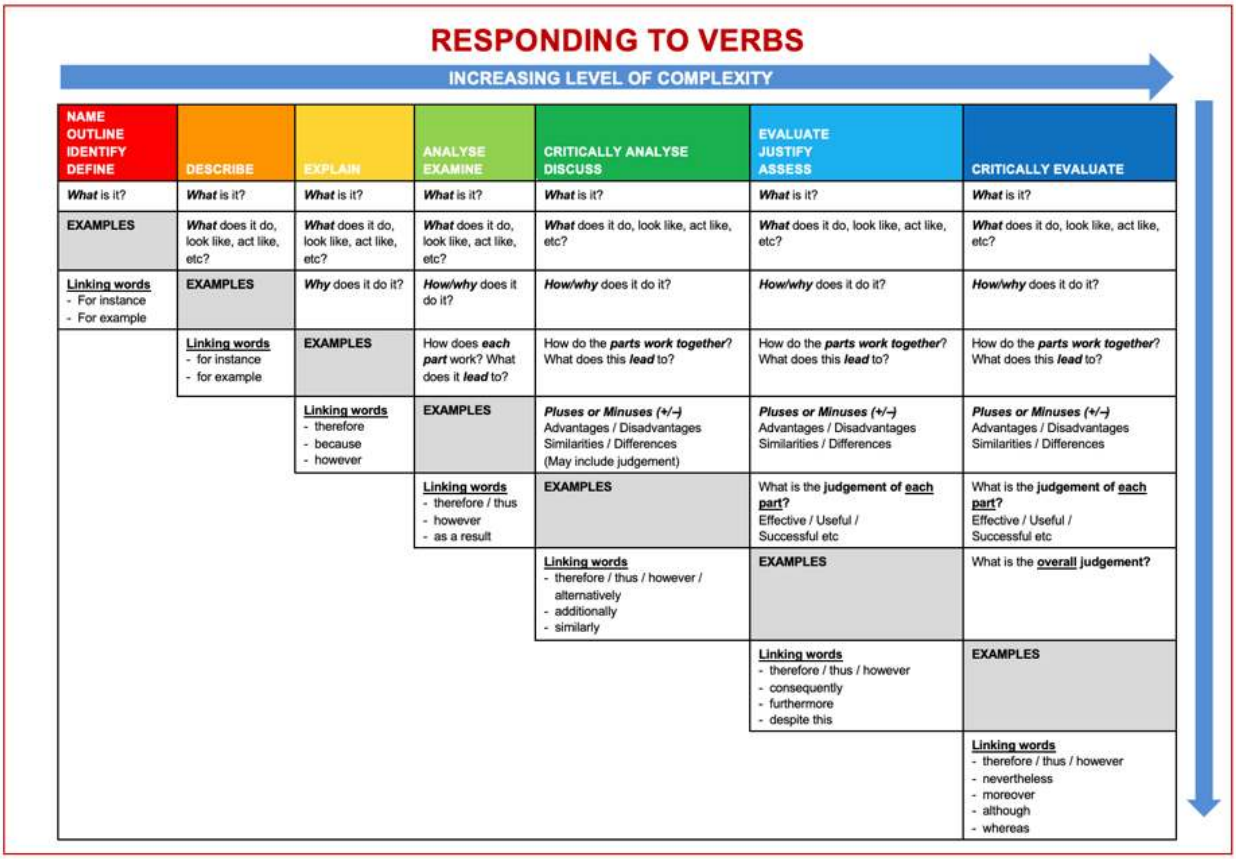
Summary Notes





Key

Purple: syllabus

Module 5 - Heredity

Outcomes

A student:

› selects and processes appropriate qualitative and quantitative data and information using a range of appropriate media BIO11/12-4
› analyses and evaluates primary and secondary data and information BIO11/12-5
› solves scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO11/12-6

› explains the structures of DNA and analyses the mechanisms of inheritance and how processes of reproduction ensure continuity of species BIO12-12

Content Focus

Life continues through the processes of reproduction and heredity. Students expand their knowledge of evolution by understanding the cellular processes involved in increasing genetic diversity. They investigate reproduction and inheritance patterns in both plants and animals as well as the role of DNA in polypeptide synthesis and the uses of technologies in the study of inheritance patterns.

Students also learn about contemporary research and the work of geneticists across a variety of industries, including medical applications and agriculture. They explore the effects on society and the environment through the application of genetic research.

Working Scientifically

In this module, students focus on processing and representing data in appropriate formats to analyse and evaluate trends, relationships and patterns. Students derive and justify valid conclusions about the processes involved in heredity. Students should be provided with opportunities to engage with all Working Scientifically skills throughout the course.

Content

Reproduction

Inquiry question: How does reproduction ensure the continuity of a species?

Students:

● 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: advantages of external and internal fertilisation
- plants: asexual and sexual reproduction
- fungi: budding, spores

- bacteria: binary fission (ACSBL075)
- protists: binary fission, budding

Sexual vs Asexual Reproduction

Terminology ...

- ★ Haploid
  - n, single set of chromosomes,
  - half no. of chromosomes as a somatic (body cell)
  - Found in gametes (sperm & eggs)
- ★ Diploid
  - 2n, two complete sets of chromosomes, one from each parent
  - Found in somatic cells

Sexual Reproduction

- ➢ Requires 2 parents. Can occur in pretty much anything except bacteria.
- ➢ 2 haploid gametes combine in fertilisation, forming a zygote
- ➢ Offspring are unique, resulting in genetic variation
- ➢ Genetic variation assists species continuity by ensuring diversity in times of selective pressure and environmental change. (Varied vulnerabilities)
- ➢ Increased variation = increased chance of some favourable characteristics present. Diseases less likely to affect whole population.
- ➢ Cons: Large time & energy investment, requires a partner, fewer offspring.

Asexual Reproduction

- ➢ Only requires 1 parent
- ➢ Many types, including
  - Binary Fission (Bacteria)
  - Budding (Fungi & certain animals)
  - Spores (Fungi)
  - Also, not on syllabus are fragmentation (many plants, some animals) and parthenogenesis (invertebrates, certain fish, amphibians & reptiles)
    - Fragmentation: organisms break into fragments which develop into a new individual eg. Starfish arm —> new starfish
    - Parthenogenesis: embryo develops from unfertilised egg. Some species use both parthenogenesis and sexual reproduction. Eg. Komodo dragon.
- ➢ Offspring are identical to parent

- ➢ Faster than sexual reproduction, no need to find a mate, lower energy requirement, no need to spend time raising offspring.
- ➢ For species continuity, asexual reproduction allows rapid population growth and can allow a species to quickly establish a position in its niche.
- ➢ Cons: no diversity, large scale extinction possible, can't adapt to selection pressure.

Methods of Reproduction

Organism	Sexual or Asexual	Method of Reproduction	Advantages	Disadvantages
Animals	Can be either, budding, fragmentation & parthenogenesis may occur in select species, but usually sexual. (Focus on sexual here)	Internal fertilisation (eg. Mammals). Inside body.	-Increased chance of success & offspring survival, egg & sperm are close by and are protected from outside environment. -Water not required. -Fewer eggs needed (still lots of sperm).	-Sexually transmitted diseases can be spread. -Fewer offspring can be produced at a time. -High energy requirement.
		External fertilisation (eg. Frogs) Outside body in watery environment.	-Large amounts of gametes produced, potentially more offspring. -No mating rituals, simpler. -Smaller energy requirement -Gametes can travel further, rapidly colonise large areas.	-Lower success rate for fertilisation. -Watery environment required. -Gametes and zygotes not as protected. -Lots of gametes must be produced. -More factors eg. environment, timing. May use synchronised, cyclical gamete production & release.

Plants	Both. Some plants like ferns may even alternate (alternation of generations) between haploid gametophytes which produce diploid sporophytes via sexual reproduction, and the diploid sporophytes which produce haploid spores which grow into gametophytes.	Sexual Reproduction (eg. Flowering plants). Flowers = reproductive organs. Flowers may have both male & female parts, or have separate flowers. See diagrams below, essentially pollen=sperm, ovules=eggs. Pollination (pollen transferred to stigma) —> fertilisation (pollen moves down into ovary, fertilising ovule, ovary grows into fruit with seeds for new plants). May also reproduce via cones (male cones release pollen which fertilises the ovules inside the scales of the female cone)	-Creates genetic diversity, = higher disease resistance & greater ability to adapt to environment as a species.	-Recessive genes may not be passed on, can be detrimental if they are favourable. -Timing of pollen release must match maturity of female organs.
		Asexual Reproduction (Eg. Vegetative reproduction). Structural modifications to the roots or stem produces new individuals without seeds or spores.	-Offspring are clones and possess all favourable traits. Useful in farming. -Less energy intensive=rapid population increase & exploitation of environment. -No issue of timing flowering -Not reliant on external factors for pollination (wind, animals).	-No genetic diversity reduces evolution and adaptation to selection pressures. -Diseases easily passed from parents to offspring.

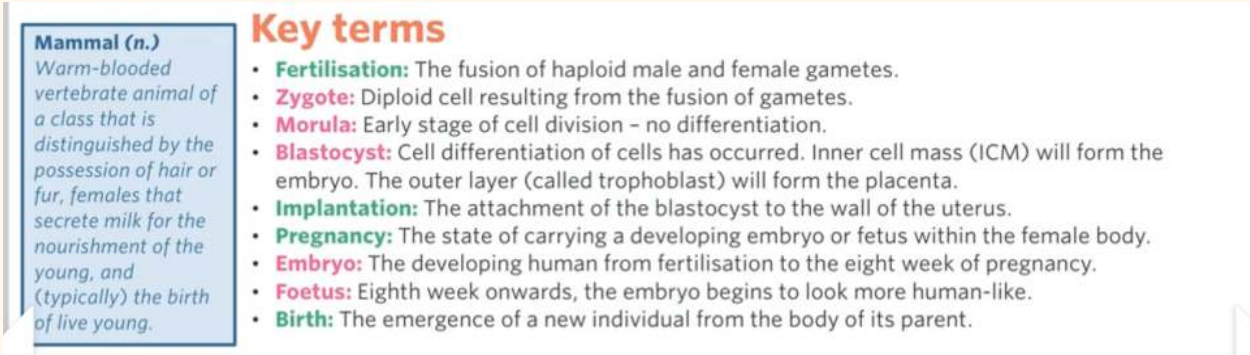
Fungi	Both. Multicellular fungi (eg. mushrooms, mould) use spores to reproduce sexually or asexually, fragmentation can also occur in the hyphae. Unicellular fungi (yeasts) reproduce both ways but usually asexually using budding or occasionally, binary fission.	Spores (eg. mushrooms & moulds). Multicellular fungi usually grow through a substrate as branching filaments (hyphae). Interwoven masses of hyphae form mycelia, and mushrooms are reproductive fruiting bodies which can release spores. Fungal cells are typically haploid, so during asexual reproduction haploid spores are produced via mitosis. These may be sporangiospores within a sac or conidia produced at the tips/sides of hyphae. Sexual reproduction via spores occurs when two fungi temporarily fuse, (plasmogamy =cells fuse, karyogamy = nuclei fuse) creating a diploid structure that produces spores by meiosis.	-Spores allow wide distribution in environment and large numbers. -Combination of sexual & asexual reproduction allows fungi to choose when/how to propagate. -Asexual reproduction is fast, low energy -Sexual reproduction increases genetic diversity.	-Somewhat dependent on environmental factors (eg. wind) -Asexual reproduction may produce offspring which are suited to only one location. -Sexual reproduction fusing of two fungi may take centuries in some species.
		Budding (eg. yeasts). Bud begins forming on side of cell, DNA is replicated, nucleus divides, cytokinesis splits cells and bud detaches from parent cell leaving	-Rapid population growth -Low energy requirement	-No genetic diversity.

		a bud scar on the parent and a birth scar on the daughter.		
Bacteria (prokaryotes including archaea)	Asexual	Binary fission (eg. *E. coli*.) Circular chromosome duplicates and moves to opposite ends of cell, cell elongates & cytokinesis occurs producing two identical daughter cells which are identical to the original parent. Mutations and horizontal gene transfer can occur which alter the genome.	-Very rapid -Only requires one organism to reproduce	-Low genetic diversity (however mutations and horizontal gene transfer in plasmids can offset this).
Protists (Protoctists)	Both. For haploid protists, 2 haploid cells fuse to form a zygote which undergoes meiosis, forming 4(?) new haploid cells. In diploid protists, mature cells undertake meiosis producing 4 gametes which fuse and form a diploid zygote —> diploid adult. Binary	Binary Fission: main form of asexual reproduction for protists. Like bacteria above, cell replicates DNA and grows, then divides into 2 cells	See bacteria above. Note that sexual reproduction may be advantageous for protists as it enables genetic diversity.	-Lower genetic diversity. Protists capable of switching to sexual reproduction may do that in adverse conditions to increase survival chances.
		Budding: new organism grows from the body of the parent organism, see fungi above.	See budding above. Rapid population growth makes protists well adapted for pathogenesis (causing disease in a host). Edrolo has good info!	See above.

	fission and budding are both asexual methods of reproduction .			

● analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals (ACSBL075)

We will focus on humans (placental mammals) but process vary between other mammals (eg. monotremes, marsupials) and timeframes also vary within placental mammals.



(Credit: Edrolo & Tim Sloane, 2018)

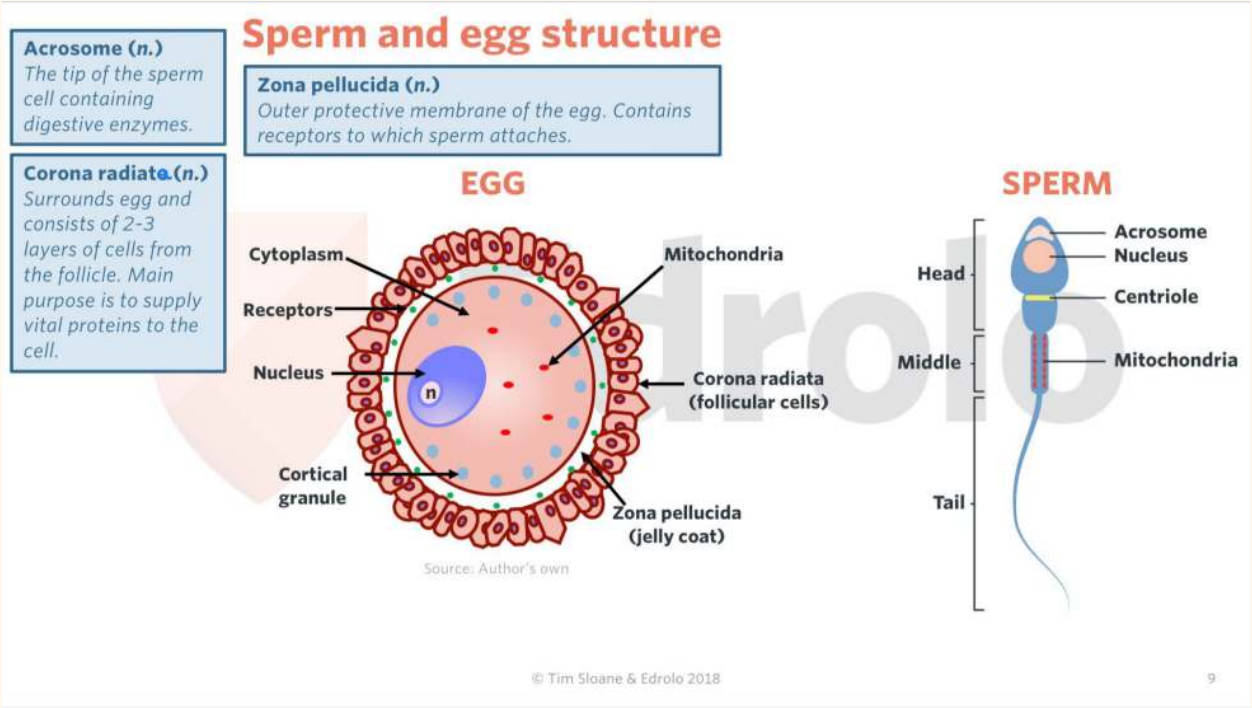
(Green are syllabus terms, pink are additional important terms.)

That fertilisation vid

Fertilisation (Mammals)

Fertilisation is the formation of a zygote in sexual reproduction by the fusing of two haploid gametes (egg & sperm). The zygote will develop into an embryo which will become a new

individual. Fertilisation creates variation which increases chances of species survival.



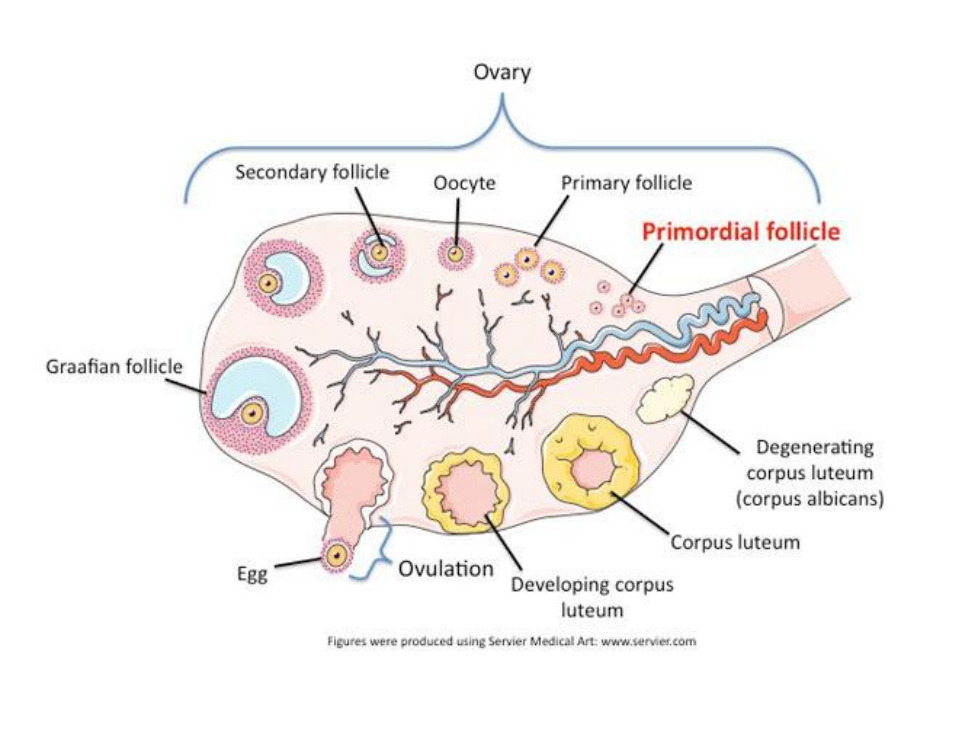
★ The female gamete, the egg (ovum) is a large and non motile cell. It contributes the organelles & cytoplasm to the zygote. The ovum is surrounded by a jelly coat, the zona pellucida, and a layer of cells from its follicle, the corona radiata. The zona pellucida serves as a barrier to sperm, and the corona radiata provides support and nourishment to the egg. Cortical granules are also present which prevent polyspermy (fertilisation by multiple sperm).

★ The male gamete, the sperm, is small, motile and only contributes its haploid nucleus and centrioles to the zygote. The sperm travels through the female reproductive tract to meet the egg and fertilise it in the Fallopian tube. The acrosome cap on the head of the sperm contains enzymes which dissolve the zona pellucida.

The Lead Up

➢ The ovarian cycle in females determines the release of ovum/eggs into the female reproductive tract. The cycle begins in puberty. Dormant follicles develop around maturing oocytes (they are actually called oocytes when still inside the ovary, ova when they enter fallopian tubes), enclosing them in a layer of follicular cells. In each roughly monthly cycle, a few follicles develop further until one becomes dominant. The most mature stage is the Graafian follicle, which then ruptures in ovulation, releasing the egg/ovum. The now empty follicle forms a corpus luteum which develops into a corpus

albicans (basically a scar). This involves all kinds of hormones, which we'll get to soon.



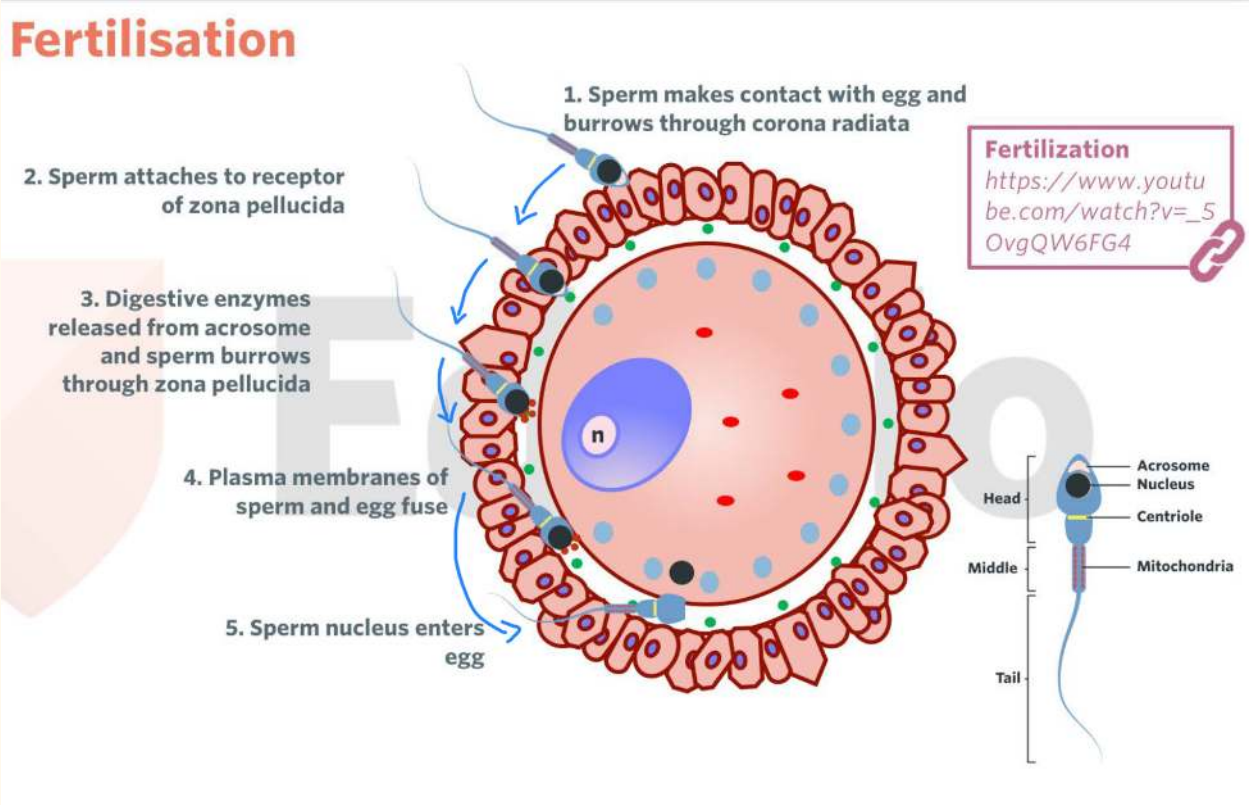
- ➢ The egg is swept down the fallopian tubes by tiny cilia towards the uterus.
- ➢ Meanwhile, if sexual intercourse occurs within around 24 hours of ovulation, the ~ 150 million (varies from about 40-500 million) sperm that are ejaculated enter the vagina and travel up. Many die travelling through the acidic environment, get trapped or attacked by immune cells, or choose the wrong fallopian tube. ~1000 reach the fallopian tube and approximately 200 reach the egg.

The Actual Fertilisation

- ➢ Sperm undergo capacitation in the fallopian tubes. Secretions from the uterine walls & tubes destabilise the sperm's membrane, making it more fluid to prepare for fertilisation and making the sperm hyperactive.
- ➢ Sperm burrow through the corona radiata and make contact with the zona pellucida, which has special receptor proteins which bind to those in the sperm, triggering the acrosome reaction.
- ➢ In the acrosome reaction, special digestive enzymes are released from the acrosome which digest a path through the zona pellucida.
- ➢ The first sperm to make contact with the plasma membrane of the egg fertilises it, as the two membranes fuse and the sperm's genetic material is drawn into the ovum.
- ➢ This fusing triggers mechanisms to prevent polyspermy, removing the ability of other sperm to enter the zona pellucida and fuse with the membrane. It also triggers meiosis II

in the female DNA, which has previously been halted. This forms the female pronucleus. The male pronucleus also forms, from the genetic material now in the ovum.

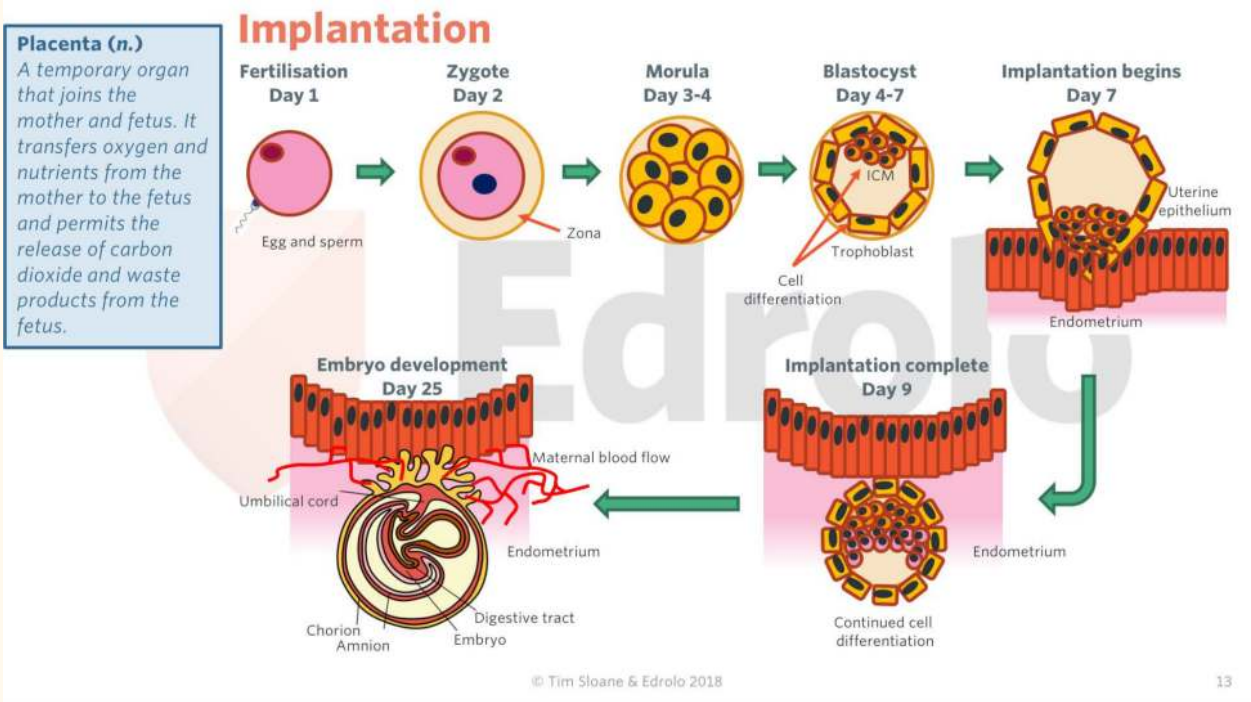
- ➢ The pronuclei combine and form the new diploid nucleus of the new individual.



(Credit: Tim Sloane, Edrolo 2018)

Implantation (Mammals)

- ➢ The zygote undergoes mitosis as it travels down to the uterus.
- ➢ While unspecialised, it is known as a morula.
- ➢ Once the Inner Cell Mass and trophoblast begin differentiating, the embryo is known as a blastocyst.
- ➢ Implantation begins at approximately day 7. At this point the endometrial lining has thickened and increased blood supply in preparation for implantation (prep known as decidualisation). If there is no implantation, the uterine lining sheds (menstruation).
- ➢ The blastocyst releases digestive enzymes that allow it to intrude down inside the uterine epithelium (outer layer) into the endometrium, at which point implantation has occurred (usually ~day 9).
- ➢ The trophoblast and the endometrium/some uterine tissue go on to form the placenta, and the inner cell mass develops into the foetus (foetus after 8 weeks).



Hormones in Pregnancy & Birth (Mammals)

Recap:

- ★ Hormone: chemical substance produced in the body which controls and regulates the activity of certain organs or cells, secreted by glands comprising the endocrine system.
- ★ Mammals have different breeding cycles. Some animals are seasonal breeders and mating occurs only in periods of female fertility when the animal is "in heat" (oestrus). Other oestrus cycles occur more frequently and so those animals are continuous breeders.
- ★ Negative feedback loops: try to return to stable state (like an equilibrium reaction!)
- ★ Positive feedback loops: amplify the change, make it happen faster until it's done.
- ★ Estrogen / Oestrogen exists in the forms estradiol (normal reproductive cycle, most potent) estriol (during pregnancy) and estrone (menopause and after).

Hormones Involved in Pregnancy

Hormones are involved in the male reproductive system to regulate spermatogenesis (sperm production) and in the female reproductive system to regulate the menstrual and ovarian cycles.

Hormone	Male/ Female	Secreted By	Role
GnRH (Gonadotropin -releasing hormone)	M & F	Hypothalamus	Causes release of FSH and LH. This sets off a negative feedback loop for sperm production.
FSH (follicle stimulating hormone)	M	Anterior pituitary	Enters testes, stimulates sperm production (spermatogenesis) in Sertoli cells
	F	Anterior pituitary	In follicular phase, stimulates growth of follicles in the ovaries. In ovulation phase, stimulates maturation of one follicle
LH (luteinising hormone)	M	Anterior pituitary	Enters testes, stimulates interstitial Leydig cells to produce testosterone
	F	Anterior pituitary	In follicular phase, causes growth of follicles, and maturation of one follicle during ovulation. Actual ovulation occurs due to a spike in LH. Also stimulates growth of a corpus luteum
Testosterone	M	Leydig cells in testes	Triggers development of secondary sex characteristics, ie signs of puberty in males. Also stimulates spermatogenesis. Additionally, inhibits secretion of GnRH by hypothalamus and LH & FSH by pituitary, stabilising sperm and testosterone production.
Inhibin	M	Sertoli cells (testes)	Released when sperm count is too high, inhibits production of GnRH & FSH by hypothalamus and pituitary. Slows spermatogenesis.
	F	Follicle cells	Inhibits FSH production.
Estrogen	F	Follicles	Female sex hormone, causes eggs to mature in puberty and triggers secondary sexual characteristics in females. Also assists with thickening endometrium. During follicular phase low levels are secreted by the follicles, inhibiting GnRH to keep FSH & LH low. High levels are secreted by the follicles before ovulation, stimulating GnRH and causing FSH and LH especially to rise, which triggers ovulation. During luteal phase secreted with progesterone by the corpus luteum, inhibiting

			GnRH, LH, FSH, until corpus luteum degrades.
Progesterone	F	Corpus Luteum (follicle)	Stabilises endometrium. Together with estrogen, inhibits GnRH, FSH and LH in luteal phase

Feedback Loops & Graphs

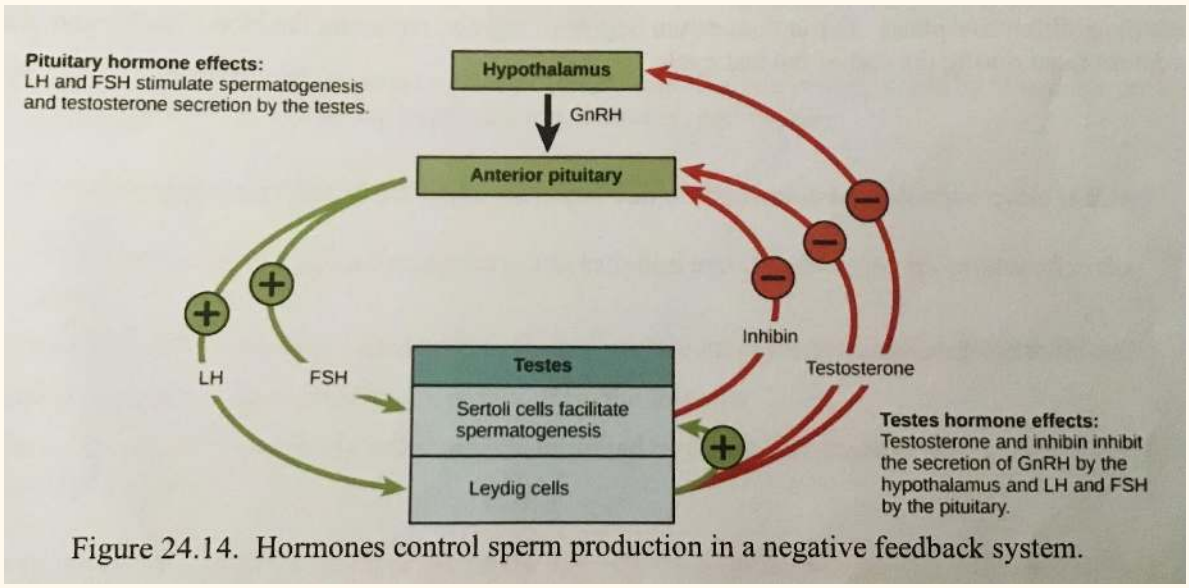


Figure 24.14. Hormones control sperm production in a negative feedback system.

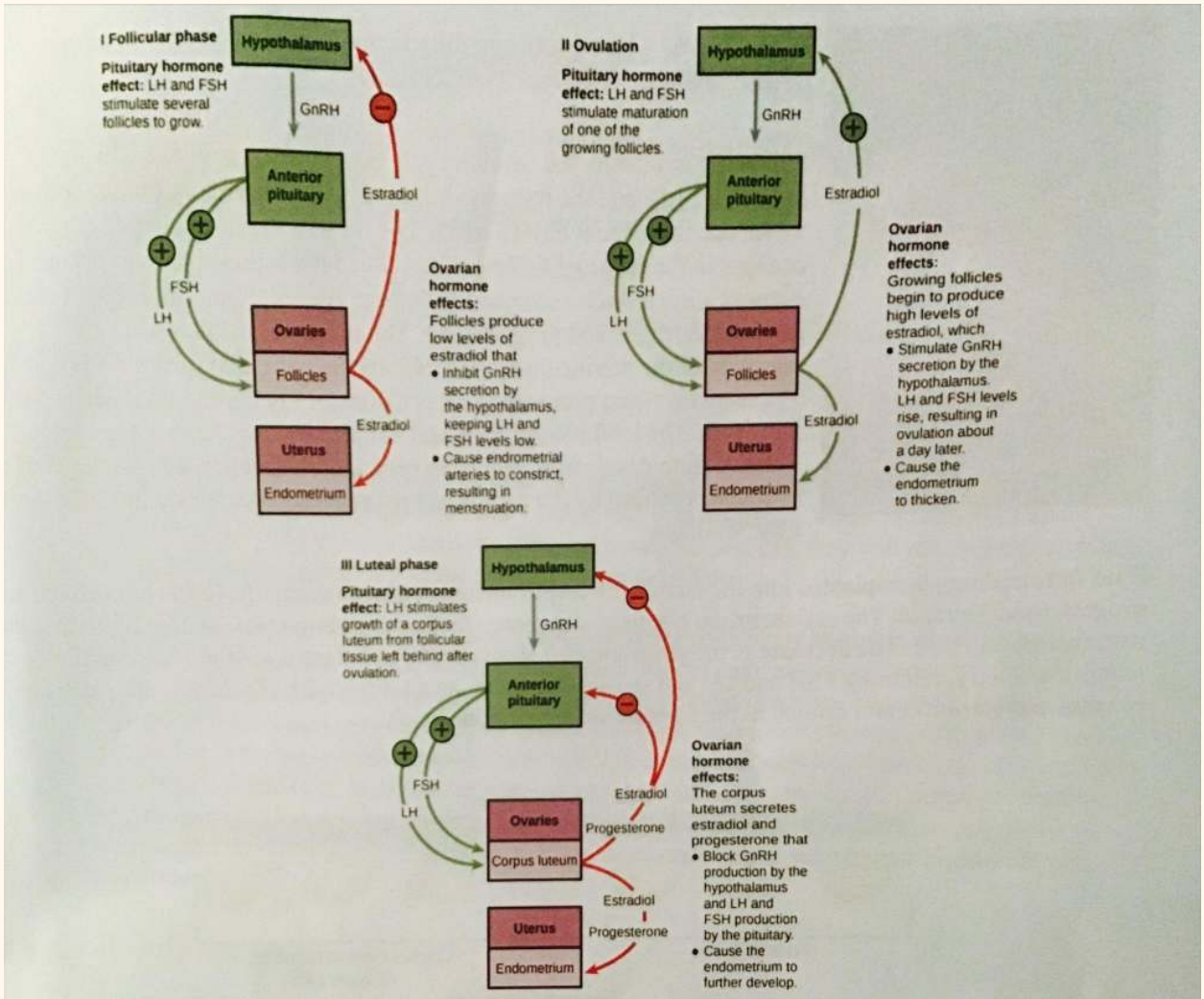


Figure 24.15. The ovarian and menstrual cycles of female reproduction are regulated by hormones produced by the hypothalamus, pituitary, and ovaries.

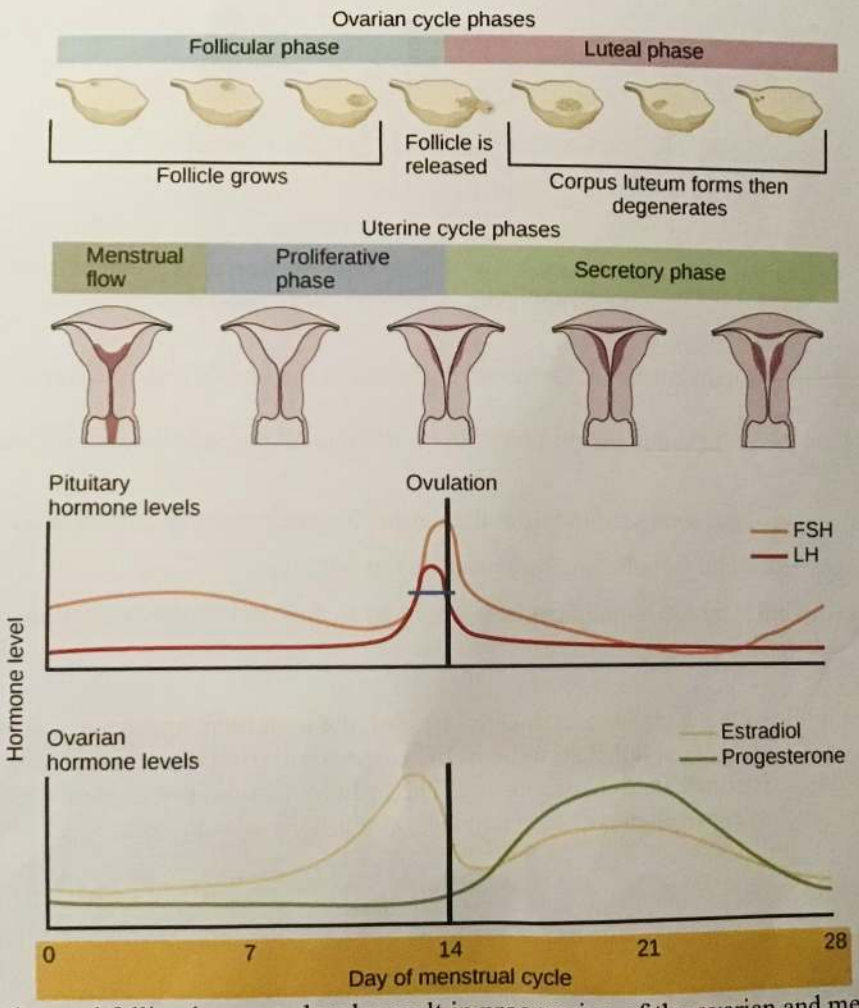
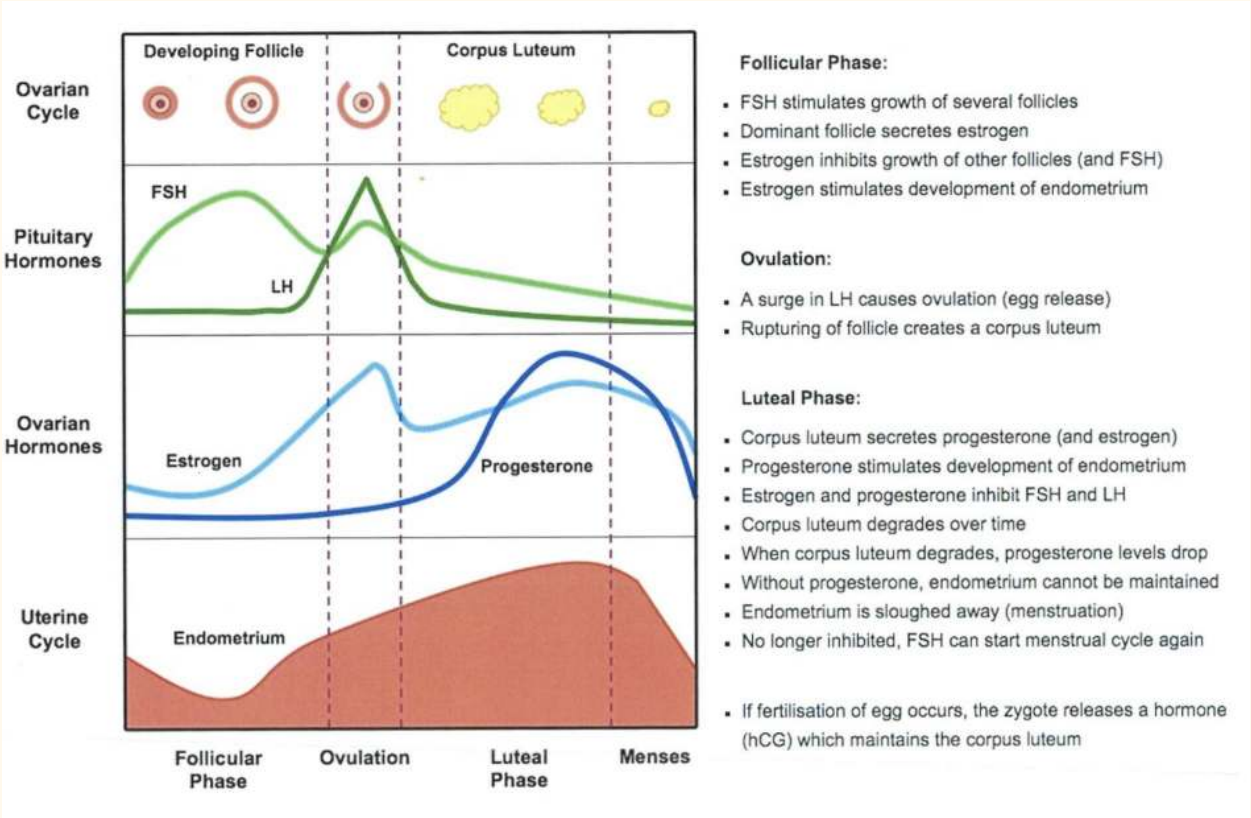


Figure 24.17. Rising and falling hormone levels result in progression of the ovarian and menstrual cycles. (credit: modification of work by Mikael Häggström)



During pregnancy, the embryo begins releasing human chorionic gonadotropin (hCG) once it has implanted in the endometrium. This maintains the corpus luteum so it can continue secreting progesterone and estrogen, which will maintain the endometrial lining. If the corpus luteum degrades like in a normal cycle, progesterone levels drop and the endometrium is shedded (menstruation). This continues for 12 weeks, at which the placenta takes over, producing hCG along with progesterone and estrogen to maintain the endometrium and also inhibit uterine contractions and prepare breast tissue for milk production respectively. Progesterone and estrogen also inhibit GnRH, FSH and LH production, preventing ovulation.

“Complex interactions of hormones control the ovarian and menstrual cycles in women, which occur at the same time and typically take 28 days. The first half of the ovarian cycle is the follicular phase. During this time, one follicle fully matures. The follicle releases estrogen, which stimulates the endometrium to thicken. In the middle of the cycle, an egg bursts out of the follicle. This is called ovulation. The second half of the cycle is called the luteal phase. The remnants of the burst follicle form the corpus luteum, which releases progesterone and estrogen. These hormones cause the endometrium to thicken and stabilise. At the end of the cycle, the corpus luteum disintegrates. This causes a decrease in progesterone and estrogen, triggering menstruation.”

(Credit: Some worksheet I did in class, probably Surfing Biology?)

Birth

Oestrogen levels continue to rise, progesterone plateaus and drops around the seventh month. Relaxin is produced by the placenta to help relax the pelvis and dilate the cervix. How exactly labour is triggered is not known, but prostaglandins secreted by the uterus play a role, and make the uterine tissue more sensitive to oxytocin, the hormone which promotes contractions. A positive feedback loop occurs during labour as the baby pushes on the cervix, sending signals to the pituitary to release more oxytocin, which causes contractions and pushes the baby further. During pregnancy and birth milk production begins in mammals. Prolactin stimulates milk production and oxytocin stimulates milk release.

• evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture (ACSBL074)

Reproductive technologies used in agriculture include

- Selective breeding (sexual)
- Artificial insemination (sexual)
- Vegetative propagation (asexual)
- Embryonic transfer (sexual)

And others such as artificial pollination and genetic engineering

Reproductive Technique	**Scientific Knowledge about Reproduction**	**How is Reproduction Manipulated? Example**	**Impact of Knowledge, Advantages/Disadvantages, Ethical Concerns**
Vegetative Propagation	Understanding of the ways plants reproduce asexually has led to wide use of plant cloning. Knowledge of plant regeneration following damage has	Plant stems can be deliberately cut and treated with hormones to stimulate root development. This creates a clone of the original plant.	All vegetative propagation methods contribute to a lack of biodiversity/genetic diversity in the long term (because they are asexual). Large numbers of plants

	led to deliberate cutting of plant tissues. Knowledge of plant tissues and their function has allowed for grafting.	Plants can be grafted onto one another by cutting and fusing the stems of two plants. Eg. Grafting stone fruit.	with desirable and predictable characteristics can be cultivated.
Selective Breeding	Organisms' phenotypes and characteristics are a result of allele combinations inherited from both parents (in sexual reproduction). Selectively breeding organisms with desired traits will result in the offspring having a higher chance of expressing the desired phenotypes . An understanding of types of dominance (Mendel, complete, incomplete and codominance) is needed to manipulate selective breeding effectively.	Involves the selection and breeding of plants/animals with desirable characteristics. Increases chances of offspring possessing these desirable characteristics. Eg. Braford cow is a combo of maternal instincts of the Hereford and bulk of the Brahman.	Reduces genetic diversity in the population, breeding individuals with similar genes results in similar genes being passed down. Diseases and weaknesses/susceptibility are likely to be passed down and spread through the population. Inbreeding can result in genetic conditions arising from recessive gene combinations.
Artificial Insemination	Understanding of the reproductive cycle, hormones and hormone control, in order to successfully inseminate female animals when the ovum is ready for fertilisation. Understanding of reproductive anatomy in order to	Involves the control of an animal's hormones, stopping females' cycles and beginning them again synthetically. Involves the collection of animal sperm and depositing it in the female's reproductive tract via the intervention of people. Results in	Advantages - control venereal (sexually transmitted) disease, especially in cattle. - No. of sires needed is reduced - Increases genetic

	ensure fertilisation occurs without a male present. Understanding of how to keep sperm in semen alive and unharmed until the time of AI.	offspring with desirable traits, without needing a male actually present. useful for animals, such as livestock. Each female can be impregnated via AI with offspring that are as genetically desirable as possible.	diversity in areas where males of a certain species may not be plentiful - Genetically desirable offspring can be produced easily. Disadvantages - expensive, time intensive - Lower chance of successful fertilisation.
Artificial Pollination	Knowledge of plant sexual reproduction and plant anatomy.	Pollen (sperm) removed from stamen of one plant. Pollen applied to stigma of another plant. Pollen fertilises ovum.	Advantages - Crossbreeding of favourable traits - Self-pollination ie. creation of genetically similar offspring - Ensures successful pollination of all plants, which translates to s high crop yields. Disadvantages - Time and labour intensive. - Expensive

Cell Replication

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

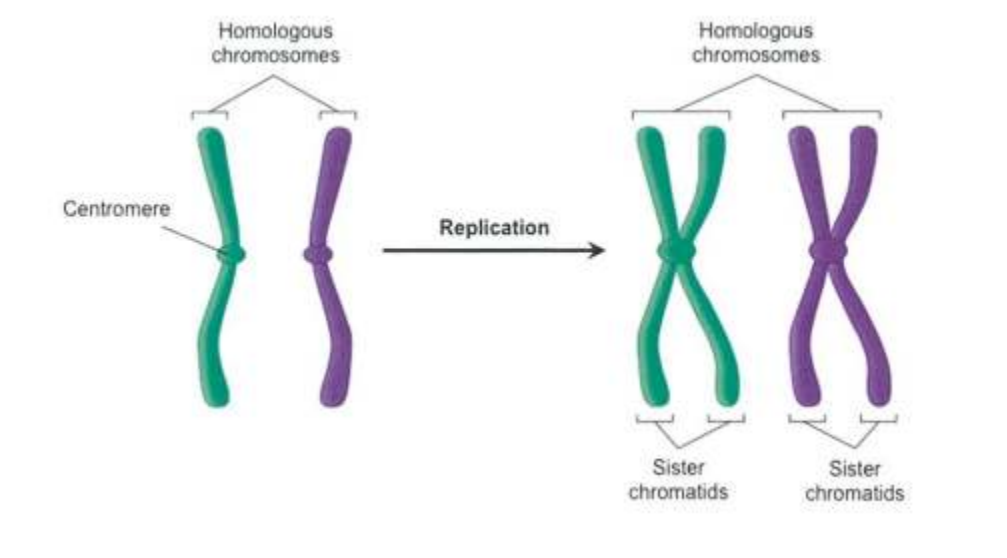
Students:

● model the processes involved in cell replication, including but not limited to:

– mitosis and meiosis (ACSBL075)

Terminology

★ Chromosomes are usually counted by their centromeres. Sister chromatids are attached via a centromere.



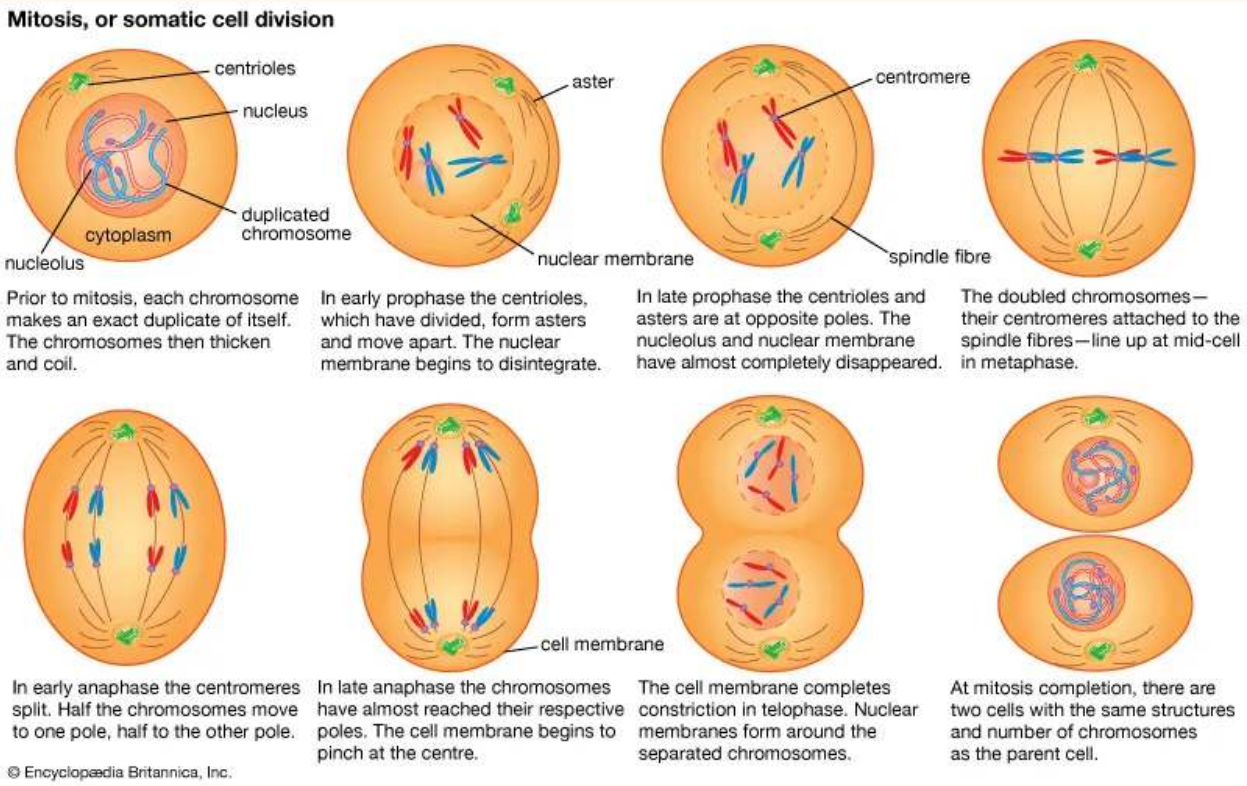
Mitosis

➢ In multicellular organisms, cell division is known as mitosis (unicellular organisms use binary fission).
➢ Mitosis creates two diploid, genetically identical daughter cells.
➢ Role and importance of mitosis includes
  ○ Growth (multicellular organisms)
  ○ Repair (of damaged tissue and worn out cells)
  ○ Asexual reproduction (eg. plant propagation)
  ○ Genetic stability - exact replication of DNA, ensures equal chromosome distribution.

Cell Cycle

- Cell division and growth occurs in a repetitive cycle, the cell cycle.
- Most time is spent in the preparatory stage, interphase.
    - G_1 Phase is the beginning of the cell cycle. Cell grows, everything except chromosomes are replicated.
    - S Phase - synthesis. All 46 chromosomes are duplicated.
    - G_2 Phase - proofreading. Cell checks for duplication errors, fixes mistakes.
- M or mitosis occurs. The nucleus divides.
- Cytokinesis - the cytoplasm splits. Two new cells are formed.
- G_0 Phase - cell cycle stops and cell returns to interphase.

Stages of Mitosis



- (Chromosomes replicate during interphase. They thicken and coil). Remember PMAT, or mitosis song.
- P - Prophase = Poles
    - Centrioles (divided in interphase) form spindle fibres and move to opposite poles. The nuclear membrane disintegrates.
- M - Metaphase = Middle

    - Chromosomes line up across the middle of the cell. Spindle fibres attach to the centromeres.
- A - Anaphase = Apart
    - Centromeres split and sister chromatids are pulled apart. Half move to each pole (now separate they are chromosomes) and the cell membrane begins to pinch at the centre.
- T - Telophase = Two
    - Two nuclear membranes are formed around the identical daughter nuclei. Spindle breaks down. Mitosis is complete.
- C - Cytokinesis = Cytoplasm chopped
    - Cytoplasm divides, separating the two nuclei into individual cells. In animals this occurs as the cell membrane constricts, in plants a cell plate is formed between the nuclei which becomes a cell wall once cellulose is deposited there. Occurs as anaphase and telophase do.

Meiosis

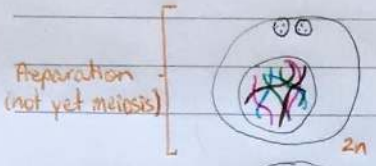
- Form of cell division occurring only in the gametes (sperm and eggs).
- Creates 4 genetically unique haploid daughter cells after two cycles (Meiosis I & II).
- Role and importance is in sexual reproduction:
    - Creates haploid gametes for sexual reproduction, ensuring chromosome number of the species is maintained.
    - Is a source of genetic variation which increases species genetic diversity and overall chance of survival.

Stages of Meiosis

Meiosis

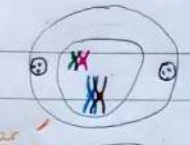
DATE M.6-Q.2-D.1.1.

- Cell division resulting in four haploid daughter cells (gametes), which are genetically unique.

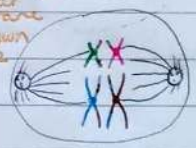


Interphase - the stage where the cell grows and replicates its DNA. Each chromosome duplicates, however remain attached to a single centromere so 46 chromosomes (humans) → still 46 chromosomes. Centrosomes also replicate.

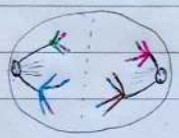
NOTES



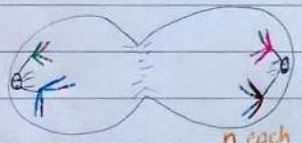
Prophase I - Chromosomes condense and homologous chromosomes form pairs. Crossing over occurs here. When chromatids of the tetrad touch at the chiasmata, they exchange genetic material between the maternal and paternal chromosomes, forming recombinant chromosomes. This ensures not all linked genes are inherited together. Centrosomes move to opposite poles.



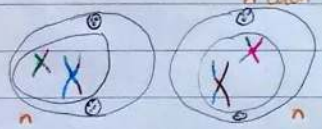
Metaphase I - Homologous pairs line up at the cell equator. Meiotic spindle extends from either pole to the chromosome centromeres.



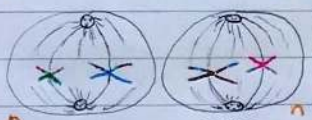
Anaphase I - Homologous pairs are separated, pulled apart to opposite poles by the meiotic spindle as the microtubules contract. Sister chromatids remain attached at the centromere.



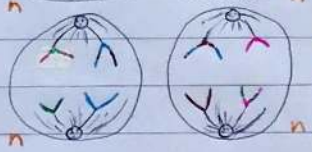
Telophase I (and cytokinesis) - chromosomes arrive at opposite ends of the cell, which pinches in the middle. Cytoplasm divides, two haploid daughter cells (with 2 sister chromatids per chromosome) are formed. Spindle disappears.



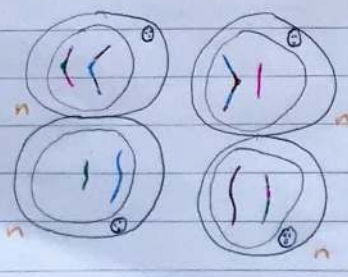
Prophase II - chromosomes condense in the two daughter cells. No crossing over. Chromosomes may not have genetically identical sister chromatids due to the previous crossing over. Spindle fibres form at right angles to previous spindle. Nuclear membrane disintegrates.



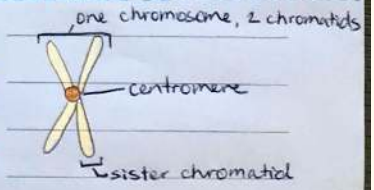
Metaphase II - chromosomes line up at the equator. Meiotic spindle extends to centromeres from poles.



Anaphase II - centromeres separate, sister chromatids pulled apart to opposite poles. Chromatids now act as individual chromosomes.



Telophase II (and cytokinesis) - chromosomes reach poles, spindle disappears. Nuclear membranes form, cytokinesis splits cytoplasm, four haploid, genetically unique daughter cells form.



Centrosome: organelle containing the centrioles which are made up of the microtubules which arrange the meiotic / mitotic spindle.

Homologous chromosomes: chromosomes of the same size and shape with the same genes in the same places. Alleles may differ.

Tetrad: four chromatids placed together when homologous pairs line up.

Chiasmata (pl.): the point of contact between two chromatids crossing over.

Meiotic Spindle: spindle apparatus that forms in meiosis from spindle fibres.

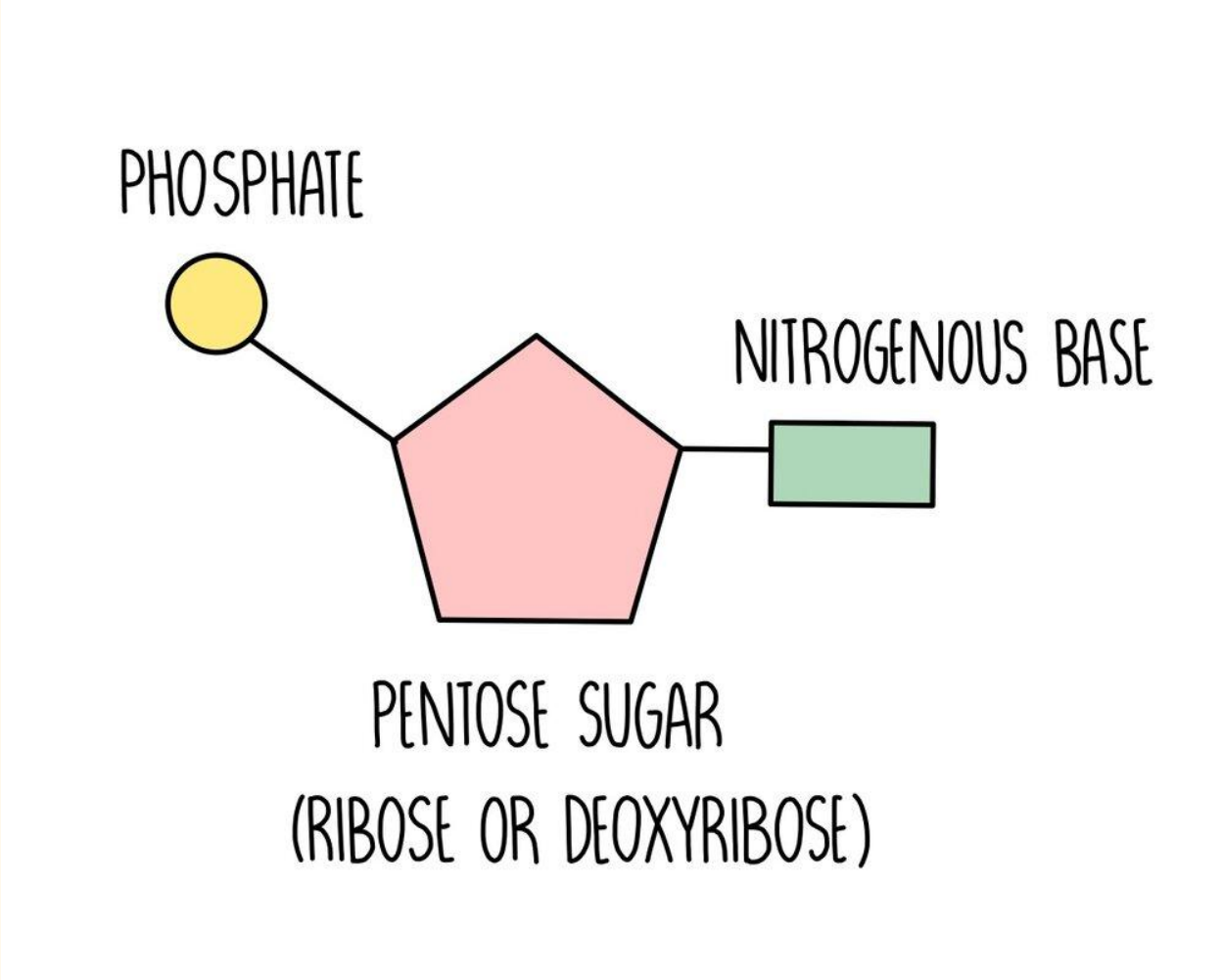
meeting pad

- DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding (ACSBL076, ACSBL077)

DNA and Components

- ➢ DNA (deoxyribonucleic acid) is a double helical nucleic acid molecule which carries genetic information in nearly all living organisms. It is comprised of two nucleotide chains which twist around each other, held together by weak hydrogen bonds.
- ➢ Larger structure is coils, chromatin, chromosomes (further detail below).
- ➢ Nucleotides, the monomers or basic building blocks of nucleic acids are made up of 3 components:
    - Deoxyribose, a 5 carbon sugar
        - We count the carbons as 1', 2', 3' etc.
    - Phosphate group
        - "The phosphate group gives DNA its negative polarity and acidic characteristics (hence the name nucleic acid), especially as it is **hydrophilic** and surrounds the **hydrophobic** bases on either side (Hughes, 2005; Nature Education, 2012)."
        - (Credit here is me, that was an excerpt from one of my own assignments)
    - One of 4 nitrogenous bases
- ➢ Nucleotides can be joined in any order (only with complementary base pairing on the opposite strand).

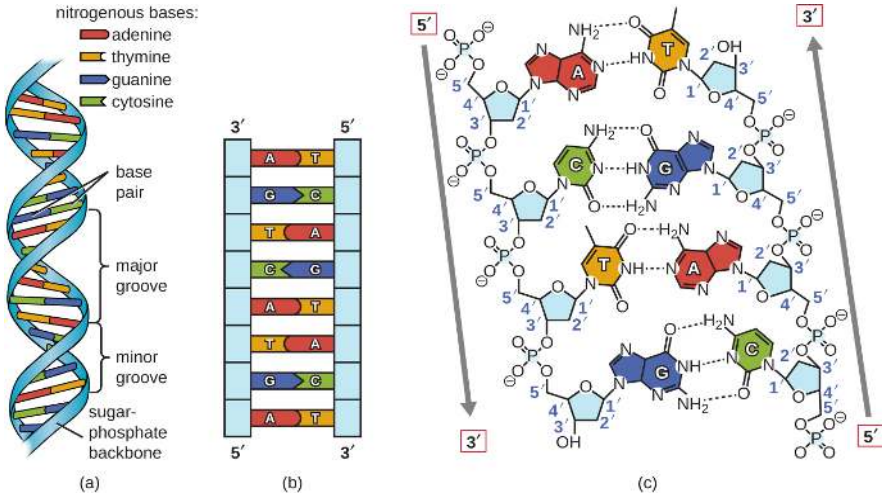
Typically nucleotides are represented like this:



Bases

- Adenine and thymine always pair together, and guanine and cytosine always pair together. (GCAT)
- From assessment 1: "The nitrogenous bases that form the "rungs" of the DNA double helix are adenine, thymine, guanine and cytosine which can be divided into purines or pyrimidines based on structure. Purines adenine and guanine are made up of two carbon-nitrogen rings. Cytosine and thymine are pyrimidines, consisting of only one carbon-nitrogen ring (Leacock, 2022). Uracil is another pyrimidine, replacing thymine in **RNA** (Leacock, 2022)."
- The equal ratios of A:T and G:C are known as Chargaff's Rules.

- This can be explained by the Watson-Crick double helix model. The hydrogen bonds in the centre holding the two strands together only form perfectly between A & T or C & G due to their chemical structure.
- James Watson and Francis Crick are attributed with the discovery of the double helix model, after they used Rosalind Franklin's x-ray diffraction patterns of DNA to build a model which explained DNA's characteristics. The two antiparallel strands which wrap around like a twisted ladder match Franklin's x-ray patterns and allow for Chargaff's rules via complementary base pairing due to the hydrogen bonding.



DNA Replication

- DNA replication is a semi conservative process where the original DNA section unwinds and the two resulting strands are used as templates for two identical double stranded DNA sequences to form. This is done in interphase prior to cell division.

In full form from Assessment 1, "Replication begins with initiation at the origin of replication, a series of nucleotides which initiator proteins bind to. This allows a helicase enzyme to start breaking the hydrogen bonds between the bases and unwind the helices, creating the replication fork (Biology Wise, 2009). Topoisomerase enzymes work ahead of the replication fork, unwinding and ensuring the DNA does not supercoil and tangle (Bailey, 2019; Chidrawi et al., 2018). Single-strand binding proteins bind to the now unbound nucleotides, preventing them from rejoining (*Chapter 9*, n.d.).

Next, RNA primers of 9-12 nucleotides made by primase are placed along each strand (Biology Wise, 2009). This allows DNA polymerase III to insert free floating complementary nucleotides onto the 3' end of the primer, as DNA can only be added in the 5' to 3' direction (Biology Wise,

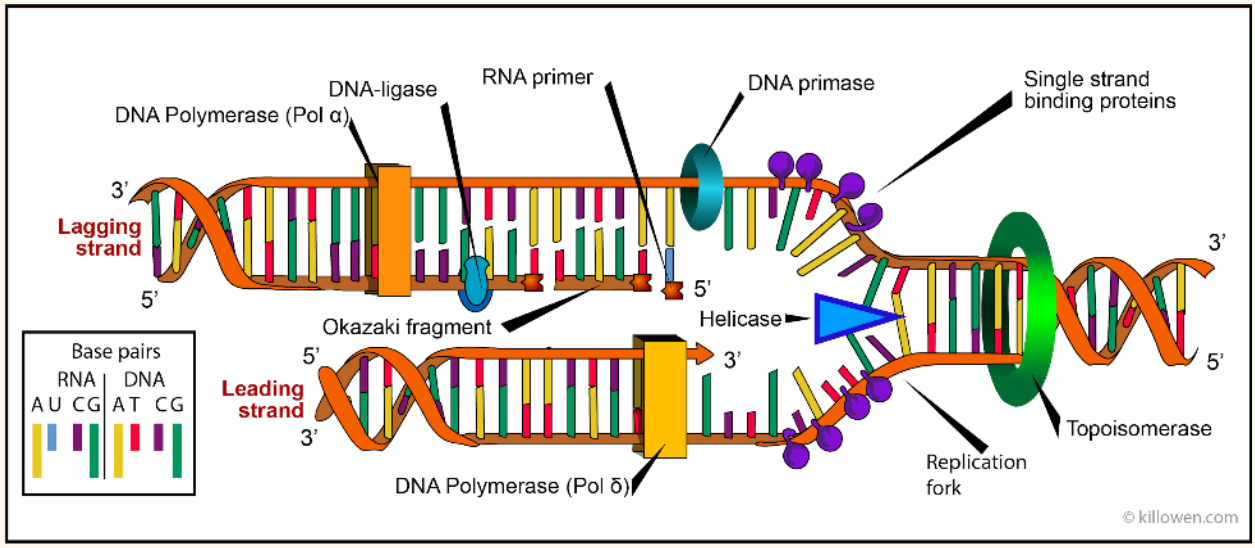
2009; Chidrawi et al., 2018). Due to the antiparallel nature of DNA, this creates a leading strand, which is the original 3' to 5' strand (towards the replication fork) and a lagging strand, oriented 5' to 3' (Bailey, 2019). Consequently, DNA can be synthesised continuously in a 5' to 3' direction along the leading strand, towards the replication fork, maintaining the antiparallel structure (Biology Wise, 2009). However, on the lagging strand, nucleotides must be added discontinuously, away from the replication fork (Biology Wise, 2009). Primase must continuously add primers closer to the replication fork so that DNA polymerase III can continue adding nucleotides as the strand is separated. This creates short, unconnected fragments of DNA, known as Okazaki fragments (Biology Wise, 2009).

The enzyme ligase joins Okazaki fragments and repairs phosphodiester bonds (Biology Wise, 2009). DNA polymerase I has an **exonuclease** and editing function to proofread bases and remove RNA primers from the lagging strand, replacing them with the correct nucleotides (Biology Wise, 2009; Chidrawi et al., 2018). DNA polymerase II also assists with editing of the lagging strand (*Chapter 9*, n.d.). This process continues until the chromosome has been duplicated or tus proteins terminate the replication, resulting in two identical DNA strands, each with one original and one new strand (Biology Wise, 2009). "

But in summary;

- DNA double helix is unwound and unzipped by the helicase enzyme at the origin of replication. This creates the replication fork.
- Topoisomerase enzymes work ahead of the fork, organising the DNA to stop tangling and supercoiling.
- Single-strand binding (SSB) proteins bind to the exposed nucleotides, preventing them from rejoining.
- The primase enzyme puts short RNA primers on the DNA strand.
- The DNA polymerase III enzyme inserts free floating complementary nucleotides next to the primer on the 3' end. This can only occur in the 5' to 3' direction.
- The original 3' to 5' strand becomes the leading strand, with continuous replication towards the fork.
- The original 5' to 3' strand becomes the lagging strand with discontinuous replication as it occurs away from the fork and must keep skipping forward, with primers being continually replaced. This forms Okazaki fragments.

- ➢ The enzyme ligase joins the Okazaki fragments, forming a complete strand.
- ➢ DNA polymerase I and II proofread and fix the lagging strand and mistakes.



• assess the effect of the cell replication processes on the continuity of species (ACSBL084)

DNA Replication

- ➢ Replication ensures genetic material is passed on to future generations.
- ➢ Without it, cells would have half the genetic information of the parent cell and eventually die due to this.
- ➢ Ensures fidelity of replication, and correct gene expression for the daughter cells or offspring.
- ➢ DNA replication is necessary for cell division.

Mitosis

- ➢ Essential for growth and development for multicellular organisms, as well as tissue repair and maintenance.
- ➢ Allows sexually reproductive organisms to grow and reach sexual maturity, eg. Humans, so we can pass on our genetics.
- ➢ In asexually reproducing organisms, mitosis creates the offspring so the species can continue.

Meiosis

- ➢ Forms gametes which create new offspring that have inherited traits from both parents.

- ➢ Allows sexual reproduction to occur and maintain chromosome numbers.
- ➢ Introduces variation through crossing over, independent assortment and random segregation (more on that later).
- ➢ Variation, along with mutation, creates genetic diversity which is important for evolution, survival and species continuity.

DNA and Polypeptide Synthesis

Inquiry question: Why is polypeptide synthesis important?

Students:

• construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076)

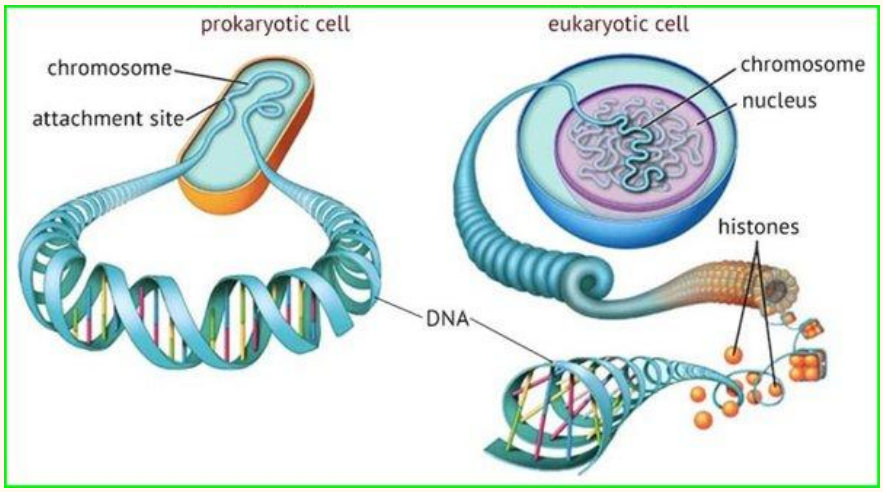
Eukaryotes

- ➢ DNA in eukaryotes is found in linear chromosomes inside the nucleus.
- ➢ From Assessment 1, "DNA in eukaryotic cells is found in the **nucleus**, within linear **chromosomes** which are made up of tight, supercoiled **chromatin** (*DNA- Is a Structure That Encodes Biological Information | Learn Science at Scitable*, n.d.). The chromatin itself is composed of **nucleosomes**, linked units of DNA strands wrapped around eight **histone** proteins (Annunziato, 2008). The tight coiling occurs due to DNA's negative **polarity**, and the positive charge of the histone proteins (Hughes, 2005)."
- ➢ DNA -> wrapped around histones = nucleosome -> coils-> supercoils -> chromosomes.
- ➢ Eukaryotic cells may also possess DNA within mitochondria or chloroplasts. Mitochondrial DNA (mtDNA) is used to study evolutionary relatedness as it comes from the mother and has a higher rate of mutation.
- ➢ Eukaryotes tend to have larger genomes than prokaryotes, including non-coding and repetitive DNA sequences.

Prokaryotes

- ➢ Prokaryotes possess free floating, singular circular chromosomes, as well as plasmids.

- From Assessment 1: "In prokaryotic cells, DNA typically forms one circular chromosome, however is not typically bound in nucleosomes (Griswold, 2008). Supercoiling with the assistance of proteins such as the HU protein allows the prokaryotic genomes to be stored compactly within the cell's **nucleoid** (Griswold, 2008)"
- Prokaryotic DNA is not membrane bound but floats in the cytoplasm in the cell's nucleoid region. Plasmids, small circular independent DNA molecules may also be present, which can be transferred between organisms.
- Prokaryotic genomes are smaller and compact in comparison to eukaryotic DNA, with little repetition.



• model the process of polypeptide synthesis, including: (ACSBL079)

– transcription and translation

Polypeptide/Protein Synthesis

- Polypeptide synthesis is the two-stage process used to turn genetic information, ie. DNA, into structural and functional molecules used inside cells (proteins).
- Reminder: amino acids make up peptide chains which make up polypeptides which make up proteins.
- Amino acids = simple organic compounds containing a carboxyl (—COOH) and an amino (—NH2) group. There are 20 (found in our genetic code).
- Gene = basic unit of heredity; consists of a segment of DNA with nucleotide bases in a specific sequence which forms a specific section of a chromosome. They code for a functional molecule. The median human gene size is 24 kilo base pairs).

★ Gene expression: ‘expression’ = whether a gene is active or ‘turned on’, or making the desired protein or not. Gene expression can also refer to the protein synthesis process.

Proteins have various functions within organisms (this will be discussed further below), but they must be produced first from genes. The process from gene to protein passes through these phases:

➔ Gene
  ◆ Segment of DNA containing the base sequence for the synthesis of a polypeptide.
➔ Amino acid
  ◆ Each codon (triplet of bases) codes for a specific amino acid which is a monomer (building block) of the polypeptide chain.
➔ Polypeptide chain
  ◆ Amino acids are joined by peptide bonds. A long chain of amino acids creates a polypeptide.
➔ Protein
  ◆ One or more polypeptides may be coiled, folded or cross-linked to form a protein.

RNA

➢ RNA (Ribonucleic acid) is required for amino acids to be made from genes.
➢ It is a single stranded nucleic acid containing a ribose sugar (in place of deoxyribose) and uses the nitrogen base uracil (U) to pair with adenine (A) instead of thymine (T) in DNA.
➢ Messenger (mRNA), ribosomal (rRNA) and transfer (tRNA) RNAs are used in polypeptide production.

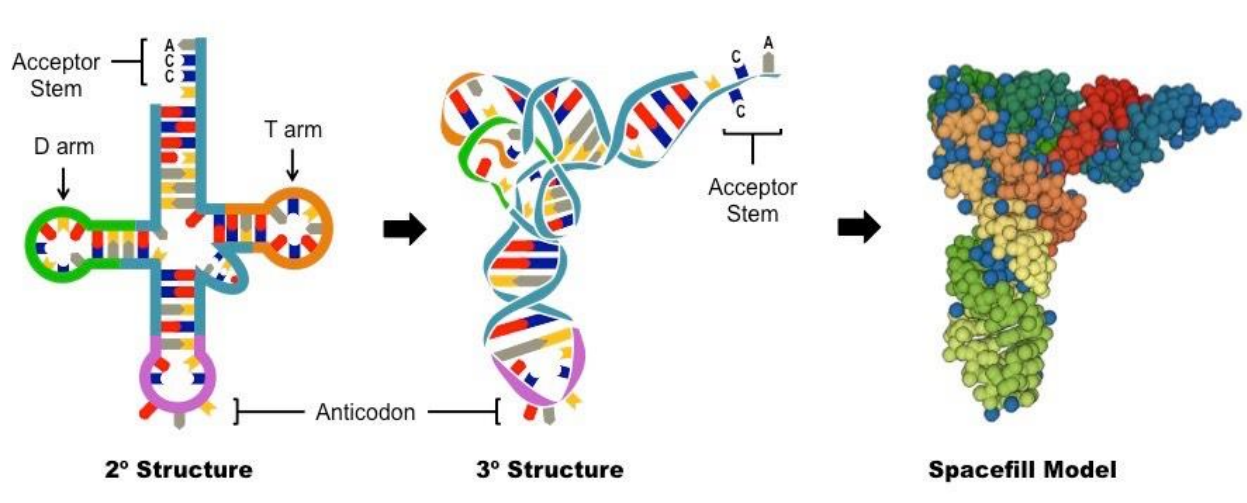
Transcription

➢ First stage; the section of DNA containing the gene is copied onto an mRNA strand.
➢ Steps:
  ○ RNA polymerase binds to the promoter sequence on the DNA strand, upstream of the desired gene. The two DNA strands ‘unzip’ and separate (only along the length of the gene).
  ○ The RNA polymerase moves along the DNA, reading the template/non coding/antisense strand and matching free floating complementary RNA nucleotides to create an mRNA strand. This mRNA has the same sequence as the coding/sense strand of DNA (except U replaces T).
  ○ Terminator sequences end the transcription and the mRNA (or pre-mRNA) is released.

- The mRNA will be modified and processed, including addition of molecules to the ends of the strand to protect against degradation and splicing (introns [non coding segments] are removed to form a mature mRNA strand). Splicing increases the variability of the information that can be expressed from one gene, as alternative splicing produces similar but not identical proteins.
- The mRNA exits the nucleus via the nuclear pores where it will find a ribosome in the cytoplasm (or in the endoplasmic reticulum).

Translation

- The second stage in protein synthesis, codons (in triplet code) on the mRNA strand are translated into amino acids, which link together to form a polypeptide.
- tRNA (transfer RNA) molecules are clover-shaped twisted molecules of RNA, 75 nucleotides long. At one end they possess three unpaired bases, forming an anticodon which will attach to the mRNA codon. The anticodon determines the amino acid that the tRNA will be able to temporarily bind to on its other end (acceptor stem).



- Codons on the mRNA correspond to amino acids, which can be determined using a codon chart. There are multiple possible codons for most amino acids. Each coding sequence will begin with AUG (start/methionine) and end with one of three stop codons

(UAA, UAG or UGA).

RNA codon table

1st position	2nd position U	2nd position C	2nd position A	2nd position G	3rd position
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	stop	stop	A
	Leu	Ser	stop	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G

Amino Acids

Ala: Alanine
Arg: Arginine
Asn: Asparagine
Asp:Aspartic acid
Cys:Cysteine
Gln: Glutamine
Glu: Glutamic acid
Gly: Glycine
His: Histidine
Ile: Isoleucine
Leu: Leucine
Lys: Lysine
Met: Methionine
Phe: Phenylalanine
Pro: Proline
Ser: Serine
Thr: Threonine
Trp: Tryptophane
Tyr: Tyrosisne
Val: Valine

➢ Steps:

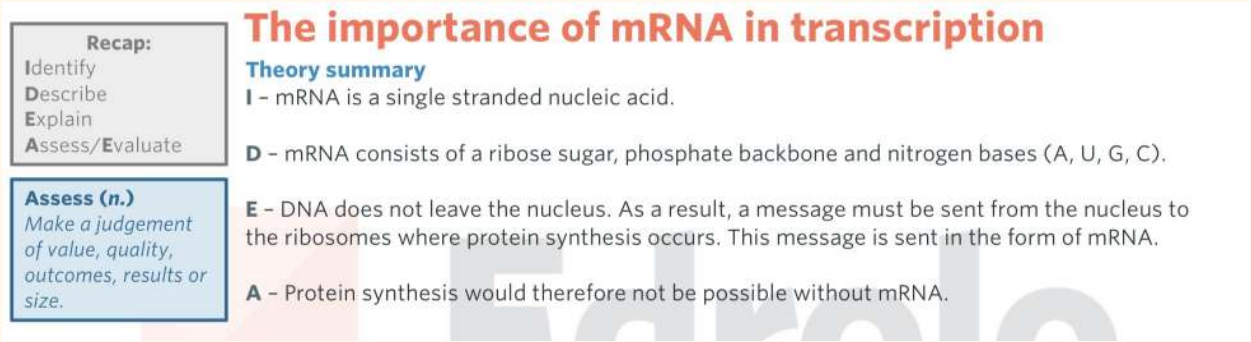
- mRNA docks to a ribosome. Ribosomes also contain RNA, ribosomal RNA (rRNA) which make up a structural component of the ribosome.
- Ribosome temporarily attaches tRNA molecules with complementary anticodons that match with the codons (sequence of 3 bases) on the mRNA.
- As subsequent tRNA molecules dock, peptide bonds form between the amino acids carried by each tRNA.

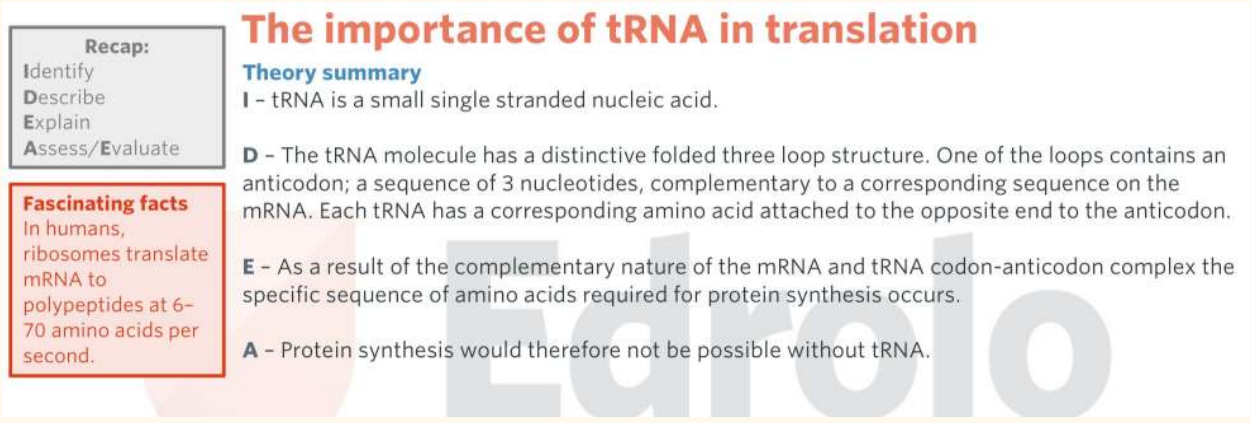
- The polypeptide chain grows as the tRNA molecules continue to dock. The amino acids are spliced off their tRNA carriers which leave, moving back to the cytoplasm where they will bind to a new amino acid and be reused.
- This continues until a stop codon is reached, upon which the ribosome releases the mRNA and the polypeptide.
- The polypeptide folds and undergoes post-translational modifications, resulting in a mature protein which can be used in the cell. It may join with other polypeptides to form the protein. The mRNA is broken down into individual amino acids to be reused.
- *Note that usually the mRNA is read by a large number of ribosomes at the same time, so that multiple copies of the polypeptide are produced from a single mRNA molecule. Each ribosome must begin at the start codon, however.

In prokaryotes the protein synthesis process is slightly different.

– assessing the importance of mRNA and tRNA in transcription and translation (ACSBL079)

Both mRNA and tRNA are essential for the expression of a gene into a functional protein.





(Credit: Edrolo & Tim Sloane, 2018)

– analysing the function and importance of polypeptide synthesis (ACSBL080)

- ➢ Polypeptide synthesis is essential for producing proteins, which the cell needs to function and survive.
- ➢ Diseases like cancer and age-related degenerative diseases like Alzheimers can arise from incorrect synthesis of proteins. Mistakes during protein synthesis are also associated with ageing and formation of toxins.
- ➢ Protein synthesis is important for increasing the complexity of organisms, especially multicellular animals. More complex organisms like humans have a higher protein to gene ratio, as variation is or can be introduced at each stage in the protein synthesis process. This means we can produce more proteins from fewer genes. Eg. *E. coli* has 4288 genes and produces around 4700 proteins, whereas humans have 20 067 genes but produce around 1 000 000 different proteins.

– assessing how genes and environment affect phenotypic expression (ACSBL081)

Key terms

- ★ Genotype: the genome or genetic makeup of an organism, determined by biological testing.
- ★ Phenotype: the physical expression of a trait which can be observed. Includes structure, behaviour and physiology.
- ★ Epigenetics: changes in an organism caused by changes in gene expression rather than alteration of the genetic code. This can occur when chemical markers bind to parts of the DNA and prevent it from being transcribed, or "switch on" other genes instead. Eg. DNA methylation. Epigenetics relates to environmental and behavioural factors, and may be inherited.

Gene expression can be measured by DNA microassay technology, giving an approximate concentration of mRNA.

Effect of Genes on Phenotypic Expression

- ➢ Phenotypic expression is a direct result of gene expression, as our genes play a major role in what proteins are produced in our bodies. Proteins control structural and functional parts of organisms, which determine our observable phenotypes.
- ➢ Our genotype states what alleles we inherit, and what traits result from those alleles.

- ➢ Genes also encode for proteins (eg. Transcription factors) that regulate the expression of other genes, influencing the differentiation of different cell types.
- ➢ Genotype determines phenotype.

To assess, define and describe genes, explain their role and link to phenotypes, define and describe phenotypes, evaluate the importance of how genes affect phenotypic expression. Eg. Cystic fibrosis, recessive single gene disorder.

Effect of Environmental Factors on Phenotypic Expression

- ➢ Environmental factors can't change a genome, but can influence which genes are expressed and therefore the phenotype of the individual.
- ➢ This is an example of epigenetics, which occurs due to the addition of chemical markers to DNA or even histones, influencing its ability to be expressed.
    - Genes may be switched off if they are packed together tightly / highly condensed.
    - The addition of methyl groups (—CH3) (methylation) blocks DNA from expression.
    - Addition of acetyl groups (—COCH3) (acetylation) promotes transcription (unblocks) so genes are expressed.
    - Promoters for each gene in eukaryotes can be activated by the presence or absence of certain chemicals. These activate RNA polymerase for transcription.
    - Translation of mRNA can also be blocked by specific proteins which prevent binding to ribosomes.
- ➢ Eg. Hydrangea flower colour is determined by soil pH.
- ➢ Eg. Identical twins - any differences are due to environmental factors, as their genetics are the same.

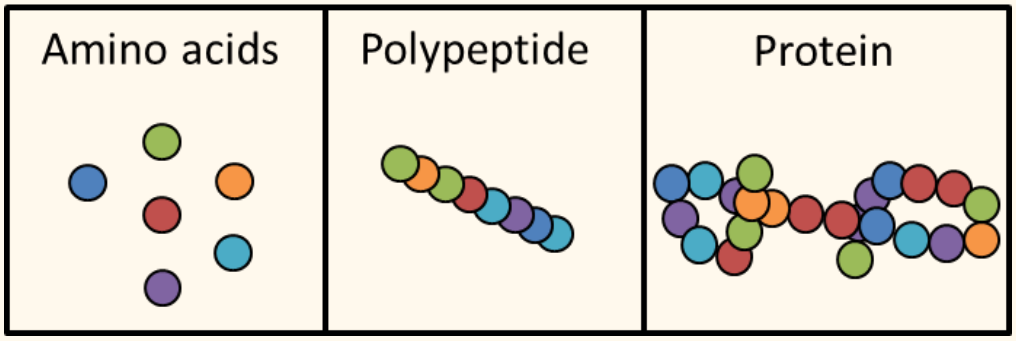
See above for using verbs accurately (assess), use examples and define/describe environmental factors in epigenetics.

• investigate the structure and function of proteins in living things

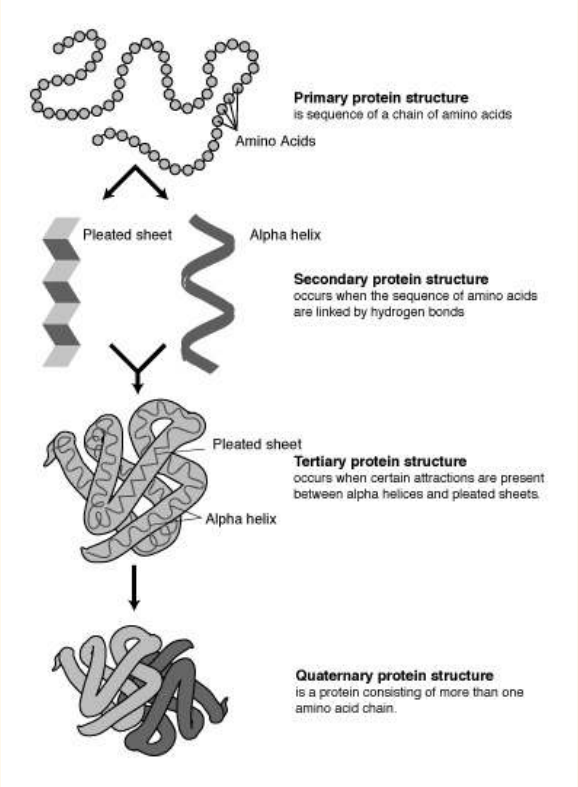
Protein Structure

- ➢ Chemically, proteins are made up of amino acids which contain carbon, hydrogen, oxygen and nitrogen (CHON) and sometimes sulphur.

- Recall amino acids link and form peptides (shorter chains) which form polypeptides (longer chains) which can combine and/or fold to form a protein.



- Recall amino acids are simple organic compounds containing an amino group, carboxyl group, hydrogen, a central carbon and a variable group (R-group) or side chain.
- The physical structure of proteins is described on 4 levels:
    - Primary structure: amino acid sequence in the polypeptide.
    - Secondary structure: pleats and spirals (alpha helix/helices) caused by hydrogen bonds.
    - Tertiary structure: folding caused by interactions between the spirals and pleated sheets.
    - Quaternary structure: two or more polypeptides fit together.



- ➢ Proteins bind with other molecules to carry out their functions, so their shape and chemical properties (eg. charge) determine their functionality. This shape is determined by the folding.
- ➢ Some proteins (conjugate proteins) are linked to a non-protein part, termed a cofactor. Tightly bound cofactors are called prosthetic groups eg. Haemoglobin with haem group containing iron. Cofactors loosely bound to an enzyme are called coenzymes.
- ➢ Other types of proteins include fibrous and globular.

Protein Function

Type of Protein	**Brief Description of Role (Humans)**	**Example**
Transport	Carries substances in the body. Can assist in removal of wastes and supply of nutrients.	Haemoglobin (carries O2 in blood).
Signalling (ie hormones, receptor proteins, antibodies)	Allows cells to communicate in order to regulate metabolic functions.	Insulin (hormone, regulates glucose uptake by cells and controls blood sugar concentration).
Storage	Stores mineral ions in the body.	Ferritin (regulates body's iron storage).
Motor	Allow movement.	Contractile proteins like actin and myosin (muscle contraction).
Defensive (Antibodies)	Form part of immune system, helps to remove foreign substances and fight against pathogens.	Antibodies (recognise and mark antigens on pathogens).
Enzymes	Globular proteins that catalyse biochemical reactions.	Lactase (helps break down lactose).
Structural	Provide support in our bodies.	Collagen (found in connective tissues eg. cartilage).
Sensory	Change activity or shape in response to stimuli.	Opsins (detect light in retinas).
DNA-associated	Regulate chromosome structure during cell division or play a role in regulating gene expression.	Histones (store DNA in nucleosomes).

Genetic Variation

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

Students:

• conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosomes, fertilisation and mutations (ACSBL084)

- ★ Variation in populations occurs due to multiple factors contributing to different or new genotypes.
- ★ *Variation* is defined as the differences in characteristics evident in individuals.
- ★ *Variability* is defined as the different forms of a gene within a population, or the total alleles present in the gene pool.
- ★ Mutation introduces new alleles. Meiosis and fertilisation introduce new *combinations* of those alleles.

Meiosis

- ➢ New genetic combinations arise from crossing over, independent assortment and random segregation. *Alphabetical order- also the order that they happen in!
- ➢ Crossing Over: occurs in prophase 1 when homologous chromosomes line up and exchange segments of DNA, producing new gene combinations with sister chromatids.
    - This mixes maternal and paternal genes across chromosomes.
    - Also ensures not all linked genes are inherited together.
- ➢ Independent Assortment
    - Occurs in metaphase I/anaphase I of meiosis
    - Homologous chromosomes separate independently of each other. The side they end up on during metaphase I is random. They are then drawn apart in anaphase I.
- ➢ Random Segregation
    - Occurs in metaphase II/anaphase II

    - Similarly to how homologous chromosomes are split up randomly in independent assortment, the separation of the sister chromatids in anaphase II is completely random, based on which side they end up on during metaphase II.

Fertilisation

➢ Essentially extra variation is introduced through the union of a genetically unique egg and a genetically unique sperm. Of the 150 million (give or take a few hundred million) sperm released in a human male ejaculate, only one fertilises the egg and this is largely due to chance.

Mutation

- See Mod 6 notes - germline mutations create new, heritable, alleles.

● model the formation of new combinations of genotypes produced during meiosis, including but not limited to:
– interpreting examples of autosomal, sex-linkage, co-dominance, incomplete dominance and multiple alleles (ACSBL085)

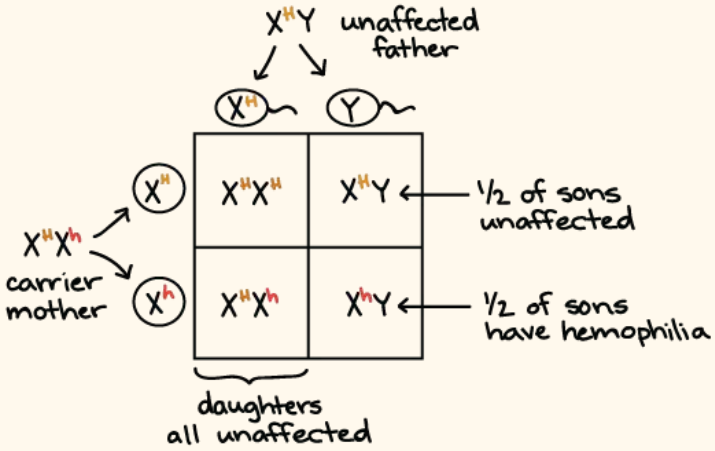
Autosomal / Mendelian

➢ Dominant/recessive traits (homozygous & heterozygous combos)
➢ Single pair of alleles
➢ Regular punnett square combinations:
    - TT = homozygous dominant
    - Tt = heterozygous dominant
    - Tt = homozygous recessive
➢ Eg. Mendel's peas, studied monohybrid crosses to track a single trait. 'Eye colour' example.
➢ If the trait is dominant: affected individuals must have at least one affected parent.
➢ Recessive traits: affected individuals may have unaffected parents. Two affected parents only have affected children.

Sex-linked

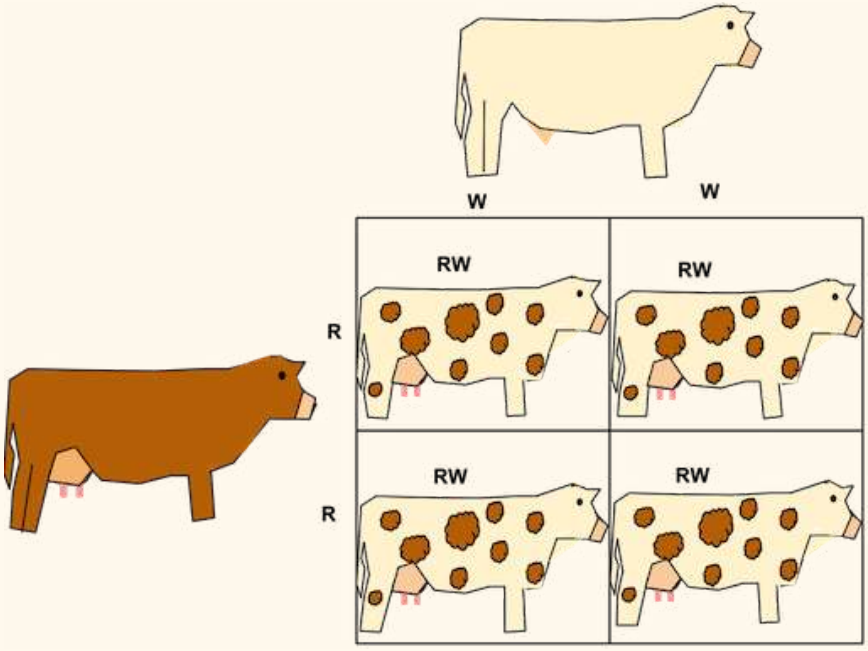
➢ Specific to X or Y chromosomes (typically X)
➢ Recall females inherit XX, males XY.
➢ Eg. Haemophilia is X-linked recessive, so males only need to inherit one recessive allele to be affected, but females must be homozygous recessive to have haemophilia. Therefore it is more common in males.

- For X-linked, occur more often in males, all sons of an affected female are affected, and male inheritance can skip a generation.



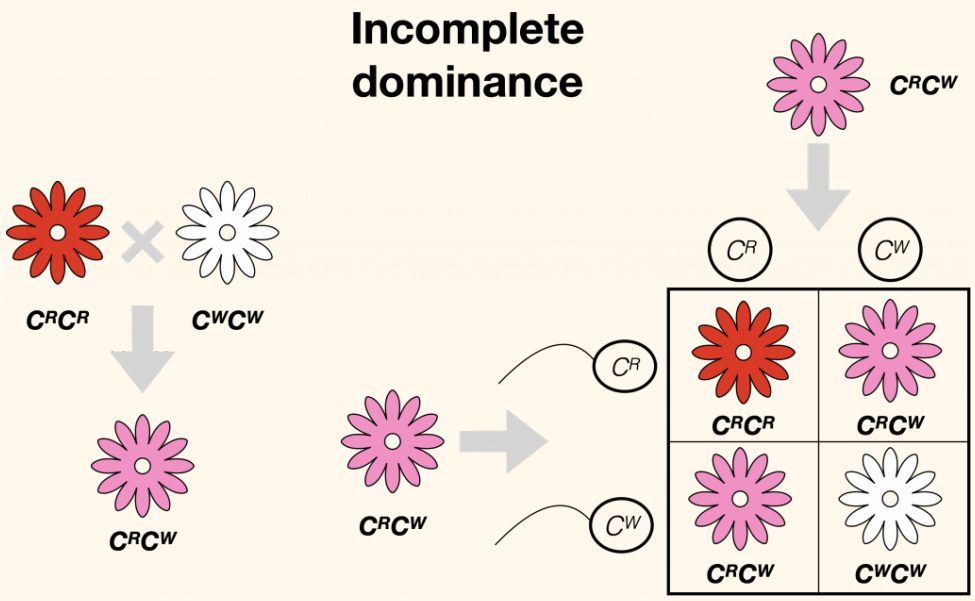
Co-dominance

- Both alleles in a gene pair are fully expressed as separate, unblended phenotypes.
- Eg. Red/White roan cattle. Hairs are alternatively red or white.



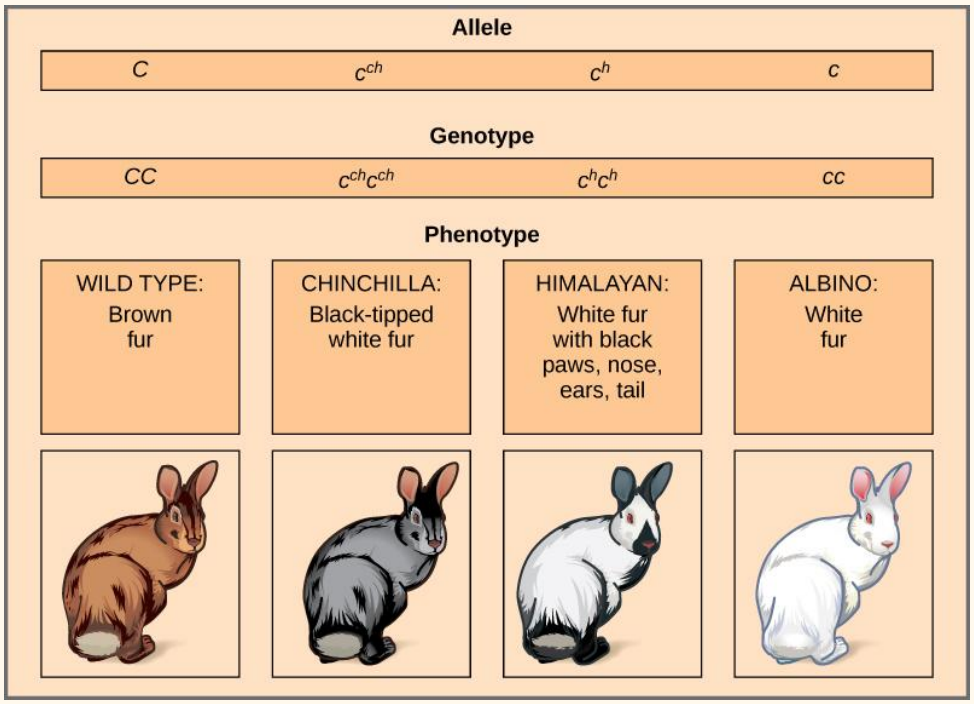
Incomplete dominance

- Neither allele is completely dominant, an intermediate blended version of the phenotypes is observed.
- Eg. Red + white flowers = pink flowers.



Multiple alleles

- ➢ Three or more alleles exist for a single trait/gene.
- ➢ Common eg. Rabbit fur colours, human blood groups.
    - May exhibit different types and hierarchies of dominance, eg. blood groups are codominant for type A and type B variant alleles but type O is recessive; rabbit fur is completely or incompletely dominant.

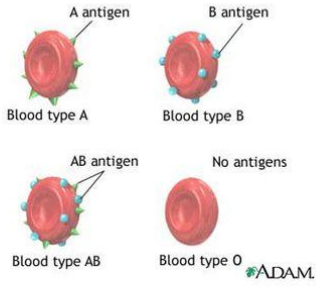


In the rabbit fur example, C is dominant over the other three alleles, c^{ch} is incompletely dominant over c^{h} which is dominant over

Multiple Alleles

- **More than two alleles can be inherited**
  - Example: Blood Types
  - Alleles= I^A, I^B, i

Blood Type (Phenotype)	Type of Antigen	Possible Genotypes
A	A	I^AI^A, I^Ai
B	B	I^BI^B, I^Bi
AB	AB	I^AI^B
O	None	ii



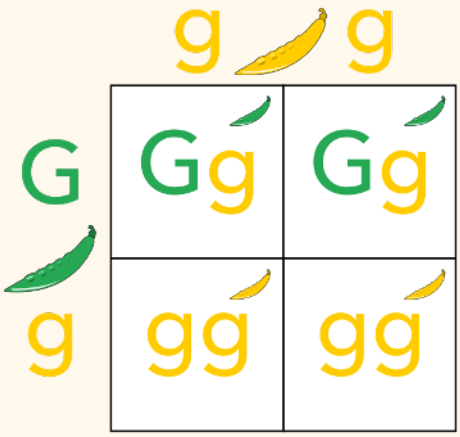
c.

Antigens found on RBCs determine blood type. +/- tive is determined by Rh (Rhesus) factor, also inherited.

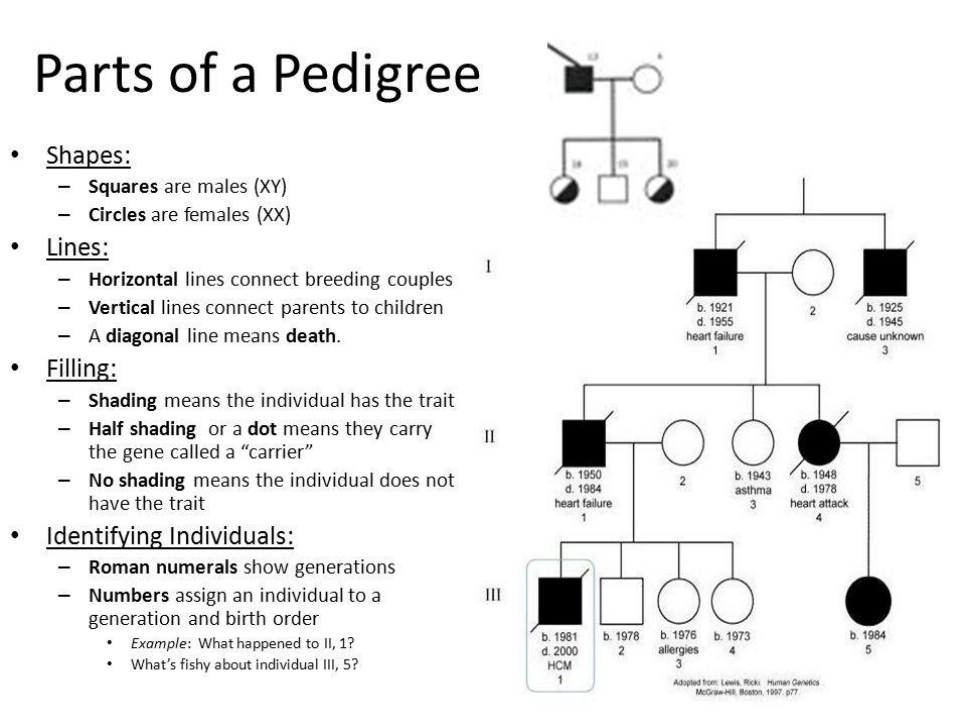
– constructing and interpreting information and data from pedigrees and Punnett squares

Punnett Squares

Basic Example



Pedigree Charts



● collect, record and present data to represent frequencies of characteristics in a population, in order to identify trends, patterns, relationships and limitations in data, for example:
– examining frequency data

- ★ Population genetics: study of how the gene pool of a population changes over time.
- ★ Gene pool: all the alleles of all of the genes in a breeding population
- ★ Polygenic: trait controlled by multiple genes, eg. human skin pigmentation
- ★ Allele frequency: measure of how common an allele is within a population
- ➢ Changes in allele frequencies are influenced by selective pressures and vary across population groups
- ➢ Combining Darwinian evolution and Mendelian genetics we can explain how changing allele frequencies leads to micro and macroevolution.
- ➢ Allele frequency = number of copies of a specific allele / total copies of that gene (total alleles).

– analysing single nucleotide polymorphism (SNP)

- ★ Polymorphism refers to the occurrence of multiple variant forms of a specific DNA sequence, i.e. multiple alleles exist for one gene locus. Each allele must occur in more that 1% of the population.

★ Single nucleotide polymorphisms are polymorphisms affecting a single nucleotide at a specific position on the genome *Owen Wilson: “wowww”*
    - SNPs account for most of the genetic differences across humans.
    - They are most common within non coding DNA (introns), but are also found in exons (coding DNA) and intergenic regions.
    - Specific SNPs are detected in populations, regions and continents.
    - They may be insertions, substitutions or deletions. Basically they’re widespread mutations. Alleles can be SNPs, but not all SNPs are coding (alleles).
    - Sickle cell disease is caused by a SNP/substitution.
    - SNPs have frequencies (eg. minor allele frequency is the frequency of the second most common allele).
    - SNPs are used as genetic markers for genetic disease screening, testing drug compatibility, in forensics and tracking human migration history.

Inheritance Patterns in a Population

Inquiry question: Can population genetic patterns be predicted with any accuracy?

Students:

● investigate the use of technologies to determine inheritance patterns in a population using, for example: (ACSBL064, ACSBL085)

- DNA sequencing and profiling (ACSBL086)

These processes require PCR and gel electrophoresis.

PCR

➢ DNA is amplified to make billions of copies of a gene or genome.
    - DNA sample is heated to denature the DNA and separate the two strands
    - Annealing: DNA primers are attached to the 3’ end of the target sequence (under reduced heat)
    - Elongation: heat resistant DNA polymerase binds to the primer and copies the strand of DNA by attaching free floating nucleotides. (Double strand now created)
    - Process is repeated.

Gel Electrophoresis

➢ Sorts DNA fragments based on size/mass. Relies on DNA’s charge.
    - After DNA extraction and PCR, samples are added to wells in a gel medium.
    - An electric current is applied.

- Smaller DNA fragments travel further through the gel and bands are separated on size.
- Staining is used to visualise bands.

DNA Sequencing

- ➢ Determines exact DNA sequence of nucleotides
- ➢ Common method is Sanger sequencing
  - DNA sequences are amplified typically with PCR, and heated to separate the strands.
  - Primers are added to the 3' end and the primed sequence is combined with a mixture of DNA polymerase, free nucleotides and one base type (A, T, G, C) which is a modified chain-terminating nucleotide, also tagged with a fluorescent dye.
  - DNA polymerase adds bases until a chain-terminating nucleotide is reached which ends the sequence. This creates fragments of different lengths, each ending with a tagged nucleotide.
  - The fragments are separated using capillary gel electrophoresis, which can differentiate mass differences in one nucleotide. Shortest fragments reach the end first.
  - A laser beam causes the nucleotides to fluoresce, and this is read in the order that they pass through the capillary tube, recreating the DNA sequence of bases!

DNA Profiling / Fingerprinting

- ➢ Used to identify and compare individuals by looking at short segments of DNA - not the whole genome or at specific nucleotides.
- ➢ Relies on the presence of VNTRs (variable number tandem repeats) or shorter STRs (short tandem repeats, 2-5 base pairs), repeating sequences of DNA located in the introns, that are highly variable across individuals. These are inherited.
- ➢ Commonly used in forensics and paternity testing.
  1. DNA is extracted, and then amplified using PCR.
  2. Restriction enzymes cut DNA into fragments at specific DNA sequences, creating a mixture of DNA fragments of different sizes.
  3. Fragments are separated by size in gel electrophoresis.
  4. The unique separation of the fragments into bands based on fragment size allows comparisons to be made.

● investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example: (ACSBL064, ACSBL073)

- the use of population genetics data in conservation management
- population genetics studies used to determine the inheritance of a disease or disorder
- population genetics relating to human evolution

Gonna skip over this dot point. Conservation, we don't want species going extinct. Genetics helps with biodiversity and selection pressures and evolution and inbreeding and stuff like that.

Module 6: Genetic Change

Outcomes

A student:

› solves scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO11/12-6
› communicates scientific understanding using suitable language and terminology for a specific audience or purpose BIO11/12-7
› explains natural genetic change and the use of genetic technologies to induce genetic change BIO12-13

Content Focus

Students learn about natural and human-induced causes and effects of genetic change, including mutations, environmental pressure and uses of biotechnology. Students investigate how the processes of inheritance and evolution are applied.

The work of scientists in various fields of work, including agriculture, industry and medicine, can be explored within the context of biotechnology. The impact of biotechnology on biological diversity is also explored in this module.

Working Scientifically

In this module, students focus on analysing trends and patterns and solving problems using evidence from data and information. Students also focus on communicating ideas about genetic change for a specific purpose. Students shouldbe provided with opportunities to engage with all Working Scientifically skills throughout the course.

Content

Mutation

Inquiry question: How does mutation introduce new alleles into a population?

Students:

- explain how a range of mutagens operate, including but not limited to:
  - electromagnetic radiation sources
  - chemicals
  - naturally occurring mutagens

★ Mutation: permanent change to the nucleotide sequence in an organism's genome.
★ Mutagen: agent causing a mutation that significantly enhances the spontaneous rate of mutation.

EMR mutagens include:

➢ UV light, which can cause pyrimidine dimers (eg. Thymine dimers), covalent bonding usually between two adjacent nucleotides that makes them unable to be read properly when DNA is replicated.
➢ X-rays
➢ Gamma radiation and other ionising radiations.

Chemical mutagens are often mis incorporated into the DNA structure or create gaps in it. Examples include radioactive agents (alpha, beta radiation), intercalating agents, some metals and nitrous acid, which may swap bases,

Naturally occurring mutagens, such as certain viruses, bacterial infections, fungi and toxins.

➢ Cycasin
➢ Mycotoxins
➢ *Helicobacter*
➢ Transposons (jumping genes)

Metabolic products and naturally occurring chemicals also come under natural mutagens.

- compare the causes, processes and effects of different types of mutation, including but not limited to:
  – point mutation

➢ Frameshift
  - Insertion
  - Deletion
  - Effects may be silent, missense, nonsense.

➢ Substitution (single nucleotide)
  - Silent
  - Missense
  - Nonsense

  – chromosomal mutation

➢ Affect whole chromosome or segment.
➢ Deletion
➢ Inversion
➢ Translocation
➢ Duplication

- distinguish between somatic mutations and germ-line mutations and their effect on an organism (ACSBL082, ACSBL083)

Yep.

- assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation (ACSBL078)

Coding encodes for proteins ie, genes. Non-coding do not encode for proteins, but may instead function as code for structural RNAs, form introns which vary expression depending on how they are spliced out, or regulatory sequences like promoters, silencers, enhancers and terminators.

- investigate the causes of genetic variation relating to the processes of fertilisation, meiosis and mutation (ACSBL078)

Literally addressed this in mod 5 😒

- evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations (ACSBL091, ACSBL092)

★ Natural selection (iykyk, right?)
★ Gene flow: transfer of different alleles from one population to another. Think migration.

★ Genetic drift: when relative frequencies of alleles change in a population, due to the disappearance of particular genes due to random, chance events.
    - Eg. Bottleneck effect (sudden loss of large numbers of population)
    - Founder effect (small group is separated and forms new population with new allele frequencies.

Biotechnology

Inquiry question: How do genetic techniques affect Earth's biodiversity?

Students:

- investigate the uses and applications of biotechnology (past, present and future), including: (ACSBL087)
  – analysing the social implications and ethical uses of biotechnology, including plant and animal examples
  – researching future directions of the use of biotechnology
  – evaluating the potential benefits for society of research using genetic technologies
  – evaluating the changes to the Earth's biodiversity due to genetic techniques

Ethics, intellectual property, biodiversity, sustainability, cost, regulation, bio hacking, ownership, labelling, monocultures, horizontal gene transfer...

Genetic Technologies

Inquiry question: Does artificial manipulation of DNA have the potential to change populations forever?

Students:

- investigate the uses and advantages of current genetic technologies that induce genetic change

➢ Reproductive technology eg A.I., IVF, pollination
➢ Cloning, stem cells, gene cloning
➢ Recombinant DNA, gene therapy, transgenic organisms.

Eg. CRISPR (clustered regularly interspaced short palindromic repeats)

- CRISPR/CRISPR-Cas9 gene editing technology, easy, precise & affordable.
- Clustered Regularly Interspaced Short Palindromic Repeats.
- Natural adaptive immune response in prokaryotes. Defence against viruses. Discovered 1987.
- CRISPR associated (Cas) enzymes cut out and copy viral "spacers" from new invading virus DNA, stored in bacterium's genome interspaced by short palindromic repeats.

- "Vaccinates" bacterium. CRISPR array is transcribed → RNA, cut into crRNA (CRISPR RNA). Combines w/ trans-activating RNA, → guide RNA. Joins Cas9 in complex.
- Next time virus invades, guide RNA matches with viral DNA, Cas9 cuts target DNA, virus neutralised.
- Guide RNA can be created by scientists- credited to Jennifer Doudna & Emmanuelle Charpentier, published 2012.
- Medical breakthrough, gene therapies, CF, Huntington's, sickle cell anaemia.
- Enviro Biotech! Plant/microbial biofuels eg. CRISPR doubled lipid production in algae. Bioplastics, bioremediation, biomining.
- Crops, disease & drought tolerance. Also increase nitrogen fixation, carbon sequestration. → lower greenhouse gases, increase soil fertility.
- Limitations: not reliable or accurate enough for widespread use. Off target effects → mutations.
- Holistic approach needed to achieve sustainable global community, not just CRISPR.

• compare the processes and outcomes of reproductive technologies, including but not limited to:
  – artificial insemination
  – artificial pollination

Kinda went over this in mod 5.

• investigate and assess the effectiveness of cloning, including but not limited to:
  – whole organism cloning

Dolly the sheep, somatic cell nuclear transfer: DNA taken from somatic cell of one sheep and placed in a denucleated egg cell of another sheep. This is then implanted into a surrogate sheep and the embryo develops into a clone of the DNA donor sheep.

  – gene cloning

Creating transgenes or multiple gene copies- protein farming.

➢ Target gene identified
➢ Bacterial plasmids isolated
➢ Restriction enzymes cut target dna and plasmids, creating 'sticky ends' which are annealed together by DNA ligase.
➢ Plasmid now contains required gene.
➢ Recombinant plasmid reinserted into the host bacteria and expressed to produce large amount of the target protein, eg. Insulin.

• describe techniques and applications used in recombinant DNA technology, for example:
  – the development of transgenic organisms in agricultural and medical applications (ACSBL087)

-Gene cloning and creation of transgenes
-Inserting the transgene into the genome by way of biolistics (tiny particles eg. Gold), micro injection into the nucleus of a single cell, viral vector transduction, electroporation (electrical current increases membrane permeability).

-Eg. Bt cotton reduced need for pesticides, Golden rice for increased vitamin A.
-Transgenic mice allow study of knockout genes.

- evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications (ACSBL086)
- evaluate the effect on biodiversity of using biotechnology in agriculture
- interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies

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Module 7 - Infectious Disease

Outcomes

A student:

› develops and evaluates questions and hypotheses for scientific investigation BIO11/12-1
› designs and evaluates investigations in order to obtain primary and secondary data and information BIO11/12-2
› conducts investigations to collect valid and reliable primary and secondary data and information BIO11/12-3
› selects and processes appropriate qualitative and quantitative data and information using a range of appropriate media BIO11/12-4
› analyses infectious disease in terms of cause, transmission, management and the organism's response, including the human immune system BIO12-14

Content Focus

This module examines the treatment, prevention and control of infectious disease both locally and globally. It includes study of the human immune system and its response to an infectious disease.

The value of studying infectious disease and its causes and effects is highlighted by the cost to humans in terms of losses in productivity and production and the impact on overall health. The module also considers medical and agricultural applications that draw on the work of a variety of scientists.

Working Scientifically

In this module, students focus on developing and evaluating questions and hypotheses when planning and conducting investigations to analyse trends, patterns and relationships in data about infectious diseases. Students should be provided with opportunities to engage with all Working Scientifically skills throughout the course.

Content

Causes of Infectious Disease

Inquiry question: How are diseases transmitted?

Students:

- describe a variety of infectious diseases caused by pathogens, including microorganisms, macroorganisms and non-cellular pathogens, and collect primary and secondary-sourced data and information relating to disease transmission, including: (ACSBL097, ACSBL098, ACSBL116, ACSBL117)
  - classifying different pathogens that cause disease in plants and animals (ACSBL117)
  - investigating the transmission of a disease during an epidemic

Recap - Terminology

★ AN EPIDEMIC is a disease that affects a large number of people within a community, population, or region.
★ A PANDEMIC is an epidemic that's spread over multiple countries or continents.
★ ENDEMIC is something that belongs to a particular people or country.
★ AN OUTBREAK is a greater-than-anticipated increase in the number of endemic cases. It can also be a single case in a new area. If it's not quickly controlled, an outbreak can become an epidemic.

(Intermountain Healthcare, 2020)

Case Study - Equine Influenza Virus (Horse Flu) Epidemic

- ➢ Horse flu is an exotic equine respiratory disease. An outbreak was observed in August 2007 in Australia.
- ➢ Cause and Transmission
    - Equine influenza is caused by Equine Influenza Virus (EIV) *orthomyxovirus* and affects horses and donkeys. Humans are unaffected by the disease (not zoonotic).
    - Two main strains exist, known as equine-1 and equine-2.
    - Equine influenza is highly contagious and spread through direct contact between uninfected and infected horses through nasal secretions and other bodily fluids.
    - Indirect contact with humans can spread the virus through contaminated shoes, feed and water buckets,