MODULE 5 – HEREDITY

Red = titles
Purple = subtitles
Green = secondary subtitles
Black = info

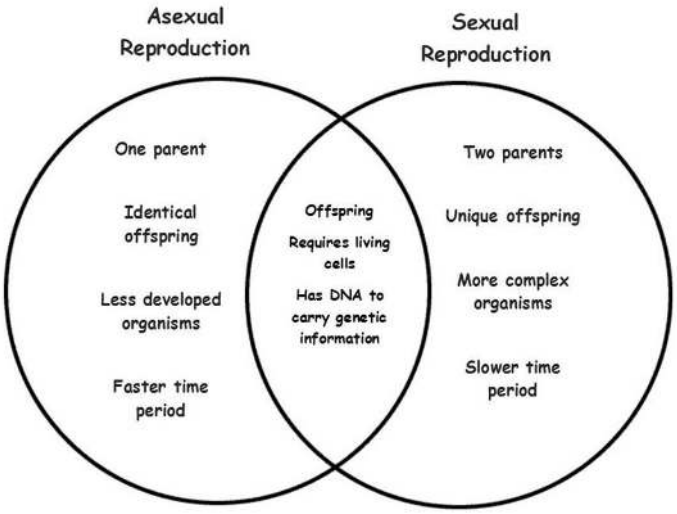
REPRODUCTION

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:

Sexual vs Asexual reproduction: THE BASICS

Sexual:
The process of forming new organisms from the fusion of the offspring's parents male and female gametes.

- Gametes are sex ells (eg, sperm and egg cells in humans) which bind
- All organisms which result have the potential to be genetically diverse, offspring are never genetically identical to either parent



Asexual:
The process of forming an offspring (usually a clone) from just one parent through cell division

- No fusion of gametes, contrast with sexual reproduction

Sexual reproduction in animals: internal vs external fertilisation

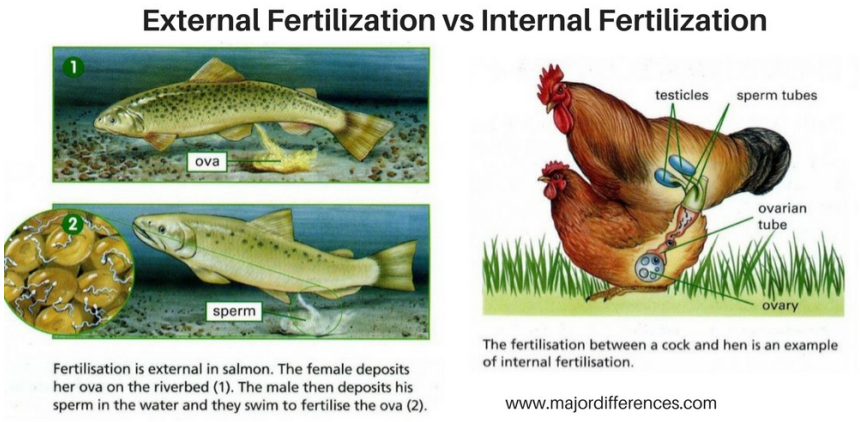
External fertilisation:

- Involves fusion of gametes *outside* the body of a parent
- Most common in aquatic animals, the water acts as a medium in which the gametes can travel
- Susceptible to environmental influences (eg; predators, pH changes)
- Hence, species that reproduce like this release large quantities of gametes to compensate for loss
- This is called ***spawning***
- Eg; corals, fish, amphibians (eg; frogs)

Internal fertilisation:

- Involves fusion of gams *inside* the body of a parent
- Gamete of one parent can be introduced inside the body of another (eg; copulation)

- Terrestrial animals typically use internal fertalisation to prevent exposure and desiccation of gametes or embryos
- Offers more protection to gametes and embryos but a potential survival cost to the parent
- Eg; humans, mammals (eg; chickens), some birds



ANIMALS: ADVANTAGES OF INTERNAL AND EXTERNAL FERTILISATION

animals: advantages of external and internal fertilisation

Internal fertilisation	**External fertilisation**
Increased possibilities of union of gametes because all conditions required for fusion of gametes is maintained inside the body	Results in the production of a large number of zygotes and thus more offspring can be produced (but at a high energy cost)
More protection against outside environments and predators, thus higher chance of survival until birth	Easier to find mates as the gametes released can drift (wind, water, etc.)
More selective of mate	More genetic variation
Less chance of desiccation of gametes	

plants: asexual and sexual reproduction

PLANTS: SEXUAL AND ASEXUAL REPRODUCTION

Sexual reproduction

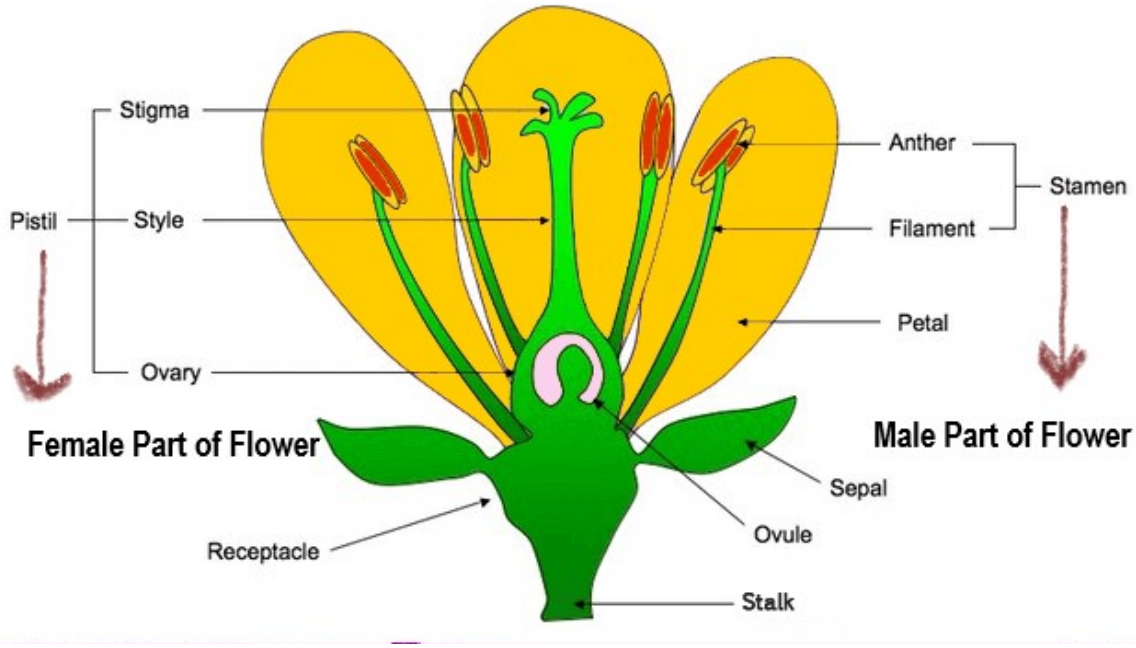
- Flowers make up the reproductive organ in plants
- Petal and nectar lure insects and other animals to assist in the delivery of pollen

Male parts

- Include filament and anther
- These make up the Stamen
- Pollen is produced in the anthers

Female parts

- Include the stigma, style, ovary and egg cell (ovule)
- These make up the Carpel
- **note that pistils are the union of one or more carpels



What is pollen?

- Microscopic structures that contain the tube cell and generative cell
- The tube cell later becomes the pollen tube
- The generative cell releases the sperm nuclei

How does pollination occur?

- It is the way in which the pollen grains reach the sticky stamen (male parts; filament and anther)
- Pollen grains may be transferred to other flowers by wind, insects and other animals

How does fertilisation occur?

- After landing on the stigma (female part), a pollen tube grows down the style (female part) into the ovary (female part)
- The male gamete moves down to fertilise the ovary

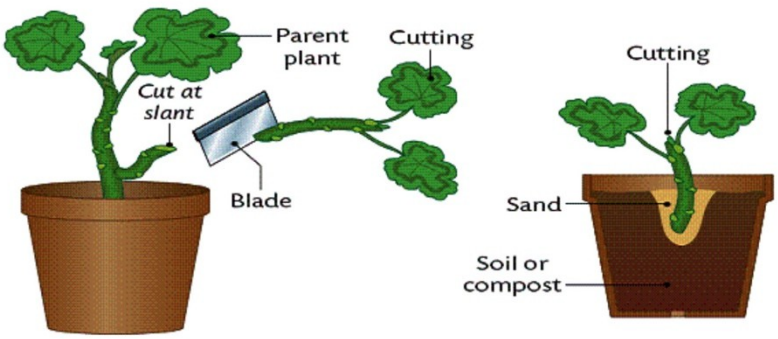
Asexual reproduction

Vegetative propagation is a type of asexual reproduction in plants, it results in genetically identical plants. Examples include cuttings, runners, bulbs and stem tubers

Cuttings

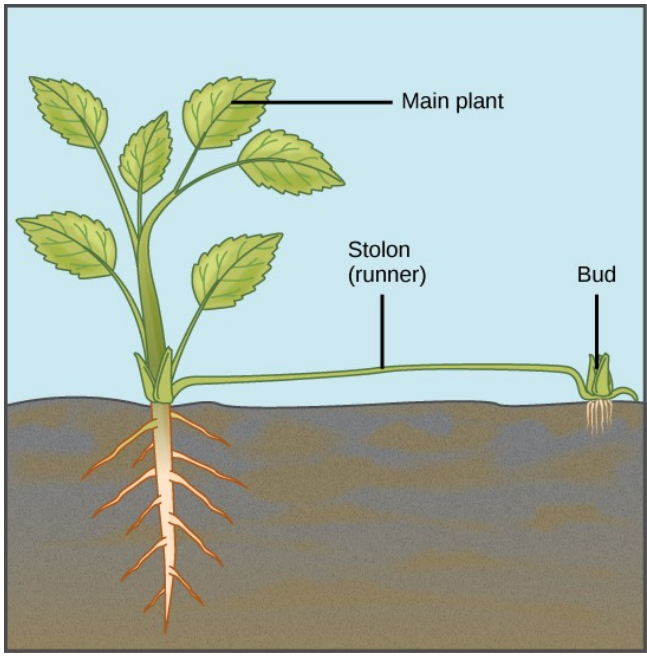
1. Stem from plant is cut
2. Stem is planted in soil
3. Soil grows into a plant

Used for plants like roses, hibiscus, sugarcane etc;



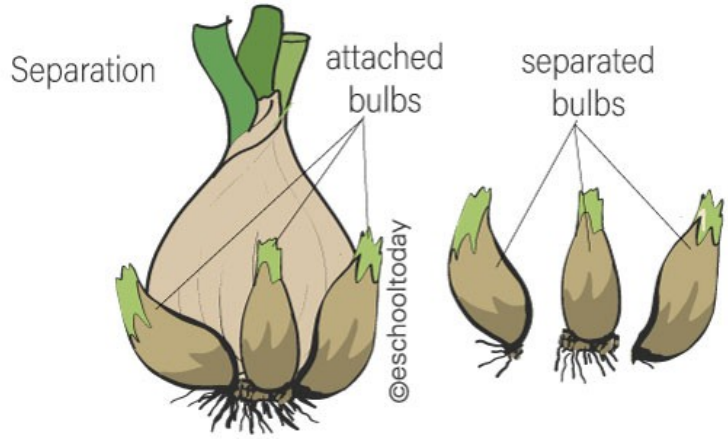
Runners

- Strawberry plants can develop runners
- Runners are stems extending from the plant along the soil
- At certain points along the runners, nodes can develop which extend in the soil
- This results in the formation of a new plants' roots at another area of the soil where a new strawberry plant can grow
- Runner joins new (genetically identical) plant to parent plant
- Eg; in mint



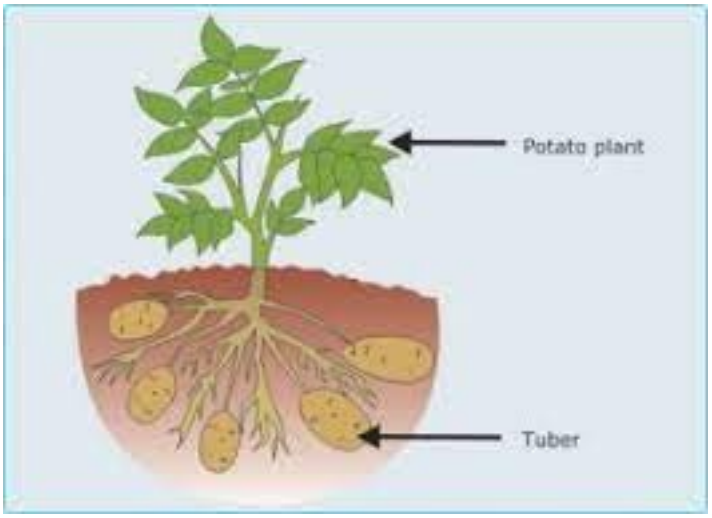
Bulbs

- Underground food storage organs
- New plant forms, underground bulb provides nutrients to the plant for its survival
- Eg; onions and garlic



Stem tubers

- 'True tubers'
- Bulbous modified stems that grow underground
- This lets the tuber store nutrients for survival



fungi: budding, spores

FUNGI - ASEXUAL

Budding

- Eg; yeast

1. Parent cell develops bud cell, containing daughter nucleus
2. Bud undergoes mitosis (cell division) while still attached to parent cell

3. This may result in a chain of bud cells
4. Bud separates from its parent fungus when it grows to become independent
5. Further cell division, more bud cells produced
6. Result is genetically identical to parent

Spores

- Eg; moulds and mushrooms
- Microscopic reproductive units (cells) formed as a result of meiosis or mitosis
- Differ from gametes as they do not need to combine with another spore to form offspring
- Eg; Hyphae
- Fine structures that branch out and have ends that are capable of producing spores called sporangia
- These Asexual spores are carried by the wind then germinate to form genetically identical new hyphae
- Spores are haploid and genetically identical to parent

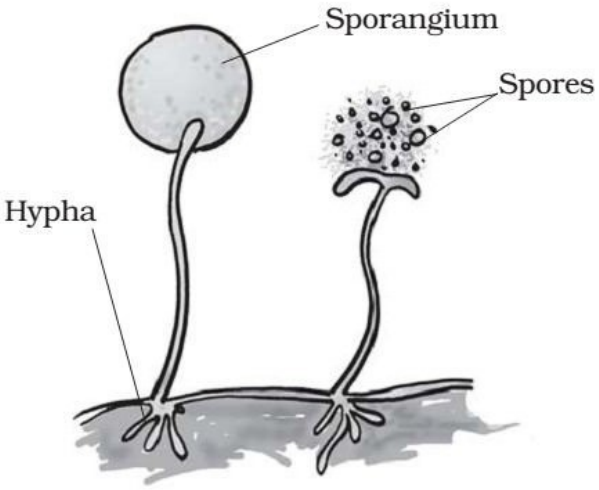


Fig. 12.7 *Reproduction through spore formation in fungus*

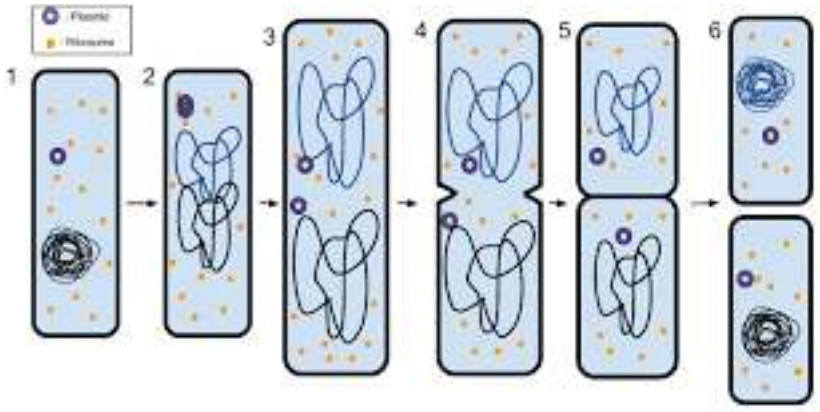
FUNGI – SEXUAL

- Opposite gender hyphae temporarily fuse together to develop a spore-producing structure known as zygospore (diploid, each of the hypha are haploid)
- **remember diploid = 2 sets of chromosomes (1 from ovum, 1 from sperm) haploid= 1 set of chromosomes per cell
- Zygosphere undergoes meiosis to produce haploid sexual spores
- These are dispersed into the environment and grow into haploid hyphae
- They are genetically different from their parents

bacteria: binary fission

BACTERIA – ASEXUAL – BINARY FISSION

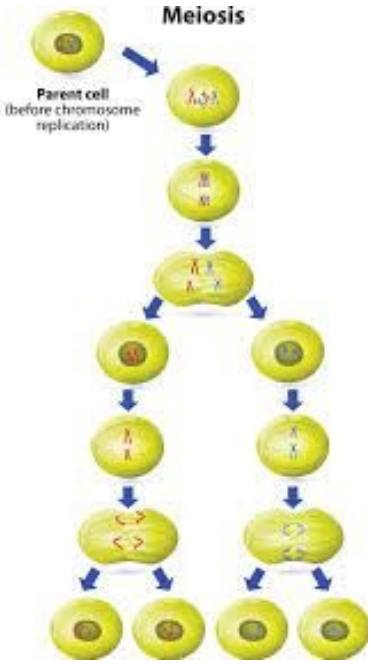
- Most bacteria have 1 chromosomes (consists of circular DNA)
- Copies the genetic material (in the form of bacterial chromosomes) of the parent cell
- There is no cell nucleus in bacteria, so no nuclear splitting occurs
- The daughter cells are genetically identical to EACH OTHER and to the PARENT



protists: binary fission, budding

PROTISTS – ASEXUAL AND SEXUAL – BINARY FISSION, BUDDING

- Eg; paramecium
- Protists (protozoa) are single celled, eukaryotic organisms
- Asexually: binary fission or budding
- Sexually: conjugation, cells fuse together briefly to exchange nuclear material



Organism	Sexual or asexual reproduction?	Method	Examples
Animal	Sexual	Internal fertilisation	Humans
Animal	Sexual	External fertilisation	Fish
Plant	Asexual	Self/cross pollination	
Plant	Asexual	Cuttings, runners, bulbs	Strawberries
Fungi	Asexual	Budding	Yeast
Fungi	Asexual	Spores	Mushrooms
Bacteria	Asexual	Binary fission	e. coli

Protists	Asexual and sexual	Budding, binary fission, conjugation (sexual)	Amoeba

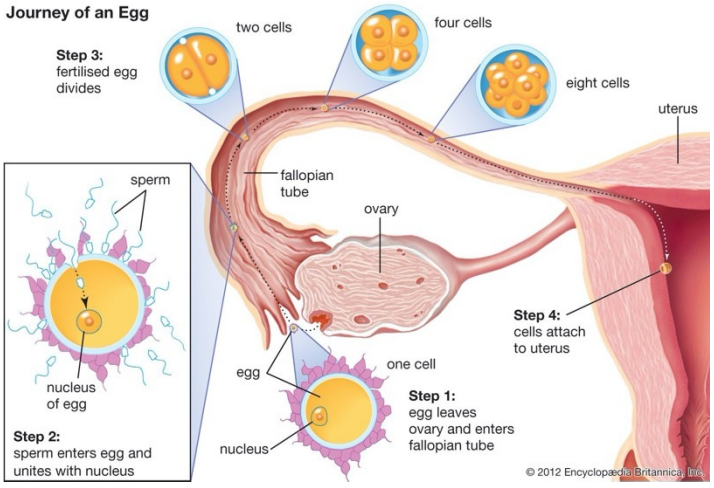
Analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals

FERTILISATION EXPLANATION

- Gametes (sperm and egg) meet and combine to form a zygote. This is called gametogenesis
- Gametogenesis can be divided into spermatogenesis (producing sperm) and oogenesis (formation of matured egg cells)
- Hormone testosterone is produced in the testes and plays a role in production sperm cells

FERTILISATION PROCESS

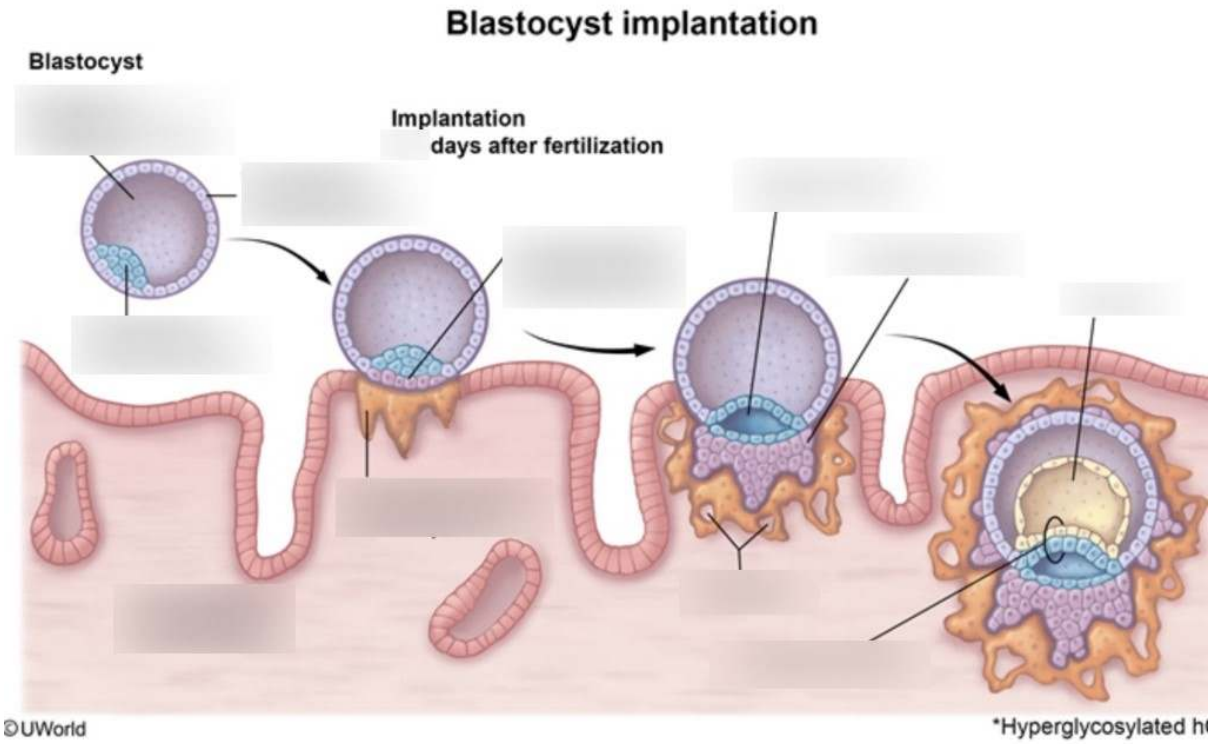
1. Occurs in 1 of the fallopian tubes
2. Sperm enters the vagina and travels up through the uterus and then along 1 of the fallopian tubes where it combines and fertilises the mature egg
3. Gametes fuse to form a zygote (single cell with 46 chromosomes, 23 from each parent)
4. During fusion, the head of the sperm cell detaches from its flagellum (tail)
5. This activates the egg cell, resulting in cell division
6. The resulting product is called a blastocyst



IMPLANTATION

- Is the process whereby a blastocyst (result of fertilisation) sticks to the walls of the uterus
- This establishes the blastocyst's access to nutrients, allowing it to develop into an embryo (blood vessels surrounding the blastocyst carry blood which has dissolved nutrients)
- If successful, the cells continues dividing
- 3 or 4 blastocyst cells develop into the inner cell mass
- Over the next FEW WEEKS, this forms into a human embryo with a head, beating heart and tiny limbs
- Some of these cells also develop into the foetal membranes that form a fluid filled protective 'bag' around the embryo

- The remaining 100 or so blastocyst cells form a structure called the trophoblast. This provides the baby's contribution to the placenta



HORMONAL CONTROL OF PREGNANCY

1. Human Chorionic Gonadotropin

- Mainly responsible for the early pregnancy symptoms ranging from missed menstruation to nausea, vomiting and fatigue
- The hormone detected by pregnancy tests

2. Progesterone

- Maintains the functionality of placenta
- Prevents sudden movement and contraction of the uterus
- Strengthens pelvic wall muscles for labour

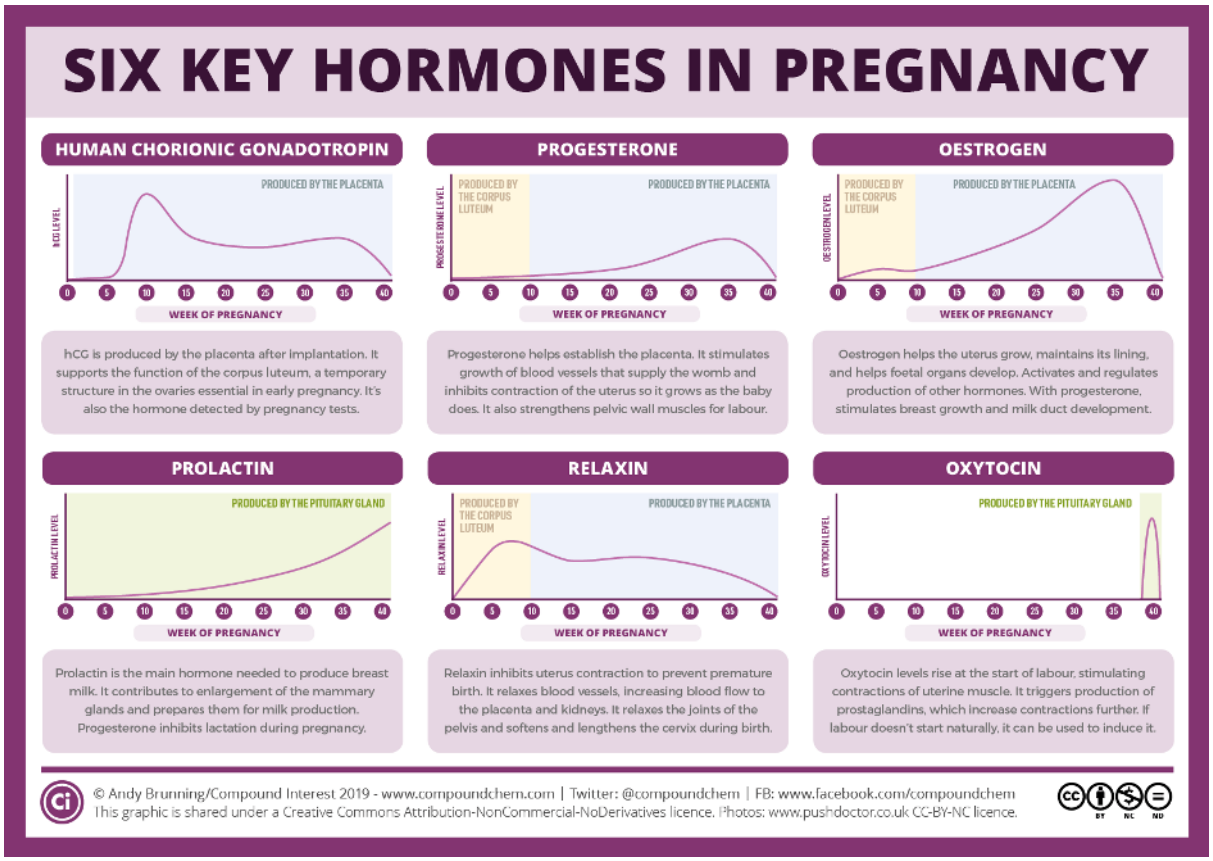
3. Oestrogen

- Ovarian hormone controlled by luteinizing hormone that triggers ovulation
- Facilitates the maturation of lungs, kidney, adrenal gland, liver and bone density of the unborn baby
- Aids the flow of blood to foetus
- Activates and regulates production other hormones (eg; with progesterone – stimulates breast growth and milk duct development)

4. Oxytocin

- Facilitates the delivery process by helping in the contractions of the uterus
- Stimulates the mammary glands to produce milk
- Hypothalamus produces oxytocin and stores it in the pituitary gland below

- Upon stimulation of the hypothalamus' neuron cells, the pituitary gland will secrete oxytocin into the bloodstream
- Can induce labour if it doesn't occur naturally



Evaluate the impact of scientific knowledge on the manipulation of plant and animals' reproduction in agriculture

Eg; BT Cotton

- Bollworm – Pest
- By genetic modification, scientists incorporate the cry1Ac gene from the soil bacterium Bacillus thuringiensis (BT) which kills the bollworm (1996-Ernt Berliner)

Eg; BT Cotton

Pros:

- reduced herbicide run-off
- increased populations of beneficial insects
- reduced pesticide use
- improved farm worker and neighbour safety (community)
- improved soil quality
- increased yield of plants

Cons:

- - An increased price of seeds
- - GM toxin harmful to humans
- - Reduces natural gene flow (genetic diversity)

Economically - positive + negative effects (developing / developed countries)
Socially - positive affects
Environmentally - positive + negative effects (less genetic diversity / fewer pesticides

CELL REPLICATION

Model the processes involved in cell replication, included but not limited to:

DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding.

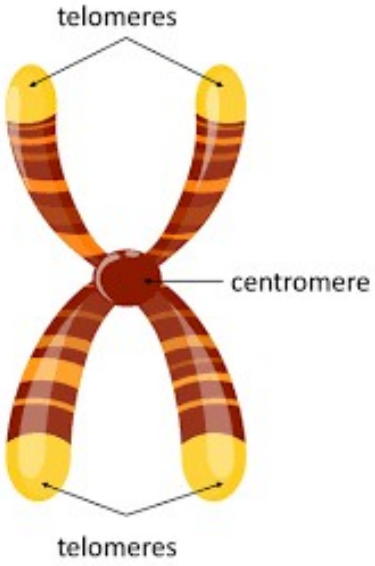
DNA REPLICATION

DNA – revision

- DNA is the genetic material that organisms inherit from their parents
- Genes are made up of DNA
- A gene is a unit of heredity information
- DNA includes an 'instruction Manuel' for coding genetics
- Chromosomes consist of 40% DNA, 60% protein (histone)
- Each coded sequence/gene is loaded is located at a particular locus on the chromosome

Chromosomes

- 'A linear sequence of genes'
- Compact coils of threadlike molecules called DNA (deoxyribonucleic acid) organised around proteins called histones (like cotton wound around a cotton reel)
- The information within genes determines phenotypes (how living things look, behave and function)
- When the cells divide the chromatin material coils into short thick rod shaped chromosomes for easy transfer
- Maternal and paternal chromosomes that carry alleles of the same genes are known as homogeneous pairs of chromosomes



Watson and Crick

- Revealed that DNA is a double Helix or twisted ladder
- A DNA molecule is made up of two chains or strands of small building blocks or '**monomers'** called nucleotides
- Each nucleotide consists of three parts
    - a phosphate
    - a sugar (deoxyribose sugar)
    - a nitrogenous base
- Each nucleotide is named after the base that it carries
    - Adenine (A)

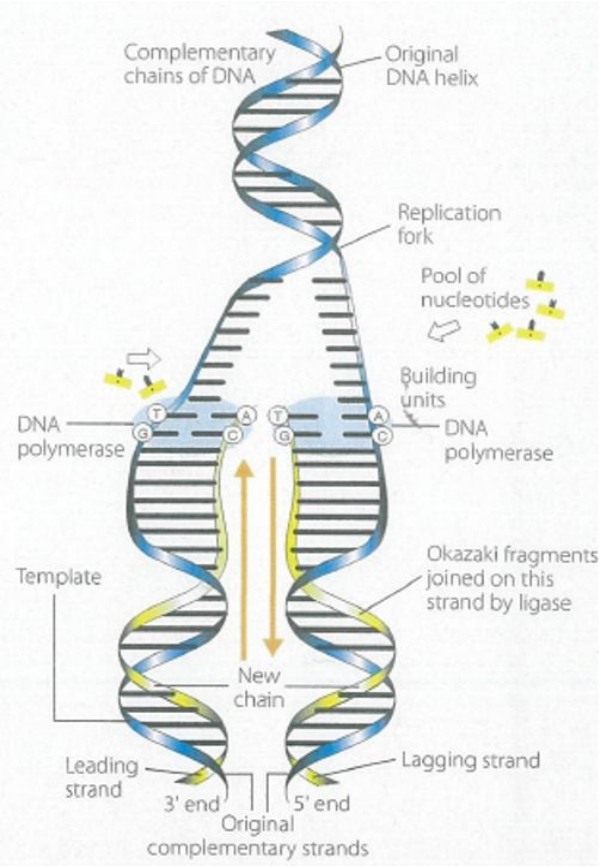
    - Thymine (T)
    - Guanine (G)
    - Cytosine (C)
- Bases are arranged in a sequence along each strand of DNA
- Each DNA molecule is thousands of bases long

Chemical structure of DNA

- The 4 bases are held together in the centre by hydrogen bonds (Adenine with Thymine, Guanine and Cytosine)
- Vertical sides are the sugar phosphate backbone, rungs are the pairs of nitrogenous bases

Process of DNA replication

1. The DNA double Helix is unwound and ***unzipped*** by the enzyme ***helicase***
2. The DNA unzips forming two single strands
3. ***Nucleotides are attached to the single strands*** resulting in two identical strands of DNA with aid of the enzyme ***DNA polymerase***
4. ***DNA fragments joined together*** by forming bonds between nucleotides with the aid of the enzyme ***DNA ligase***
5. The two double stranded molecules are now called chromatids and twister form double Helix



The significance of DNA replication

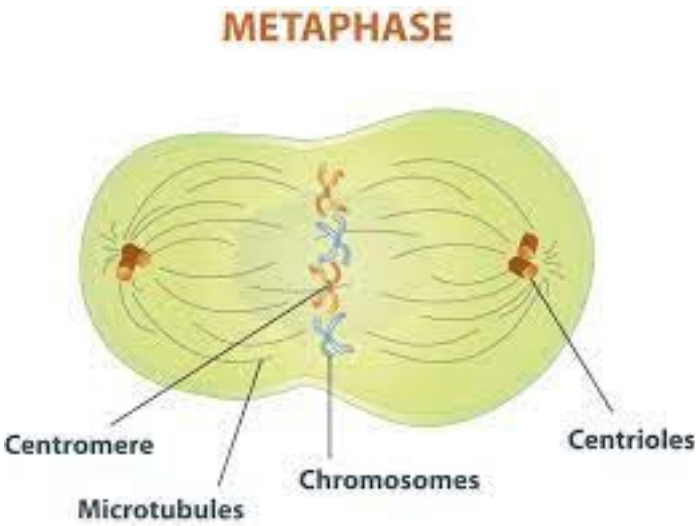
- ***Genetic information is passed on*** from generations

- During sexual reproduction, the genetic code is copied, and then half of the genetic information passes into the next organism
- This ***organism has half the genetic material from each parent***
- The DNA in a single cell contains the genetic information to make an entire organism
- When a cell divides it takes with it an exact copy of the genetic code of that organism

Mitosis and meiosis

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



The cell cycle

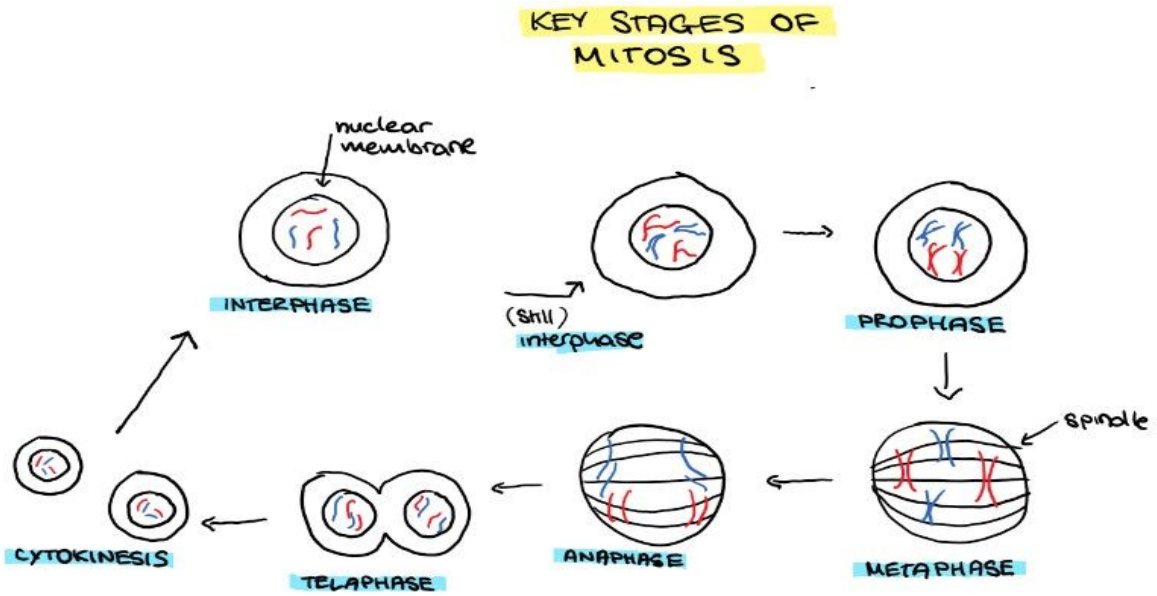
- The cell cycle is repeated throughout the life of a cell
- This includes cell growth, activities needed to survive and a vision to create new cells
- Eukaryotic cells may divide via either mitosis or meiosis

Mitosis

- The cell nucleus divides into two
- It is essential for the maintenance of organisms both for ***continued growth and repair of damaged or worn-out cells***
- One cell replicates its genetic new clear content and then divides to become two cells, distributing one full set of genetic material to each cell
- If cells divide in an uncontrolled manner, tumours or cancer may result

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	- Spindle fibres attach to centromeres of chromosomes - Chromosomes align at equator of cell
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)



Cytokinesis

- ***Last stage of my mitosis***
- Process of ***splitting daughter cells apart***
- Each daughter cell contains the same number and same quality of chromosomes
- In plant cells cytokinesis involves formation of a cell plate while the nucleus is still in telophase

Meiosis

- ***Reduces the number of chromosomes in the parents cell by half and produces four gamete cells***
- Required to produce egg and sperm cells for sexual reproduction
- Two consecutive nuclear divisions (meiosis 1 and meiosis 2) leading to the production of four haploid gametes with the maternal and paternal chromosomes being distributed randomly between the cells

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 o 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

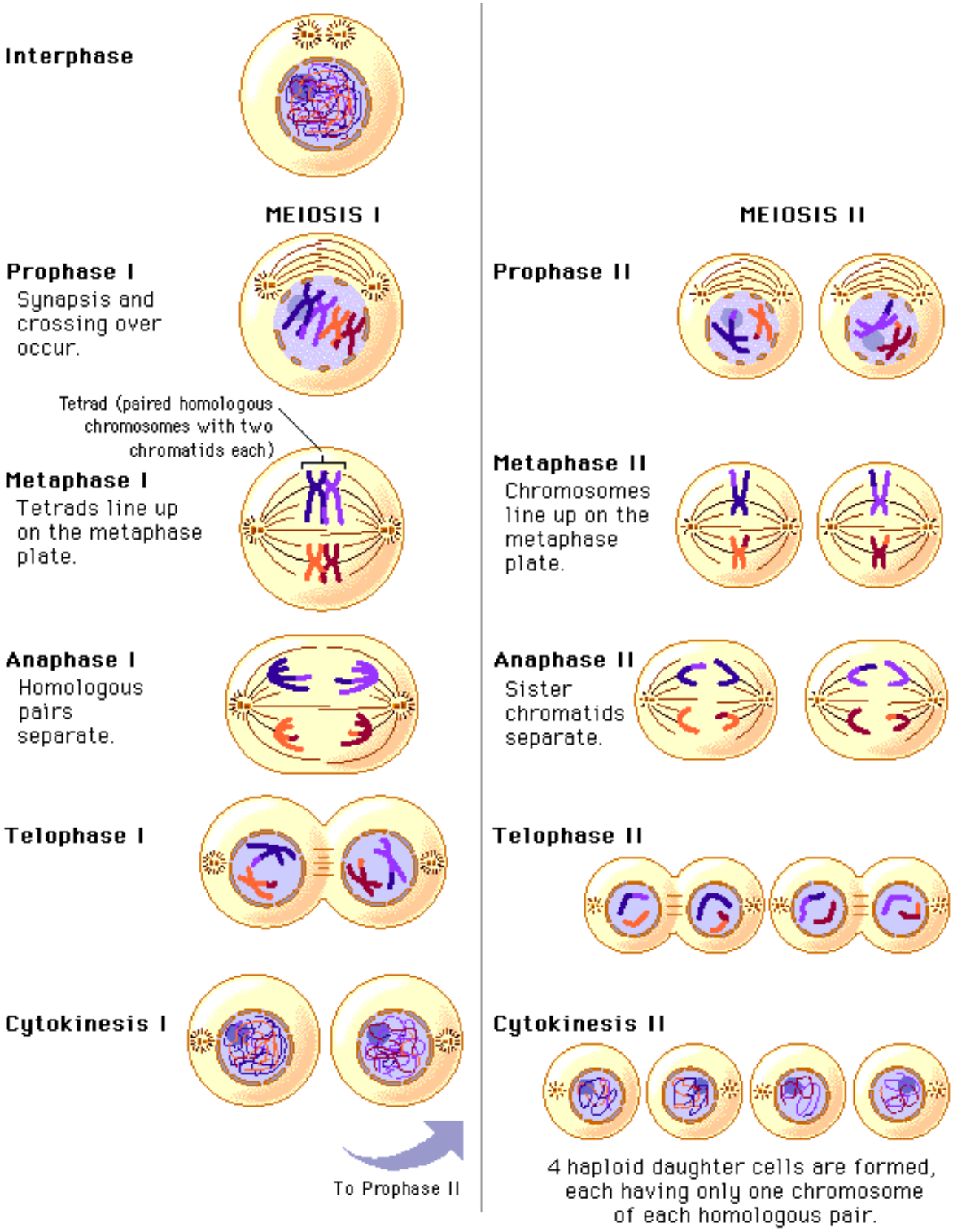
● PROPHASE II
- Nuclear membranes break down.

● METAPHASE II
- Chromosomes align at the centre of the cell.

● ANAPHASE II
- Network of spindle fibres separate SISTER CHROMATIDS o 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).



Assess the effect of the cell replication processes on the continuity of a species

GENETIC VARIATION

Arises as a result of chromosomes at 2 stages

1. during ***crossing over***

2. When chromosomes ***randomly segregate*** and paternal and maternal chromosomes assort independently of each other

During Meiosis I

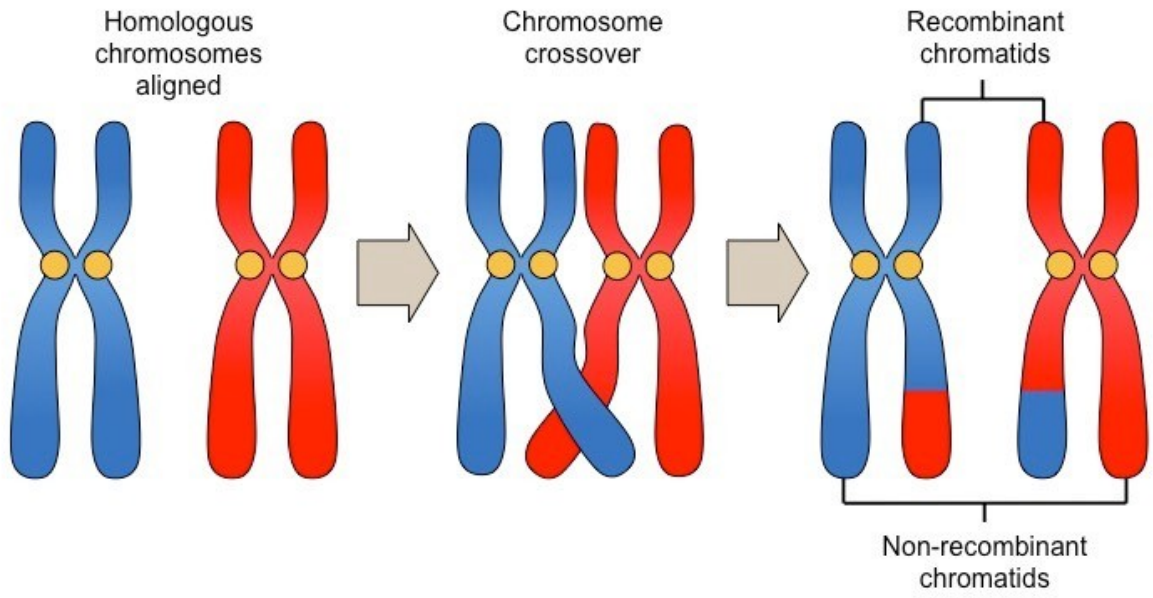
a. ***Chromosomes line up*** in homologous pairs during ***prophase I***

b. ***Crossing over*** occurs arms of homologous chromosomes exchange genetic materials during ***metaphase***

- Crossing over ensures that linear genes can be inherited independent of each other
- Exchange of genetic material causes the mixing of paternal and maternal genes
- This results in high genetic variation (increased number of combination of genes that may be transmitted by gametes)

c. ***Chromosomes in each pair separate during anaphase I***

- One chromosome of each pair moves into a daughter cell
- This 'random segregation' ensures the chromosome number in resulting gametes will be half of the original cell i
- Independent assortment occurs (paternal and maternal chromosomes saw themselves independently)



During Meiosis II

- 2 daughter cells resulting from meiosis I undergo meiosis II (similar to mitosis)
- Behaviour of chromosomes doesn't further affect gene variation

Consequences:

- Produces 4 half cells
- Combination of parent genes

Role of gamete formation and sexual reproduction in variability of offspring

- Gamete formation results in having of a chromosome number diploid to haploid
- Sexual reproduction results in combined gametes (haploid to diploid) (**n)** to produce a new diploid Organism (2N)
- Sex cells have a random assortment of dominant and recessive genes
- Resulting embryo has a completely different set of genes from either of their parents

DNA AND POLYPEPTIDE SYNTHESIS

Construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes

Prokaryotes	**Eukaryotes**
DNA in cytoplasm, specifically at nucleoid	DNA in nucleus, some located in mitochondria, some in chloroplast (for plants, protists)
Circular chromosome contains DNA DNA not wrapped around proteins	Linear chromosome in nucleus DNA in mitochondria are circular DNA in chloroplast can be linear or circular
DNA sequences aren't repetitive	DNA sequence is more repetitive
Transcription and translation stages of protein synthesis happen at the same time in the cytoplasm	Transcription (in nucleus) and translation (in cytoplasm) does NOT occur simultaneously
Contains plasmids (DNA molecules)	Does not contain plasmids
Most prokaryotes only contain one allele per gene	Most eukaryotes contain 2 alleles per gene

Protein synthesis

Intro to genes

- Order of bases in DNA is ***instructional manual*** for when cell wants to make proteins
- ***Gene = a portion of DNA***, determines order of amino acids (made up of polypeptide chains)
- Each set of 3 bases codes for 1 amino acid in the polypeptide chain (same triplet=same amino acid)
- Order of codons and bases within determine which amino acids are produced and the order
- ***Translating DNA is like code breaking and the code is the template***
- Specific codon ALWAYS corresponds to a specific amino acid BUT different codons can code for the same amino acid (only 20 amino acids, 64 possible codons)

Model the processes of polypeptide synthesis including: Transcription and translation

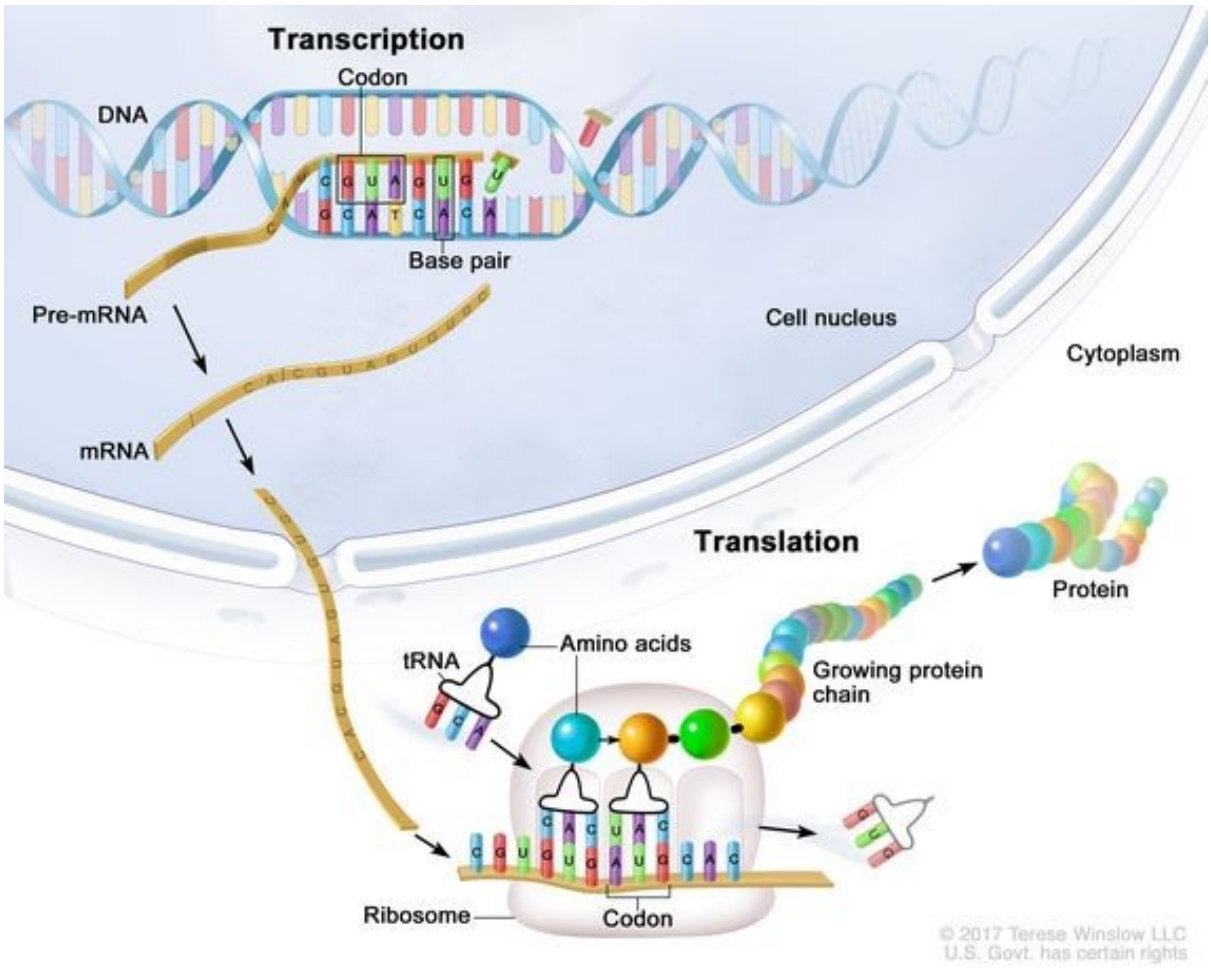
Transcription

- ***mRNA copy of a gene is made, DNA is used as a template***
- Occurs in ***Nucleus of cell***
  1. ***RNA polymerase attaches to DNA*** at desired gene and ***separates strands*** to expose the nucleotides in that region
  2. ***1 strand is used as a template to make mRNA strand identical to the other strand***
  3. Free floating nucleotides ***pair*** with complementary bases
  4. A-T, G-C, ***Uracil replaces thymine to bond with Adenine*** (this is because it is making an mRNA strand not a DNA strand)
  5. DNA strands re-join
  6. ***DNA polymerase adds RNA nucleotides one at a time***

7. In eukaryotes, introns are spliced out of the strand and exons are stuck together to form final mRNA strand (introns don't code for proteins so are swapped with exons)
8. ***mRNA*** molecules ***leave the nucleus*** and enter the cytoplasm through a nuclear pore (small gap in nuclear envelope).
9. This begins stage 2: translation

Translation

- Occurs in ***cytoplasm***
- ***Nucleic acid is translated into a protein, a polypeptide chain is formed***
  1. ***mRNA attaches*** to a ribosome at a particular start codon
  2. ***tRNA molecules with correct anticodon (***triplet of bases that corresponds to specific mRNA codon) ***pair up with mRNA*** in the ribosome
  3. 2nd tRNA molecule attaches to the next codon on the mRNA strand
  4. Ribosome catalyses ***formation of a peptide bond between 2 amino acids (***1st tRNA molecule moves away from ribosome, leaving amino acid)
  5. Ribosome continues to move along mRNA until it reaches a stop codon and is instructed to finish
  6. Polypeptide chain and mRNA strand are ***released*** from the ribosome (lion king moment lol)



mRNA= protein= Uracil replaces thymine, bonds with Adenine

DNA= mitosis/meiosis = G-C, A-T

HELICASE

- Enzyme that separates DNA strands (thus creating the replication fork)

RNA PRIMASE

- Initiates replication of DNA

DNA POLYMERASE

- Knits nucleotides together

Assessing the importance of mRNA and tRNA in transcription and translation

Importance of mRNA

- Correct gene allows the current mRNA to specify the correct tRNA.
- This way it carries a specific amino acid to bind with the matching mRNA codon
- Ensures the right amino acid sequence of the resulting polypeptide chain, hence the ***correct protein***

Importance of tRNA

- Ensures the anticodon specifies and binds to the correct amino acid
- Ensures that the resulting polypeptide chain will have the right amino acid sequence, allowing the ***protein folding process*** to occur correctly
- Thus: ***essential for shape and function***
- Eg; enzymes are made of protein. Without them, metabolic processes (eg; cellular respiration) would not occur

mRNA	VERSUS	tRNA
mRNA serves as the messenger between genes and proteins		tRNA carries specified amino acid into the ribosome for protein synthesis
Functions at the nucleus and the cytoplasm		Functions at the cytoplasm
Carries a codon sequence which is complementary to the codon sequence of the gene		Carries an anticodon which is complementary to the codon on the mRNA
Carries an order of sequential codons		Carries individual anticodons
Is a linear, single-stranded molecule		Is a L-shaped molecule
Size depends on the sizes of the protein coding genes		About 76 to 90 nucleotides long
Destroyed after the transcription		Reactivated by attaching to a second amino acid specific to it after releasing its first amino acid during translation

Analysing the function and importance of polypeptide synthesis

Function and importance of polypeptide synthesis

- Provides main component for ***proteins***
- Produces ***highly specific enzymes***
- Produces ***highly specific antibodies*** (necessary for defending against pathogens such as bacteria)
- Produces ***haemoglobin*** (a protein molecules which increases the amount of oxygen our red blood cells can carry to our cells for cellular respiration)

Assessing how genes and environment affect phenotypic expression

The influence of the environment on hydrangeas

- pH of soil they are grown in determines colour
- blue if pH is less than 5 (acid soils)
- pink if pH is greater than 7
- this is due to the availability of other ions in the soil
- other example: growth conditions influencing height of plants

Investigate the structure and function of proteins in living things

STRUCTURE AND FUNCTION OF PROTEINS

- a chain of amino acids joined by polypeptide bonds.

Function:

- Major component of every cell
- Proteome
- Main groups are structural proteins, enzymes, hormones and immunity
- **Structural proteins** : maintain cell shape, important for growth, repair and maintenance of tissue (eg; collegian)
- **Enzymes**: biological catalysts, living proteins which speed up chemical reactions. Act on specific substrates to either break it down or combine them (eg; sucrase breaks sucrase down into glucose and fructose)
- **Hormones**: proteins secreted into blood by endocrine cells (eg; glands)
- **Immunity**: antibodies which react with antigens to help remove them from the body and fight off infection

Test for proteins:

- Biuret test
  1. Add sodium hydroxide to sample
  2. Add copper sulphate
  3. If it is purple, then a protein is present
  4. If blue, there is no protein

4 LEVELS OF STRUCTURE

Primary structure

- Amino acids held together by peptide bonds

Secondary structure

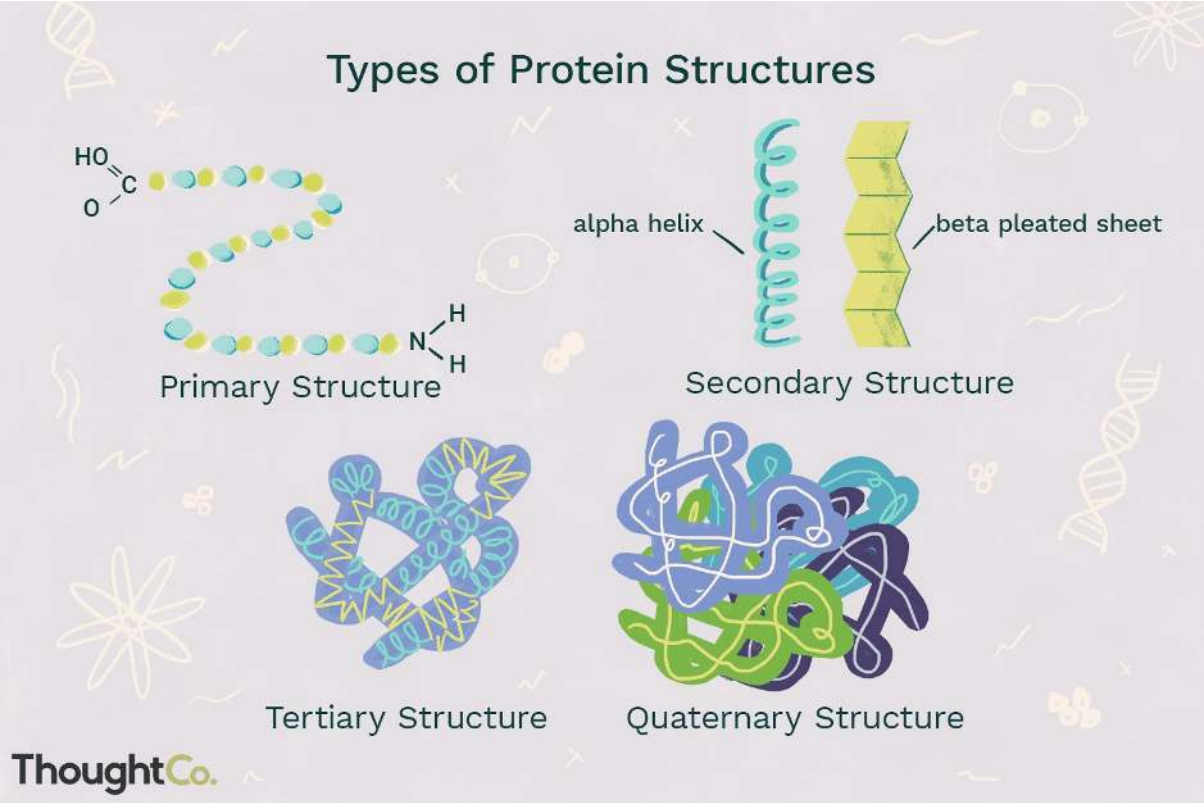
- Each polypeptide chain will ***coil up*** into helixes by forming more hydrogen bonds between carboxyl and amine groups
- Coiling up occurs due to ***hydrogen bonds***

Tertiary structure

- The coiled-up polypeptide chains further coil up to form an ***irregular three-dimensional structure***
- Provides shape and thus function

Quaternary structure

- Different polypeptides interact with other polypeptides ***forming a functional protein*** such as an enzyme
- The bonding involves ***hydrogen bonding***



GENETIC VARIATION

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

Gregor Mendel

- From Austria in 1840s
- 1957, began breeding garden peas
- 1860s, formulated principles of genetics
- Chose garden peas because they were easy to grow, produced new generations quickly and had easily distinctive characteristics

His experiments with garden peas

- Ensured self-fertilisation by placing bag over flowers to ensure pollen from stamen landed on carpel of same flower
- Ensured cross fertilisation by cutting off stamen before pollen was produced then dusting carpel with pollen from other plant
- Ensured reliability by using thousands of plants in each experiment
- Worked with true-breeding plants (self-fertilising, identical offspring)
- Cross-fertilised 2 true-breeding plants, produced F1 generation
- F1 generation was then self-fertilised or cross fertilised to produce F2

The results

- Each traits had dominant and recessive factor
- 2 true breeding = dominant factor present in first generation.
- Recessive factor present n second generation in a 3:1 relationship

	Flower Colour	Plant Height	Seed Color	Seed Shape	Pod Colour	Pod Shape	Flower Position
Dominant Trait	 Purple	 Tall	 Yellow	 Round	 Green	 Inflated (full)	 Axial
Recessive Trait	 White	 Short	 Green	 Wrinkled	 Yellow	 Constricted (flat)	 Terminal

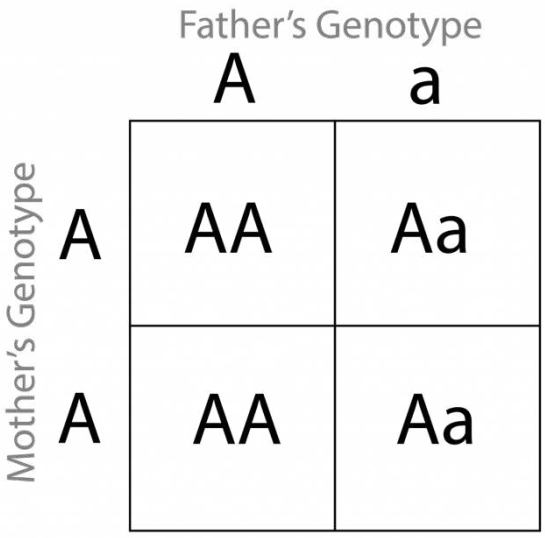
Constructing and interpretating information and data from pedigrees and punnet squares

Outcomes of monohybrid crosses

- Involve 1 factor (eg 1 tall and 1 short plant)

Homozygous vs heterozugous

- Homo=same
- Hetero=different
- Fully expressed = dominant
- No effect = recessive



Allele vs gene

- Allele= alternative form of gene
- Gene = section of DNA coding for proteins, expressed as phenotype
- Sometimes mean the same thing

Phenotype = outward appearance (blue eyes)
Genotype = allele present (TT)

Model the formation of new combinations of genotypes produced during meiosis, including but not limited to:

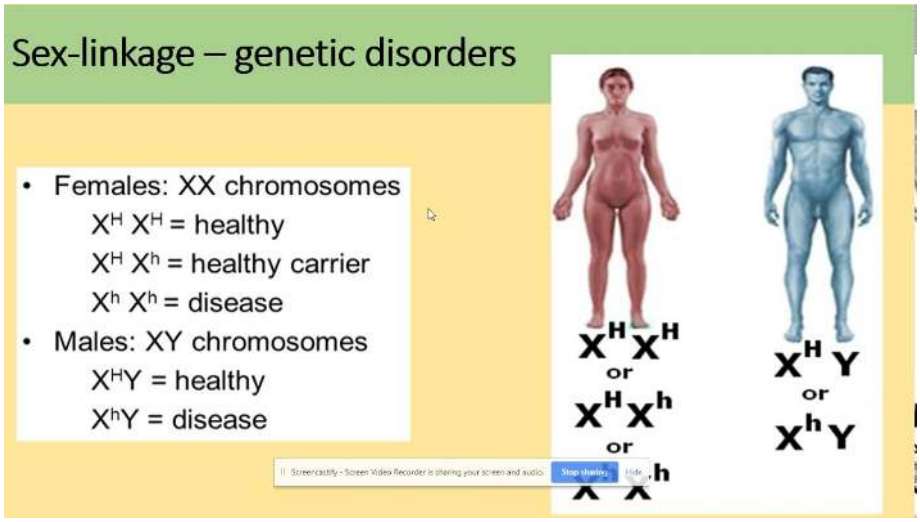
- **Interpreting examples of autosomal, sex-linkage, co-dominance, incomplete dominance and multiple alleles**

Sex linkage

- Every cell in the human body contains 23 pairs of chromosomes (22 autosomes and 1 pair of sex chromosomes)
- Females = XX (homogametic)
- Males = XY (heterogametic)

Sex linked genes

- Males lack 1 X chromosome and thus have 1 allele for each sex linked gene (females have a pair of alleles)
- Eg; red-green colour blindness is sex linked
    - Gene carried on X chromosome, no supporter on Y
    - Males only need 1 allele
    - Females require 2 to have the trait
    - Thus; more males are colourblind

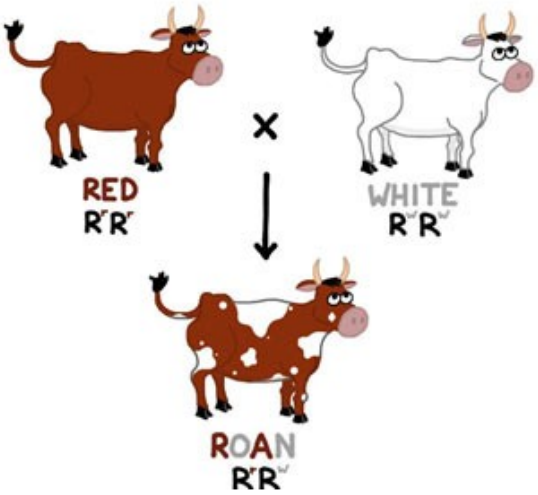


Co-dominance

- If the individual has 2 different alleles (heterozygous) for a characteristic, both alleles are expressed (upper case letters!!!)
- Eg; cattle
  - R^rR^r and R^wR^w = R^rR^w
  - (Shorthorn cattle have alleles for both red and white hair thus they are red with white patches, called Roan)
- Eg; Human Blood Types

ALLELES PRESENT	BLOOD TYPE
AA or A0	A
BB or B0	B
00	O
AB	AB

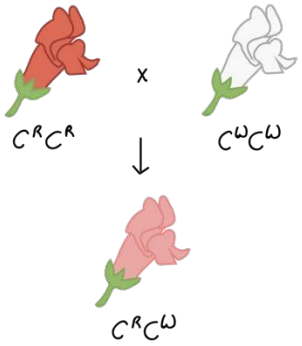
  - O is recessive to A and B, A and B are co-dominant and form AB



Blood Type	Genotype	
A	i^Ai i^Ai^A	AA AO
B	i^Bi i^Bi^B	BB BO
AB	i^Ai^B	AB
O	ii	OO

Incomplete dominance

- Both alleles are dominant
- Blends the 2 traits
- Eg; Pink Snapdragon (red allele and white allele, makes pink flower)

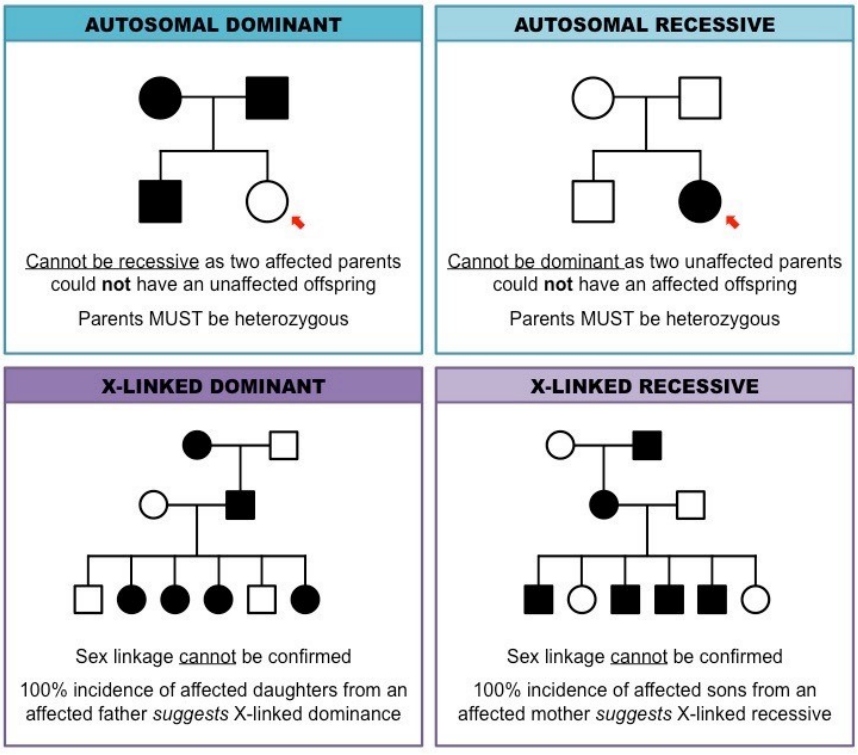


Constructing and interpreting information and data from pedigrees and punnet squares

- Circle = female
- Square = male
- Filled in = displays phenotype being studied
- Likely dominant if:
  - Heterozygous individuals affected
  - 2 heterozygous parents produce unaffected child (50% chance)
    - Affected offspring must have AT LEAST 1 affected parent
  - Dad and daughter have trait
- Likely recessive if:
  - The trait skips a generation
  - Mother and son have trait

Autosomal or x-linked?

- X-linked recessive = males more commonly affected
- Autosomal = males and females are equally affected
- NEVER passed from father to son

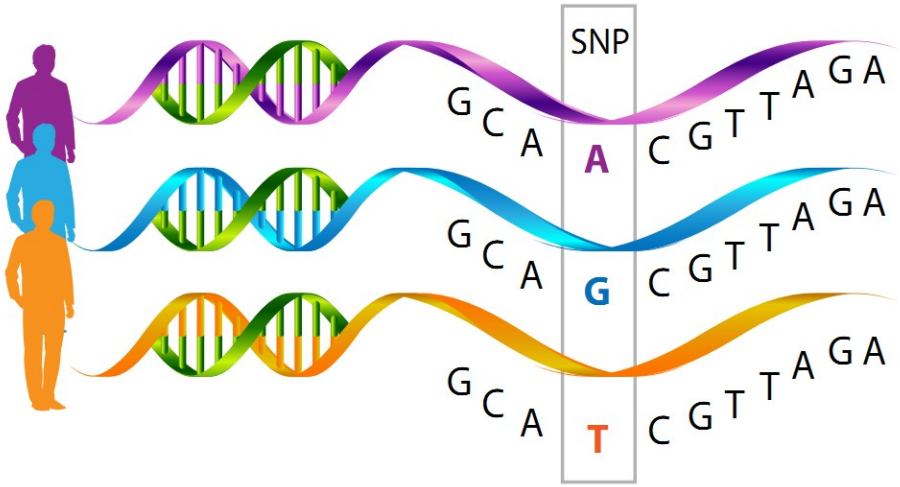


INHERITENCE PATTERNS IN A POPULATION

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.

Analysing single nucleotide polymorphism (SNP 'snips')

- SNP's can generate biological variation by causing differences in genes coding for specific proteins
- These can influence traits such as appearance, disease susceptibility or response to drugs
- Most SNP's have no observable difference
- SNPs versions are inherited frm your parents. Siblings are matched
- Each SNP represents a difference in a single DNA building block called a nucleotide
- A gene sequence is considered an SNP if it occurs in at least 1% of the population
- SNP occurs where 1 nucleotide is different to the rest of the nucleotides at the same locus of the choromosome
- This is due to the new nucleotide randomly replicating or substituting an existing nucleotide in the organism's DNA sequence
- SNPs are responsible for over 90% of human genetic variation

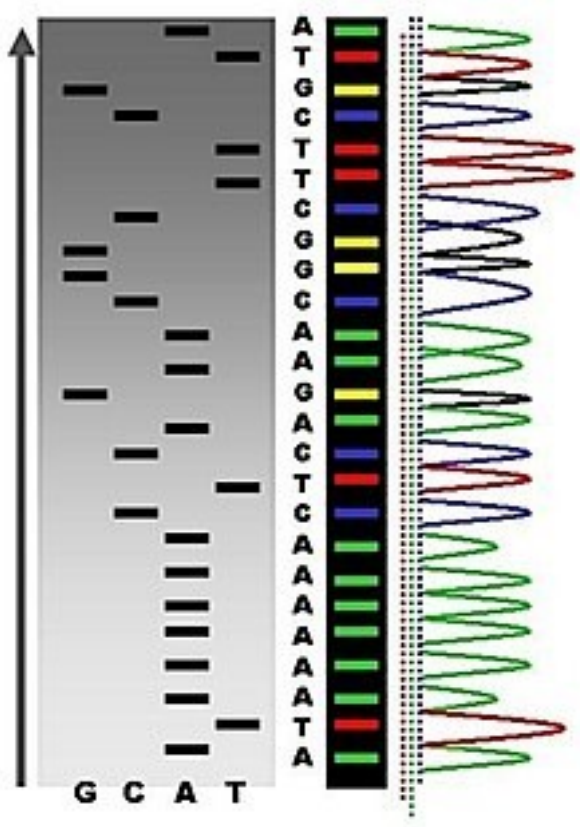


How can SNPs be useful?

- Scientists need to identify SNPS that correlate with a particular effect in a pattern
- Reliable SNPs could serve as predictive markers. They inform our decisions about numerous aspects of medical care, including specific dieases, effects of various drugs and advanced reactions to specific drugs
- This can save time, money by matching patients with appropriate medication based off diagnosis

DNA sequencing

- Scientists take piece of DNA and determine the sequence of letters it contains in an effort to use it/find out more about its function
- Complete sequence is called your genome (uniqye)
- Provides insight into causes of human disease, essential for identification of nucleotide sequence
- This allows scientists to determine if there is a mutation responsible for the diseas
- Eg; 20 possible genes assocated with late stage Alzhemer's
- By identifying the exact nucleotide sequence of these 20 genes any SNP or mutation can be identified and analysed as possible causes
- Also identifies organisms, their evolving relationship with each other and potential drug target
- Basic method is Sanger sequencing (uses natural process of DNA replication to identifies the precise order of nucleotide)



DNA profiling

- Does not attempt to determine sequence
- The goal is to determine whether a sample of DNA containing material like blood came from a given individual

Techniques in DNA profiling

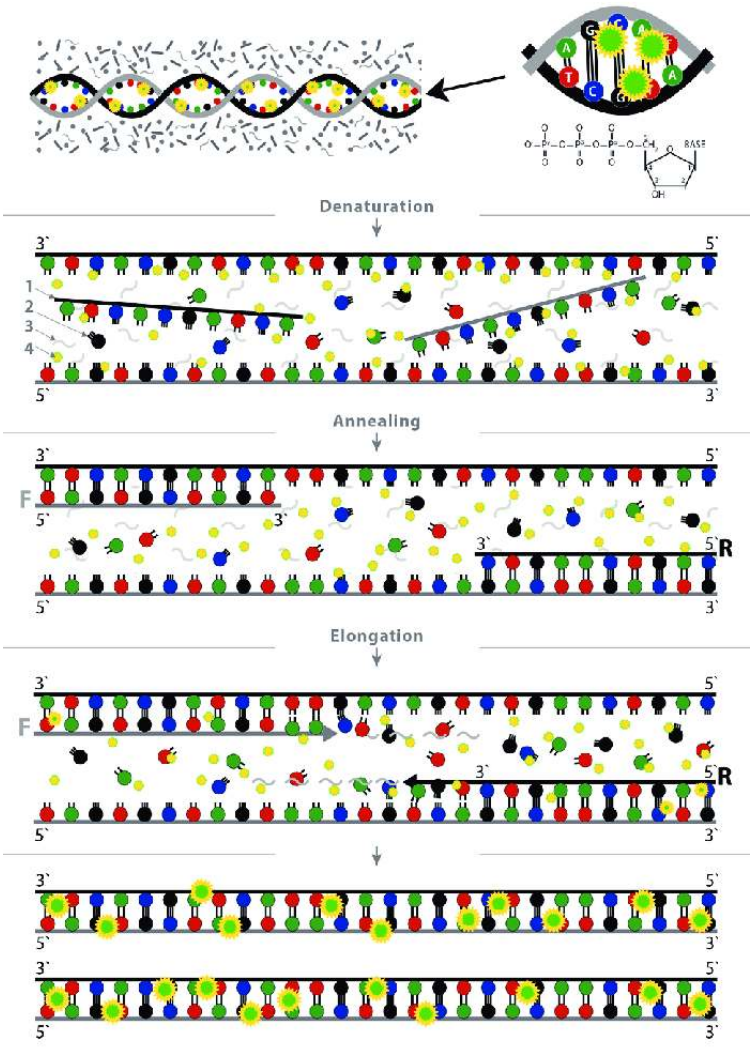
- Fragmentation
- PCR
- Gel Electrophoreiss

Fragmentation of DNA using restriction enzymes

- Within non coding regions of an individual's genome there are long stretches of DNA made up of replicated sections called short tandem repeats (STRs)
- These repeats generate unique DNA profiles
- Every human may have the sequence CAGT but it definitely differs in quantity (for example I could have it 10 times and my friend could have it 15)

Polymerase Chain Reactions (PCR's)

- Makes millions of copies of a gene, outside the cell, emplifies sequences through cycles of replication, heating and cooling

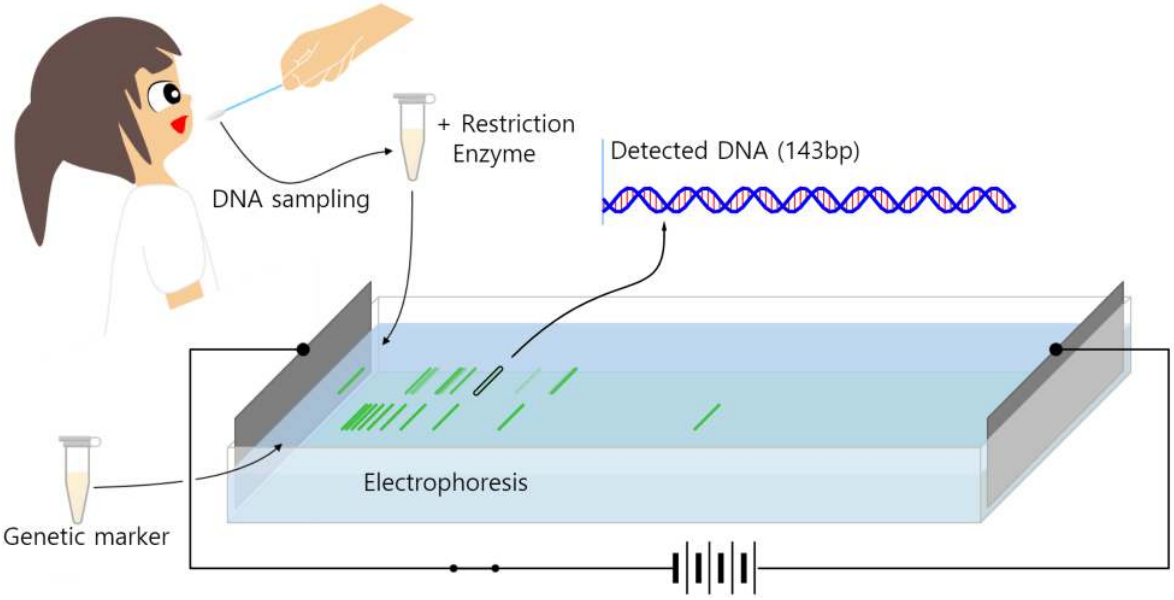


1. Denutaration
   - 96 degrees
   - Heat the reaction strongly to separate/denature the DNA strands, provides single stranded tamplate
2. Annealing
   - Cool the reaction so the primers can bind to complementary sequences
3. Extension/elongation
   - 72 degrees
   - Raise the reaction temperature so TAQ polymerase extends the primers synthesising new strands of DNA

This repeats hundreds of thousands of times (taking days or weeks depending on length of DNA region being copied)
It is used in both sequencing and profiling and is practical in forensics, genetics and diagnostics

Electrophoreiss

- DNA samples into an agrose gel, electric current through the DNA samples
- Fragments are attracted to the negative terminal of the machine (DNA is polar, backbone is positive)
- Scientsts can analyse the ways fragments move
- Shorter segments move farther away from their original location or longer ones stay closer



Conservation management

- Scientists study frequency of alleles for one or more genes in a population
- Objective is to ensure that the species of conern can adapt to changing selective pressures
- Lack of genetic diversity is directly proportional to probability of extinction
- Example: ALBANY CYAD PLANTS
    - Habit is being eaten and disturbed, now in low numbers
    - Purpose was to study determination of genetic structure and decide on degree of susceptibility to extinction
    - DNA profiling showed little genetic variation between subpopulations thus susceptible to extinction

- Example: KOALA GENE DECODED
    - Population threatened by disease and climate change
    - Cross over between regions
    - Aim was to decrease dog attacks and track genome and vaccination success

Population genetic studies used to determine inheritance of a disease

- Measure allele frequencies, giving image of number of alleles lost during evolution and number transferred through gene flow
- Gives image of probabilities of inheritance disorders within a population
- EXAMPLE: ALZHEIMER'S

- More than 20 new genes are possible causes of late development of Alzheimer's disease (AD)
- Highlighted similarities between early onset and late onset AD (suggesting beneficial treatment for one can be beneficial for other)
- Autosomal dominant, one copy of altered gene in each cell is sufficient to ause the disorder
- mainly inherited from 1 affected parent
- International Genomics of Alzheimer's Project (IGAP)

Population genetics relating to human evolution

- Anthropological genetics combines population genetics (eg; DNA analysis) with historical and linguistic evidence
- Determines pathways of human evolution and explains causes of human diversity (eg; mutation, natural selection, genetic drift, gene flow)
- Objective is to determine origin of modern human civilisation
- EXAMPLE: ANTHOPOLIGICAL GENETICS
    - 98.8% DNA shared with chimpanzees and bonobos
- Mitochondrial DNA (mtDNA) traces evolutionary relationships within a species
- Helps divide human population into distinct haplogroups
- Humans can trace origins back to one of 7 super haplogroups origination from an original 'Mitochondrial Eve'