

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
- analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals (ACSBL075)  
- evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture (ACSBL074)  

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 (ACSBLO75)  
 - DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding (ACSBLO76, ACSBL077)
- assess the effect of the cell replication processes on the continuity of species (ACSBLO84) 

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 (ACSBLO76) 
- model the process of polypeptide synthesis, including: (ACSBLO79)
 - transcription and translation
 - assessing the importance of mRNA and tRNA in transcription and translation (ACSBLO79)
 - analysing the function and importance of polypeptide synthesis (ACSBLO80)
 - assessing how genes and environment affect phenotypic expression (ACSBLO81)  
- investigate the structure and function of proteins in living things 

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 (ACSBLO84)
- 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 (ACSBLO85) 
 - constructing and interpreting information and data from pedigrees and Punnett squares
- 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
 - analysing single nucleotide polymorphism (SNP)

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: (ACSL064, ACSL085)
- DNA sequencing and profiling (ACSL086) 
- investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example: (ACSL064, ACSL073)    
- 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 

MODULE 5: HEREDITY

ASEXUAL - 1 Parent, genetically identical
SEXUAL - 2 Parents, genetically unique

Mechanisms of reproduction:

Animals: Sexual Reproduction

Internal Fertilisation -

- + more likely to occur
- + embryo protected from predators
- + higher chance of survival for offspring
- more energy required
- less offspring produced
- more energy to care for young
eg mammals, reptiles

External Fertilisation

- + little energy for mating
- + large amount of offspring
- + can spread widely → less competition
- many unfertilised gametes
- lesser chance of survival for offspring
eg. fish, amphibians

Asexual Reproduction

- Budding - coral
- Regeneration + fragmentation - sea sponges
- Parthenogenesis (female gamete without male) - honey bees

Plants

Sexual Reproduction

- genetically unique offspring + increases variation
 - flowers - pollination → male pollen grains from Stamen combine with female ovum from Pistil

Asexual Reproduction

- + will be well suited to environment
- Tuber, rhizomes, vegetative propagation, cuttings
- Bulbs - storage organ consists of short stem + fleshy leaves
 - ↳ onions
- Runners - modified stems that grow close to the ground
 - ↳ strawberries + spinifex

Fungi

Asexual Reproduction

- Budding — the nucleus divides & a bulge forms on side of the cell which is split by cytokinesis. the bud detaches itself from parent cell
 - + energy efficient, rapid growth
 - ↳ yeast cells
- Spores — unicellular reproductive cells — mitosis produces genetically identical cells which are distributed in environment by wind or vectors.
 - ↳ puffball mushroom
 - + survive adverse conditions, no sexual interaction needed.
- Also through fragmentation

Protists

— eukaryotes that are not animals, plants or fungi eg. mould, amoeba.

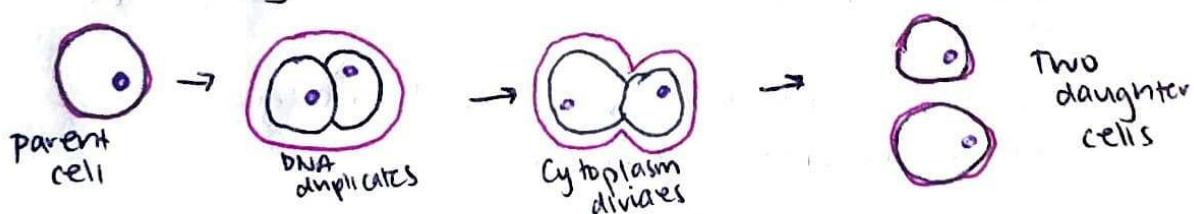
Asexual Reproduction: • Binary Fission — nucleus divides into two, cell grows until divides into half for two daughter cells — amoeba.

- Also by budding or conjugation (sexual reproduction)
- Budding — new organism grows from body of parent organism to form a colony — protzoa
 - + exponential growth, adapted to environment.

Bacteria

Asexual Reproduction: Binary Fission

• Parent cell divides to 2 daughter cells, DNA replicated and divided into two. Cell elongates + splits into 2 daughter cell. — staph, e-coli



- Also by conjugation — one bacterium transfers gene copies to another through direct contact. — e-coli
 - + time efficient, exponential growth

Manipulation of Reproduction in Agriculture

- using fundamental principles of reproduction to produce desired outcomes.

Selective breeding — choosing ^{animals /} plants to breed with specific phenotypic traits that are favoured.

- ↳ producing favourable offspring

Artificial Insemination (Animals) — inseminating the semen of a male w/ specific traits into a female - cows

- ↳ allowed by understanding of fertilisation

Artificial Pollination (Plants) — fertilising the ovules of one plant with pollen from another plant - flowers.

- ↳ produce desired traits.

◦ Genetic engineering — manipulate organisms due to DNA knowledge.

Genetic Material Replication

Effect on continuity of species:

- can help save fertile life - continue species
- brings genetic stability
 - ↳ new cells / organisms have enough genetic information to survive
- way of preserving genes across generations
 - lack of genetic continuity may result in disease

Watson + Crick Model

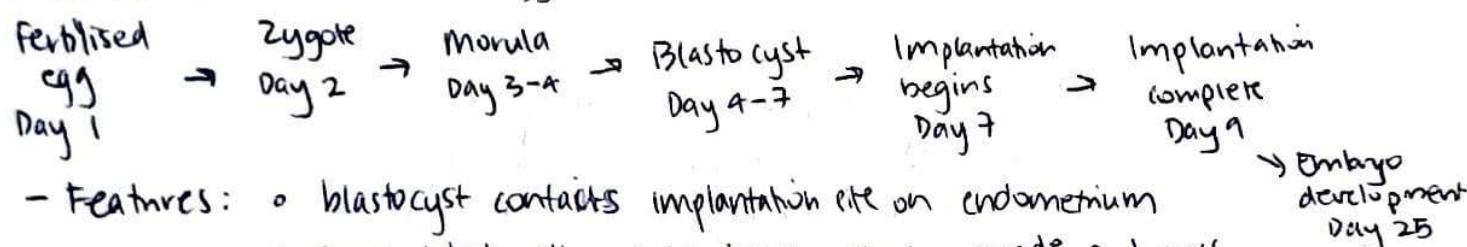
- Double helix twisted model - template for each other

Mammal Reproduction

Fertilisation - fusion of sperm + egg in fallopian tube

- Features:
 - sperm makes way through layers of egg
 - reaction occurs within egg, plasma membranes fuse
 - sperm nucleus enters egg
 - vitelline layer becomes impenetrable to unsuccessful sperm.

Implantation - fertilised egg adheres to wall of uterus



- Features:
 - blastocyst contacts implantation site on endometrium
 - trophoblast cells of blastocyst attach, invade and cross endometrial membrane + stroma.

Hormone Control in Pregnancy

- human chorionic gonadotrophin (hCG) produced after implantation - supports corpus luteum - peaks around 10 weeks
- Relaxin inhibits premature contractions
- Oestrogen helps thicken uterine lining + foetal growth
- Progesterone - maintains placenta function + prevents uterine contractions / movement (too early)

Hormone Control in Birth

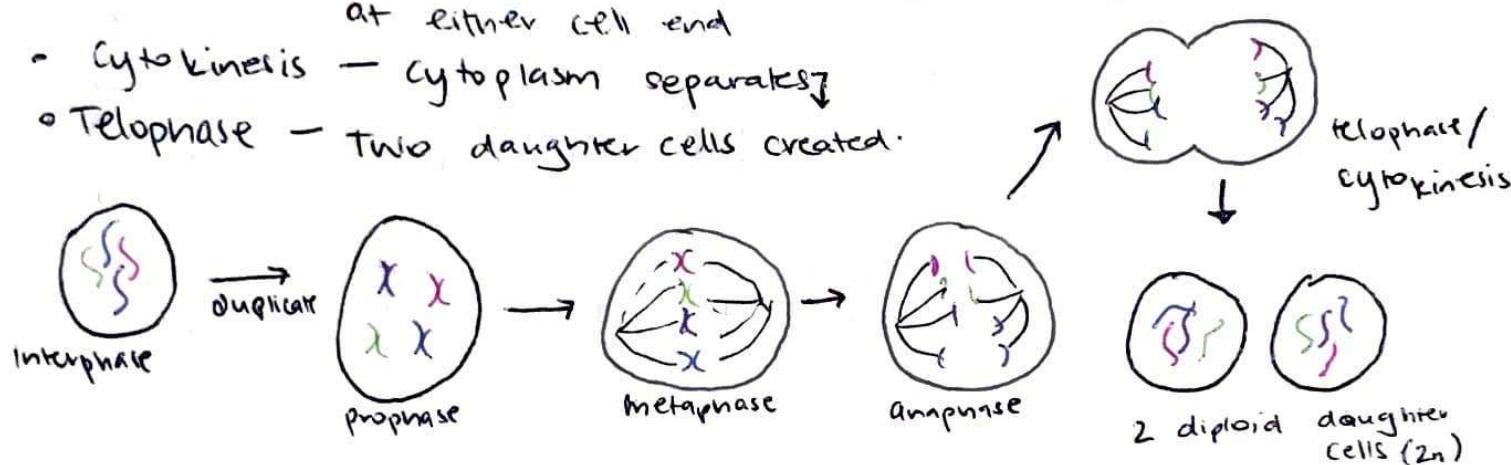
- Progesterone strengthens pelvic muscles for labour
- Oestrogen stimulates milk duct development
- Prolactin needed to produce breast milk
- Relaxin lengthens/softens cervix for birth
- Oxytocin stimulates uterine contractions.

Cell Replication:

Mitosis:

- cell division resulting in two genetically identical daughter cells - diploid ($2n$) \rightarrow somatic cells.

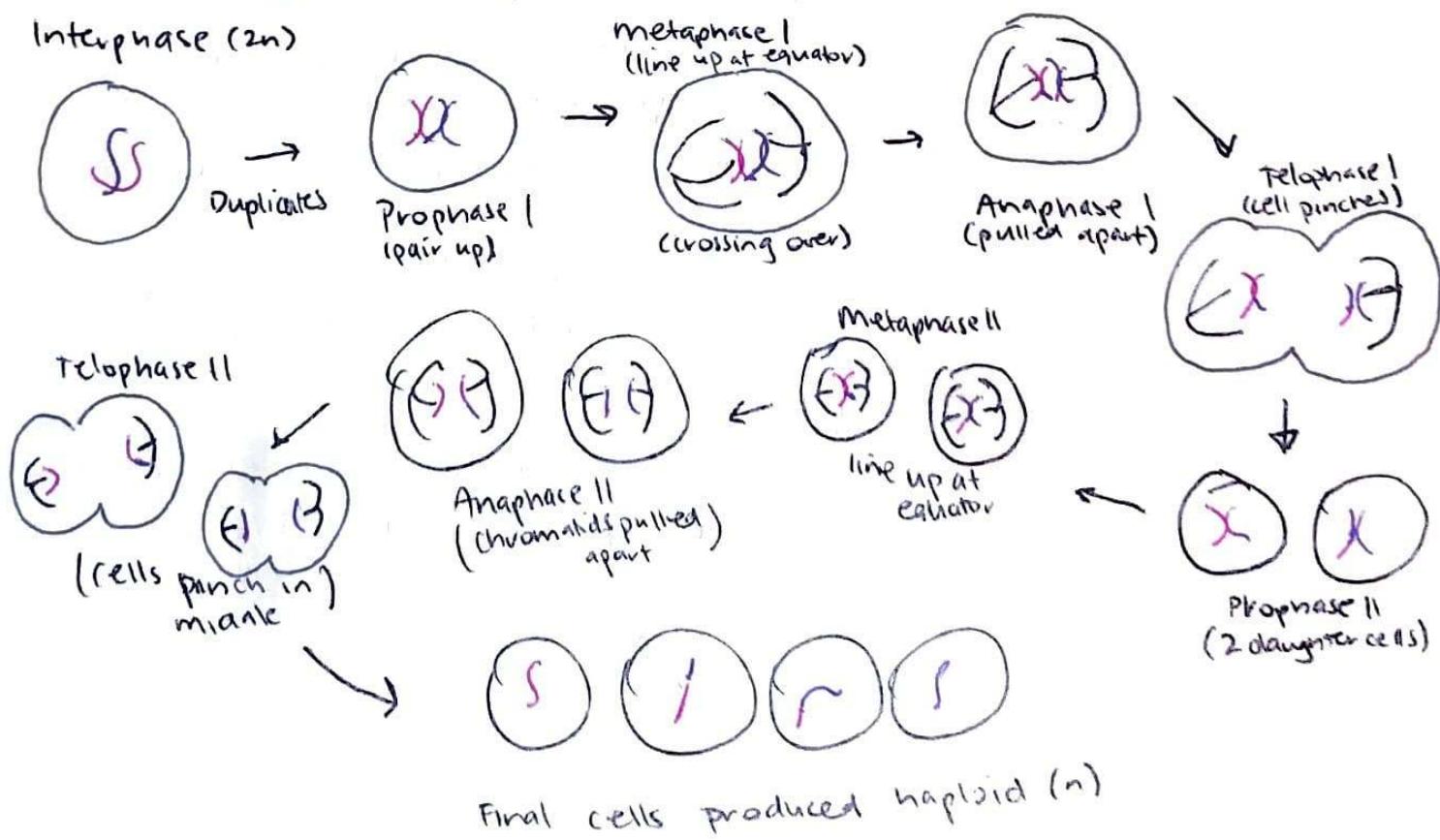
- Interphase - two copies produced of each chromosome
- Prophase - duplicated chromosomes condense, spindle forms at each end to pull chromatids apart
- Metaphase - chromosomes align in single file in centre of cell
- Anaphase - chromatids separate so only one copy of chromosome at either cell end
- Cytokinesis - cytoplasm separates
- Telophase - two daughter cells created.



Mitosis:

- cell division in gamete formation resulting in unique haploid (n) cells \rightarrow reproductive cells.

Interphase ($2n$)



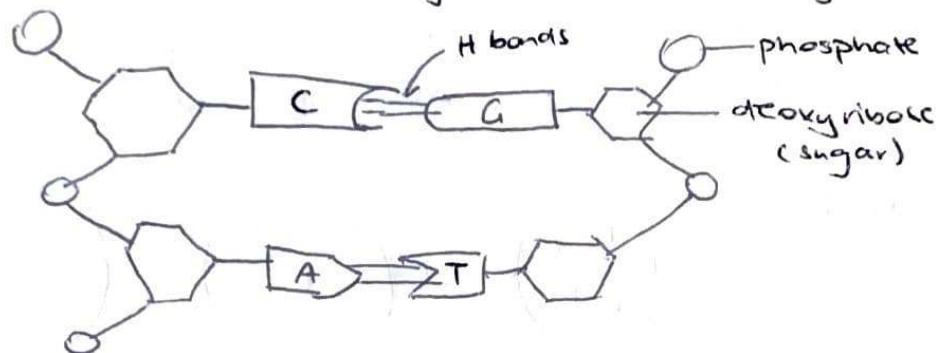
• Genetic variation in Meiosis by:

- Random Segregation \rightarrow pair of alleles segregate + each gamete receives one allele.
- Independent assortment \rightarrow alleles for each trait separate independently from other alleles.

DNA Replication

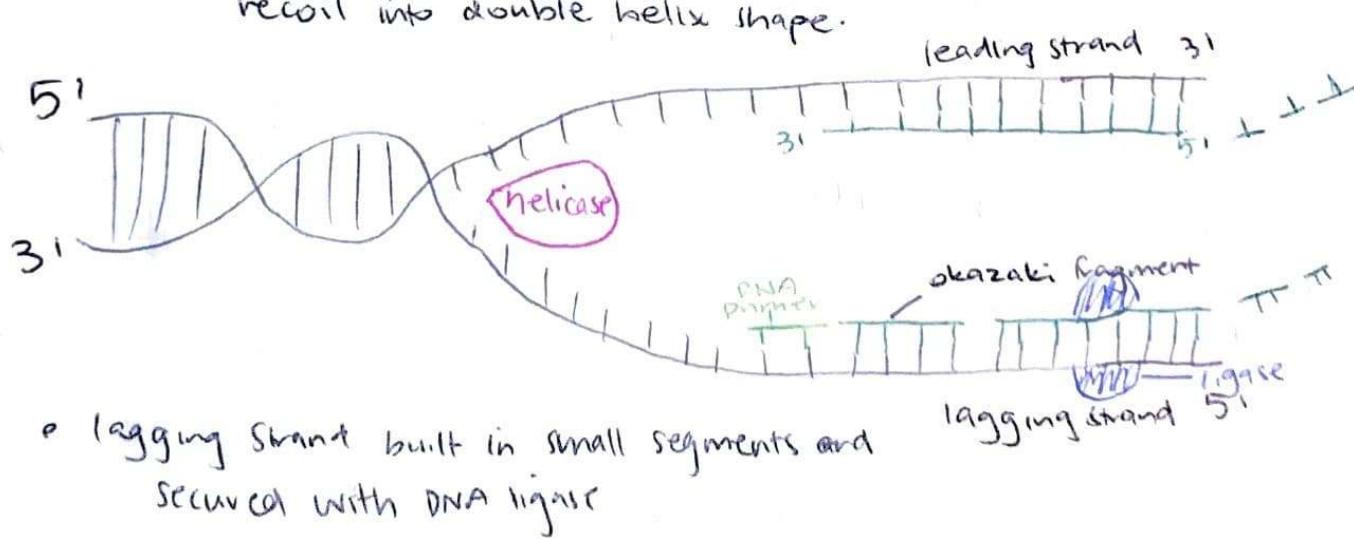
- Watson + Crick Model - Double helical nucleic acid model - carries code in nucleotide bases - A G C T

- Sugar - phosphate backbone, nitrogenous bases w/ hydrogen bonds



Replication:

- Helicase (enzyme) unwinds by breaking hydrogen bonds to 'unzip' two strands from each other
- Primers bond to end of strand to signal start point. DNA polymerase bonds to separate strands at primer site and adds new bases (complementary) \rightarrow 5' to 3' direction
- When end of molecule is reached, two strands produced recoil into double helix shape.



Eukaryotic vs Prokaryotic DNA

<u>Feature</u>	<u>Prokaryotes</u>	<u>Eukaryotes</u>
Location	<ul style="list-style-type: none">freely found in cytoplasm or nucleoid region	<ul style="list-style-type: none">contained within a membrane bound nucleus
Packaging	<ul style="list-style-type: none">not bound with histone proteins or tightly coiled	<ul style="list-style-type: none">bound to histone and tightly coiled to fit in nucleus
Shape of chromosome	<ul style="list-style-type: none">circular shape, can form loops	<ul style="list-style-type: none">linear
Genome	<ul style="list-style-type: none">compact, contains small repeats + no introns	<ul style="list-style-type: none">large amounts of repeats and introns
Chromosome number	<ul style="list-style-type: none">one which may have multiple copies	<ul style="list-style-type: none">variant number dependent on organism
Number of nucleotide base pairs	<ul style="list-style-type: none">160,000 - 12.2 million	<ul style="list-style-type: none">depend on organism e.g. humans $\rightarrow 23 \rightarrow 2.9$ Billion
Replication process	<ul style="list-style-type: none">simple + fast	<ul style="list-style-type: none">more complex + slower

Polypeptide Synthesis:

Transcription: process where complementary copy - mRNA - of a gene is made in the nucleus

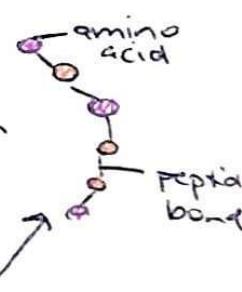
Translation: process where mRNA is converted into specific sequence of amino acids - carried by tRNA - in the ribosomes.

- proteins fundamental to all cell structure + function
 - needed to express genes
 - if error occurs, can lead to disease.

Phenotype: genes + environment

- environment does not effect/modify genotype but sometimes influences phenotype via chemical markers or tags added to DNA.
 - eg. Hydrangea flower colour + soil pH
 - blue = alkaline
 - pink = acidic
- genotype - DNA responsible for phenotypic trait, always influences phenotype coded for by DNA
 - eg. Gene for Blood type, Haemophilia or Cystic Fibrosis not at all effected by environment only genotype.
 - can combine to make phenotype

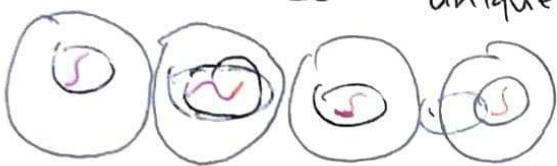
Protein Structure + Function

- amino acids → peptides → polypeptide chain → protein
- 4 levels of structure:
- Primary - linear sequence making polypeptide chain
 - Secondary - regular patterns of folding
 - eg. beta sheet
 - alpha helix
 - Tertiary Structure - overall folding to 3D shape
 - Quaternary Structure - protein of more than one amino acid chain.
- ~~less~~ function:
- essential to all biological processes
 - Structure always essential for function
 - eg. enzymes, antibodies, hormones or transport.
- 

Genetic Variation

Variation due to meiosis:

- Mutations — permanent DNA change → if occur in gametes can be inherited by offspring + form new alleles.
 - ↳ introduce new genotype/allele
 - ↳ alter certain traits by silencing particular gene
 - ↳ produce different products from similar mRNA transcripts.

eg. Sickle-cell Anaemia — autosomal recessive
— autoimmune disorder or malaria resistance.
- Crossing Over — exchange of alleles between homologous chromosomes → unique gene combination in gametes

4 unique daughter cells [haploid (n)]
- Fertilisation — each ejaculate contains ~ 300 million sperm all different due to crossing over, independent assortment & random segregation
 - ↳ only one fertilised egg → by chance.

New combinations of genotypes produced during meiosis

- Autosomal Genotype: gene combination found in non-sex chromosomes.

eg

	T	t
T	TT	Tt
T	TT	Tt

dominant eg T or
recessive eg t
50% dominant homozygous
50% dominant heterozygous.
- Sex-Linked — phenotypic expression dependent on the sex chromosome eg. Haemophilia, colourblind

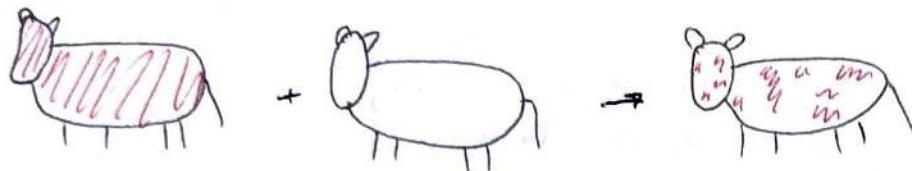
eg

	X^B	X^b
X^B	$X^B X^B$	$X^B X^b$
Y	$X^b Y$	$X^B Y$

50% heterozygous dominant - carrier female
50% homozygous recessive - affected male

- Co-Dominance - both alleles fully expressed resulting in combined phenotype.
 - ↳ heterozygous - neither dominant nor recessive

eg. White cow + Red cow \rightarrow Roan Cow



- Incomplete Dominance - one allele not completely expressed over another

↳ blending of phenotype

eg. White flower + Red flower \rightarrow Pink flower

- Multiple Alleles - Three + alternative forms of an allele can create a variant number of phenotypes / code for same gene.

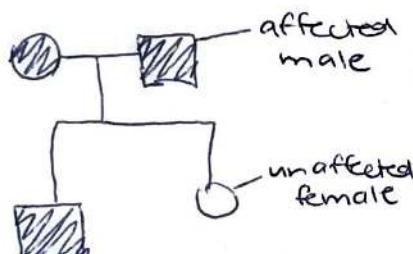
eg. Blood type

	A	B
A	AA	AB
O	AO	BO

AA + AO - type A
 AB - type AB
 BO + BB - type BO

Pedigrees:

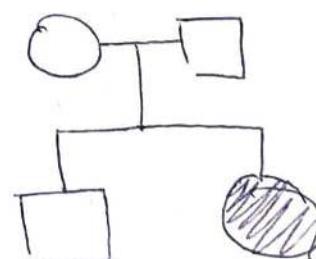
Autosomal dominant:



Parents must be heterozygous
Cannot be recessive

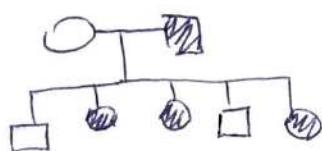
Punnett Square

Autosomal recessive

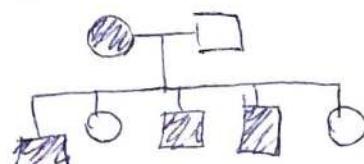


cannot be dominant

Sex-linked dominant



Sex-linked recessive



Population Frequencies

$$\text{Allele frequency} = \frac{\text{number of copies of allele in population}}{\text{total number of gene copies in population}}$$

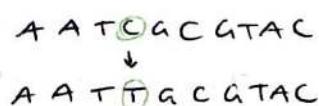
Eg. Population of 100 : 200 genes

50 are heterozygous allele A

10 are homozygous allele A

$$\text{Total } A = \frac{50 + 20}{200} = \frac{70}{200} = 35\% \text{ A allele.}$$

Single Nucleotide Polymorphism (SNP)



- change of a single nucleotide in a specific position on the genome. - Insertion, substitution or deletion.
 - ↳ must occur in at least 1% population
- Most in introns so no cell function impact
 - If in exons - may cause disease

Inheritance Patterns in a Population:

DNA Sequencing

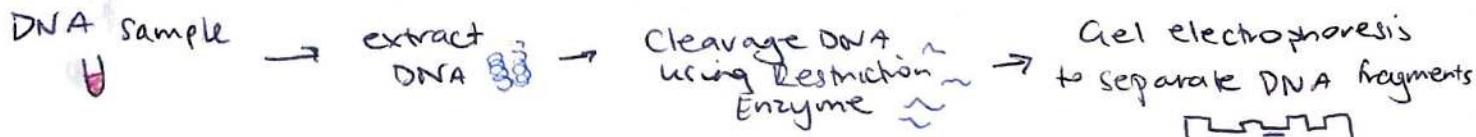
- Determines the sequence of bases.

- Methods:
- Maxim Gilbert method: chemicals used to identify specific base → electrophoresis compare patterns
 - Sanger Method: DNA is isolated + replicated using a polymerase chain reaction. The sequence is graphed by a computer.

DNA profiling

- analysing DNA using gel electrophoresis. Testing short Tandem repeats (STRs)

- used to: - confirm relation; trace inheritance or solve crimes.



Develop/visualise finger prints

$= = =$

Auto radiography
← to obtain DNA bands



Southern blotting technique



Data Analysis for trends/patterns

Conservation Management

- preserve species through maintaining genetic variation

- usually studies endangered species.

Methods:

- Field observation - distribution / abundance

- DNA analysis (SNPs, genome-wide studies)

e.g. African Cheetah

- only 7000 left in wild

- low genetic diversity

→ worldwide breeding initiatives to widen gene pool.

Inheritance of Disease / Disorder

- Technology has allowed to identify genes w/ specific diseases.

- predict possibility of offspring inheriting

e.g. SNPs for NSW newborn genetic screening - Cystic fibrosis.

Human Gene Mutation Database - store germline mutations

associated with inherited diseases. e.g. 35 genes linked to PD.

Human Evolution

- explain cause of human diversity over time.

- Study human genome

- Human migration theories:

- Multiregional - suggests gene flow

- all human life to Africa 2M y.a.

- Replacement - left Africa + second migration

- happened 100,000 y.a.

- modern humans result of interbreeding

- + out-competing.

Uncertainty:

absolute uncertainty measures range of values in which the true value lies in

$$\text{abs. uncertainty} = \frac{\text{highest possible value} - \text{lowest possible value}}{2}$$

Relative Uncertainty is absolute uncertainty as a percentage of the measured value

$$\text{rel. uncertainty} = \frac{\text{absolute uncertainty}}{\text{measurement}} \times 100$$

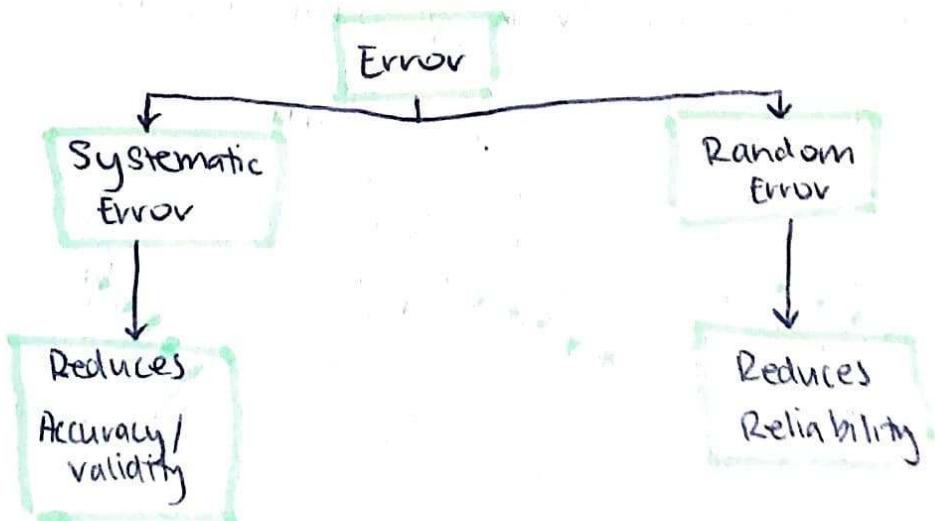
Error

Random error: shifts from true value by random amount/direction each time

- Eg \Rightarrow Reaction time, measurement errors from insufficient / not precise enough equipment.
↳ effects Precision + reliability

Systematic error: Shifts from true value by same amount/direction each time

- Eg \Rightarrow Scale error, zero/tare error, parallax error, equipment not calibrated



Reliability

- is the repeatability of the scores

- ↳ consistent when repeated under same conditions - random error will cause unreliability

Can be assessed by

- repeating experiment 4-7 times w/ same method + compare results

Improved by:

- repetition of the experiment
- Reducing random error - finding average of results/data.

Validity

- extent the results measure what was intended

- ↳ is the dependent variable being caused by the independent variable.

Can be assessed by

- comparing data with standard value
- a control variable

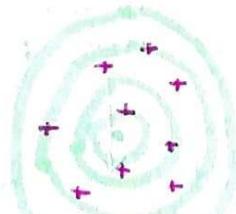
Improved by:

- Holding all other variables constant
- Use appropriate equipment
- NO plausible reason for DV change other than IV

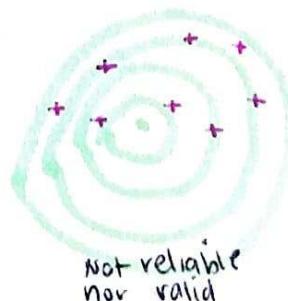
Validity / Reliability: Cannot be valid unless reliable



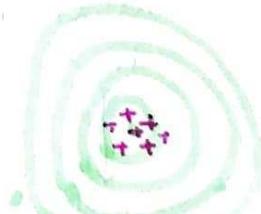
Reliable not valid



low reliability
low validity



Not reliable
nor valid



Reliable and valid

Accuracy

◦ how close the final result is to the true value

Improved by:

- making single measurements more accurate
- Reducing systematic error