Chapter 10 —> Genetics

11.1 Meiosis

Interphase:

- Period preceding meiosis and involves events needed to prepare the cell for successful division
- DNA replicated in S phase —> results in chromosomes that contain two identical DNA strands
  —> sister chromatids —> the genetically identical strands —> centromere holds them
  —> separate during meiosis II —> become independent chromosomes
- If DNA replication did not occur there would be no need for 2 meiotic divisions —> benefit is that increases genetic recombination that occurs
- Interkinesis —> may occur between meiosis I and II, but no DNA replication occurs

Stages of meiosis:

Meiosis I:

- P I —> chromosomes condense, nuclear membrane dissolves, hom. chromosomes form bivalents and crossing over occurs
- M I —> spindle fibres from opposing centrosomes connect to bivalents and align at the equator
- A I —> spindle fibres contract and split the bivalent, hom. Chromosomes move to opposites
- T I —> chromosomes decondense, nuclear membrane reform, cytokinesis (two haploid cells)

Meiosis II:

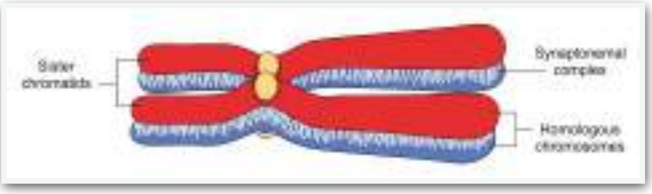
- P II —> chromosomes condense, nuclear membrane dissolves, centrosomes move to opposites
- M II —> spindle fibres from opposing centrosomes attach to chrom. and align at the equator
- A II —> spindle fibres contract and separate the sis chromatids bringing them to opposites
- T II —> chromosomes decondense, nuclear membrane reforms, cytokinesis (four haploid cells)

Random assortment:

- Independent assortment describes how pairs of alleles separate independently from one another during gamete formation —> gene inheritance is independent from other genes
- Due to the random orientation of homologous chromosomes in meiosis I —> metaphase I
- Independent assortment won't occur if two genes are on the same chromosome (linked genes)

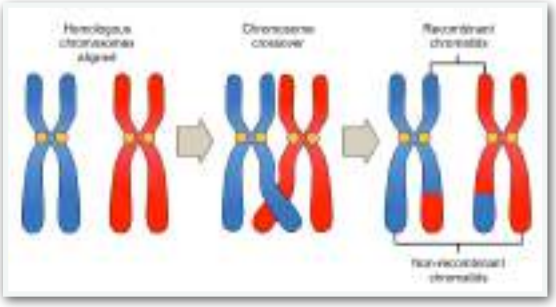
Chiasmata:

- Synapsis —> when homologous chromosomes become connected —> during prophase I
  —> bivalent —> two chromosomes or tetrad —> four chromatids



- Synaptonemal complex —> protein-RNA complex that connects chromosomes
- Chiasmata —> where non-sister chromatids remain physically connected at —> hold hom. chromosomes together as a bivalent until anaphase I
  —> forms as a result of crossing over and non-sister chromatids can show an exchange of genetic material

Crossing over:



- DNA can be exchanged between non-sister homologous chromatids —> if it happens the chromosomes become recombinant chromosomes
- Produces new allele combinations on the chromosomes
- Increases the genetic diversity of potential offsprings

10.2 Inheritance

Dihybrid crosses:

- Determines the genotypic and phenotypic combinations of offspring for two unlinked genes
  —> four different gamete combinations
- The independent segregation of unlinked genes results in a greater number of potential gamete combinations, as well as a greater variety of possible phenotypes
- Dihybrid cross formation:
  1) Designate characters to represent the alleles
  2) Write down the genotype and phenotype of the parents (P generation)
  3) Write down all potential gamete combinations for both parents
  4) Use a Punnett square to work out potential genotypes of offspring
  5) Write out the phenotype ratios of potential offspring

Linked genes:

- A linkage group is a group of genes whose loci are on the same chromosome and hence don't independently assort —> tend to be inherited together
  —> phenotypic ratio similar to a monohybrid cross
  —> may become separated via recombination —> due to crossing over
- Thomas Hunt Morgan —> proved that linked genes were not independently assorted
- Sex linkage —> clear bias in phenotypic distribution —> may be X-linked
- Gene linkage —> certain phenotypic combinations occurred in much lower frequencies than was to be expected —> due to —> alleles for these traits were located on a shared chromosome
  —> linked alleles could be uncoupled via recombination
- The amount of crossing over between linked genes differed depending on traits combination
  - Crossover frequency may be a product of the distance between two genes on a chromosome
  - The further apart, the higher the crossover frequency
  - Used to show the relative positions of genes on a chromosome

Recombinations:

- Results in combinations of genes not found in the parents
- The frequency of recombinant phenotypes within a population will be lower than that of non-recombinant phenotypes —> because crossing over is random and chiasmata do not form at the same locations with every meiotic division

- The relative frequency of recombinant phenotypes will be dependent on the distance between linked genes —> more possible locations where a chiasma could form between the genes when they are more apart
- Recombinant phenotypes can be identified by performing a test cross (crossing with a homozygous recessive for both traits)

Chi Squared Test:

- Offspring with unlinked genes have an equal possibility of inheriting any potential phenotypic combination
- Offspring with linked genes will only express the phenotypic combinations present in either parent unless crossing over occurs —> 'unlinked' recombinant phenotypes occur less frequently
- Are a statistical measure that are used to determine whether the difference between an observed and expected frequency distribution is statistically significant
- If observed frequencies are not as expected for unlinked dihybrid cross:
  —> genes are linked —> do not independently assort
  —> inheritance of traits not random —> affected by natural selection

1) Identify hypotheses (null versus alternative)
2) Construct a table of frequencies (observed versus expected)
3) Apply the chi-squared formula —> $\chi^2 = \Sigma \frac{(O-E)^2}{E}$
4) Determine the degree of freedom (df) —> df = (m - 1)(n - 1) —> m = n. rows , n = n. columns
5) Identify the p value (should be <0.05)

Polygenic traits:

- Variation in phenotypes for a particular characteristic can be either discrete or continuous
- Monogenic traits —> (characteristics controlled by a single gene loci) tend to exhibit discrete variation —> individuals express one of a number of distinct phenotypes
- Polygenic traits —> (characteristics controlled by more than two gene loci) tend to exhibit continuous variation —> individual's phenotype is on of a spectrum of potential phenotypes
  —> More number of loci responsible for trait more possible phenotypes
  —> follow Gaussian distribution —> bell shape
- Maize grain colour —> example of polygenic trait —> controlled by three gene loci
  —> each gene has two possible alleles

- Phenotypic characteristics aren't only determined by genotype, but also environmental factors
- Human height is controlled by multiple genes —> Environmental factors such as diet and health (disease) can further influence an individual human's height
- Skin colour is controlled by multiple melanin producing genes, but is also affected by factors such as sun exposure

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10.3 Speciation

Evolution:

- Gene pools —> the sum total of alleles for all genes present in a sex reproducing population
  —> if large, high genetic diversity and chances of biological fitness and survival
  —> if small, low genetic diversity, reducing biological fitness, more chances of extinction

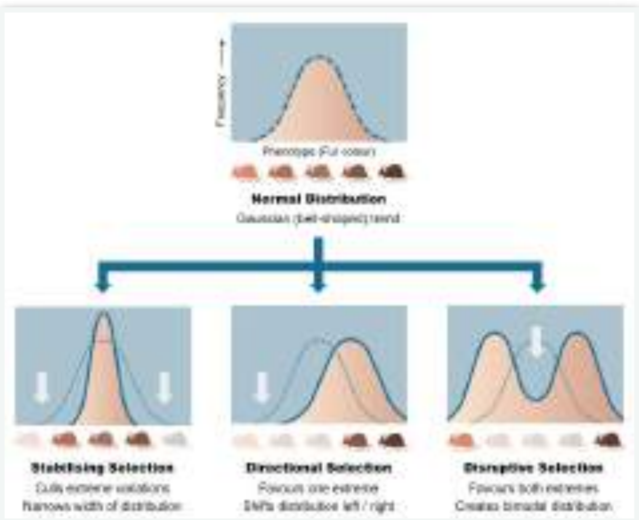
- Can be used to determine allele frequency within a population
- Evolution —> the cumulative change in the heritable characteristics of a population across successive generations —> allele frequencies change
- Mechanisms of change —> mutation —> random change in the genetic composition
  —> gene flow —> immigration or emigration affecting alleles
  —> sexual reproduction —> new gene combinations
  —> genetic drift —> change in gene pool composition —> random event
  —> natural selection —> as a result of different environmental pressures

Allele distribution:

- Genetic drift —> change in composition of a gene pool as a result of chance or random events
  —> smaller populations will be more affected by event
- Allele frequencies change significantly when a large population is reduced to a small population
  —> occurs due to two mechanisms —> population bottlenecks and founder effect

Population bottlenecks:
- Occur when an event reduces population size by an order of magnitude (~ >50%)
- May result from natural occurrence (fires, …) or be human induced (overhunting, …)
- Results in a higher level of genetic drift and newly developing gene pool will be different from the original

Founder effect:
- Occurs when a small group breaks away from a larger population to colonise a new territory
- Subject to more genetic drift as smaller population —> gene pool will change accordingly
- Original population remains largely intact —> differently from population bottlenecks



Types of selection:

-Natural selection —> the change in the composition of a gene pool in response to a differentially selective environmental pressure

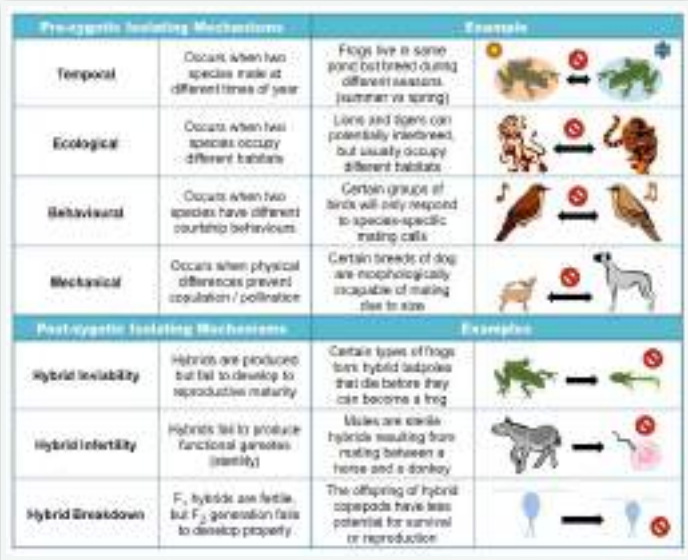
-**Stabilising selection** —> an intermediate phenotype is favoured at the expense of both phenotypic extremes
  —> results in the removal of extreme phenotypes
  —> env. conditions are stable + low competition

-**Directional selection** —> one phenotypic extreme is selected at the cost of the other phenotypic extreme
  —> phenotypic distribution shifts in one direction
  —> in response to changes in env. conditions
  —> typically followed by stabilising selection

- **Disruptive selection** —> phenotypic extremes favoured at expense of intermediate phenotypes
  —> results in a bimodal spread
  —> occurs when fluctuating env. conditions (e.g. seasons) favour two different phenotypes
  —> separation may eventually split the population into two distinct species —> speciation

Isolation barriers:

- Reproductive isolation —> when barriers prevent two populations from interbreeding
  —> pre zygotic isolation —> before fertilisation —> no offspring
  —> post zygotic isolation —> after fertilisation —> offspring infertile



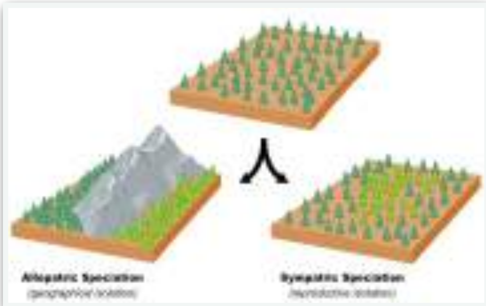
Pre-zygotic Isolating Mechanisms		Example	
Temporal	Occurs when two species mate at different times of year	Frogs live in same pond but breed during different seasons (summer vs spring)	
Ecological	Occurs when two species occupy different habitats	Lions and tigers can potentially interbreed, but usually occupy different habitats	
Behavioural	Occurs when two species have different courtship behaviours	Certain groups of birds will only respond to species-specific mating calls	
Mechanical	Occurs when physical differences prevent copulation / pollination	Certain breeds of dog are morphologically incapable of mating due to size	
Post-zygotic Isolating Mechanisms		**Examples**	
Hybrid Inviability	Hybrids are produced but fail to develop to reproductive maturity	Certain types of frogs form hybrid tadpoles that die before they can become a frog	
Hybrid Infertility	Hybrids fail to produce functional gametes (sterility)	Mules are sterile hybrids resulting from mating between a horse and a donkey	
Hybrid Breakdown	F_1 hybrids are fertile, but F_2 generation fails to develop properly	The offspring of hybrid copepods have less potential for survival or reproduction	

Pre zygotic isolation:

- Temporal —> when two populations differ in their periods of activity or reproductive cycles
- Behavioural —> when two populations exhibit different specific courtship patterns
- Geographical —> when two populations occupy different habitats within a common region

Speciation:

- Evolutionary process that results in the formation of a new species from a pre-existing species
- Occurs when reproductive isolating mechanisms prevent two breeding organisms from producing fertile, viable offspring —> can be allopatric or sympatric



Allopatric speciation:

- Occurs when a geographical barrier physically isolates populations of an ancestral species
- Populations begin to evolve separately due to cumulative mutation, genetic drift and natural sel.
- Eventually populations won’t be able to interbreed no more

Sympatric speciation:

- The divergence of species within the same geographical location (i.e. without a physical barrier)
- May result from the reproductive isolation of two populations as a result of genetic abnormalities
- Chromosomal error may arise + prevent successful reproduction with organisms lacking the error
- Most commonly caused by meiotic failure during gamete formation —> can cause polyploidy

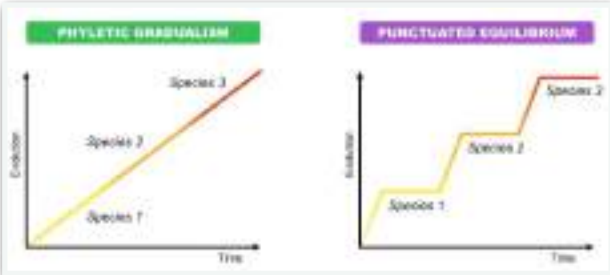
- Polyploidy —> meiotic cells fail to do cytokinesis, chromosomal number doubles in gamete
- Fertile polyploid offspring will typically require two polyploid parents —> if not it will result in an uneven number when forming gametes
- More common in plants as they lack separate sexes, can reproduce asexually and self fertilise
  —> polyploid crops —> allow for the production of seedless fruits
  —> typically grow larger, live longer and are disease resistant
- Gene allium —> monocotyledonous flowering plants and includes onions
  —> many of these species are polyploid —> resulted in distinct phenotypes

Pace of speciation:

- Evolution occurs both within a species (microevolution) and across the species barrier (macroevolution = speciation)
- Via speciation may occur by two models: phyletic gradualism or punctuated equilibrium

Phyletic gradualism:

- Generally occurs uniformly, via the steady and gradual transformation of whole lineages
- Speciation is seen as a smooth and continuous process —> fossil record of the horse is a proof



Punctuate equilibrium:

- Species remain stable for long periods before undergoing abrupt and rapid change (speciation)
- Speciation seen as periodic process —> supported by lack of transitional fossils for most species

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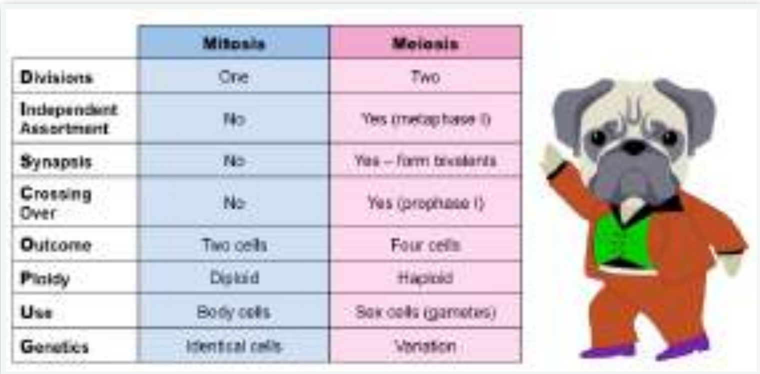
Extra:

Meiosis vs Mitosis:



	Mitosis	Meiosis
Divisions	One	Two
Independent Assortment	No	Yes (metaphase I)
Synapsis	No	Yes – form bivalents
Crossing Over	No	Yes (prophase I)
Outcome	Two cells	Four cells
Ploidy	Diploid	Haploid
Use	Body cells	Sex cells (gametes)
Genetics	Identical cells	Variation

Stages of prophase:

- **Leptotene** —> chromosomes condense and attached to the nuclear membrane by telomeres
- **Zygotene** —> synapsis form with a synaptonemal complex between homologous chromosomes
- **Pachytene** —> crossing over of genetic material occurs between non-sister chromatids
- **Diplotene** —> synapsis ends with disappearance of synaptonemal complex; homologous pairs remain attached at chiasmata
- **Diakinesis** —> chrom. fully condensed + nuclear membrane disintegrates prior metaphase I

Mendel and meiosis:

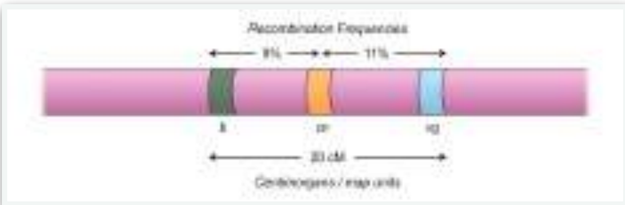
- **Law of Segregation:** Each hereditary characteristic is controlled by two alleles which separate into different gametes

- **Law of Independent Assortment:** The separation of alleles for one gene is independent to allele separation for another gene
- **Principle of Dominance:** In pairs of alleles that are different, one allele will mask the effect of the other allele
- Exceptions —> linked genes will not undergo independent assortment
  —> not all genes display a dominance hierarchy —> codominance, …

Test cross:

- Involves mating an unknown genotypic individual with a known homozygous recessive
  —> as recessive alleles are masked by dominant alleles
  —> phenotype of any offspring will reflect genotype of unknown parent
- Also used to determine if two genes are linked or unlinked —> mate with known heterozygote
  —> if equal ratio of the four potential phenotypes —> two genes are likely unlinked
  —> if two phenotypes in high amounts and two phenotypes in low amounts (recombinants), the two genes are likely linked

Centimorgan:



- Unit of measure used to approximate the distance between genes —> 1 map unit = 1% recombination *f*
- No longer used to measure distance —> Genome mapping allowed scientists to determine specific distances between genes in kilo bases (kb)
- Thomas Morgan demonstrated that genes that were further apart on a chromosome were more likely to recombine —> more potential sites for crossing over to occur between distant genes

Species caveats:

- Members of a species are unable to produce fertile, viable offspring different species
- Certain organisms do not reproduce sexually but can transfer genetic material via plasmids
- Breeding capacity unestablished —> no contact between populations
  —> no living representatives
- May be physically impossible for certain members of the same species to interbreed
- Ring species —> species spread around an area to form interlinked populations, but population 'ends' cannot interbreed

Allopolyploidy:

- Autopolyploidy occurs when a polyploid offspring is derived from a single parental species
- Allopolyploidy occurs when a polyploid offspring is derived from two distinct parental species
- Allopolyploids are more prevalent than autopolyploids as they do not show polysomic inheritance and have better fertility rates

Extinction:

- The total cessation of a species or higher taxon level —> reduces biodiversity
- Phyletic extinction —> results gradually —> organisms progressively evolve into something else
- Abrupt extinction —> species may not leave any identifiable descendants and cease to exist

- Caused by —> habitat degradation, predation, disease, natural disaster, …
  —> over 99% of species that have ever existed on Earth are now extinct
- Mass extinction events —> categorised by an unusually high number of species dying out in a relatively short period —> 5 to this moment

Polymorphisms:

- The occurrence of two or more clearly different phenotypes within same population of a species
- Are the individual components of a trait —> involve more than one allele for any given gene
- Transient poly. —> when there are two alleles in the gene pool, and one is replacing the other
  —> due to a strong environmental pressure causing directional selection
- Balance poly. —> when two alleles in the gene pool have non-changing frequencies of the alleles
  —> due to selective pressures promoting the coexistence of two alleles

Hardy-Weinberg principle:

- To predict the frequency of two alternate alleles within a population
- For two alleles —> possible genotypes are AA, Aa, aa —> A with frequency p and a with q
- Total frequency of both alleles will be 100 % —> $p + q = 1$
- Equation must be squared as genotypes have two alleles each —> $(p + q)^2$
- Population must be large and with random mating
- No mutation or gene flow must be present
- No natural selection or allele-specific mortality
