- **Genetic variation**

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

❖ *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)*

Meiosis

How meiosis generates variation

- **Crossing over**
  - Crossing over occurs when homologous chromosomes exchange genes ∴ resulting combinations of alleles on chromatids differ from original parent chromosomes
  - Involves the swapping of chromatid parts of homologous chromosomes
- **Independent assortment**
  - The assortment of maternal and paternal chromosomes (during metaphase) independent of how other pairs of homologous chromosomes have aligned themselves during metaphase.
  - This increases genetic variation by resulting in highly varies assortments of maternal and paternal DNA in the gametes
  - i.e. all maternal chromosomes will not necessarily align all one side
- **Random segregation**
  - The random separation of chromatids from every homologous pair (i.e. there are 4 chromatids per homologous pair so one of each in every gamete segregates the homologous chromosomes).
  - This increases genetic variation by ensuring that inherited homologous chromosomes come from both maternal and paternal genetic information

Fertilisation

- Each gamete formed from meiosis contains different combinations of alleles for each gene.
- During fertilisation, a male gamete (sperm) fuses with a female gamete (egg) to form a zygote
- Not only does each gamete contain different, recombined genetic material, but there are many different possibilities for which male and female gamete will fuse.

<u>Mutation</u>

- Mutation in meiosis is fundamental to variation in the genotypes of offspring, as it is the source of all variation.
- Mutation that occurs during replication of chromosomes in the initial phase of meiosis is called a ***germline mutation***, meaning it has the potential to be passed onto offspring.

- *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*
  - *Constructing and interpreting information and data from pedigrees and Punnett squares*

Autosomal

- Humans have 23 homologous pairs of chromosomes.
- Of the 23 pairs, 22 are autosomal chromosomes
- If inheritance is 'autosomal', then the trait is carried on one of the 22 autosomal chromosomes, and will also either be recessive or dominant

Example: Oculocutaneous albinism is inherited in an autosomal recessive pattern
Parents genotype: Rr x Rr – i.e. both parents are carriers of the defective allele
Parents phenotype: do not have disease

	R	**r**
R	**RR**	**Rr**
r	**Rr**	**rr**

Offspring genotype: 1RR: 2Rr: 1rr
Offspring phenotype: 3:1(affected)

Sex-linkage

- The 23rd chromosome in humans is the sex chromosome. If the sex is a girl, they will have two X chromosomes, but if the sex is male, they will have an X & Y shaped chromosome.
- Thus, in sex-linkage, one sex- usually the male will have only one copy of the gene due to the shorter Y chromosome.
- This means that if the gene is on the X chromosome, the boy will have only one copy of the gene, whilst the female will have two copies since she has 2 X chromosomes.

Example: Red-green Colour-blindness is a recessive sex-linked trait, that is more common in males because they only need one copy of the affected allele to have the trait, as they have no potential for a dominant gene on the Y chromosome to counter its effect.

- If R is the dominant allele, and r is the recessive, affected allele then;
  - X^RX^R is a normal female (does not have trait)
  - X^RY is a normal male (does not have trait)
  - X^RX^r is a carrier female (does not have trait)
  - X^rY is an affected male (has trait)

Co-dominance

- Occurs when the effect of both alleles appears in a heterozygous offspring
- Neither allele in the heterozygous genotype is dominant over the other, and thus both alleles are expressed and are visible in the phenotypes of heterozygotes

Example: Co-dominance can be exhibited in Roan cattle where

- RR= red coat
- RW = roan coat (red and white patches- NOT blending)
- WW=white coat

Incomplete dominance

- Occurs when neither allele of a gene is dominant over the other, so organisms with a heterozygous genotype will express and have a phenotype which is a blend of the two alleles.

Example: Snapdragon flowers exhibit incomplete dominance.

- R = red flower allele
- W = white flower allele
  - RR = red
  - RW = pink (blend of red and white alleles)
  - WW = white

Multiple alleles

- Multiple alleles (i.e. not just 2) may exist for a particular gene and thus the relationships between alleles are not as simple

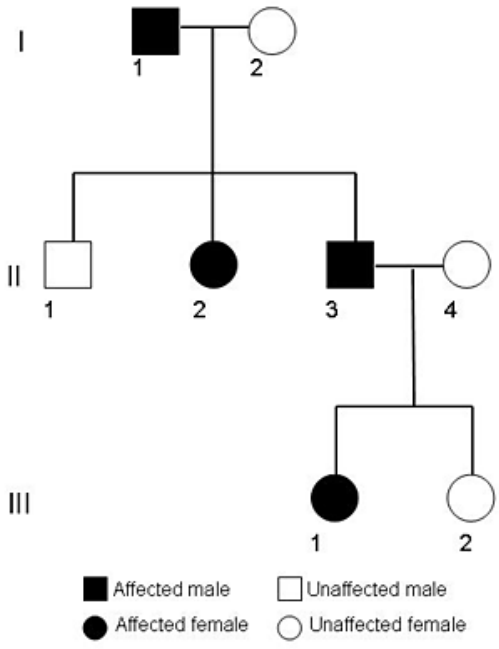
Example: Blood type is an example of a trait with multiple alleles. There are 3 different alleles for blood type- A, B & O

- A is dominant to B
- B is dominant to O
- A & B are co-dominant- producing type O blood

Phenotype	Genotype
O	AB
A	AA or AO
B	BB or BO

Pedigrees

- Pedigrees are a graphical representation of inheritance patterns of a particular trait (phenotype) in related individuals over a number of generations
- We can use pedigrees to determine if a trait is autosomal or sex linked, and if the trait is dominant or recessively inherited



This pedigree demonstrates an autosomal dominant condition

Punnet squares

Example: Pea plants- for the characteristic of height
T= tall
T=short

Parents phenotype: Tall x short
Parents genotype: Tt x tt

	T	t
t	T t	t t
t	T t	t t

Offspring genotype ratio: 2 Tt: 2 tt
Offspring phenotype ratio: 50% Tall: 50% short

- *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*

  - *Examining frequency data*

Frequency Data

- Blood type is determined by 2 separate genes:
  - The gene for blood antigens (the A, B, AB or O part)
    - A and B are co-dominant while O is recessive
  - The gene for rhesus factor (+ or -)
    - + is dominant and – is recessive
- The frequency of blood groups is different around the world:
  - A is common in Europe and with indigenous Australians
  - B is most frequent in South Asia

  - *Analysing single nucleotide polymorphism*

Single Nucleotide Polymorphism

- A SNP refers to a change in a single nucleotide at a specific position (due to a mutation)
- SNP's result in over 90% of differences between humans and may drive human evolution
- Different SNP's appear more frequently in different places around the world