• **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 varied 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.

**Mutation**

- 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.

<table>
<thead>
<tr>
<th></th>
<th>R</th>
<th>r</th>
<th>Offspring genotype: 1RR: 2Rr: 1rr</th>
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<tbody>
<tr>
<td>R</td>
<td>RR</td>
<td>Rr</td>
<td>Offspring phenotype: 3:1(affected)</td>
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<td>r</td>
<td>Rr</td>
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**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;
  - \( X^R X^R \) is a normal female (does not have trait)
  - \( X^R Y \) is a normal male (does not have trait)
  - \( X^r X^r \) is a carrier female (does not have trait)
  - \( X^r Y \) 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
- R= 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

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Genotype</th>
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<tr>
<td>O</td>
<td>AB</td>
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<tr>
<td>A</td>
<td>AA or AO</td>
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<tr>
<td>B</td>
<td>BB or BO</td>
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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

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

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

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