TOPIC: MODES OF INHERITANCE

Key Knowledge:

- The use of symbols in the writing of genotypes for the alleles present at a particular gene locus
- The expression of dominant and recessive phenotypes, including codominance and incomplete dominance
- Patterns of inheritance, including autosomal and sex-linked inheritance

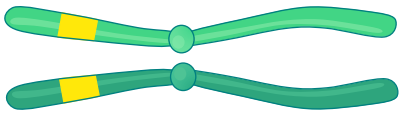
TYPES OF ZYGOSITY

Gametes are sex cells formed by the process of meiosis – males produce sperm and females produce ova. The gametes are haploid, meaning they possess one copy of each chromosome. When egg and sperm fuse during fertilisation, the resulting offspring will contain two copies of each chromosome (i.e. it is diploid). These homologous chromosomes have identical genes, but the alleles that they possess may be different.

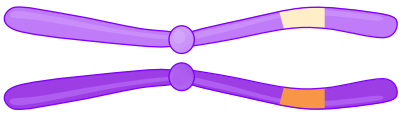
For any given gene, the combination of alleles (genotype) can be categorised as follows:

- **Homozygous:** If maternal and paternal alleles are the **same**, the genotype is said to be homozygous
- **Heterozygous:** If maternal and paternal alleles are **different**, the genotype is said to be heterozygous

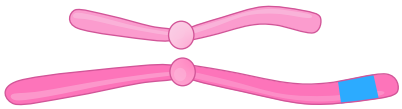
Males have different sex chromosomes (XY), meaning they have only one allele for each sex-linked gene. When there is only **one copy** of an allele for a gene, the individual is said to be **hemizygous** for that gene. Individuals who are hemizygous cannot be carriers of recessive disease conditions – this is the reason why X-linked recessive disorders occur more commonly in males.



HOMOZYGOUS
Alleles are the same



HETEROZYGOUS
Alleles are different



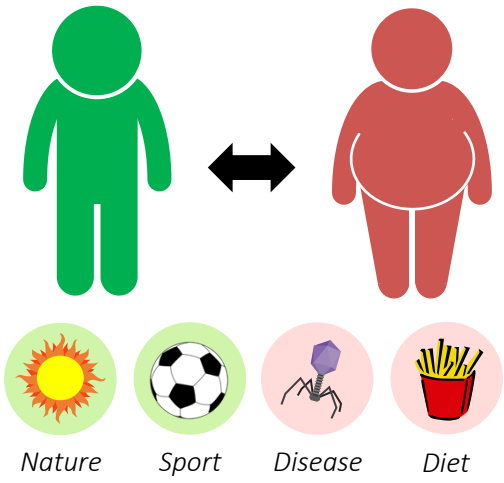
HEMIZYGOUS
Only one allele (males)

EPIGENETICS

While the genotype is an important factor in determining the physical expression of inheritable characteristics (phenotype), it is **not** the only contributing factor. A phenotype can also be influenced by the patterns of expression of particular genes.

Epigenetics describes the differences in a phenotype that are caused by changes in gene expression, rather than by changes within the allele sequence. There are a variety of factors that will determine whether a particular gene is switched on or off:

- **Diet:** Certain foods trigger specific metabolic responses
- **Lifestyle:** Gene expression is influenced by activity levels
- **Environment:** External conditions (UV) can activate genes
- **Pathogens:** Diseases result from a loss of homeostasis



Epigenetics can influence phenotype

MODES OF INHERITANCE

Two alleles may interact in different ways to affect the physical expression of the trait (phenotype). The way in which alleles are expressed in combination is called the mode of inheritance. Different expression patterns can occur, including a dominant / recessive hierarchy, codominance or sex-linked characteristics.

1. COMPLETE DOMINANCE

Most traits follow a classical dominant / recessive pattern of inheritance, whereby one allele is expressed over the other allele. The dominant allele will mask the recessive allele when in a heterozygous state and the homozygous dominant and heterozygous forms will therefore be phenotypically indistinguishable (the recessive allele will only ever be able to be expressed within the phenotype when in a homozygous state).

When representing alleles, the convention is to capitalise the dominant allele and use a lowercase letter for the recessive allele (same letter must be used as the alleles are alternative versions of the same gene).

- **Example:** Mouse colour coats – black fur coats (BB or Bb) are dominant to brown fur coats (bb)

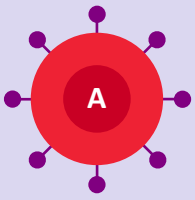
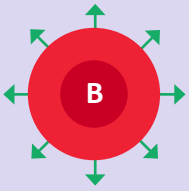
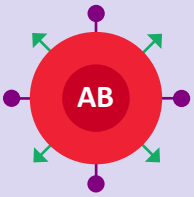
PHENOTYPE (Coat colour)			
GENOTYPE	BB (black)	Bb (black)	Bb (brown)

2. CODOMINANCE

Codominance occurs when pairs of alleles are *both expressed equally* in the phenotype of a heterozygote. Heterozygous individuals therefore have a different phenotype in comparison to homozygous dominant individuals as the alleles are having a joint effect.

When representing alleles, the convention is to use a common capital letter with **superscripts** to represent the different versions of the dominant allele (recessive alleles are still represented with a lowercase letter).

- **Example:** Human blood groups – I^A and I^B alleles are codominant, allowing for an AB blood group ($I^A I^B$)

BLOOD TYPE	A	B	AB	O
GENOTYPE	$I^A I^A$ or $I^A i$	$I^B I^B$ or $I^B i$	$I^A I^B$	ii
PHENOTYPE				

Codominance can influence a phenotype in two different ways:

- Both traits may be seen equally within the heterozygous phenotype (i.e. forms a mosaic phenotype)
- The traits may mix to create a new outcome within the heterozygote (i.e. forms a blended phenotype)

Expression of both traits is classical codominance, while blending of traits is called **incomplete dominance**

3. SEX LINKAGE

Sex linkage refers to when a gene is found on a sex chromosome (X or Y). Because males and females have different combinations of sex chromosomes (males = XY ; females = XX), the patterns of inheritance will be different according to the sex of the offspring. Sex linkage can involve either X-linked or Y-linked genes. Sex linked alleles are represented as superscripts attached to the relevant sex chromosome (example: X^A).

Y-Linked Traits:

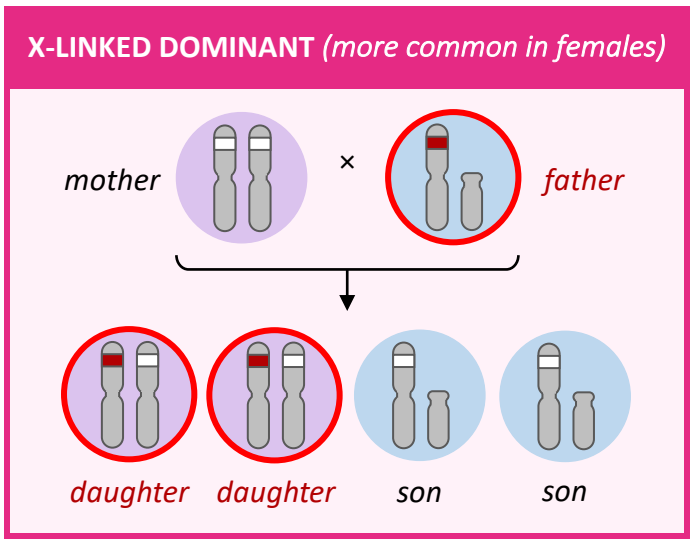
- Only males possess a Y chromosome and so Y-linked traits are only ever present in men (not women)
- As males only have one Y chromosome, the trait is not dominant or recessive (it is always expressed)
- A Y-linked trait will always be inherited by the sons and will never be inherited by the daughters

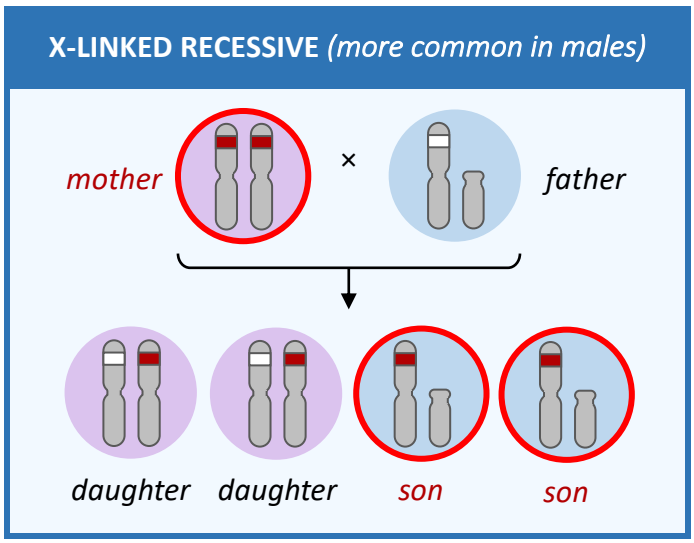
X-Linked Dominant Traits:

- As females possess two X chromosomes, an X-linked dominant trait will be more common in women
- An affected father always has affected daughters (daughters must inherit the father's X chromosome)
- Sons only inherit the condition if the mother is affected (sons inherit their X chromosome from mother)

X-Linked Recessive Traits:

- As males only possess one X chromosome, an X-linked recessive trait will be more common in men
- This is because males cannot be carriers for X-linked recessive traits (they cannot be heterozygous)
- Affected mothers always have affected sons, while unaffected fathers cannot have affected daughters





POLYGENIC INHERITANCE

Some characteristics are determined by a single gene (monogenic), while other traits may be controlled by multiple genes (polygenic). **Monogenic traits** will produce limited versions of a given trait due to being encoded by a single gene with a discrete number of alleles. The presence or absence of a widow's peak (hairline) is monogenic. **Polygenic traits** will exhibit a far greater spectrum of allelic variation due to being encoded by multiple genes whose alleles may combine in any number of different combinations (i.e. **continuous variation**). Traits that exhibit continuous variation will be normally distributed (i.e. they will exhibit a bell-shaped distribution pattern). Polygenic traits include human height, weight and overall skin colouration.