YEAR 10 - GENETICS

A <u>pedigree</u> is a symbolic family tree showing males and females who show a particular characteristic or disease. Geneticists use the information from a pedigree to determine the <u>mode/pattern of inheritance</u> and therefore can calculate <u>probability</u>. This is particularly useful when it comes to <u>genetic counselling</u> – where couples (who have a known family history of diseases) wish to have a children get the chance to see the probability of having an affected child and can therefore make a decision whether to go ahead.

Symbols used in Pedigree Charts:

Male	
Female	
Affected Male	
Affected Female	
Mate between Male and Female	
Offspring produced	
Identical Twins	A A LANDA LA
Non-identical Twins	
Deceased	
Generations	

Determining the mode of inheritance: You will need to be able to look at a pedigree and determine if Inheritance is **AUTOSOMAL** or **SEX-LINKED** and then if it is **DOMINANT** or **RECESSIVE**.

AUTOSOMAL VS. SEX-LINKED

- <u>Autosomal</u> traits that occur on the first 22 pairs of chromosomes (not the sex chromosomes). Responsible for many traits such as eye colour, skin pigment and hair colour.
- <u>Sex-Linked</u> (X-Linked) traits that are located/expressed on the <u>Sex Chromosomes</u> (pair 23). They are called the sex chromosomes because they determine the genetic sex of the individual. Remember the <u>Y chromosome</u> is small compared to the X and therefore carries fewer genes. It is the <u>X chromosome</u> that carries the trait. Remember females are <u>XX</u> and males are <u>XY</u>. The ova produced by the woman always carries the <u>X</u>, and the sperm carry either a <u>X</u> or <u>Y</u> chromosome which therefore determines the sex of the offspring.

COMPARISON TABLE (to help recognise inheritance patterns in pedigrees):

Autosomal Recessive Inheritance

- · Often skips a generation
- Affected individuals can have unaffected parents
- Almost equal number of males and female affected
- If both parents affected all offspring produced are affected
- In most cases when an unaffected individual mates with an affected individual, all children produced are unaffected. When at least one child is affected (indicated a unaffected parent is heterozygous for the condition), approx half the children are affected
- Often found where incestuous mating occurs
- Examples: Albinism, Curly Hair, Cystic Fibrosis, Thalassaemia, Tay-Sach's disease, haemochromatosis

Autosomal Dominant Inheritance

- Trait does not usually skip a generation
- An affected person mating with an unaffected person produces approx 50% affected offspring (indicates the affect person is also heterozygous for the condition)
- Almost equal number of males and females affected
- Examples: Tongue rolling ability, the type of ear wax and Huntington's disease

Sex-Linked Recessive Inheritance

- Most affected individuals are Males
- Affects males result from mothers who are also affected or are carriers (heterozygotes), and have affected brothers, fathers or maternal uncles.
 - The sons of affected mothers should be affected
 - Approx half the sons of carrier females should be affected
- Affected females come from affected fathers and affected or carrier mothers
- Examples: Haemophilia, Duchenne Muscular Dystrophy, Colour blindness

Sex-Linked Dominant Inheritance

- Traits do not skip generations
- Affected sons must come from affected mothers
- Approx half of the children of an affected heterozygous female are affected
- Affected females come from affected mothers or fathers
- All the daughters, but none of the sons, of an affected father are affected.
- Examples: Very few, but Rett Syndrome is one example