23/11 GENETIC DISORDER

Physical and chemical disturbances, as well as errors during meiosis, can damage chromosomes in major ways or alter their number in a cell.

The phenotype of an organism can also be affected by small scale changes involving individual genes.

Random mutations are the sources of all new alleles, which can lead to new phenotypic traits

- Alterations in chromosomal number
- Alterations in chromsomal structure.

ALTERATIONS IN CHROMOSOMAL ~~STRUCTURE~~ NUMBER

- Members of a pair of homologous chromosomes do not move apart properly during meiosis I or sister chromatids fail to seperate during meiosis II
- If either of the aberrant gametes unites with a normal one at fertilization, the zygote will also have an abnormal number of a particular chromosome known as aneuploidy.
- Fertilization involving a gamete that has no copy of a particular chromosome will lead to a missing chromosome in the zygote (2n-1) is said to be monosomic (2n-1), trisomic (2n+1) polyploidy (3n, 4n...)

eg Down syndrome

Down syndrome is often called trisomy 21.

ALTERATIONS IN CHROMOSOMAL STRUCTURE

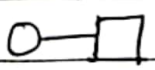
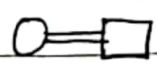
- Deletion
- Duplication
- Inversion.
- Translocation.

Autosomal Recessive inheritance.
Autosomal Dominant inheritance.
X-linked inheritance (dominant & recessive)
Y-linked inheritance

Albinism is a recessive trait.
Achondroplasia is a dominant trait (Dwarfism)

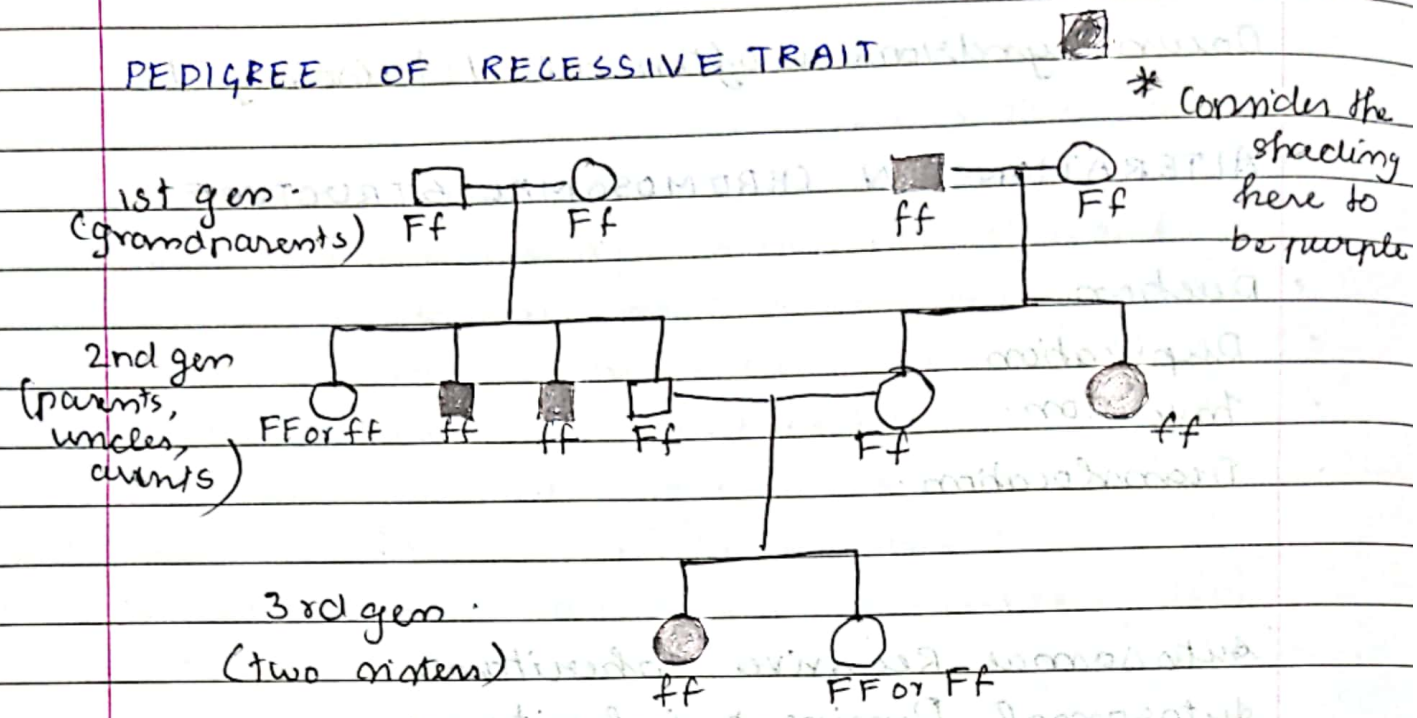
Pedigree Analysis -
Collecting the information about a family's history for a particular trait and assembling this information into a family tree describing the traits of parents and children across the generations is the family pedigree.

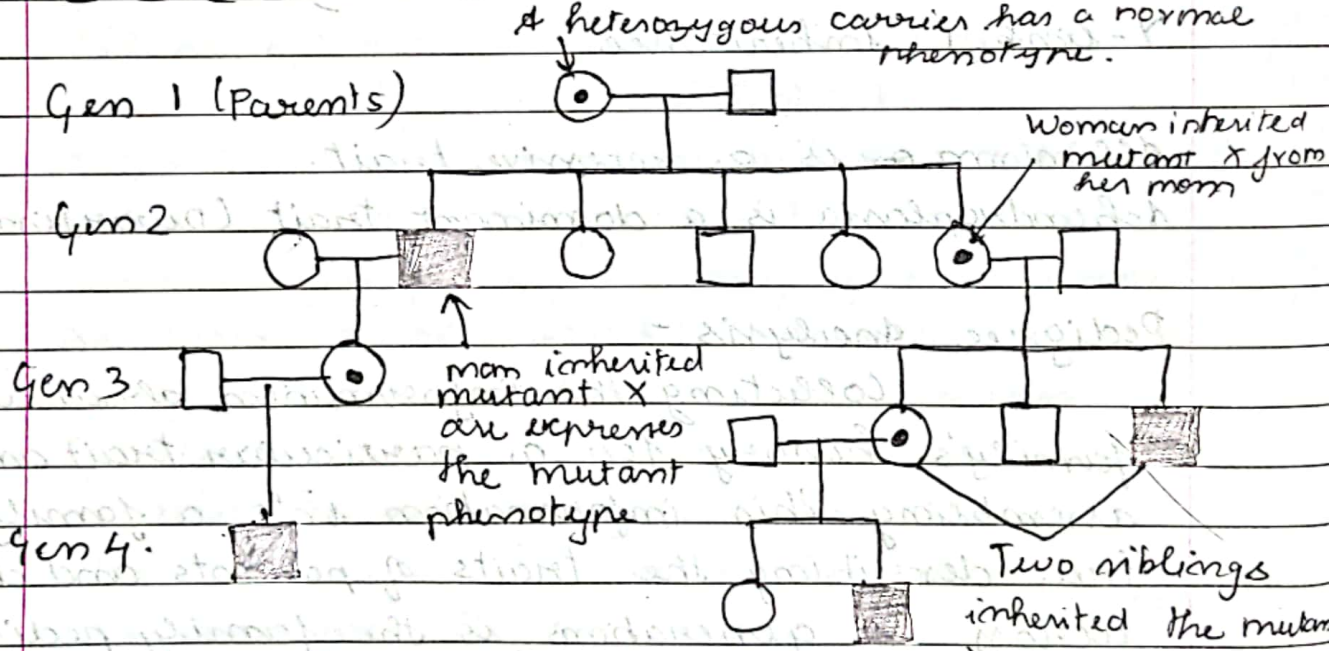
Pedigree chart rules.

	Unaffected	Affected	Heterozygote (Unaffected phenotype)
Female			
Male			
Mating			
Mating b/w relatives			

PEDIGREE OF RECESSIVE TRAIT

* Consider the shading here to be purple





Autosomal recessive trait

- Appears in both sexes with equal frequency
- Trait tends to skip generations
- Affected offsprings are usually born to unaffected parents
- When both parents are heterozygous, approx 1/4 of the offspring are affected.
- Appears more frequently among the children of consanguine marriages.

Autosomal dominant trait

- Appears in both sexes with equal frequency
- Both sexes transmit the trait to their offspring
- Does not skip generations.
- Affected offspring must have an affected parent, unless they possess a new mutation.
- When one parent is affected (heterozygous) and the other parent is unaffected. and approx 1/2 of offspring will be affected.
- Unaffected parents do not transmit the trait.

X-linked recessive trait.

- More males than females affected.
- Affected sons are usually born to unaffected mothers. Thus trait skips generations
- A carrier (heterozygous) mother produces approx 1/2 affected sons.
- Never passed from father to son.
- All daughters of affected fathers are carriers.

X-linked dominant trait:

- Both males and females are affected. More females than males affected.
- Does not skip generations. Affected sons must have an affected mother; affected daughters must have either affected mo or an affected father.
- Affected fathers will pass the trait on to all their daughters.
- Affected mothers (ie heterozygous) will pass the trait on to ½ of sons and ½ of daughters.

Y-linked trait

- Only males are affected.
- Is passed from fathers to all sons.
- Does not skip generations.

ANALYSIS OF SEX-LINKED TRAITS IN HUMANS.

- X-linked traits, like autosomals ones, can be analysed using pedigrees.
- Human pedigree analysis, however is complicated by several factors:
  a) Data collection often relies on family recollection
  b) If the trait is rare and the family small, there may not be enough individuals to establis a mechanism of inheritance.
  c) Expression of the trait may vary, resulting in affected individuals being classified as norm
  d) More than one mutation may result in the same phenotype and comparison of different pedigrees may show different inheritance for the "same"