

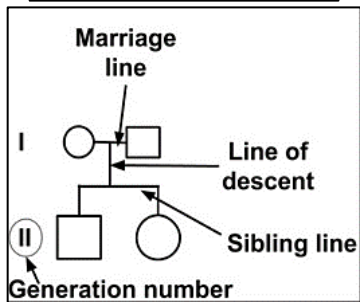
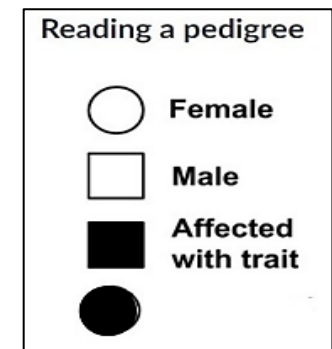
GENETICS: MUTATIONS AND PEDIGREE DIAGRAMS

1. TERMINOLOGY

- 1.1 A change in the sequence of the nitrogenous bases of nucleotides.
- 1.2 A change in the normal structure or number of chromosomes.
- 1.3 A genetic disorder characterised by the absence of blood clotting factor.
- 1.4 A human disorder caused by the non-disjunction of chromosome pair 21.
- 1.5 A diagram showing the inheritance of genetic disorders over many generations.
- 1.6 The inability to perceive certain colours due to a gene mutation

2.1 PEDIGREE DIAGRAMS (GENETIC LINEAGE)

A pedigree diagram s used to study the inheritance of characteristics in a family over a number of generations



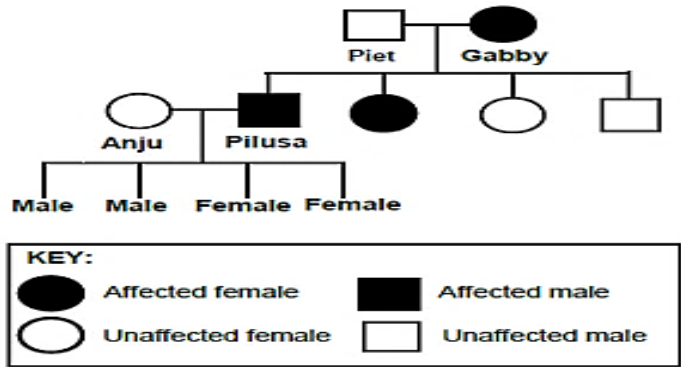
3.1 EFFECTS OF MUTATIONS – harmless, harmful, useful

- Harmless mutations do not affect the structure or functioning of the cell/organism.
- Examples
- A harmful mutation
- Examples:
- Useful mutations are beneficial to an organism and increase an organism’s chance of survival.
- Examples:

3.2. GENE AND CHROMOSOMAL MUTATIONS

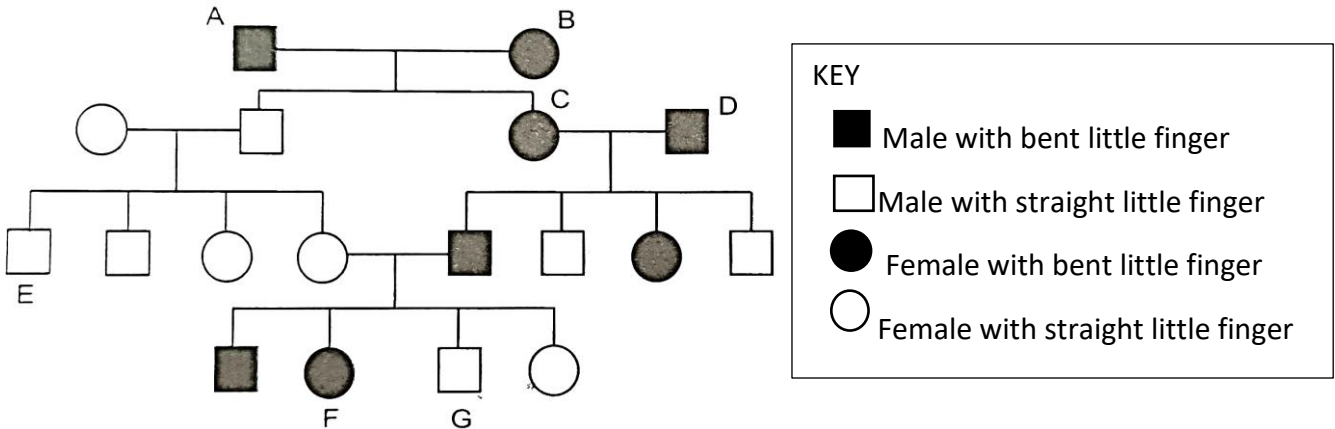
- Gene mutations occur
- Examples: Haemophilia, colour-blindness, sickle cell anaemia, albinism.
- Chromosome mutations occur
- Example: Down Syndrome

2.2 Goltz syndrome is a sex-linked genetic disorder. It is caused by a dominant allele X^G. The diagram below shows the inheritance of Goltz syndrome in a family.



- 2.2.1 Name the type of diagram shown. (1)
- 2.2.2 How many:
(a) Females are in this?..... (1)
(b) Males in the F1-generation have Goltz syndrome?.....(1)
- 2.2.3 Give Gabby’s genotype..... (2)
- 2.2.4 Anju and Pilusa have four children. Give the phenotype of their sons. (2)
- 2.2.5 Explain your answer to QUESTION 2.2.4. (4)

4.Study the diagram below which shows the inheritance of the type of little finger over four generations of a family.



- 4.1 Name the type of diagram shown. (1)
- 4.2 How many members of the family have straight little fingers? (1)
- 4.3 Is person F homozygous or heterozygous for the type of little finger?..... (1)
- 4.4 Which type of little finger is controlled by a dominant gene? (1)
- 4.5 Explain your answer in Question 6.4. (3)
- 4.6 In the fourth generation, what proportion will be female with a straight little finger? (2)
- 4.7 Persons F and G are twins? Were they produced from a single fertilised egg cell? (1)
- 4.8 Give TWO reasons based on the phenotype to support your answer to QUESTION 47. (2)

5 . Study the table below which shows the relationship between the age of a mother and the risk of having a Down syndrome baby.

Age of mother (years)	Risk of Down’s syndrome baby (per 10000 births)
25	8
35	25
45	200

- 5.1 What is the relationship between the mother’s age and the chance of having a baby with Down’s syndrome? (2)
- 5.2 How many chromosomes are present in a cell of the body of a baby with Down’s syndrome?..... (1)
- 5 .3 Explain why a person with Down’s syndrome has an abnormal number of chromosomes. (4)
- 5.4 How many times does the risk of having a baby with Down’s syndrome increase between 35 and 45 years of age? (1)