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Genetic Disorders

- Include Mendelian disorders and chromosomal disorders

Mendelian Disorders

- Characterized by mutation in a single gene
- Their mode of inheritance follows the principles of Mendelian genetics.
- Mendelian disorders can be
    - autosomal dominant (muscular dystrophy)
    - autosomal recessive (sickle cell anaemia)
    - sex linked (haemophilia)
- Haemophilia
    - Sex-linked recessive disease
    - Transmission − From unaffected female (carrier) to male progeny
    - Females act as carriers of disease, but rarely suffer from haemophilia since for a female to become haemophilic, the mother should be carrier and father should be haemophilic.
    - In this disease, protein involved in blood clotting is affected. Therefore, even a simple cut results in uncontrolled bleeding.
- Sickle cell anaemia
    - Autosomal recessive disease
    - Transmission − From parent to offspring when both parents are carriers of disease
    - Pair of alleles Hb^A and Hb^S controls the expression of this disease.

      Hb^A and Hb^A – Normal
      Hb^A and Hb^S – Carrier of disease
      Hb^S and Hb^S – Diseased

- Cause of the disease − Change in gene causes the replacement of GAG by GUG leading to the substitution of Glu by Val at sixth position of beta globin chain of haemoglobin.
- The mutant haemoglobin so formed polymerises at low oxygen tension, resulting in change in shape of RBC to sickle-like.