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# Lecture – 15

## Genetic disorders

by

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- **Pedigree analysis:** Analysis of traits in a several generation of a family
- **Genetic disorders are two types**
- 1–Mendelian disorder  
(Alteration/Mutation in single gene)
- 2- Chromosomal disorder



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# Mendelian disorder

- 1. Haemophilia
- 2. Sickle cell anaemia
- 3. Phenylketonuria



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# Chromosomal disorder

- 1. Down syndrome
- 2. Klinefelter syndrome
- 3. Turner syndrome



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# Haemophilia

- Sex linked recessive
- Unaffected female is carrier to male
- Female rarely affected
- Single protein part of cascade protein involve in clotting of blood is affected
- Ex Queen Victoria



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# Sickle cell anemia

- Autosomal recessive pattern of inheritance
- GAG codon changing to GTG) of the  $\beta$ -globin gene, which results in glutamic acid(E/Glu) being substituted by valine (V/Val) at position 6 of B globin chain
- under low Oxygen concentration shape of RBCs appear like sickle.



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# Phenylketonuria

- Inborn error of metabolism
- Autosomal recessive pattern of inheritance
- Lack an enzyme which convert phenylalanine into tyrosine
- Phenylalanine converts into pyruvic acid
- Poor absorbed by Kidney
- Mental retardation



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# Down syndrome

- Langdon down 1866
- Trisomy
- Presence of additional copy of chromosome 21
- Short stature
- Small round head
- Furrowed tongue
- Partially open mouth
- Broad palm
- Psychomotor, mental development retard





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# Kleinfelter syndrome

- Caused due to presence of additional copy of X chromosome
- 47 – XXY
- Affected individual
- Sterile
- Overall masculine development



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# Turner syndrome

- Absence of X chromosome
- 45 +XO
- Affected individual
- Sterile
- Ovaries are rudimentary



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**THANK YOU**