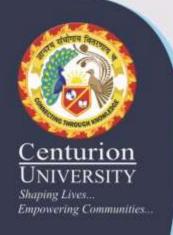


Lecture – 15 Genetic disorders by

Dr. Praveen Kumar

Asst. Prof. GPB

MSSSoA, CUTM, Odisha



- Pedigree analys: 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



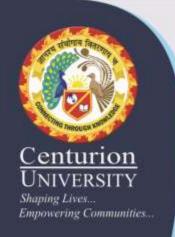
Mendelian disorder

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

Chromosomal disorder

Shaping Les Down syndrome

- 2. Kleinfeltor syndrome
- 3. Turner syndrome



Haemophilia

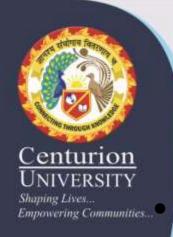
- Sex linked recessive
- Unaffected female is carrier to male
- Female rarely affected
- Single protein part of cascade protein involve in clotting of blood is affected
- Ex Queen Victiria



Sickle cell anemia

Empowering Communities Autosomal recessive pattern of inheritance

- GAG codon changing to GTG) of the β-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.



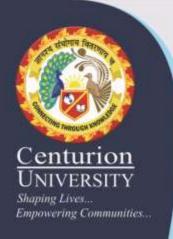
Phenylketonuria

Inborn error of metabolism

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

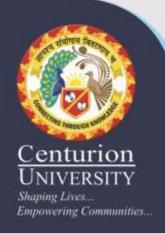
Down syndrome

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



Kleinfeltor syndrome

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



Turner syndrome

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



Shaping Lives... Empowering Communities...

THANK YOU