04 TNPSC GROUP II / IIA MAINS SCERT - SCIENCE & TECHNOLOGY QUESTION WITH ANSWER

4. Explain about Genetic disorders

- "Genetic Disorder - Disease or syndrome caused by an abnormality in an individual DNA. Small mutation in a single gene / addition or subtraction of an entire chromosome or even set of Chromosomes".

Types :

1. Mendelian Disorders
2. Chromosomal Disorders

1. Mendelian Disorder :

a) Atteration or mutation in a single gene

b) Transmitted to offsprings - Mendelian pattem of Inheritances

c) Disorders - dominant / recessive & autosomal / sex linked

1. Thalassemia
2. Albinism
3. Phenylketonuria
4. Huntington's Chorea

1. Thalassemia :

- Autosomal recessive disorder causing production of abnormal haemoglobin molecules resulting in anaemia
- Types - α & β thalassemia - mutation / deletion of alpha or Beta globin chains.
- α & β thalassemia controlled by HBA1 & HBA2 genes on Chromosome 16.
- Cooley's Anaemia - Common type also called as Beta thalassemia.

2. Albinism : Absense of Melatin

- Inborn error of metabolism caused by autosomal recessive gene.
- Absense of pigment responsible for skin colour (Melanin).
- Melanocytes present in normal numbers but lack pigment.

3. Phenylketonuria :

- Inborn error of Phenylalanine Metabolism
- Caused due to pair of autosomal recessive genes.
- Mutation in gene PAH - Phenylalancine hydroxy lase on Chromosome 12
- Effects :
  1. Severe mental retardation
  2. Light pigmentation of skin & hair

4. Huntington's Chorea :

1. Inherited as autosomal dominant lethal gene in man.
2. Characteristices - Involuntany gerking of body.
3. Effects :
   - Degradation of Nervous system
   - Gradual mental & Physical deterioration
   - Patients die between age of 35 & 40

2. Chromosomal Abnormalities :

a) Error in number (28 pairs) or structure of Chromosome

b) Anamolies occur during cell division

c) Aneuplaidy - failure of chromatide to segrogate

Syndromes in Human beings

1. Autosornal Aneuploidy

1. Down's Syndrome
2. Patau's Syndrome

2. Allosornal Abnormalities

1. Klienfester's Syndrome
2. Turners Syndrome

1. Down's Syndrome

a) Trisomic condition of Chromosome - 21

b) Characteristics

1. Severe Mental Retardation
2. Defective development of central nervous system
3. Eyes, Nose, Ears are malformed

2. Patau's Syndrome

a) Trisormic Condition of Chromosome - 13

b) Cause - Meiotic non disjuction

c) Characteristics - Multiple & Severe body malformations

d) Symptoms - Small head, Small eyes, Porain male foramtion

3. Klienfelter's Syndorme : (XXY males)

a) 47 Chromosomes (44AA + XXY) in males.

b) Characteristics

- Sterile Males, under developed genetalia.
- Feeble breast development.

4. Turner's Syndrome (XO Females)

a) Loss of X Chromosomes - 44AA + XO in females

b) Characteristics

- Sterile females, underdeveloped breasts
- Lack of menstural cycle during puberty