Chapter 8 – Genetic Disorder

Exercise

Q1. Define following terms: dominant, recessive, homozygous, heterozygous, phenotype and genotype.

Ans- There are various terms related to the experiment of inheritance. Some of them are:

1. Dominant character- It is the character that can be expressed even if only one pair of dominant allele is crossed.

2. **Recessive character**- Recessive characters are visible when two pairs of recessive alleles are crossed together.

3. Homozygous- It is a condition when the individual contains two identical alleles.

4. Heterozygous- It is a condition when an individual contains different types of alleles.

5. Phenotype- It represents the physical characteristics of the progeny.

6. Genotype- It represents the genetic characters of the progeny.

Q2. Describe the origin, symptoms and treatment of Down syndrome.

Ans- Down's Syndrome.

Origin- It is a genetic disorder that occurs due to the presence of an extra chromosome 21. This chromosome is repeated three times because of error in the cell division process. It was first discovered by John Langdon Down in 1866.

Symptoms- Some of the most common symptoms of down's syndrome are: Slanting eyes, flat face, small mouth, protruding tongue, short neck, short limbs, low IQ levels, under-developed genitals.

Treatment- No treatment is there to treat this disorder by date. By observing the characteristics of the children in younger age, can benefit in speech by speech therapy. Also physiotherapy and nutritional.

Q3. Describe the origin, symptoms and treatment of Klinefelter syndrome.

Ans- Klinefelter's Syndrome

Origin- It occurs in approximately one in 1000 persons. Males are affected. It occurs due to the presence of an extra X chromosome that arises due to the inability of detachment during the time of meiosis. Instead of X and Y fertilization, XX and Y fertilize into XXY chromosome.

Symptoms- The general symptoms of this disorder are: unusually tall height, reduced facial and body hair, smaller genitals, enlarged breast and coarse voice.

Treatment- Patients can be treated with testosterone to look man-like. Psychological counseling is also necessary for preventing depression.

Q4. Describe the origin, symptoms and treatment of Turner syndromes.

Ans- Turner's Syndrome

Origin- Affects in approximately one out of every 2500 girls. In this, the absence of the X chromosome takes place in the females due to the error in the cell division process during meiosis. It was first reported by the doctor Henry Turner in 1938.

Symptoms- The common symptoms of this disorder are: short stature, small breast, webbed neck, swollen hands and feet, underdeveloped genitals, absence of menstrual cycle.

Treatment- No known cure. However, administration of hormones like androgen and estrogen can strengthen the growth and functions of ovaries.

Q5. Describe various structural chromosomal abnormalities.

Ans- There various types of structural chromosomal abnormalities known by far. some of them are given as:

1. **Deletion**

- In this a portion of chromosome is broken that leads to the shortening of the chromosome.
- The retinoblastoma is a disease caused by the deletion of a segment of chromosome.

2. **Duplication**

- In this, a portion of chromosome gets repeated and leads into a longer chromosome.
- The Charcot-Marie-Tooth disease is a result of Duplication of chromosome segment.

3**. Inversion**

- In this, a portion of chromosome breaks then reverses and attaches again to the chromosome. The orientation gets distorted.
- The RCD syndrome is an example of inversion of chromosomal segment.

4. **Translocation**

- In this, a portion of chromosome breaks and attaches itself with another chromosome.
- Burkitt's lymphoma is an example of translocation of gene segment from one chromosome to another.