

| SI.No | CHROMOSOMAL DISORDERS    | KARYOTYPE   |
|-------|--------------------------|---|
| 1.    | Down's Syndrome          | XXY, 47 chromosomes   |
| 2.    | Klinefelter's syndrome   | Trisomy of Chromosome No- 21, 47 chromosomes  |
| 3.    | Turner's syndrome        | XO, 45 chromosomes  |
| SL.NO | MENDELIAN/GENE DISORDERS | CAUSE   |
| 4.    | Phenylketonuria          | Mutation in sex linked recessive genes causing reduction in proteins needed for blood clotting.   |
| 5.    | Colour Blindness         | Mutation in autosome linked recessive genes causing substitution of Glutamic acid to valine in the beta globin chain of haemoglobin that results in R.B.C turning sickle shaped, because of poor quality haemoglobin. |
| 6.    | Thalassemia              | Mutation in autosome linked recessive gene that codes for phenylalanine hydroxylase enzyme  |
| 7.    | Sickle cell anaemia      | Mutation in Sex linked recessive genes, causing defect in red and green cones of eye  |
| 8.    | Haemophilia              | Mutation in autosome linked recessive genes causing reduction in the number of haemoglobin chains   |