8. CHROMOSOMAL BASIS OF INHERITANCE

Each species possesses a specific and a characteristic number of chromosomes in its somatic cells. In a diploid cell, the chromosomes occur in pairs . called homologous pairs. The two chromosomes of a pair resemble in structure and genetic content. One member of a chromosome pair is derived from the mother while the other is derived from the father. Each chromosome is a double structure consisting of a pair of chromatids. During mitosis, the two chromatids separate into two daughter nuclei and then into cells.

The two alleles of an allele pair of the gene are also equally distributed in the daughter cells. At Meiosis, the members of a chromosome pair are separated and are distributed equally in daughter cells. Similarly, each member of an. allele pair goes to each haploid cell (gamete). Hence, a gamete contains onlv one allele. When the two haploid gametes unite during fertilisation, the diploid chromosome number and the allele pair are restored. It was William Flemming who discovered the chromosomes in the nuclei- of Salamander cells.

The Concept of Linkage : This deals with the seggregation and recombination of chromosomes. When genes are located on the same rhromosoni^s, there can be c[r]ossing over between them or no crossing over. Crossing over simply means exchange. When two non-sister chromatids undergo a crossing over, two new allele combinations can be created. Only two "non-sister chromatids participate in crossing over at any given point The crossing ever takes place during Meiesis-[r]. When the crossing over takes place, each member of a homologous pair consists of two sister chromatids. When crossing over takes place two types of gametes are formed. If there is no crossing over between genes, $_0$they are transmitted intact: This will lead to the formation of only parental gametes. Tliis is called complete linkage. The gametes that result from crossing over are called the recombinant gametes..

If two genes are located on the same chromosome, and if they are very far from each other, crossing over is possible in all meiotic cells. In such cases, 50% of the gametes are recombinants and 50% are parental. If the two genes are located on the same chromosome and are very close to each other, crossing over takes place. But in the gametes that result from this, the parentals dominant over the recombinants. All genes located on the same chromosome show some linkage between them. The strength of the linkage is determined by the distances between them. The more farther apart they are, the weaker the linkage. Genes located on the same chromosome constitute, a linkage group.

Recombination : This is a meiotic process that generates a haploid product whose genotype is different from the two haploid genotypes constituting the diploid. Recombination occurs due to crossing over during Meiosis-I. The rejoining takes place between non-sister chromatids. The recombination frequency is governed by the distance between the genes. That is, closer the distance between the two genes, less is the probability of crossing over and recombination.

Sex-Linked Inheritance : In many sexually differentiated organisms, each cell contains a pair of chromosomes known as sex chromosomes. The other chromosomes are called autosomal chromosomes or autosomes. In humans, the male produces only two types of gamete while the female produces only one type of gamete. The chromosome of the male carries a gene called SRY (sex determining region) which codes for a product called testis-determining factor (TDF) which is required for the development of male sexual characters. However, in the fly Drosophila. the sex determining factor is the ratio between x-chromosomes and autosomes.

Genetic Variation: These result due to different mechanisms such as recombination, gene-mutation, gene-environment interactions and by chromosomal observations. Gene mutation was a term introduced by Hugo de Vries to explain the variations in the plant Evening Primrose (Oenothera Lamarckina). Gene mutation is a sudden discrete change in the genetic material which is heritable. Mutation serves as the source of most of the genetic variability, in a population. This variability provides the basis for natural selection and hence biological evolution. Mutations in gametic cells can get transmitted to the next generation but those iri the somatic cells are confined only to the individual.

Chromosonir.-l Aberrations : In addition to gene mutations, alterations in chromosomes can also occur. In gene mutations, there is no change in the chromosome. In chromosome aberrations, there can either be a change in the structure of the chromosome or a change in the chromosome number. The alterations in me chromosome can be in the nature of deletion, duplication, inversion, and translocation. When a part of the chromosome is lost, it is known as deletion. Duplication is the repetition of a chromosome. When a segment of the chromosome breaks but later rejeins after rotating by loO degrees, it is called inversion. If a segment of a chromosome breaks and

relocates within a non-homologous chromosome it is called translocation.

There can be changes in the chromosome number. Each species has a characteristic number of chromosomes which is a reflection of a monoploid number. Those individuals having multiples of monoploid number are called euploids. Euploids having more than twice the number of monoploid are called Polyploids! Organisms with changes that involve individual chromosomes are referred to as Aneuplolds.

Prokaryotic Chromosomes: The prokaryotic celi lacks a nuclear membrane and the genetic material is found in a compact structure called the nucleoid. The chromosome is made up of DNA with associated proteins.- These proteins have close similarities with histones found in Eukaryotes.

Eukaryotic Chromosomes: Unlike the prokaryotic chromosome, the eukaryotic chromosome **is** complex in structure because of greater amounts of DNA per chromosome, more number of chromosomes and the large number of DNA related proteins. The DNA of eukaryotes is associated with postiively charged histones. The .histones and DNA form linear arrays of spherical structures called nucleosomes.

Genetic Disorders in Humans: In 1902, Archibald Garrod and William Bateson reported several disorders that appeared to be inherited. Some of these disorders are :

1. **Sickle Cell Anaemia** : This is caused by the expression of a recessive gene. It is linked to an autosomal chromosome (chromosome 11). In individuals afflicted by this disorder, the red blood cells become elongated and curved- a condition called sickling of the erythrocytes. The red blood cells in such individuals aggregate in the capillary systems of veins and several tissues suffer damage due to shortage of oxygen. These erthrocytes are destroyed more rapidly than the normal red blood cells hence leading to anaemia.
2. **Phenylketonuria** : This is also due to the expression of a recessive gene. It is autosomal and the disorder leads to metabolic errors. It leads to failure of brain to develop in infancy and hence mental retardation. The metabolism in affected individuals fails to convert the amino acid, phenylalanine into the amino acid, tryosine. This leads to overproduction of phenylalanine. This accumulates in the cerebrosopinal fluid leading to mental retardation.
3. **Down**'s **Syndrome** : This is due to imbalance in the chromosome number, first described by Langdon Down in 1866. The afflicted individuals display prominent folding at the corner of the eyes and have short statures. They have small round heads and protruding tongues which causes the mouth to be partially open. They have short, broad hands. In such individuals, the physical, psychomotor ancl mental development is retarded and the life expectancy is shortened. These individuals have an additional chromosome on chromosome 21. That is, there are three copies of chromosome 21 instead of the normal two. Such a condition is called Trisomy. This disorder is produced due to a rare meotic abnormality and hence does not run in families.
4. **Alzheimer's Disease:** It results due to the accumulation of amyloid proteins in the brain leading to degeneration of the neurons. The individuals suffer loss of memoiy as well as general physical impairment. The disease is common among individuals with Down's Syndrome.
5. **Genetic Disorders linked to Sex-Chromosomes :** These are Klinefelter's Syndrome and Turner's Syndrome. In Klinefelter's Syndrome, there, can be 47 or 48 chromosomes. If it is XXY (47 chromosomes) or XXXY (48 chromosomes) sexual differentiation leads to development of males. The males with XYY type of Klinefelter's Syndrome show above average height and subnormal intelligence and are prone to psychopathic tendencies. If the chromosome composition is XXX or XXXX (i.e., 47 and 48 chromosomes) sexual differentiation leads to the development of females who have short statures, poor development of breasts and rudimentary ovaries. If the chromosomes composition is XO (i.e., only 45 chromosomes) the sexual differentiation leads to development of females. This condition is called Turner's Syndrome. Females with Turner's Syndrome have short stature, webbed-neck, lack of secondary sexual characteristics and sterility.