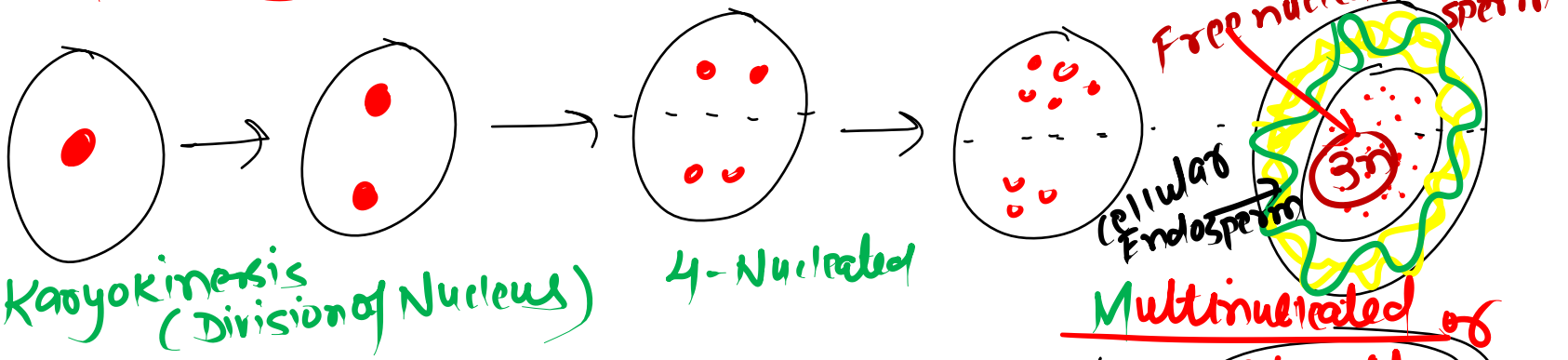
Mitosis

Karyokinesis → Division of Nucleus

Cytokinesis → Division of Cytoplasm

ABLES KOTA

1.

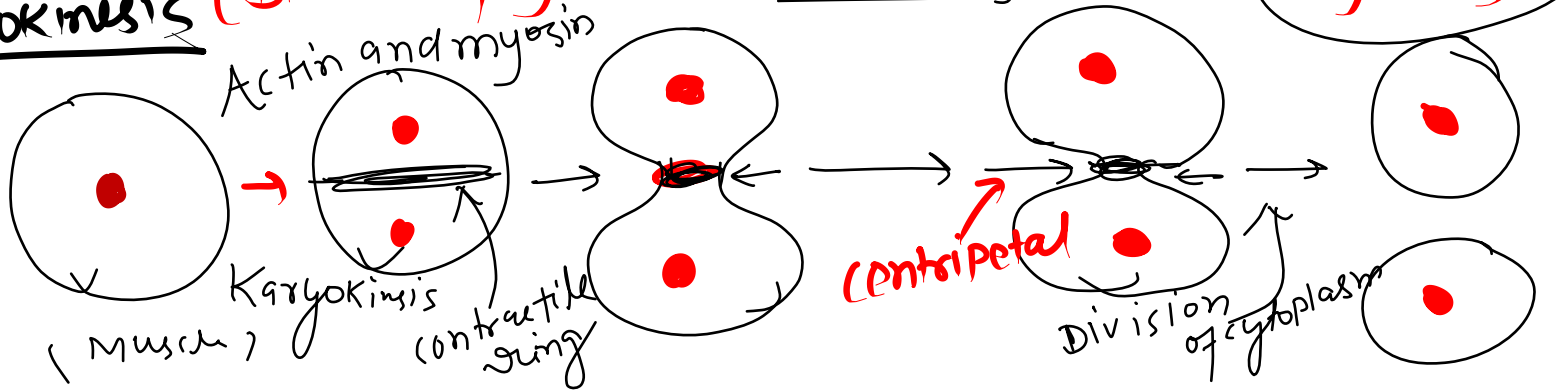


Karyokinesis (Division of Nucleus)

4-Nucleated

Multinucleated of Polyploidy

2. **Cytokinesis** (Division of cytoplasm) Furrow formation



Haemophilia : This sex linked recessive disease, which shows its transmission from unaffected carrier female to some of the male progeny has been widely studied. In this disease, a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this, in an affected individual a simple cut will result in non-stop bleeding. The heterozygous female (carrier) for haemophilia may transmit the disease to sons. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life). The family pedigree of Queen Victoria shows a number of haemophilic descendents as she was a carrier of the disease.

Sickle-cell anaemia : This is an autosome linked recessive trait that can be transmitted from parents to the offspring when both the partners are carrier for the gene (or heterozygous). The disease is controlled by a single pair of allele, Hb^A and Hb^S. Out of the three possible genotypes only homozygous individuals for Hb^S (Hb^SHb^S) show the diseased phenotype. Heterozygous (Hb^AHb^S) individuals appear apparently unaffected but they are carrier of the disease as there is 50 per cent probability of transmission of the mutant gene to the progeny, thus exhibiting sickle-cell trait (Figure 5.15). The defect is caused by the substitution of Glutamic acid (Glu) by

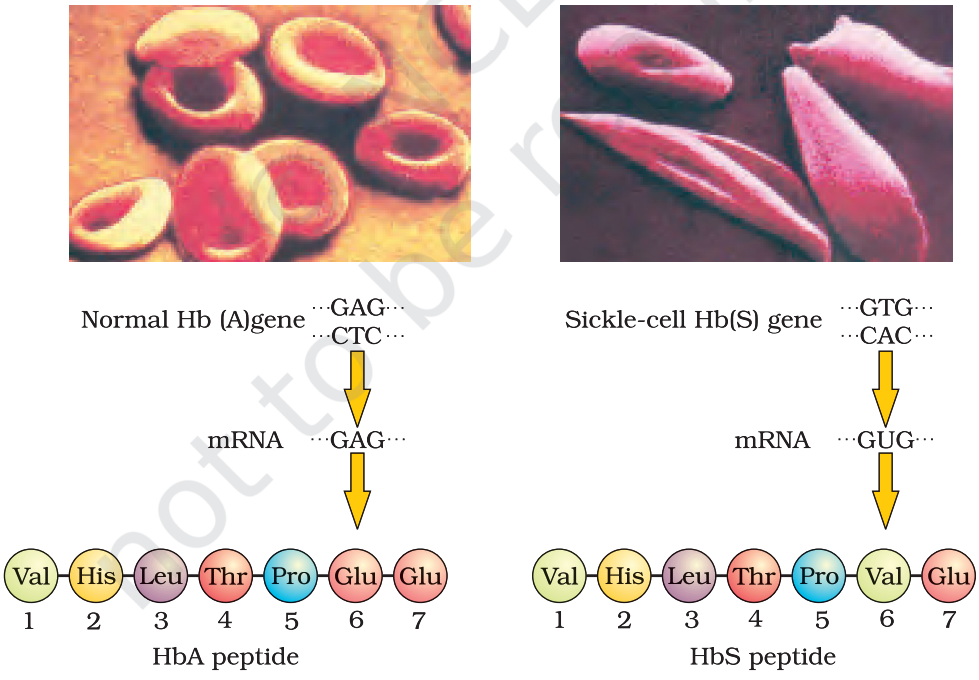


Figure 5.15 Micrograph of the red blood cells and the amino acid composition of the relevant portion of β-chain of haemoglobin: (a) From a normal individual; (b) From an individual with sickle-cell anaemia

Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG. The mutant haemoglobin molecule undergoes polymerisation under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure (Figure 5.15).

Phenylketonuria : This inborn error of metabolism is also inherited as the autosomal recessive trait. The affected individual lacks an enzyme that converts the amino acid phenylalanine into tyrosine. As a result of this phenylalanine is accumulated and converted into phenylpyruvic acid and other derivatives. Accumulation of these in brain results in mental retardation. These are also excreted through urine because of its poor absorption by kidney.

5.6.3 Chromosomal disorders

The chromosomal disorders on the other hand are caused due to absence or excess or abnormal arrangement of one or more chromosomes.

Failure of segregation of chromatids during cell division cycle results in the gain or loss of a chromosome(s), called **aneuploidy**. For example, Down's syndrome results in the gain of extra copy of chromosome 21. Similarly, Turner's syndrome results due to loss of an X chromosome in human females. Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism and,

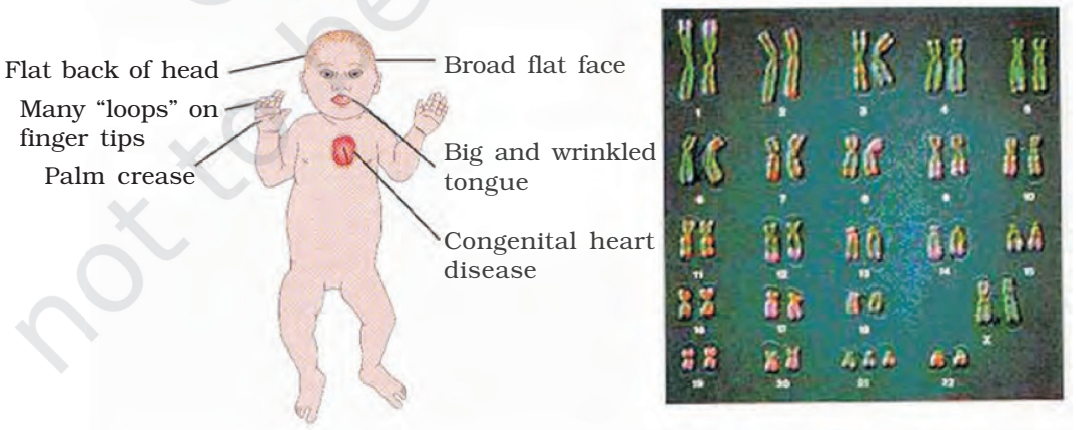


Figure 5.16 *A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual*

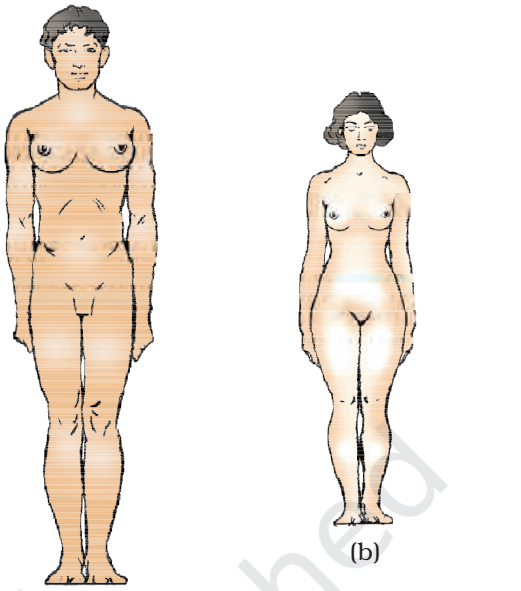
this phenomenon is known as **polyploidy**. This condition is often seen in plants.

The total number of chromosomes in a normal human cell is 46 (23 pairs). Out of these 22 pairs are autosomes and one pair of chromosomes are sex chromosome. Sometimes, though rarely, either an additional copy of a chromosome may be included in an individual or an individual may lack one of any one pair of chromosomes. These situations are known as trisomy or monosomy of a chromosome, respectively. Such a situation leads to very serious consequences in the individual. Down's syndrome, Turner's syndrome, Klinefelter's syndrome are common examples of chromosomal disorders.

Down's Syndrome : The cause of this genetic disorder is the presence of an additional copy of the chromosome number 21 (trisomy of 21). This disorder was first described by Langdon Down (1866). The affected individual is short statured with small round head, furrowed tongue and partially open mouth (Figure 5.16). Palm is broad with characteristic palm crease. Physical, psychomotor and mental development is retarded.

Klinefelter's Syndrome : This genetic disorder is also caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, XXY. Such an individual has overall masculine development , however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed (Figure 5.17 a). Such individuals are sterile.

Turner's Syndrome : Such a disorder is caused due to the absence of one of the X chromosomes, i.e., 45 with X0, Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters (Figure 5.17 b).



Tall stature with feminised character

Short stature and underdeveloped feminine character

Figure 5.17 Diagrammatic representation of genetic disorders due to sex chromosome composition in humans : (a) Klinefelter Syndrome; (b) Turner's Syndrome

SUMMARY

Genetics is a branch of biology which deals with principles of inheritance and its practices. Progeny resembling the parents in morphological and physiological features has attracted the attention of many biologists. Mendel was the first to study this phenomenon systematically. While studying the pattern of inheritance in pea plants of contrasting characters, Mendel proposed the principles of inheritance, which are today referred to as 'Mendel's Laws of Inheritance'. He proposed that the 'factors' (later named as genes) regulating the characters are found in pairs known as alleles. He observed that the expression of the characters in the offspring follow a definite pattern in different–first generations (F_1), second (F_2) and so on. Some characters are dominant over others. The dominant characters are expressed when factors are in heterozygous condition (Law of Dominance). The recessive characters are only expressed in homozygous conditions. The characters never blend in heterozygous condition. A recessive character that was not expressed in heterozygous conditon may be expressed again when it becomes homozygous. Hence, characters segregate while formation of gametes (Law of Segregation).

Not all characters show true dominance. Some characters show incomplete, and some show co-dominance. When Mendel studied the inheritance of two characters together, it was found that the factors independently assort and combine in all permutations and combinations (Law of Independent Assortment). Different combinations of gametes are theoretically represented in a square tabular form known as 'Punnett Square'. The factors (now known as gene) on chromosomes regulating the characters are called the genotype and the physical expression of the chraracters is called phenotype.

After knowing that the genes are located on the chromosomes, a good correlation was drawn between Mendel's laws : segregation and assortment of chromosomes during meiosis. The Mendel's laws were extended in the form of 'Chromosomal Theory of Inheritance'. Later, it was found that Mendel's law of independent assortment does not hold true for the genes that were located on the same chromosomes. These genes were called as 'linked genes'. Closely located genes assorted together, and distantly located genes, due to recombination, assorted independently. Linkage maps, therefore, corresponded to arrangement of genes on a chromosome.

Many genes were linked to sexes also, and called as sex-linked genes. The two sexes (male and female) were found to have a set of chromosomes which were common, and another set which was different. The chromosomes which were different in two sexes were named as sex chromosomes. The remaining set was named as autosomes. In humans, a normal female has 22 pairs of autosomes