***GENETICAL DISORDER.***

*This report is prepared by*

**NAME: ADITIYA KUMAR**

**UNIVERSITY ROLL NO:**

**UNIVERSITY REGISTRATION NO:**

**DEPARTMENT OF CSE (AI & ML)**

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* **ABSTRACT:**
* A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Most cells in the body contain long strands of DNA that provide the cell with instructions. Each DNA strand is tightly coiled around a protein called a histone. This coiled structure is called a chromosome. Chromosomes contain small sections of DNA called genes. These genes provide the body with a specific set of instructions. Each human cell normally contains 23 pairs of chromosomes, with one of each pair provided by each parent. Therefore, a person has two copies of every gene.A change or fault in the DNA can cause a genetic condition. Since genes pass from parent to child, these disorders may be heritable. However, not everyone with a genetic condition in their family will experience symptoms of the disorder. Genetic conditions can affect any gene or chromosome. This means that there are a wide range of genetic disorders, each causing various symptoms. There are many types of genetic disorders, they include-

1. Mendelian Disorders

* Haemophilia
* Cystic fibrosis
* Sickle-cell anemia
* Colour blindness
* Phenylketonuria
* Thalassemia, etc.

1. Chromosomal Disorders

* Down Syndrome
* Turners Syndrome
* Klinefelter Syndrome
* Triple-X Syndrome, etc.
* **INTRODUCTION:**

Broadly, genetic disorders may be grouped into two categories-**1.Mendelian disorders** Mendelian disorders are mainly determined by alteration or mutation in the single gene. These disorders are transmitted to the offspring on the same lines. The pattern of inheritance of such Mendelian disorders can be traced in a family by the pedigree analysis.

A. HAEMOPHILLIA: 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 female has to be at least is extremely rare because mother of such 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.

B. 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^ and Hbs. Out of the three possible genotypes only homozygous individuals for Hbs (HbSHbs) show the diseased phenotype. Heterozygous (Hb^Hbs) 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 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 polymerization under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.

C. PHENYLKETONURIA: This inborn error of metabolism is also inherited as the autosomal resessive trait. The affected individual lacks an enzyme the 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.

**2. 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.

A. DOWN 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. Mental Physical. Psychomotor and development is retarded.

B. 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 express. Such individuals are sterile.

C. TURNER’S SYNDROME: Such a disorder is caused due to the absence of one of the X chromosomes, i.e., 45 with XO. Such females are sterile as ovaries are rudimentary besides other features including lack of other secondary sexual characters

* **SUMMARY:**
* Genetic disorders occur as a result of a mutation to DNA. This mutation may affect whole chromosomes or the specific genes within chromosomes. DNA mutations may also happen within the DNA of mitochondria, which power a person’s cells.
* Most genetic conditions are heritable, but some can occur for the first time within the person who experiences the disorder.
* Genetic disorders are lifelong conditions. For this reason, treatments tend to focus on helping a person manage the symptoms, preventing complications, and improving quality of life.
* In some cases, there may be medications available to help slow the progression of a particular disease.