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Importance of Human Genetic Disorders and its Symptoms

Campos Coleman*

Department of Genetic Medicine, Vanderbilt University Medical Center, Nashville, United States of America

***Corresponding author:** Campos Coleman, Department of Genetic Medicine, Vanderbilt University Medical Center, Nashville, United States of America; E-mail: coleman.c@gmail.com

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Description

Genetic disorders are a wide range of conditions caused by errors in an individual's DNA. These disorders can range from mild to severe and can be inherited or acquired. They can affect any part of the body and may appear at any age. Genetic disorders can be caused by various factors such as mutations, deletions, or duplications of genes, chromosomal abnormalities, or changes in the number of chromosomes. There are thousands of recognized genetic disorders, and new systems are being discovered all the time. Some of the most common genetic disorders include Down syndrome, cystic fibrosis, sickle cell anemia, Huntington's disease, and muscular dystrophy. Many of these disorders are inherited from one or both parents and can be passed down from generation to generation. Inherited genetic disorders are caused by mutations in specific genes or changes in the number or structure of chromosomes. There are three types of inherited genetic disorders: Single-gene disorders, chromosomal disorders, and multifactorial disorders.

Single-gene disorders are caused by a mutation in a single gene and are usually inherited in a dominant or recessive manner. Dominant disorders occur when a person inherits a mutated gene from one parent and a normal gene from the other parent. In contrast, recessive disorders occur when a person inherits two copies of a mutated gene, one from each parent. Chromosomal disorders are caused by abnormalities in the number or structure of chromosomes. Down syndrome is an example of a chromosomal disorder and it is caused by an extra duplicate of chromosome. Other chromosomal disorders can result from missing or duplicated chromosomes or from changes in chromosome structure. Multifactorial disorders are caused by a combination of genetic and environmental factors. These disorders are often complex and can be influenced by multiple genes and environmental factors such as diet, lifestyle, and exposure to toxins.

Genetic disorders can affect any part of the body, and the symptoms and severity of these disorders can vary widely. Some genetic disorders may cause physical abnormalities such as facial abnormalities or skeletal deformities, while others may affect the function of internal organs such as the heart, lungs, or liver. Some genetic disorders may also affect cognitive function, resulting in intellectual disability or developmental delays. Diagnosing genetic disorders typically involves a combination of medical history, physical examination, and genetic testing. Genetic testing can identify specific mutations or chromosomal abnormalities that may be responsible for a person's symptoms or condition. Genetic testing can also be used for prenatal diagnosis to determine if a fetus has inherited a genetic disorder.

Treatment for genetic disorders varies depending on the specific disorder and the severity of the symptoms. Some genetic disorders have no cure, and treatment is focused on managing symptoms and improving quality of life. Other genetic disorders may be treated with medications, surgery, or gene therapy. Gene therapy is an emerging field that holds a lot of potential for the treatment of genetic disorders. This process requires implementing a healthy version of a mutated gene into the body in order to repair the underlying genetic defect. Gene therapy has shown stimulating results in the treatment of some genetic disorders such as spinal muscular atrophy and severe combined immunodeficiency.

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