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Editorial

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Medical Genetics and Their Current Medical Practice

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Introduction

Medical genetics is a field of medicine that deals with the diagnosis and treatment of hereditary diseases. Human genetics is a branch of scientific inquiry that may or may not apply to medicine, whereas medical genetics refers to the use of genetics to medical care. Genetic medicine is a newer term for medical genetics that encompasses fields like gene therapy, personalized medicine, and predictive medicine, a rapidly growing new medical specialty. Health disorders like birth defects and dysmorphology, intellectual disabilities, autism, mitochondrial disorders, skeletal dysplasia, connective tissue disorders and cancer genetics comes under the field of medical genetics. Within the perspective of nondirective counselling, genetic counselling is the act of delivering information regarding genetic diseases, diagnostic testing, and dangers in other family members. Depending on the illness, the genetic counselor's specific job differs. Genetic counsellors usually work alongside geneticists and specialise in paediatric genetics, which focuses on developmental problems in newborns, infants, and children. Pediatric counseling's main purpose is to explain the genetic foundation for a child's developmental difficulties in a sympathetic and articulate manner that helps possibly anxious or frustrated parents to grasp the information.

Diagnosis

Each patient will have a diagnostic evaluation that is personalised to their specific signs and symptoms. A differential diagnosis will be made by the geneticist, and necessary tests will be recommended. These tests could look for chromosomal abnormalities, metabolic inborn defects, or single gene illnesses.

Chromosome Studies

In a general genetics clinic, chromosome studies are done to discover the aetiology of developmental delay/mental retardation, birth abnormalities, dysmorphic characteristics, and/or autism. Prenatal chromosome analysis is also used to identify whether a foetus is impacted by aneuploidy or other chromosome rearrangements. Finally, cancer samples frequently reveal chromosomal abnormalities. Karyotype chromosomal analysis, Fluorescence in situ hybridization (FISH), Chromosome painting, Array comparative genomic hybridization are different developmental methods for chromosomal analysis.

Biochemical Studies

Biochemical tests are used to look for metabolite abnormalities in biological fluids such as blood (plasma/serum) or urine, but also in cerebrospinal fluid (CSF). Under rare circumstances, specific enzyme function tests are also used. The newborn screen in the United States includes biochemical tests to look for curable disorders like galactosemia and phenylketonuria (PKU). Quantitative amino acid analysis, Urine organic acid analysis and acylcarnitine combination profile are few tests performed to identify the disorders like Urea cycle disorder, cystinuria, in detection of excretion of unusual organic acids etc.

Molecular Studies

DNA sequencing is a technique for determining a gene's genomic DNA sequence. Only the exons that code for the expressed protein, as well as a tiny portion of the flanking un-translated regions and introns, are examined in general. As a result, despite their great specificity and sensitivity, diagnostic tests do not always detect all of the mutations that potentially cause disease.

Treatment and Management of The Symptoms

The genetic information (DNA) is wrapped up in structures called chromosomes in each cell of the body. Because genetic disorders are usually the consequence of chromosome or gene changes, there is presently no medication that can cure genetic anomalies in every cell of the body. As a result, there is no "cure" for genetic illnesses at this time. However, there is medicine available to manage the symptoms of many hereditary disorders.

The cause of disease is well recognized in some situations, particularly inborn errors of metabolism, and gives the possibility of nutritional and medicinal intervention to avoid or lessen long-term problems. Infusion therapy is utilized to replace the lost enzyme in other circumstances. The use of gene therapy or other novel drugs to treat certain genetic problems is currently being researched. Enzyme shortages disturb normal metabolic processes, which causes metabolic diseases.

Diet and Medication

In some well-known metabolic disorders, dietary restriction and supplementation are major therapies performed. The diet will fluctuate depending on the caloric needs of the growing child, and if a woman has one of these illnesses, particular attention will be required during pregnancy.

Enhancements of residual enzyme activity, inhibition of other enzymes in the biochemical route to prevent hazardous compound buildup, or redirection of a poisonous substance to another form that may be expelled are all medical treatments.

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Infusions of a recombinant enzyme (made in a laboratory) are used to treat some lysosomal storage illnesses, which can minimise the buildup of chemicals in various tissues.

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