Commentary

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Genetic Disorders: A literature Review

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Introduction

What is a genetic disorder?

A genetic disorder is a disease caused wholly or partly caused by changes in a person's DNA i.e. sequence different from the normal sequence. It can be caused by a mutation in one gene, multiple genes, by a combination of gene mutations and environmental factors, or by damage to chromosomes. Mutations can be caused due to error in DNA replication or due to environmental factors (cigarette smoke, UV light) which leads to changes in the DNA sequence.

DNA provides the code for making proteins (molecules that performs most of the functions in human body). When a section of DNA is changed in some way, the protein it codes for also gets affected and may not be able to carry out its normal function. Depending on the source of the mutation occurrence, there might be no effect or little or may also highly alter the biology of cell, which results in a genetic disorder.

Some diseases are caused by mutations which a person inherits from parents since birth. Other diseases can be caused by acquired mutations in a gene or group of genes occurring during life-stages. Such mutations occur either randomly or due to environmental exposure.

Genetic disorders are grouped in 4 categories

Single gene disorders: This type disorders are caused mainly due to the defects in one particular gene. It is also known as Mendelian or monogenetic disorder. It has 3 different patterns of inheritance

- it occurs when an individual has one copy of a defective gene.
- it occurs when an individual has two copies of a defective gene.
- it occurs when the defective gene is present on X-chromosome. It is more common in males as they have only one X chromosome, as a result a single defective gene can cause the symptoms to occur.

Few examples of single-gene disorders are: hemochromatosis, cystic fibrosis, alpha- and beta-thalassemias, Marfan syndrome, fragile X syndrome, Huntington's disease, and sickle cell anaemia.

Multifactorial disorders: This type disorders are caused by defects in multiple genes, often due to complex interaction with environmental and lifestyle factors (diet). It is also known as complex or polygenic disorder.

Few examples of multifactorial disorders are: cancer, heart disease, diabetes, Alzheimer's disease, high blood pressure, arthritis, and obesity.

Chromosome disorders: This type disorders are caused due to changes in the number or structure of the chromosomes, as chromosomes are the carriers of the genetic material, so it can result in disease. This type of disorder mainly arises due to problem in cell division. It may also due to chromosomal translocation, in which portions of two chromosomes are exchanged.

Few examples of chromosome disorders are: Down syndrome, Turner syndrome, Cri du chat syndrome, Klinefelter syndrome.

Mitochondrial genetic disorders: It is caused by mutations in the non-nuclear DNA of mitochondria. Each mitochondrion contains 5-10 circular pieces of DNA. As egg cells, keep their mitochondria during fertilization. Hence, this type of disorder is always inherited from female parent.

Few examples of mitochondrial genetic disorders are: mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS), a rare form of dementia; Leber's hereditary optic atrophy (LHON), an eye disease; and myoclonic epilepsy with ragged red fibers (MERRF).