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Editorial Note on Genetic Diseases

Jiang SW*

Professor, Mercer University School of Medicine, USA

Editorial

A genetic disease is any disease which is caused by an abnormality in the genetic makeup of an individual. The genetic abnormality can range from minuscule to major from a discrete mutation in a single base in the DNA of a single gene to a gross chromosomal abnormality involving the addition or subtraction of an entire chromosome or set of chromosomes. Few people inherit genetic disorders from the parents; some acquired changes or mutations in a pre-existing gene or group of genes cause other genetic diseases. Genetic mutations can occur either randomly or due to some environmental exposure.

There are various types of genetic disorders which includes single gene inheritance, multifactorial inheritance, chromosome abnormalities and mitochondrial inheritance. All genetic disorders are present before birth, and some genetic disorders produce birth defects, but many birth defects are developmental rather than hereditary. The opposite of a hereditary disease is an acquired disease. Most cancers involve genetic mutations to a small proportion of cells in the body are acquired diseases. Some family cancer syndromes such as BRCA mutations are hereditary genetic disorders.

Genetic disorders may be complex, multifactorial, or polygenic, meaning they are likely associated with the effects of multiple genes in combination with lifestyles and environmental factors. A multifactorial disorder includes heart disease and diabetes. Although complex disorders often cluster in families, they do not have a clear-cut pattern of inheritance which makes it difficult to determine a person's risk of inheriting or passing on these disorders. A chromosomal disorder is a missing, extra, or irregular portion of chromosomal DNA. It can be from an atypical number of chromosomes or a structural abnormality in one or more chromosomes.

Gene therapy refers to a form of treatment where a healthy gene is introduced to a patient. This should alleviate the defect caused by a faulty gene or slow the progression of the disease. A major obstacle has been the delivery of genes to the appropriate tissue, cell and organ affected by the disorder. Researchers have investigated how they can introduce a gene into the potentially trillions of cells which can carry the defective copy.

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***Address for Correspondence:** Dr. Shi Wen Jiang, Professor, Mercer University School of Medicine, USA, E-mail: jiang_s@mercer.edu*

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