About Genetic Diseases

Genetic diseases are defined as diseases caused by aberrations of genetic material. Therefore, these diseases can potentially be passed from generation to generation. However, not every patient has a family history of a similar problem. This is because new mutations can occur when an individual inherits his/her genetic material from his normal parents.

To understand genetic diseases, we must first learn what chromosomes and genes are. Chromosomes are the “visible” genetic materials present in the cell nucleus and are mainly composed of DNA and histone proteins. With the exception of a few cell types, such as red blood cells, sperms and eggs, every human cell carries 23 pairs of chromosomes, including 22 pairs of autosomes and one pair of sex chromosomes. Genetic diseases occur when there is an abnormal number or structure of the chromosomes. Gene is the basic unit of genetic information and is composed of deoxyribonucleic acid (DNA). There are over 20,000 genes distributed over the 23 pairs of chromosomes. Each gene defines different function, controlling the various activities of the human body. The relationship between the cell nucleus, chromosomes, genes and DNA, can be illustrated by an analogy to a small library. The nucleus is the library and each chromosome is a book of the library. Each of the genes is a chapter in a book and DNA is the text of each chapter.

Genetic diseases could be broadly divided into three groups.

1. Chromosomal abnormalities

These are caused by abnormal number or structure of the chromosomes. A well-known example of this group is Down syndrome.

2. Single gene disorders

These are caused by abnormalities or mutations in individual genes. An example is the Thalassemia. These disorders follow specific modes of inheritance: autosomal dominant, autosomal recessive, sex-linked dominant and sex-linked recessive.

3. Multi-factorial disorders

These diseases are caused by a combination of multiple genetic and environmental factors. Examples of these diseases include diabetes mellitus, hypertension, mental illnesses and cancer.

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