Know More About Genetic Disease

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1. What is a genetic disease?

Most people have the concept that a genetic disease must be one that is transmitted from one generation to the next. Actually this is not totally correct. In medicine, a genetic disease refers to one that is caused by abnormalities of the genetic material at the stage of germ cell or early embryo.

2. Do all genetic diseases have a family history?

Genetic diseases often have the characteristic of being heritable through generations, but it does not mean every case must have a positive family history. It is not uncommon to see sudden appearance of a genetic disease in a family without precedents. The affected one becomes the first case in the family. One reason is that new mutations can arise when the genetic material is passed on from one generation to the next. Another reason is autosomal recessive inheritance.

3. Are genetic diseases necessarily congenital?

Congenital diseases are those that are evident at birth or early infancy. Generally speaking, a lot of genetic diseases do manifest at birth and thus are congenital. On the other hand, a lot of congenital diseases are hereditary or have a significant genetic factor. Nevertheless, quite a number of congenital diseases are not at all hereditary. For instance congenital defects or malformations caused by intrauterine infection or maternal consumption of drugs that are harmful to the developing embryo or growing fetus. Finally, genetic diseases do not necessarily manifest at birth or in early infancy. Examples are colour blindness and Duchenne muscular dystrophy, both of which do not manifest until the baby reaches a certain age. There are even genetic diseases like Huntington's disease that do not usually manifest until adulthood.

4. Are familial disease necessarily genetic?

Familial diseases are those that demonstrate clustering within families. In other words, these diseases often affect more than one members within a family. Genetic diseases by nature are often familial, due to sharing of common genetic material among family members. However, familial clustering does not necessarily indicate that the disease is genetic. For example, hepatitis B shows familial clustering but is not a genetic disease; familial clustering sometimes originates from the common living environment and similar lifestyles.

5. What is a gene?

Gene is the unit for genetic information. Human being has about 30,000 genes distributed over 23 pairs of chromosomes. Every gene has a different function, controlling all kinds of biological activities including embryological development, fetal growth, metabolism, personality, cognition and proliferation.

6. What are Chromosomes?

Chromosomes are the genetic material within a cell nucleus. Except some specialized cells like red blood cells and gametes, every cell in our body carries 23 pairs of chromosomes. Twenty-two pairs are known as autosomes and show no sex difference. The remaining pair are known as sex chromosomes; females have a pair of X, while males have both an X- and a Y-chromosomes. When the number or structure of chromosomes goes wrong, a genetic disease is the result. A classic example is Down syndrome, which is caused by trisomy (having 3 copies instead of 2) of chromosome 21. Another example is Turner syndrome, in which case affected females have monosomy (having 1 copy instead of 2) of X chromosome.
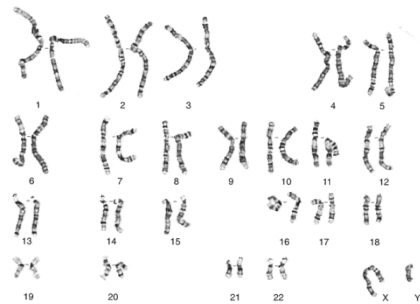
7. What is DNA?

DNA (deoxyribonucleic acid) is a long and thin molecule with a double helix structure, which is the result of two complementary chains twisted about each other. The chains are composed of four different nucleotides, namely Adenine, Guanine, Cytosine and Thymine. Every gene is a strand of DNA with a unique sequence (the order of the nucleotides) that specifies the genetic information. If the DNA sequence of a gene goes wrong, the information conveyed by the gene will be faulty, resulting in a genetic disease.

8. The relationship between DNA, genes and chromosomes.

One can imagine that each person has 46 books, each book consisting of thousands of chapters, which are written in words. In this analogy, the words are equivalent to DNA, the chapters are equivalent to the genes, and the books are equivalent to the chromosomes.

9. Categories of genetic diseases.

Genetic diseases can largely be divided into three categories:

A. **Chromosomal disorders** - those that are caused by numerical or structural abnormalities of the chromosomes, e.g. Down syndrome, Turner syndrome.

B. **Single gene disorders** - those that are caused by defects of individual genes, e.g. thalassaemia, haemophilia. Gene defects are not detectable by chromosome analysis.

C. **Multifactorial disorders** - those that are the result of the combined effect of genetic factors and environmental factors, e.g. diabetes mellitus, hypertension, psychiatric diseases and cancer.

10. Is chromosome analysis useful?

By means of chromosome analysis, we can detect numerical or structural abnormalities of the chromosomes. Chromosomal disorders such as Down syndrome or Turner syndrome can then be diagnosed. Nevertheless, a normal result of chromosome analysis does not mean exclusion of genetic disease, because abnormalities with DNA sequence cannot be detected by chromosome analysis. To use the previous analogy, even if the whole set of books are present and all the pages intact, typing errors can occur that lead to erroneous interpretation of the information.