Genetic disease

A genetic disease is any disease 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. Some people inherit genetic disorders from the parents, while acquired changes or mutations in a preexisting gene or group of genes cause other genetic diseases. Genetic mutations can occur either randomly or due to some environmental exposure.

There are a number of different types of genetic disorders (inherited) and include:

1. Single gene inheritance
2. Multifactorial inheritance
3. Chromosome abnormalities
4. Mitochondrial inheritance

Gene inheritance disorders

Single gene inheritance is also called Mendelian or monogenetic inheritance. Changes or mutations that occur in the DNA sequence of a single gene cause this type of inheritance. There are thousands of known single-gene disorders. These disorders are known as monogenetic disorders (disorders of a single gene).

Single-gene disorders have different patterns of genetic inheritance, including

- autosomal dominant inheritance, in which only one copy of a defective gene (from either parent) is necessary to cause the condition;
- autosomal recessive inheritance, in which two copies of a defective gene (one from each parent) are necessary to cause the condition; and
- X-linked inheritance, in which the defective gene is present on the female, or X-chromosome. X-linked inheritance may be dominant or recessive.

Some examples of single-gene disorders include

1. Cystic fibrosis,
2. alpha- and beta-thalassemias,
3. sickle cell anemia (sickle cell disease),
4. Marfan syndrome,
5. fragile X syndrome,
6. Huntington's disease, and
7. Hemochromatosis.

Multifactorial genetic inheritance disorders

Multifactorial inheritance is also called complex or polygenic inheritance. Multifactorial inheritance disorders are caused by a combination of environmental factors and mutations in multiple genes. For example, different genes that influence breast cancer susceptibility have been found on chromosomes 6, 11, 13, 14, 15, 17, and 22. Some common chronic diseases are multifactorial disorders.

Examples of multifactorial inheritance include

Heart disease, high blood pressure, Alzheimer's disease, arthritis, diabetes, cancer, and obesity.

Multifactorial inheritance also is associated with heritable traits such as fingerprint patterns, height, eye color, and skin color.

Chromosomal abnormalities

Chromosomes, distinct structures made up of DNA and protein, are located in the nucleus of each cell. Because chromosomes are the carriers of the genetic material, abnormalities in chromosome number or structure can result in disease. Chromosomal abnormalities typically occur due to a problem with cell division.

For example, Down syndrome (sometimes referred to as "Down's syndrome") or trisomy 21 is a common genetic disorder that occurs when a person has three copies of chromosome 21. There are many other chromosomal abnormalities including:

1. Turner syndrome (45,X0),
2. Klinefelter syndrome (47, XXY), and
3. Cri du chat syndrome, or the "cry of the cat" syndrome (46, XX or XY, 5p-).

Diseases may also occur because of chromosomal translocation in which portions of two chromosomes are exchanged.

Mitochondrial genetic inheritance disorders

This type of genetic disorder is caused by mutations in the non-nuclear DNA of mitochondria. Mitochondria are small round or rod-like organelles that are involved in cellular respiration and found in the cytoplasm of plant and animal cells. Each mitochondrion may contain 5 to 10 circular pieces of DNA. Since egg cells, but not sperm cells, keep their mitochondria during fertilization, mitochondrial DNA is always inherited from the female parent.

Examples of mitochondrial disease include

1. Leber's hereditary optic atrophy (LHON), an eye disease;
2. myoclonic epilepsy with ragged red fibers (MERRF); and
3. Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS), a rare form of dementia.