# GENETIC DISEASES

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# ГЕНЕТИЧЕСКИЕ ЗАБОЛЕВАНИЯ

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**Abstract.** This article deals with thegenetic diseases, the causes of their appearing and thestructure of the DNA molecule.

**Аннотация.** В данной статье пойдет речь о генетических заболеваниях, причинах их вызывающих истроении молекулы ДНК.

**Keywords:** genetic diseases, genetic mutations, DNA, heredity.

**Ключевые слова:** генетические заболевания, генетические мутации, ДНК, наследственность.

When most people consider the genetic basis of disease, they might think about the rare, single gene disorders, such as cystic fibrosis (CF), phenylketonuria or haemophilia, or perhaps even cancers with a clear heritable component (for example, inherited predisposition to breast cancer).

However, although genetic disorders are individually rare, they account for approximately 80% of rare disorders, of which there are several thousand. The complete number of rare disorders means that, collectively, approximately 1 in 17 individuals are affected by them.

We can find a lot of differences in our DNA. Some of these differences might render an individual more susceptible to one disorder (for example, a type of cancer), but could render the same individual less susceptible to develop an unrelated disorder (for example, diabetes). The environment (including lifestyle) plays a significant role in many conditions (for example, diet and exercise in relation to diabetes).

In fact, most cancers result from an accumulation of genetic changes that occur through the lifetime of an individual, which may be influenced by environmental factors. Clearly, understanding genetics and the genome as a whole and its variation in the human population, are integral to understanding disease processes and this understanding provides the foundation for curative therapies, beneficial treatments and preventative measures.

We would like to say about DNA and it’s changes in details.DNA- is a polymer composed of two polynucleotide chains that coil around each other to form a double helix.The two DNA strands are known as polynucleotides as they are composed of simpler monomeric units
called nucleotides. Each nucleotide is composed of one of four nitrogen-containing nucleobases (cytosine, guanine, adenine, thymine), a sugar called deoxyribose, and a phosphate group. The nucleotides are joined to one another in a chain by covalent bondsbetween the sugar of one nucleotide and the phosphate of the next.

A geneticist’s characterization of mutation is ‘any heritable change to the DNA sequence’, where heritable refers to both somatic cell division (the proliferation of cells in tissues) and germline inheritance (from parent to child). Such changes to the DNA may have no consequences but sometimes lead to observable differences in the individual (the ‘phenotype’). So, in the past such alterations in the human population, particularly when they were associated with a disease state, were referred to as ‘mutations’.

Single-gene syndromes have different patterns of genetic inheritance, includingautosomal 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; andX-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 : cystic fibrosis,alpha- and beta-thalassemias, sickle cell anemia, Marfan syndrome, fragile X syndrome, Huntington's disease, and haemochromatosis.

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

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 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:Turner syndrome (45, X0),Klinefelter syndrome (47, XXY) and "Cry of the cat" syndrome (46, XX or XY).

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