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GENETICS OF NEUROMUSCULAR DISEASES COMPLICATIONS

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Abstract

Neuromuscular diseases are one of the numerous groups of hereditary diseases, characterized by muscle diphunctions, a decrease or loss of control of movements. The occurrence of these diseases is associated with a defect in embryonic development or genetically determined pathology. The purpose of this article is to highlight neuromuscular diseases in a new interpretation and to take measures to prevent it.

Keywords: Neuromuscular, hereditary diseases, peripheral neuron, motor neuron, myopathy, myoplegia, myasthenia gravis, diagnosis, treatment usils.

Introduction

Neuromuscular diseases affect the peripheral nervous system and muscles. Therefore, the main effect of neuromuscular diseases depends on the ability to perform voluntary movements. Neuromuscular diseases lead to significant disability, including, in the most extreme case, almost complete paralysis. Neuromuscular diseases include some of the most devastating diseases that affect humanity, such as motor neuron disease. Neuromuscular diseases appear at any time, that is, from the development of the child in the uterus to old age. Most often they have genetic peculiarities. The last 25 years have been the Golden Age of genetics, with disease genes now identified that are responsible for many genetic neuromuscular diseases. Neuromuscular disorders can progress to avlots as autosomal dominant, autosomal recessive, or X-linked signs. They can also be caused by mitochondrial DNA mutations or de novo mutations that are not present in your parent's peripheral blood DNA.

For Neuromuscular Diseases, the following symptoms are characteristic: weakness, muscle atrophy, spontaneous muscle twitching, spasms, insomnia, etc. If the neuromuscular connections are impaired, patients can experience several manifestations of eyelid drooping, bilateral vision and muscle weakness, which only intensify during the day. In some cases, there may be a violation of the function of swallowing and breathing.

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It can be divided into four main groups, depending on the location of Neuromuscular Diseases.

- muscles;
- neuromuscular nodes;
- peripheral nerves
- motor neuron

Myopathy

The term myopathy (myodystrophy) combines a very large group of diseases combined with a common symptom: primary damage to muscle tissue. The development of myopathy can be caused by various factors: heredity, viral damage, metabolic disorders, etc. Inflammatory myopathies (myositis) include diseases that arise from the inflammatory process. They develop as a result of autoimmune diseases and can be accompanied by other diseases of a similar nature. This is dermatomyositis, polymyositis, myositis with various additives.

Mitochondrial myopathies. The cause of the disease is structural or biochemical mitochondria. This type of disease includes:

Cairns-Sayre syndrome;

- Mitochondrial encephalopathy

In addition to these diseases, there are a number of rare types of myopathies that affect the central nucleus, endocrine system, etc.

MYOPLEGIA

Like myopathy, these are hereditary neuromuscular diseases characterized by muscle weakness or paralysis of the limbs. The following types of myoplegia exist:

- - hypokalemic;
- hypercalemic;
- normokalemic..

The myoplegia attack is caused by the redistribution of organisms calci - its sharp decrease in intercellular fluid and plasma, and proliferation (proliferation) in cells is observed. In muscle cells, membrane polarization is disrupted, the electrolytic properties of muscles change. During an attack, a sharp weakness of the limbs or trunk develops in the bavmor, can affect the tomog, larynx and respiratory tract. It can be fatal.

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MYASTHENIA GRAVIS

The disease is most often affected by women (2/3 of patients). It has two forms-congenital and acquired. In this disease, there is a violation of the transmission of nerve impulses, as a result of burning, the linear muscles are weakened. The disease is associated with a change in the functions of the neuromuscular system. Muscle weakness affects the normal functioning of organs: be morning eyelids can always be semi-closed, urination is impaired, chewing and walking are more difficult. As a result, the disease can lead to disability and even death.

MOTOR NEURON DISEASES

Motor neuron diseases are characterized by damage to motor neurons in the brain and spinal cord. The gradual death of cells affects the work of the muscles: they gradually weaken, and the affected area increases. Brain neurons responsible for movement are located in the cerebral cortex. Their branches - axons-fall into the area of the spinal cord, where communication with the neurons of this section occurs. This process is called Synapse. As a result, a neuron in the brain emits a special chemical (transmitter) that transmits signals to the neurons of the spinal cord.

Emeri-Dreyfus myodystrophy. X is a gradually progressive form of linked recessive inheritance type myodystrophy, which is mainly caused by a mutation in the cytoskeletal muscle protein-emerin gene, which forms in skeletal, smooth muscles and cardiomyocytes. The disease begins at the age of 5 to 15 years. The earliest and typical symptoms are increased flexion contractures in the extensors of the elbow joints and arms, and retarded Achilles tendons. As a rule, at the age of 12, patients are diagnosed with significant contracture in the knee, ankle and elbow joints. Then the weakness and atrophy of the biceps and triceps muscles of the shoulder, and later - the deltoid and other muscles of the shoulder girdle are weakened states. In some cases, walking along the toes and outer edges, which occurs at about 5 years of age, is noted as the first symptom. Until this point, the motor development of children is usually not disturbed. Muscle weakness is not felt and grows slowly. At about 20 years of age, relative stability appears. The ability to walk and climb stairs is preserved. The facial muscles are not affected. Muscle weakness is present in the arms (scapular-humeral) and legs (peroneal). Technique and pseudohypertrophy of the gastrocnemius muscles may be absent. Tendon reflexes are not triggered. The muscles of the back cervix often contract, restrictions are noted. Frequent and prognostic important symptoms of the

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disease are cardiac conduction disorders and enlarged or hypertrophic cardiomyopathy. Cardiomyopathy can be complicated by the development of atrial paralysis due to fibrosis of the pacemakers of the sinus node. In such cases, an urgent implantation of an artificial pacemaker is indicated. Bradycardia attacks and seizures can in some cases precede muscle weakness, but often occur in the third decade of life. Changes in the cardiac conduction system are not always detected in a standard ECG study, but monitoring can detect atrioventricular blockages and Samoylov-Venskebax periods. Irreparable arrhythmia with the implantation of an artificial pacemaker can lead to stroke and death of the patient. The vital prognosis for Emery-Dreyfus myodystrophy depends entirely on the extent of the heart injury.

Diagnostics

In order to make the correct diagnosis, it is important to carry out the following studies:

- biochemical. Determination of muscle enzymes, first of all creatine phosphokinase (CPK). Myoglobin and aldolase levels are determined;
- electrophysiological. Electromyography (EMG) and electroneuromiography (ENMG) help differentiate between primary and secondary myopathy. They will help determine, first of all, what affects it - the spinal cord or peripheral nerve;
- pathological and morphological. They consist of a muscle biopsy. Studying the material also helps to distinguish between primary and secondary myopathy. Determination of dystrophin content makes it possible to distinguish myopathy of Duchenne from Muscular Dystrophy of Becker, which is important for proper treatment;
- DNA Diagnostics.The study of DNA leukocytes makes it possible to identify hereditary diseases in 70% of patients.

TREATMENT OF NEUROMUSCULAR DISEASES

When one of the diagnoses associated with neuromuscular diseases is made, in each case, treatment is selected individually, taking into account all the analyzes taken. The patient and his relatives should initially understand that this is a long and very complex process that requires significant financial costs. Difficulties in prescribing treatment are also associated with the fact that it is not always possible to accurately determine the primary metabolic defect. In this case, the disease develops constantly, which means that treatment should be aimed

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primarily at slowing down the development of the disease. This helps to maintain the patient's ability to care for himself and affects his quality of life.

METHODS OF TREATMENT OF NEUROMUSCULAR DISEASES

- Correction of skeletal muscle metabolism. Drugs that stimulate metabolism, potassium preparations, vitamin complexes, anabolic steroids;
- Stimulation of the Segment apparatus. Neurostimulation, myostimulation, reflexology, balneotherapy, physiotherapy exercises (exercises and load are selected individually);
- Correction of blood circulation. Various types of massage, thermal treatment of certain areas, oxygen barotherapy;
- Diet and parenteral nutrition provide the body with all the necessary nutrients - protein, potassium salts, vitamins of the desired group;
- Correction classes with orthopedics. Correction of contractures, deformities of the chest and spine, etc.

No medicine has been invented today, any person from taking it will instantly become absolutely healthy. For all the complexity of the situation, it is important for a patient with neuromuscular disease to continue his best life. An example of Hawking, who has been confined to a wheelchair for over 50 years but continues his research, suggests that illness is not a reason to give up.

Conclusion

The conclusion is that there are genetic and organized forms of Neuromuscular Diseases, which should be given special importance in their treatment. If the treatment work is not carried out on time, it leads to very severe symptoms, as well as cases of evn scientist.

Morphological, immunohistochemical and electron microscopy methods of studying biopsy samples play a large role in diagnostics. During mild biomicroscopy, the state of muscle fibers helps distinguish primary myogenic atrophy from secondary denervation (neurogenic or myelogenous) amyotrophy. Histochemical analysis of biopsy is necessary to identify specific metabolic defects in muscle tissue. Electron microscopy discovered a whole class of diseases combined with the concept of" systemic myopathy". An etiological and pathogenetic treatment has been developed for many diseases of muscles, neuromuscular synapses, peripheral nerves and motoneurons. In other cases, therapy is aimed at slowing the progression of the disease, prolonging the duration of remission and improving the patient's quality of life. The treatment of

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neuromuscular diseases requires the joint efforts of neurologists and rehabilitologists. Treatment tactics depend on the severity of the disease and the rate of development.

References

1. O`zbekiston milliy ensiklopediyasi.
2. ibbiyot Ensiklapediyasi
3. NEUROMUSCULAR DISEASES DURING DEVELOPMENT 1. Cornelio. F. Lanzi. G and Fedrizzi. E
4. ttps://www.mayoclinic.org
5. https://buhconsul.ru/uz/nervno-myshechnyezabolevaniya-chto-takoe-nervno-myshechnye/