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Diagnostics of the Origin of Myopathies and Modern Clinical Diagnostic Methods

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Abstract: One of the congenital forms of myopathy is progressive Duchenne muscular dystrophy, which manifests itself in preschool children and is characterized by a deficiency of dystrophin function, manifested by a decrease in the strength, stability and functionality of myofibrils. The information in this section cannot be used for self-diagnosis and self-treatment. In case of pain or other exacerbation of the disease, diagnostic tests should be prescribed only by the attending physician. You should consult your doctor for a diagnosis and the appointment of the correct treatment.

Key words: nemaline myopathy, multinucleated myopathy, centronucleated myopathy, merosin deficiency muscular dystrophy, glycogenosis, mitochondrial myopathies.

The causes of the acquired form of myopathy may be the following.

inflammatory diseases of the muscles (myositis, polymyositis);

endocrine diseases (Itsenko-Cushing's syndrome, hyperaldosteronism, hyperparathyroidism, hypothyroidism and hyperthyroidism);

intoxication (high doses of alcohol, drug use, occupational hazards);

somatic pathology accompanied by chronic liver, kidney, heart failure, chronic obstructive pulmonary disease;

malignant neoplasm.

Irreversible changes occur in the affected muscle fibers (myofibrils) - atrophy, which is then replaced by fat and connective tissue (the muscle itself decreases in size);

Symptoms of myopathy

The main complaint of a patient with myopathy is muscle weakness (there are 3 degrees of myopathy, depending on its severity). This may initially manifest as a violation of normal load tolerance: fatigue when walking, performing routine household chores, the need for additional rest, heaviness in the legs.

Over time, dystrophic changes develop, muscle tone decreases further and weakness increases. It becomes difficult for a person to run, jump, climb stairs, stretch, and stand on tiptoe. Muscle atrophy and a decrease in volume are noted, which is accompanied by deformation of body parts (mainly this

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occurs in the area of the forearms and lower legs). A characteristic phenotype of the patient is formed:

drooping shoulders;

pterygoid blades;

lumbar lordosis, protruding abdomen;

gait disturbance, frequent falls while walking.

Pain occurs in the back and limbs, flat feet develop, and contractures form in the joints, which limits the range of motion.

Symptoms of facial muscle myopathy include difficulty curling your lips, baring your teeth, puffing out your cheeks, and pronouncing vowel sounds.

With myopathy of the eye, there is a decrease in visual acuity.

When the respiratory muscles and diaphragm are involved in the pathological process, ventilation of the lower parts of the lungs decreases, which contributes to the development of pneumonia. Laryngeal paresis may develop.

When the myocardium is damaged, myocardial dystrophy develops, which is manifested by heart failure, shortness of breath, increased edema, palpitations, and pain in the heart region.

If the disease develops in childhood, the child may experience delays in mental and physical development, speech delays and disorders, and learning difficulties.

Diagnosis of myopathy

Diagnostic measures are aimed at identifying the cause of the disease, identifying signs of complications, and conducting differential diagnostics.

The doctor will listen to the patient's complaints and determine whether there are any diseases in the relatives. During the physical examination, the following will be assessed:

a. anthropometric indicators;
b. walking;
c. muscle size, including the tongue;
d. muscle tone and strength;
e. tendon, abdominal reflexes;
f. sensitivity;
g. vegetative state;
h. cognitive functions, emotional state;
i. the state of the cardiovascular, respiratory, digestive systems.
j. Your doctor may order the following tests:
k. general blood test with determination of leukocyte formula, ESR;
l. general urinalysis;
m. biochemical study with the determination of total protein, its fractions, total and direct bilirubin, ALT, AST, creatinine, urea;
n. blood glucose, glycated hemoglobin;
o. hormonal study of the thyroid gland (TSH, T3, T4), parathyroid gland, adrenal cortex;

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p. 25-OH-vitamin D;
q. blood electrolytes, pH;
r. coagulogram;
s. lipid profile;
t. creatine phosphokinase, lactate dehydrogenase;
u. molecular genetic studies.
v. Instrumental diagnostics includes:
w. 12-lead ECG;
x. EchoCG;
y. chest x-ray;
z. electromyography;
aa. electroneurography;
a. biopsy with subsequent pathohistological and immunohistochemical examination;
b. goniometry (study of the range of motion of joints);
c. Ultrasound, MRI of muscles;
d. Ultrasound examination of the abdominal organs;
e. MRI of the brain;
f. densitometry;

Spirometry.

According to the results of electromyography, a decrease in the strength and duration of muscle potentials is noted. Special scales are used to assess the motor function of the upper and lower extremities.

Consultations with a rehabilitation specialist, orthopedist, cardiologist, pulmonologist, endocrinologist, nutritionist, speech therapist may be prescribed. A complete examination plan is determined by the doctor, taking into account the clinical picture and the presence of concomitant diseases.

Treatment of myopathy

Treatment of the congenital form of the disease is symptomatic (eliminating the manifestations of the disease), since it is impossible to eliminate the causes of the disease (genetic mutations).

For secondary forms of the disease, therapy is aimed at eliminating damaging factors by correcting concomitant endocrine disorders, stopping alcohol and drug use, changing the workplace, and eliminating the inflammatory process.

Pharmacotherapy involves taking the following medications:

a. anabolic hormones;
b. anticholinesterase drugs ("Proserin", "Neuromidine");
c. multivitamins (including vitamins E, B, C, nicotinic acid), levocarnitine, coenzyme Q10, calcium preparations;
d. non-steroidal anti-inflammatory drugs;

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Glucocorticoids.

Metabolic therapy helps to improve muscle trophism, muscle cell growth, improve neuromuscular transmission, slow down dystrophic and atrophic changes. Treatment with anabolic steroids has a number of side effects with long-term use (baldness, in women - growth of facial hair, deepening of the voice, menstrual irregularities, acne, decreased skin elasticity, changes in the male body). Anticholinesterase drugs can have a sedative and analgesic effect.

a. Physical treatments may include:
b. massage;
c. physical therapy, breathing exercises;
d. electrophoresis;
e. iontophoresis with calcium;
f. ultrasound.

When engaging in physical exercise, it is important to take into account the initial level of physical fitness and avoid overworking the muscles, performing eccentric exercises and exercises with high resistance (walking, swimming, cycling are recommended).

The diet includes enriching the diet with nuts, vegetables, fruits, herbs, dairy products, grains, proteins and foods containing carnitine. It is not recommended to drink coffee and caffeinated drinks, alcohol and spices.

a. Surgical treatment includes the following operations:
b. joint correction;
c. spinal stabilization;
d. gastrostomy placement (in case of malnutrition), tracheostomy (in case of respiratory failure).
e. According to clinical guidelines, the use of depolarizing muscle relaxants during surgery for Duchenne myopathy is contraindicated.

Prognosis for myopathy

Congenital forms of the disease have been reported to have a more unfavorable prognosis. Secondary myopathies may regress in the absence of a damaging factor and therefore have a more favorable course. Severe myocardial myopathy and muscles involved in respiration have a poor prognosis.

Myopathies are a group of diseases based on various disorders in the metabolism and structure of muscle tissue, leading to a decrease in the strength of the affected muscles and a limitation of motor activity. Typical symptoms of myopathy: progressive muscle weakness, the development of muscle atrophy, decreased tendon reflexes and muscle tone. The results of electrophysiological studies, biochemical tests of blood and urine, molecular genetic and histochemical analysis of samples obtained as a result of muscle biopsy help to diagnose myopathy. Treatment involves the complex use of metabolic drugs in courses 3 times a year.

Causes of myopathy

Myopathies belong to the group of neuromuscular diseases. They are characterized by dystrophic damage to muscle tissue (mainly skeletal muscles), selective atrophy of individual fibers (myofibrils) with full functional preservation of the nervous system of animals. They are characterized by a chronic, steadily developing course. As a rule, the manifestation of clinical manifestations of myopathy occurs in childhood and adolescence. Most cases of the disease are caused by genetic pathology - primary myopathies. Acquired myopathies are less common - secondary or symptomatic.

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Myopathies

Primary myopathies are based on genetically determined disorders in the activity of ion channels of mitochondria and myofibrils, in the synthesis of muscle proteins or enzymes that regulate the metabolism of muscle tissue. The inheritance of a defective gene can occur recessively, dominantly and in a X-linked manner. At the same time, external factors often act as a trigger that initiates the development of the disease. Such "trigger" factors can be various infections (chronic tonsillitis, frequent acute respiratory viral infections, bacterial pneumonia, salmonellosis, pyelonephritis, etc.), nutritional dystrophy, severe injuries (pelvic fractures, polytrauma, head injury, etc.). physical overstrain, intoxication.

Acquired myopathies can develop against the background of endocrine diseases (hyperparathyroidism, Itsenko-Cushing's disease, hyperaldosteronism), chronic intoxication (drug abuse, drug addiction, alcoholism, occupational hazards), malabsorption and vitamin deficiency, severe chronic diseases (CKD, chronic liver failure, heart failure, COPD), tumor processes.

Pathogenesis

The presence of genetically determined or acquired defects in metabolites involved in metabolism and the construction of muscle fibers leads to the emergence and development of degenerative changes in the latter. Myofibrils atrophy develops, and they are replaced by adipose and connective tissue. Muscles lose their ability to contract, which leads to muscle weakness and limits the ability to perform active movements.

Recent studies have identified dysfunction of both the central (diencephalic) and peripheral parts of the autonomic nervous system, which play an important role in the pathogenesis of the disease in patients with various forms of myopathy. This may explain the typical predominant lesion of myopathies in the proximal parts of the extremities, which have autonomic innervation.

Classification

Specialists in the field of neurology have developed several classifications of myopathies. The most popular among clinicians is the etiopathogenetic principle of separation, according to which hereditary, inflammatory, metabolic, membrane, paraneoplastic and toxic myopathies are distinguished. Among hereditary myopathies, 3 types are most common: Erb's juvenile/adolescent form, Duchenne's pseudohypertrophic form and humeroscapulofacial form. Less common are scapuloperoneal, oculopharyngeal, distal and other forms. A separate group includes congenital myopathies: central nuclear disease, nemaline and myotubular myopathy, myofibril type imbalance.

Inflammatory myopathies are classified as infectious - arising as a result of infectious-inflammatory damage to muscle tissue in various infectious processes: bacterial (streptococcal infection), viral (enteroviruses, influenza, rubella, HIV), parasitic (trichinosis, toxoplasmosis) and idiopathic - dermatomyositis, inclusion body myositis , polymyositis, myopathies due to collagenosis.

Metabolic myopathies are divided into those associated with disorders of lipid metabolism in muscles (acetyl-CoA dehydrogenase deficiency, carnitine deficiency), disorders of glycogen metabolism (Andersen's disease, Pompe disease, glycogenosis type III, McArdle's disease, phosphorylase β-kinase deficiency), purine metabolism (MADA enzyme deficiency) and mitochondrial myopathies (reductase, ATP, cytochrome b, β1 deficiency).

Symptoms of myopathy

Most myopathies begin gradually, with mild muscle weakness in the limbs, rapid fatigue from walking and other physical activity. Over the years, weakness increases, muscle atrophy appears and progresses, and deformities of the limbs appear. Due to significant muscle weakness, patients have difficulty getting up from the floor and climbing stairs, jumping or running is impossible. They must use special techniques to get out of a chair. The patient's appearance is characteristic: wing-shaped

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shoulder blades, sagging shoulders, protruding abdomen, and increased lumbar lordosis. A "duck gait" is observed - the patient sways from side to side.

Pathological changes in myopathy occur symmetrically in the muscles of the limbs and trunk. Typically, muscle atrophy is observed in the proximal arms and legs. In this regard, the muscles of the distal limbs may appear hypertrophied. This myopathic pseudohypertrophy is most pronounced in the muscles of the legs. Along with the increase in muscle weakness, there is a gradual fading of tendon reflexes and a progressive decrease in muscle tone, that is, peripheral flaccid paralysis develops and worsens. Over time, the result of a sharp limitation of active movements is joint contractures.

Myopathies can be accompanied by damage to the facial muscles, which is manifested by the inability to stretch the lips, whistle, raise the eyebrows, or smile. Damage to the orbicularis oris muscle leads to dysarthria, which is associated with difficulty pronouncing vowel sounds.

The clinical presentation of some myopathies includes damage to the respiratory muscles, leading to congestive pneumonia and the development of respiratory failure. Pathological changes in the heart muscle with the development of cardiomyopathy and heart failure, and in the muscles of the pharynx and larynx, dysphagia and myopathic paresis of the larynx can develop.

Features of individual forms of myopathy

Juvenile Erb's myopathy is inherited in an autosomal recessive manner. Pathological processes begin to appear at the age of 20-30. First of all, they cover the muscles of the pelvis and thigh, and then quickly spread to other muscle groups. The involvement of facial muscles is not typical. The younger onset of myopathy leads to early immobility of patients. If the disease develops in old age, its course is less severe: patients retain the ability to move for a long time.

Pseudohypertrophic Duchenne myopathy is inherited in a sex-linked recessive manner. Only boys are affected. As a rule, it manifests itself in the first 3 years of life, less often - in the period from 5 to 10 years. Usually, it begins with atrophic changes in the muscles of the pelvis and proximal legs, accompanied by pseudohypertrophy of the calf muscles. Contracture and curvature of the spine (kyphosis, scoliosis, hyperlordosis) appear early. Oligophrenia may occur. The disease occurs with damage to the respiratory muscles and heart (cardiomyopathy is observed in 90% of patients with Duchenne myopathy), which leads to early death.

Landouzy-Dejerine myopathy has an autosomal dominant inheritance. It manifests itself at the age of 10-20 years with damage to the facial muscles. Gradually, weakness and atrophy cover the muscles of the shoulder girdle, shoulders and chest. The muscles of the pelvic girdle are usually not affected. It is characterized by a slow course with long-term preservation of performance, without reducing life expectancy.

Scapuloperoneal myopathy is an autosomal dominant disorder characterized by the development of atrophy in the muscles of the distal leg and proximal arm, as well as the presence of mild sensory disturbances in the distal parts of the lower and upper extremities.

Oculopharyngeal myopathy is characterized by weakness of the tongue and pharyngeal muscles with extraocular muscle involvement. It usually presents with bilateral ptosis, followed by dysphagia. A characteristic feature of this myopathy is its late onset - in the 4th to 6th decade of life.

Distal late myopathy is inherited in an autosomal dominant manner. It is characterized by the development of weakness and atrophy in the distal parts of the extremities: first in the legs and arms, and then in the feet and wrists. It is characterized by a slow course.

The characteristics of the clinical manifestations of various forms of congenital, hereditary, and metabolic myopathies have been described in independent reviews.

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Diagnostics

Electrophysiological examination methods help the neurologist to diagnose myopathy: electroneurography (ENG) and electromyography (EMG). They allow to exclude damage to the peripheral motor neuron and thus to distinguish myopathy from infectious myelopathy, disorders of the cerebral circulation, myelitis and tumors of the spinal cord. EMG data show changes in muscle potentials characteristic of myopathies - a decrease in their amplitude and a decrease in their duration. The presence of a large number of short peaks indicates a progressive process.

Biochemical blood tests for myopathy show an increase in the level of aldolase, CPK, ALT, AST, LDH and other enzymes. Biochemical analysis of urine indicates an increase in the concentration of creatinine. Muscle biopsy is of great importance in determining the form of myopathy. Morphological examination of muscle tissue samples reveals the presence of atrophied myofibrils randomly distributed among practically intact and hypertrophied muscle fibers, as well as the replacement of areas of muscle tissue with connective or fatty tissue. A final diagnosis is possible only after comparing the results of histochemical, immunobiochemical and molecular genetic studies.

A patient with myopathy may be referred to a cardiologist, have an ECG, or have an ultrasound of the heart to look for damage to the heart muscle; if pneumonia is suspected, consult a pulmonologist and have a chest X-ray.

Treatment of myopathies

Currently, pathogenetic treatment of myopathies is at the stage of scientific experiments in the field of genetic engineering. In clinical practice, symptomatic therapy is mainly used, which consists in improving the metabolism of muscle tissue. For this purpose, vitamins E, B1, B6, B12, ATP, neostigmine, amino acids (glutamic acid, hydrolysate from pig brain), anticholinesterase drugs (ambenonium, galantamine), anabolic steroids (nandrolone decanoate, methandienone), potassium and calcium preparations. Thiamine pyrophosphate is used. A combination of several drugs is prescribed for a course of 1-1.5 months. 3 times a year.

Drug treatment of myopathies is supplemented with physiotherapy (electrophoresis with neostigmine, iontophoresis with calcium, ultrasound), light massage and exercise therapy. Exercise therapy can be carried out in a swimming pool. The set of exercises should be selected so as not to overload the weakened muscles. In some cases, patients need to consult an orthopedist and select orthopedic corrective devices (corsets, shoes).

The basis for the treatment of acquired forms of myopathies is the therapy of the underlying disease: correction of endocrine disorders, elimination of toxic effects and detoxification of the body, elimination of the infectious process, transition of a chronic disease to a stable remission stage, etc.

Prognosis and prevention

The most unfavorable prognosis is hereditary myopathies that manifest themselves in early childhood. Otherwise, the prognosis depends on the form of myopathy and the involvement of the heart and respiratory muscles in the process. The prognosis of secondary myopathies is more favorable if the underlying disease is successfully treated.

Prevention of primary myopathies is a complete collection of family history and mandatory consultation with a geneticist for couples planning pregnancy. Prevention of secondary myopathies is the elimination of toxic effects on the body, timely treatment of infectious and endocrine diseases, and correction of metabolic disorders.

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