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MODERN SOLUTIONS FOR THE DIAGNOSIS AND CLINICAL DIAGNOSIS OF AMYOTROPHIC LATERAL SCLEROSIS

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Abstract: Amyotrophic lateral sclerosis (ALS) is an incurable disease that destroys the cells that transmit motor nerve impulses. As a result, muscles stop receiving commands from the brain, gradually weaken and atrophy. Over time, more and more motor neurons are involved in the process, which leads to paralysis of the limbs, body, swallowing disorders and respiratory arrest.

Key words: Motor neurons - motoneurons, mechanism and causes of ALS development, ALS symptoms, ALS types and stages of development, ALS complications, ALS diagnosis

Disease definition

Amyotrophic lateral sclerosis (ALS) is a rare disease, also known as Lou Gehrig's disease, motor neuron disease, motor neuron disease, or Charcot disease.

French psychiatrist Jean-Martin Charcot first described ALS in 1869. However, the disease became widely known after the famous American baseball player Henry Lou Gehrig was diagnosed with amyotrophic lateral sclerosis in 1939.

Famous American baseball player Henry Lou Gehrig retired from the sport at the peak of his career, at the age of 36. The reason was ALS

Amyotrophic lateral sclerosis was once found in composer Dmitry Shostakovich and Chairman of the People's Republic of China Mao Zedong. But perhaps the most famous person with ALS is British physicist Stephen Hawking.

Hawking

Stephen Hawking, an English theoretical physicist and author of numerous scientific works, lived with ALS for over 50 years.

Amyotrophic multiple sclerosis occurs in approximately 2-3 out of 100,000 people worldwide. In 90% of cases, the disease occurs sporadically, and only 5-10% are hereditary forms.

ALS develops more often in men. The disease usually begins at the age of 58-63 - in the sporadic form and at the age of 47-52 - in the familial form. The average life expectancy of patients with this diagnosis is only 2-3 years. Only 7% of people live more than 5 years.

ALS that occurs in children and young adults under the age of 25 is called juvenile ALS. It often progresses very slowly and does not affect natural lifespan.

In the International Classification of Diseases, 10th Edition (ICD-10), ALS is coded G12.2 (“Motor neurone disease”).

Motor neurons - motoneurons

Motor neurons, or motor neurons, are large cells of the nervous system with long (up to 1 meter) processes (axons) located in the brain and spinal cord. Their main function is to transmit signals from the brain to the muscles, thereby ensuring muscle contraction.

There are upper and lower motor neurons. Upper motor neurons are located in the cortex and brain stem. They activate lower motor neurons (in the lower brain and spinal cord). These, in turn, send impulses - "commands" - directly from the brain to the muscles.

Motor neurons

Upper motor neurons transmit signals from the brain to lower motor neurons. And they send them directly to the muscles, causing them to contract.

If upper or lower motor neurons die for any reason, then the signals from the brain to the muscles stop flowing. As a result, the muscles stop contracting. Muscle atrophy develops.

ALS is characterized by simultaneous damage to both upper and lower motor neurons. In practice, there are cases of isolated damage to only one group of motor neurons, but less often. Combined damage to both upper and lower motor neurons leads to the development of specific motor disorders - spastic-atrophic paresis, muscle atrophy develops against the background of their exacerbation.

Mechanism and causes of ALS development

Amyotrophic lateral sclerosis is a multifactorial disease that develops as a result of the simultaneous influence of hereditary factors, environmental conditions, and lifestyle.

The mechanism of the development of the disease is not fully understood. However, it is believed that for unknown reasons one of the genes associated with ALS is disrupted (most often the occurrence of the disease is associated with mutations in the SOD1, TARDBP and C9orf72 genes). As a result, incorrect proteins begin to be synthesized, which accumulate inside the motor neurons, forming entire clusters.

Such accumulations prevent the normal functioning of motor neurons, and over time they die. In order to somehow compensate for the lack of motor neurons, the body begins to synthesize a huge amount of monosodium glutamate - "fuel" for the nervous system. For some time, this substance actually "feeds" the remaining neurons and keeps them working. But over time, excess glutamate leads to the destruction of other cells of the nervous system.

However, long-term studies have identified non-genetic factors that increase the risk of developing amyotrophic lateral sclerosis.

Factors leading to the development of the disease:

cigarette smoking, including passive smoking;

contact with agricultural fertilizers: formaldehyde, herbicides, insecticides, pesticides;

contact with heavy metals, including lead (for example, when welding metals);

injuries in professional sports;

previous infections affecting the nerve cells of the brain and spinal cord;

unhealthy lifestyle: frequent stress, lack of sleep, little physical activity.

Welding

People who have frequent contact with lead, a heavy metal, are at increased risk of developing amyotrophic lateral sclerosis.

Symptoms of ALS

The initial symptoms of the disease are non-specific: they are easily confused with manifestations of other pathologies, and sometimes with banal fatigue or emotional stress.

The first symptoms of amyotrophic lateral sclerosis:

fasciculations - muscle twitching;

muscle weakness;

incompetent, awkward hand movements;

muscle spasms, muscle twitching after strenuous physical activity, cramps;

speech impairment (due to weakening of the tongue muscles).

As the disease progresses, muscle weakness increases. Various muscle groups are involved in the pathological process.

Fasciculations - muscle twitching

Muscle twitching is one of the first signs of amyotrophic lateral sclerosis. Initially, fasciculations affect only individual muscles, but then spread to entire muscle groups in the arms, legs, and body.

Muscle weakness and stiffness

In ALS, the number of signals that travel from the brain to the muscles decreases. As a result, muscle tissue atrophies and the range of motion decreases. At the same time, the person experiences a lack of strength. He moves and speaks more slowly, his voice becomes quieter, and his breathing becomes faster.

Muscle cramps and spasms

As mentioned above, with amyotrophic lateral sclerosis, impulses from the brain periodically enter the muscles. This leads to muscle tension and cramps, often painful. Many patients note that such unpleasant sensations can appear suddenly and even interrupt sleep.

Swallowing problems

Due to the simultaneous atrophy and spasm of the muscles of the face, larynx and oral cavity, a person has difficulty swallowing food. As a result, he receives fewer nutrients. Over time, this can lead to weight loss, and then to cachexia, extreme fatigue. In addition, the inability to chew and swallow food normally, combined with breathing problems, can provoke pneumonia - pneumonia: it develops due to the ingress of food or liquid into the respiratory tract during breathing.

Excessive salivation

Weakness in the muscles of the mouth causes large amounts of saliva to accumulate in the mouth, which a person with ALS cannot swallow. The consistency of the saliva can be very sticky - making it difficult to remove.

Coughing and choking

Many patients with amyotrophic lateral sclerosis experience coughing and choking. This is caused by a large amount of saliva or phlegm in the mouth, which the person is unable to swallow due to weakness and spasm of the oropharyngeal muscles.

Speech disorder

Weakening of the facial and laryngeal muscles leads to speech problems.

Initially, the person can speak independently, although the words may be slurred. As the disease progresses, this ability is lost.

Difficulty breathing

Sooner or later, with ALS, the respiratory muscles atrophy. The person stops breathing on their own and needs artificial lung ventilation (ALV) devices to maintain life.

Emotional instability

Sometimes people with ALS become overly emotional - they laugh or cry for no reason.

Dementia

Dementia is not typical of spontaneous ALS, but occurs in 10-15% of patients with familial cases of the disease. In addition, more than half of people with amyotrophic lateral sclerosis experience apathy or depression.

The disease progresses steadily. Death occurs from respiratory paralysis approximately 3-5 years after the onset of the disease.

Types and stages of development of ALS

In the domestic literature, six forms of amyotrophic lateral sclerosis are distinguished depending on the location of the pathological process.

The main forms of ALS are:

cervix;

cervicothoracic;

bulbar (neurons responsible for speech, swallowing, chewing are affected);

lumbosacral;

high (central motor neuron affected);

primary generalized (initial combination of bulbar symptoms with signs of damage to central and peripheral motor neurons).

In the English-language literature, there are spinal forms of ALS (corresponding to the cervicothoracic and lumbosacral) and bulbar, as well as with the onset of respiration (corresponding to the cervical form).

Stages of ALS development according to local classification:

prodromal symptoms (indicate the onset of the disease before the first specific signs appear);

first local symptoms;

extended stage (generalization stage);

final stage;

extended life stage.

The identification of the fifth stage was made possible by artificial lung ventilation (ALV), which can maintain respiratory function for long periods of time in patients who are unable to breathe on their own.

mechanical ventilation

A ventilator is a device that allows oxygen to be forcibly delivered to the lungs.

Types of ALS according to the rate of progression:

rapidly progressive (life expectancy from the onset of the first symptoms is less than 2 years);

with an average development rate (average life expectancy - from 2 to 10 years);

slowly progressive (average life expectancy is more than 10 years).

Complications of ALS

Muscle atrophy and spasticity lead to contractures - permanent restrictions in joint mobility.

Complete paralysis of the affected part of the body develops - the neck, arm or leg. At the same time, the person loses the ability to walk, eat, take care of himself and perform simple daily tasks.

Patients with the bulbar form of ALS may develop aspiration pneumonia - a lung disease caused by the entry of contents from the oropharynx, nasopharynx, and stomach into the respiratory tract when they regurgitate into the esophagus.

The most common complications of ALS are:

weakness combined with spasticity, up to complete paralysis with the development of contractures;

inability to hold the neck upright, drooping head;

deep vein thrombosis;

difficulty swallowing food, then inability to swallow solid and liquid food;

urine and feces retention;

foot deformity;

apnea - involuntary holding of breath during sleep;

ALS-related cachexia - extreme fatigue;

respiratory failure up to respiratory arrest.

ALS diagnosis

A neurologist diagnoses amyotrophic lateral sclerosis. At the appointment, the specialist asks what complaints the patient has, how long they have been present, and what concomitant diseases he has. The doctor also finds out what diseases are present in the family history. Then he proceeds to the examination - examines the muscles of the body, assesses their strength and tone, checks reflexes using a special hammer. With ALS, the muscles become thin and lose strength, but at the same time they are tense and fasciculated.

To exclude or confirm the diagnosis, the doctor uses the following criteria for the probability of the disease:

clinically proven ALS: there are signs of damage to central and peripheral motor neurons in three of the four possible parts of the central nervous system (CNS);

clinically possible ALS: signs of damage to central and peripheral motor neurons in two parts of the central nervous system;

clinically probable, laboratory-confirmed ALS: signs of motor neuron damage in one area, laboratory-confirmed signs of chronic and acute denervation in two or more muscles or limbs;

Possible ALS: signs of damage to central and peripheral motor neurons in one part of the central nervous system, or signs of damage to a central motor neuron in two or more parts of the central nervous system.

An important diagnostic step is to distinguish amyotrophic lateral sclerosis from other diseases that have similar symptoms.

Diseases with symptoms similar to ALS:

spinal form of multiple sclerosis;

cerebellar ataxia - lack of coordination caused by pathology of the cerebellum;

brain and spinal cord tumors;

Myasthenia gravis - an autoimmune neuromuscular disease;

chronic neuroinfections, permanent consequences of acute neuroinfections.

One of the main tools for diagnosing ALS is electromyography (EMG). During this procedure, a specialist uses a special electromyograph device that applies an electrical current to the muscles. In response to the current, the muscles usually contract.

The device assesses the speed of impulse transmission from the nerves of the arms and legs to the motor neuron located in the spinal cord.

In the early stages of the disease, the speed of muscle conduction and the response of the muscles to impulses can be very high. This is explained by the fact that the body tries to compensate for the death of neurons and produces a large amount of monosodium glutamate - a type of fuel that activates the nervous system. However, as the disease progresses, the signal from the muscles to the brain is transmitted worse and worse, and the EMG readings worsen.

EMG

Electromyography is one of the main studies in the diagnosis of amyotrophic lateral sclerosis

In some cases, magnetic resonance imaging (MRI) may be indicated. This test allows for visualization of internal structures of the body and can rule out or confirm tumors in the spinal cord and brain. Genetic testing can confirm or rule out a familial form of ALS.

If the study shows a mutation in the gene in the patient and relatives, then a diagnosis of "familial ALS" is made. If the patient's genes are damaged, but there is no damage in the genetic material of the relatives, this is a "sporadic hereditary form of ALS." If no abnormality is detected in the genes, this is simply a "sporadic form."

The doctor may also order tests that help assess the patient's condition and identify abnormalities in the functioning of internal organs.

Complete blood count with leukocyte count and ESR, smear microscopy for pathological changes in leukocyte count (venous blood)

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