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SPINAL MUSCLE ATROPHY

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Spinal muscular atrophy is a rare genetic disease, its occurrence mechanism and treatment has not been studied sufficiently. The SMA is characterized by gradual hypotrophy of the muscles, which eventually leads to problems with breathing and swallowing and, consequently, without proper therapy, to the imminent death of patients with such a diagnosis. This pathology is a consequence of mutation in the SMN1 gene (located in the long shoulder of the fifth chromosome), the products of which ensure the survival of motor neurons. SMA manifests at a frequency of one case per 6-10 thousand. people. The carrier of the mutant gene is every 40-50 inhabitants of the Earth.

According to international standards, there are four types of spinal muscular atrophy: SMA I or Vernig-Hoffman disease (demonstration age 0-6 months), SMA II or Dubovica disease (first symptoms 7-18 months), SMA III or Kygelberg-Velander disease (disease debut after 18 months), SMA IV or «adult SMA» (this type usually manifests itself after 35 years). Each type is characterized by its own disease flow and prognosis.

It is proved that early diagnosis of this disease contributes significantly to the success of the treatment of spinal muscle atrophy. Currently, there are three main diagnostic methods (genetic testing, newborn screening and prenatal screening). Earlier, the initiation of therapy and constant work on those functions that were lost due to pathology, contribute to the fact that patients with SMA can develop physically, keeping up with their peers. Currently, there are three drugs that either contribute to the production of a protein that is not produced in patients with this pathology (due to a defect in the gene SMN1), or completely eliminate the cause of the disease (replacement of defective SMN1 gene with normal).

In our country the number of patients with SMA increases every year. In 2021 the first RB drug against SMA (Spinraza) was registered.

We should move in the following directions to minimize the mortality from this pathology: It is necessary to agitate genetic testing actively to be able to carry the mutant SMN1 gene before planning conception, provide a wide range of society with information about the mechanisms of occurrence of this pathology in order to avoid incorrect interpretations of the causes of the SMA. An important direction is to increase the availability of drugs, which are aimed at fighting disease, as well as increasing the availability of genetic testing of children with SMA symptoms from sparsely populated regions.