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THE CASE OF LATE DIAGNOSIS OF NEUROFIBROMATOSIS

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ABSTRACT

Neurofibromatosis is a genetic disorder that causes benign tumors, which developed on nerves, on the spinal cord or in the brain; in rare cases, tumors can become cancerous. Neurofibromatosis is usually diagnosed in childhood or early adulthood, and characterized by neurological manifestations, specific skin changes and orthopedic defects. Complications of neurofibromatosis can include cardiovascular problems, hearing loss, dizziness, loss of vision, weakness of the muscles, severe pain, convulsion. We present a girl 14-year-old patient with late diagnosed neurofibromatosis, who developed central tetraparesis, torticollis and dysphonia.

Keywords: neurofibromatosis, tumor, mutation, tetraparesis, deformation of bones.

INTRODUCTION

For an appointment with a neurologist, a girl E.M., born in 2003 - 14 years old, complaints of severe weakness, curvature of the spine, weakness of the limbs, large area, torticollis, quiet voice.

From Anamnesis vitae: Child from 3 pregnancies, 3 births. The mother's pregnancy proceeded normally. I went to school at the age of 7. School performance is good. In mental development, he does not lag behind his peers.

From the anamnesis morbi: according to the father, at the age of one year, the child was diagnosed with a bilateral dislocation of the hip joint. At the age of 10 months, the girl walked on tires, from the age of 2 she began to walk on her own. Speech from 1.5 years. Up to 9 years, the development corresponded to the age, she ran, jumped, studied at a comprehensive school. From the age of 9-10, she began to stumble, her gait changed, her figure and spine began to change. The girl was examined many times in Fergana, Bukhara, and Tashkent. A genetic counselor was consulted. As a result, the girl was diagnosed with “Hereditary degenerative disease of the nervous system. Myopathy".

For many years she received outpatient and inpatient treatment, but with a temporary effect, the patient's condition worsened. A tumor-like formation began to appear in the cervical region, general muscle weakness increased, and the voice became dysphonic. In 2014, MRI revealed an extramedullary intradural tumor of the cervical region. In 2016, the patient was operated on with a diagnosis of Multiple neurofibromatosis. Schwann removal. The operation was carried out in India.

In the neurological status: clear consciousness. The position is active. The musculoskeletal system is deformed, duck gait, right-sided torticollis, slight hoarseness of voice, central tetraparesis, predominantly right-sided. There are traces of scars in the cervical region at the back, the right shoulder girdle is almost absent, the movement of the arm is limited to the horizontal to the side, but forward in full. Atrophy of the muscles of the shoulder girdle, pterygoid scapulae, pronounced scoliosis in the cervical and thoracic region. Muscle strength in the proximal sections 4 b, on the right - 4.3 b., in the distal 5 b. Tendon hyperreflexia with expansion of reflexogenic zones, D>S, "+" pathological reflexes on the hands of Yakobson Lask on the right, hyperreflexia with an expanded reflexogenic zone on the legs, PR, AR high, D>S, spontaneous Babinski reflex on the right. There are no sensory disorders, he performs coordinating tests correctly, in the Romberg position he sways slightly due to weakness.

After the examination, the diagnosis was "Condition after removal of multiple neurofibromatosis of the cervical region and cervical thickening, with central tetraparesis, predominantly right-sided, mild bulbar disorders, torticollis and dysphonia."

Recommended: corset, massage, baklosan, elkar and observation in dynamics.

Thus, neurofibromatosis is a hereditary autosomal dominant disease caused by a mutation in the gene of one of the proteins, such as neurofibromin, merlin, etc., which are tumor growth suppressors, and is characterized by the development of tumors of predominantly ectodermal origin with damage to the nerves, skin, and central nervous system. It is not uncommon for a gene mutation to occur de novo. It is usually found in childhood or adolescence, less often after 20 years and is characterized by a steadily progressive course, the presence of multiple, usually symmetrically located tumor-like formations along the peripheral nerves, nerve roots and cranial nerves, anomalies in the development of the bone skeleton, as well as typical pigment spots on the skin of color "café au lait", sometimes angiomas of skin vessels and papillomas, the appearance of freckles in unusual places, such as in skin folds.[1,2,3]

Lisch nodules on the iris and specific small nodules on the fundus are pathognomonic signs. Possible manifestations of the disease epileptic convulsions. Fibromatosis of vascular adventitia contributes to the formation of aneurysms and the possibility of their rupture. Bilateral acoustic neuroma is manifested by bilateral hearing loss and vestibular disorders. Tumors of the brain and spinal cord may develop. Brain tumors may not manifest themselves for a long time. Spinal cord tumors can cause back pain, muscle weakness, numbness, and tingling in the extremities [4,5]. Complications are varied and include blindness due to optic nerve tumors, tumor malignancy, development of pheochromocytoma with symptomatic malignant arterial hypertension, renal artery stenosis and coarctation of the aorta associated with neurofibromatosis-specific vasculitis, neurofibromatosis-associated vasculopathy with coronary and cerebral artery disease, decreased or loss of peripheral nerve function due to prolonged compression by tumors, cosmetic defects, macrocephaly, scoliosis, chest deformity, deformity and pathological bone fractures, growth retardation[7,8].

Difficulties and errors in diagnosis can occur when differentiating from other tumors of the nervous system, especially when the patient does not have characteristic skin manifestations. Moreover, it must be remembered that neurofibromatosis is primarily a genetic defect that predisposes a person to an increased risk of developing tumors of the nervous system. An important role in establishing the correct diagnosis is played by the detection of Lisch nodules

by an ophthalmologist, the presence of bone dysplasia, and a thorough questioning of the pedigree. Neuroimaging techniques and genetic research are of decisive importance.

Conclusion. The above clinical case of a sick girl who, before establishing the correct diagnosis of neurofibromatosis, was treated under different diagnoses for as long as 3 years without a positive effect, and received neurological drugs that enhance tumor growth, is a vivid example of the fact that every doctor must always be alert about tumor process.

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