Edun Oluwatomisin, Damoah Lenards Obeng, Owolabi Ayoade Adeniyi

DUCHENNE MUSCULAR DYSTROPHY

Department of Pediatrics No.1 with Neonatology
Kharkiv National Medical University
Kharkiv, Ukraine
Scientific advisor: Omelchenko Olena

Introduction. Muscular dystrophies are a group of genetic conditions characterized by progressive muscle weakness and wasting (atrophy). The Duchenne and Becker types of muscular dystrophy are two related conditions that primarily affect skeletal muscles, which are used for movement, and heart (cardiac) muscle. These forms of muscular dystrophy occur almost exclusively in males.

Materials and methods: clinical case.

Results. Patient F., 3 years old, was hospitalized with complaints of progressive weakness in the legs, impaired walking (he cannot independently climb stairs, get up from the floor, sit down). From the anamnesis of life and illness, it is known that the child is from 3 pregnancies, proceeding against the background of the threat of termination, 3 urgent births. At the age of 24 months, the child was diagnosed with hepatosplenomegaly; a blood test showed a persistent increase in the activity of transaminases (5 norms), γ-glutamyl transpeptidase (4 norms), and CPK (20 norms). ELISA and PCR diagnostics methods made it possible to exclude hepatitis. After 2.5 years of life, the boy developed stiffness in his legs in the morning and unsteadiness when walking, and frequent falls.

Neurological status: Reduced muscle tone in the lower extremities. Bicipital and carporadial, knee, and Achilles reflexes are reduced and symmetrical. Muscle strength in the upper and lower extremities is reduced to 4 points, a positive symptom of Govers: he rises from a lying position and crouches, resting his hands on the floor and knees, walking on toes, the child climbs the steps with an added step. The functions of the pelvic organs are not impaired.

The results of electroneuromyography: axonal neuropathy of the peroneal nerve on the left.Echocardiography left-right shunt of the central part of the MPP up to 1.5 mm, minimal aortic dilation at the level of the Valsalva sinuses. The carried out enzyme diagnostics (in spots of dried blood) made it possible to exclude some hereditary

lysosomal storage diseases. Molecular genetic research - deletion of exon 52 of the dystrophin gene;

Discussion: A 3-year-old child was diagnosed with Duchenne-Becker muscular dystrophy, the stage of preserved ability to move independently, based on complaints, anamnesis, neurological status, results of laboratory instrumental and molecular genetic studies. The child was prescribed treatment: prednisone at a dose of 0.5 mg/kg/day. Physiotherapy with dosed physical activity is recommended.

Conclusion. To verify the diagnosis of progressive Duchenne-Becker muscular dystrophy in the presence of clinical symptoms characteristic of this disease in a patient, it is necessary to conduct a molecular genetic study to search for a mutation in the DMD gene.

Ezzahidi Manal, Outti Hajar, Al Saleh Ahmad

CLINICAL CASE OF PATIENT WITH FAMILIAL MEDITERRANEAN FEVER

Department of Propedeutic of Pediatrics No.2
Kharkiv National Medical University
Kharkiv, Ukraine
Scientific advisor: As. Lupaltsova Olga

Introduction. Familial Mediterranean fever (FMF) is a genetic autoinflammatory disorder that maker recurrent fevers and painful inflammation of the abdomen, lungs and joints. Also known as recurrent polyserositis. It is characterized recurrent episodes of peritonitis, pleuritis, and arthritis, usually with accompanying fever.

The common polymorphisms in the MEFV (Mediterranean fever) gene acting the role of pyrin in disease pathogenesis. Mutations in the MEFV gene are caused the disease. Gene is localized on 16p13.3 and more than 70 mutations have been described in pathogenesis. In the various ethnic groups the date of literature is described differences in the gene mutations of FMF.

Aim. To improve the diagnosis of primary immunodeficiencies in children with atypical symptoms.

Material and methods. In this article the clinical case is performed the familial Mediterranean fever in child with pulmonary manifestations.