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Genetic heterogeneity of the hyperkinetic form of cerebral palsy

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Abstract:
Introduction
In more than half of full-term children with a hyperkinetic form of cerebral palsy, it is not possible to identify the etiological factor in the development of the disease.

Patients and methods
Under observation was a girl of 12 years old with a hyperkinetic form of cerebral palsy. The neurological examination, brain MRI and the NGS-based “Inherited Epilepsy” gene panel was performed

Results.
Diffuse muscle weakness with subsequently appeared dystonic attacks and facial myokimia presented at birth. At date of examination a girl had also delayed mental and motor development, dysarthria and hypersalivation. Structural pathology was not detected on brain MRI. By NGS-testing a heterozygous mutation c.919G>A in ADCY5 gene was reported. According recommendations of ACMG this variant classified as a variant of uncertain clinical significance. The database of the Genomed laboratory contains information about 5 patients with mutations in the ADCY5 gene. Of these, in one patient the mutation was previously described as pathogenic, causing familial dyskinesia with facial myokymia (OMIM: 606703). For other patients, mutations in the ADCY5 gene are considered as possible causes of the disease and require additional testing.

Conclusion.
When confirming the clinical significance of mutations in the ADCY5 gene in the described patient, this clinical case will be a vivid example of allelic genetic heterogeneity. The polymorphism of clinical manifestations in patients with mutations in the same gene confirms the effectiveness of using multi-gene panels in patients with motor disorders.

Key words:
cerebral palsy, genes, determinations, next generation sequencing.

Biography:
Currently, Elizaveta Volodyaeva is the head of the department of rehabilitation of children of the Rehabilitation Center for the Disabled "Tsaritsyno", Moscow. Graduated from the Evdokimov Moscow State Medical University. After graduation, she studied in clinical residency in neurology at the Central Medical Academy of the Presidential Administration of the Russian Federation. She worked at the Scientific and Practical Center of Child Psychoneurology. Collaborates with the Department of Pediatric Neurology of the Russian Medical Academy of Postgraduate Education, Moscow, under the guidance of Professor Valery Zykov. Works with Natalia V. Chebanenko, Associate Professor of the Department of Pediatric Neurology. Studies neurogenetics: genetic heterogeneity of cerebral palsy, genetics of epilepsy, genetics of autism, the role of genetic factors for the rehabilitation of children with cerebral palsy and hereditary diseases of the nervous system. Elizaveta Volodyaeva is the author of scientific publications and articles in journals.

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