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Familial Dyskinesia with facial Myokymia mutation adcy5 de novo

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Abstract:
Introduction:
Familial dyskinesia with facial myokymia, first described in 2001 by Fernandez et al., is caused by a mutation in the adenylate cyclase gene (ADCY5). Adenylate cyclase is involved in the transformation of ATP into a cyclic form of AMP, which regulates a number of cellular processes: stimulates phosphorylation of enzymes, receptors and ion channels. The main clinical features of this nosology are: early debut in the form of diffuse muscle hypotonia with the appearance of hypercinesis in the form of chorea, dystonia, myoclonia, tremor.

Patients and methods:
A 13-year-old boy with a rare clinical case of a de novo mutation of familial dyskinesia with facies myokymia was observed.
Results: A feature of our observation is the attachment of hyperkinesis with maximum severity to 7 years, the dominance of dystonia in impaired motor stereotype and the absence of myokymia. Structural pathology was not detected on MRI of the brain. A typical option was the lack of effect on valproate and antipsychotics. Probably, one should start with clonazepam or clobazam, which have an indirect inhibitory effect on the activity of adenylate cyclase-5, taking into account the literature data (Shaw C., Hisama F., Friedman J., Bird T. D., 2014). At the same time, most authors agree that any drug therapy has a variable effect in this disease, which is advisable to inform parents and patients about.
Conclusion: The prescribed therapy for children with familial dyskinesia with facies myokymia should be selected individually, taking into account concomitant diseases, individual tolerance, interactions with other drugs taken.
Keywords: familial dyskinesia with facial myokymia, ADCY5 mutation, adenylyl cyclase 5, chorea, dystonia.

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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