34th Conference on
Clinical Neuroscience and Neurogenetics
Feb 24th, 2023 | Webinar

Genetic Factors of Cerebral Palsy with Epilepsy

Elizaveta I. Volodyaeva
Head of the Department, Tsaritsyno Rehabilitation Center for the Disabled of the Department of Social Protection of the Population, Russia

Natalia V. Chebanenko
Candidate of Medical Sciences, Associate Professor of the Department of Pediatric Neurology of the Russian Academy of Medical Sciences

Pavel L. Sokolov
Leading researcher, Russia

Valery P. Zykov
The Russian Academy of Medical Sciences, Russia

Abstract:
Purpose:
Study of genetic abnormalities in patients with cerebral palsy suffering from epilepsy.

Method:
The next generation sequencing (NGS) study was conducted in 373 patients with cerebral palsy and epilepsy. In 136 (36.5%) patients identified genetic variants were validated by Sanger sequencing and classified as pathogenic. The pathogenic variants were detected in 91 genes. The distribution of genes into groups of determinants was carried out (Sokolov PI et al. Russian Journal of Child Neurology. 2020;15:65-77).

Result:
There were more genes in the determinant groups ENM (regulation of neuronal membrane excitability) 20,5%, CMTR (control of chromatin modifications, transcription and replication processes) 14,7%, CS (regulation of cytoskeleton formation and functioning) 13,2%, NTS (regulation of neurotransmitter metabolism and synapse functioning) 10,3%. The distribution of genes according to the degree of motor deficiency was specific: in all groups, except for canalopathy genes (ENM). Brain defects revealed in the CMTR (control of chromatin modifications, transcription and replication processes) 25,5%, CS (regulation of the formation and functioning of the cytoskeleton) 9,6% and ENM (regulation of the excitability of the neuronal membrane) 9,6% groups. The RMF group (regulation of the functions of the mitochondrial apparatus) was characterized by the highest resistance to epilepsy. In cases from the group with the canalopathy genes (ENM), the epileptic process was not the most refractory.

Conclusions:
According to the contribution to the pathogenesis of cerebral palsy with epilepsy, the distribution of determinants for the provision of excitability and conduction of the nervous tissue (ENM and NTS), the regulation of neuroontogenesis processes (NOG and CMTR), and the predetermination of enzymatic defects leading to storage diseases (GSD) are permissible. The determinant ENM is responsible for both the formation of motor deficits and the formation of the epileptic process. At the same time, its influence on motor deficit is nonspecific.

Received: January 13, 2023; Accepted: January 15, 2023; Published: Feb 24, 2023

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

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.

Received: January 13, 2023; Accepted: January 15, 2023; Published: Feb 24, 2023