DETERMINATION THE MAIN GENE MUTATION OF ORPHAN NEUROLOGICAL DISEASES IN CHILDREN

A. Karabekova
+ 7(727)3387090 (ex.7099), aizhankarabekova@gmail.com

Kazakh National Medical University named after S.D. Asfendiyarov (Almaty, Kazakhstan)
Atchabarov Research Institute of Fundamental and Applied Medicine, KAZNU

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Introduction. Rare or orphan diseases (OD) are diseases which affect a small number of people compared to the general population and specific issues are raised in relation to their rarity. Most rare diseases are genetic, and thus are present throughout the person's entire life. Many OD appear early in life, so that we to determine spread of the orphan neurological diseases in children under 3 years old. Children's early ages are seen neuromuscular diseases: spinal muscle atrophy, myasthenia, myopathies, and mitochondrial diseases.

The main goal of this investigation is to determine of genetic markers of the following orphan diseases with myopathy and myoclonic signs in children of Kazakh population: spinal muscle atrophy; Leigh syndrome; GM1u GM2-gangliosidosis; Dravet Syndrome.

Spinal muscular atrophy (SMA) involves the loss of nerve cells called motor neurons in the spinal cord and is classified as a motor neuron disease; Genes are involved: SMA, TYPE I, II , III; Leigh syndrome is an under-recognized inherited neurometabolic disorder that affects the central nervous system; NDUFS8, COX10; GM1 and GM2 - gangliosidosis is an inherited disorder that progressively destroys nerve cells (neurons) in the brain and spinal cord. HEXA, HEXB; Dravet syndrome, also known as severe myoclonic epilepsy of infancy (SMEI), is a type of epilepsy that begins in the first year of life with frequent and/or prolonged seizures; SCN1A.

Methods. For detection of SNP polymerases (SMA (SNP -13); Leigh syndrome (SNP-15); GM1; GM2 - gangliosidosis (SNP-15); Dravet syndrome(SNP- 4)) we will use the TaqMan® OpenArray® Real-Time PCR Plates (Life Technologies, USA) by QuantStudio 12K Flex.

Results. We plan to determine spread of the orphan nerurological diseases with myopathy and myasthenia based on genetic and clinical signs in Kazakh child population We plan to define clinically relevant mutations of the genes responsible for the spinal muscle atrophies (SMN1, 2), GM1, 2 -gangliosodosis 1 и 2 types (GLB1, HEXA, HEXB), Leigh syndrome (NDUFS8, COX10), Dravet syndrome (NDUFS8, COX10).

Conclusion. Kazakhstan can start a production of the specific medication for the enzyme replacement therapy. Collected data will be highly interesting and importance for the wide spectrum of the specialists and scientists. We will develop a system improving a children's health quality of our country.