

Genetic screening in a population of bulgarian children with autism using exome and chromosome sequencing

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Autism is a genetic neurobehavioral disorder with onset in infancy associated with developmental impairments in social communication skills and stereotypic repetitive deportments. Its etiology is multigenic and highly heterogeneous, with very few of the same pathogenic variants present in a significant percentage of affected individuals. The average case of autism is a product of many susceptibility increasing variations. The purpose of the study is to obtain genetic data on the presence of polymorphisms to be used in the analysis of the specific characteristics of different autism subtypes. A cohort of 14 Bulgarian patients with clinical diagnosis of autism (8 male and 6 female) from 14 unrelated families were analyzed. A whole exome and chromosome sequencing was performed. We identify variants in known genes or regions, such as microduplication (in one patient), single nucleotide substitutions (in nine individuals), and large deletions (in four patients). Moreover, homozygous variants were found in 2 individuals, and de novo variants – in 4. The obtained results provide an important step forward in the characterization of the subtypes of autism and in defining the morbidity risk in the families. Sequencing technology is a candidate to be applied in the appropriate and timely diagnosis, which is essential for selecting adequate therapeutic approaches. Combining knowledge of the molecular mechanisms of mutagenesis and proper molecular-genetic testing approaches is a promising source of knowledge to be used in the management of autistic patients.

Biography

Associate Professor Lyudmila Belenska is a scientist with more than twenty years of experience in conducting research in the field of molecular biology and immunology. At the same time, she is a lecturer at the Medical faculty of Sofia University "St Kliment Ohridski" in human biology, genetics, immunology, cell biology, parasitology and developmental biology. The broad range of research and teaching experience provides her with an in-depth insight into the genetic basis and molecular mechanisms of neurodevelopmental disorders of the autism spectrum.

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