

STUDY OF THE GENETIC SUSCEPTIBILITY TO AUTISM SPECTRUM DISORDERS IN THE VOLGA-URAL REGION OF RUSSIA

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Autism is a socially significant neurological disorder, which has three main signs of behavioral lesions: in social interaction, speech and range of interests. Autism spectrum disorders (ASD) have a significant connection with the genetic component, which was confirmed in families and twin studies in particular. A bunch of genetic alterations presumably involved in the ASD pathogenesis is known to date including chromosomal rearrangements, CNVs and SNVs as well as hundreds of candidate genes and this complicates the analysis of their biological basis. Study of genetic susceptibility to ASD in Russia is carried out by several groups but it was not implemented in the Volga-Ural region before.

Blood samples were collected in Volga-Ural region from 107 individuals with the established diagnosis "autism" with subsequent DNA extraction. MLPA analysis in 35 individuals using SALSA MLPA probe mix (MRC-Holland) designed to analyze the chromosomal regions 15q11-q13 (including UBE3A, GABRB3 and the 15q13 micro deletion region with CHRNA7), the 16p11 micro deletion region and the SHANK3 gene at 22 q13 allowed us to detect a deletion in the 6 exon of the GABRB3 gene (15q12) in two probands and a deletion in the 6 exon of the MAZ gene (16p11) in one proband. We also found the lack of significant difference comparing 107 children with autism with the control group (94 healthy children) in the allele frequency of the polymorphic variant rs9616915 in the 6 exon of the SHANK3 gene (p.Ile245Thr) as well as rs2196826 in the PLD5 intron. We also found an absence of the changes in five exons of the NLGN3 gene in the studied individuals.

Thus, we found rare deletions in three individuals from Volga-Ural region in the chromosomal regions that showed involvement in autism earlier.

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STUDY OF THE GENETIC SUSCEPTIBILITY TO AUTISM SPECTRUM DISORDERS IN THE REPUBLIC OF BASHKORTOSTAN, RUSSIA

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Autism is a socially significant neurological disorder, which has three main signs of behavioral lesions: in social interaction, speech and range of interests. Autism spectrum disorders (ASD) have a significant connection with the genetic component, which was confirmed in families and twin studies in particular. A bunch of genetic alterations presumably involved in the ASD pathogenesis is known to date including chromosomal rearrangements, CNVs and SNVs as well as hundreds of candidate genes and this complicates the analysis of their biological basis. Study of genetic susceptibility to ASD in Russia is carried out by several groups but it was not implemented in Republic of Bashkortostan before.

Blood samples were collected from 42 individuals in Republic of Bashkortostan with the established diagnosis "autism" with subsequent DNA extraction. The lack of significant difference with the control group in the allele frequency of the polymorphic variant rs9616915 in the 6 exon of the SHANK3 gene which leads to the substitution of isoleucine with threonine (p.Ile245Thr) as well as rs2196826 in the PLD5 intron was revealed. We also found an absence of the changes in three exons of the NLGN3 gene in the studied individuals. However, a MLPA analysis allowed us to detect a deletion in the 6 exon of the GABRB3 gene in two probands and a deletion in the 6 exon of the MAZ gene in one proband.