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Analysis of Potential Associations of Single Nucleotide Polymorphisms in Patients with Schizophrenia Spectrum Disorders

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Abstract : Relevance. The genetic risk of developing schizophrenia is determined by two factors: single nucleotide polymorphisms and gene copy number variations. The search for serological markers for early diagnosis of schizophrenia is driven by the fact that the first five years of the disease are accompanied by significant biological, psychological, and social changes. It is during this period that pathological processes are most amenable to correction. The aim of this study was to analyze single nucleotide polymorphisms (SNPs) that are hypothesized to potentially influence the onset and development of the endogenous process. Materials and Methods It was analyzed 73 single nucleotide polymorphism variants. The study included 48 patients undergoing inpatient treatment at "Psychiatric Clinical Hospital No. 1" in Moscow, comprising 23 females and 25 males. Inclusion criteria: - Patients aged 18 and above. - Diagnosis according to ICD-10: F20.0, F20.2, F20.8, F21.8, F25.1, F25.2. - Voluntary informed consent from patients. Exclusion criteria included: - The presence of concurrent somatic or neurological pathology, neuroinfections, epilepsy, organic central nervous system damage of any etiology, and regular use of medication. - Substance abuse and alcohol dependence. - Women who were pregnant or breastfeeding. Clinical and psychopathological assessment was complemented by psychometric evaluation using the PANSS scale at the beginning and end of treatment. The duration of observation during therapy was 4-6 weeks. Total DNA extraction was performed using QIAamp DNA. Blood samples were processed on Illumina HiScan and genotyped for 652,297 markers on the Infinium Global Chips Screening Array-24v2.0 using the IMPUTE2 program with parameters Ne=20,000 and k=90. Additional filtration was performed based on INFO>0.5 and genotype probability>0.5. Quality control of the obtained DNA was conducted using agarose gel electrophoresis, with each tested sample having a volume of 100 µL. Results. It was observed that several SNPs exhibited gender dependence. We identified groups of single nucleotide polymorphisms with a membership of 80% or more in either the female or male gender. These SNPs included rs2661319, rs2842030, rs4606, rs11868035, rs518147, rs5993883, and rs6269.Another noteworthy finding was the limited combination of SNPs sufficient to manifest clinical symptoms leading to hospitalization. Among all 48 patients, each of whom was analyzed for deviations in 73 SNPs, it was discovered that the combination of involved SNPs in the manifestation of pronounced clinical symptoms of schizophrenia was 19±3 out of 73 possible. In study, the frequency of occurrence of single nucleotide polymorphisms also varied. The most frequently observed SNPs were rs4849127 (in 90% of cases), rs1150226 (86%), rs1414334 (75%), rs10170310 (73%), rs2857657, and rs4436578 (71%). Conclusion. Thus, the results of this study provide additional evidence that these genes may be associated with the development of schizophrenia spectrum disorders. However, it's impossible cannot rule out the hypothesis that these polymorphisms may be in linkage disequilibrium with other functionally significant polymorphisms that may actually be involved in schizophrenia spectrum disorders. It has been shown that missense SNPs by themselves are likely not causative of the disease but are in strong linkage disequilibrium with non-functional SNPs that may indeed contribute to disease predisposition.

Keywords : gene polymorphisms, genotyping, single nucleotide polymorphisms, schizophrenia.
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