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Analysis of cohort of patients with 22q11.2 deletion syndrome - a single-center experience from Serbia

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Neurodevelopmental disorders (NDDs), such as autism spectrum disorders (ASD), intellectual disability (ID), schizophrenia, and bipolar disorder, are caused by disruption of brain development. They affect approximately 4% of the European population. However, molecular mechanisms underlying NDDs are still unknown. One of the syndromes with a high risk for NDDs is 22q11.2 Deletion Syndrome (22q11.2DS) caused by microdeletion 22q11.2. 22q11.2DS is the most common microdeletion in humans; approximately, 25% of patients with 22q11.2DS develop schizophrenia compared to 1% in the general population, while an ID is detected in approximately 45% of patients and ASD in 14-50% of cases. We analyzed genomic and clinical findings in our cohort of 35 patients with 22q11.2DS. The majority of patients have 3 Mb deletion and nine patients have inherited 22q11.2 microdeletion from their parents. Twenty-one different clinical presentations are revealed in the cohort with developmental delay detected in about 50% of patients. Approximately 80% of patients have heart malformations, palatal clefts/velopharyngeal insufficiency was detected in about 30% of them, facial dysmorphism in approximately 80% and hypocalcemia was seen in about 20% of patients. Here we presented a cohort of patients with 22q11.2DS which represents a good system for modeling NDDs in vitro.

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