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Content

GENERATION OF INDUCED PLURIPOTENT STEM CELLS DERIVED FROM PATIENTS WITH 22Q11.2 DELETION SYNDROME AS A TOOL FOR STUDYING NEURODEVELOPMENTAL DISORDERS

Ivana Simeunović,[1] Goran Cuturilo,[2,3] Nataša Kovačević-Grujičić,[1] Olena Petter,[4] Mina Perić,[1] Jovana Kostić,[1] Adrian J. Harwood,[4,5] Milena Stevanović,[1,6,7] Danijela Drakulić[1]

[1]Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia; [2]University Children's Hospital, Belgrade, Serbia; [3]Faculty of Medicine, University of Belgrade, Belgrade, Serbia; [4]Neuroscience and Mental Health Innovation Institute, School of Medicine, Cardiff University, Cardiff, Wales, United Kingdom; [5]School of Biosciences, Cardiff University, Cardiff, Wales, United Kingdom; [6]Faculty of Biology, University of Belgrade, Belgrade, Serbia; [7]Serbian Academy of Sciences and Arts, Belgrade, Serbia

Introduction: Neurodevelopmental disorders (NDDs), such as autism spectrum disorders (ASD), intellectual disability (ID), schizophrenia, and bipolar disorder, are caused by the alterations in early brain development. They affect approximately 4% of the European population and represent a high socio-economic impact and financial burden. Treatments of NDDs are focused on symptoms since 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. 22q11.2 microdeletion is the most common microdeletion in humans; it is one of the strongest known risk factors for development of psychiatric illness and the highest known genetic risk for schizophrenia (approximately, 25% of patients with 22q11.2DS develop schizophrenia compared to 1% in the general population).

Methods: Genomic and clinical findings in 35 patients with 22q11.2DS were analyzed and peripheral blood mononuclear cells of patients with 22q11.2DS and healthy controls were reprogrammed.

Results: The majority of patients have 3 Mb deletion and nine of them have inherited 22q11.2 microdeletion from parents. Twenty-one different clinical presentations are revealed in the cohort with developmental delay detected in about 50% of patients. iPSCs were generated from four patients with 22q11.2 microdeletion and five healthy controls.

Conclusion: Cohort of patients with 22q11.2DS is form and iPSCs were generated which enable research of molecular mechanisms underlying NDDs.

Key words: 22q11.2 Deletion Syndrome; neurodevelopmental disorders; iPSCs

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