Trends in **Molecular Biology** • Special issue

Abstract Book

CoMBoS2

2nd Congress of Molecular Biologist of Serbia

ISBN-978-86-82679-15-8

Belgrade • 2023

CoMBoS2 – the Second Congress of Molecular Biologists of Serbia, Abstract Book – Trends in Molecular Biology, Special issue
06-08 October 2023, Belgrade, Serbia
Online Edition
https://www.imgge.bg.ac.rs/lat/o-nama/kapacitet-i-oprema/istrazivacka-delatnost
https://indico.bio.bg.ac.rs/e/CoMBoS2

IMPRESSUM

PUBLISHER:
Institute of Molecular Genetics and Genetic Engineering (IMGGE), University of Belgrade

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Ivan **Strahinić**

Institute of Molecular Genetics and Genetic Engineering (IMGGE),
University of Belgrade
Belgrade, 2023
ISBN 978-86-7078-173-3

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Content

ESTABLISHMENT OF INDUCED PLURIPOTENT STEM CELLS FROM PATIENTS WITH 22Q11.2 DUPLICATION SYNDROME AS A MODEL SYSTEM FOR STUDYING NEURODEVELOPMENTAL DISORDERS

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Introduction: Neurodevelopmental disorders (NDDs), such as autism spectrum disorders (ASD), schizophrenia, and intellectual disability, represent important public health challenge in modern societies with a prevalence of about 10 to 15% of all births and the tendency of increasing worldwide. They are caused by disruption of early brain development. Treatments of NDDs are focused on symptoms due to a limited understanding of underlying pathophysiological mechanisms. Individuals with the 22q11.2 Duplication Syndrome (22q11.2dup), caused by heterozygous 22q11.2 microduplication, have an elevated risk of developing NDDs. Literature data revealed that ASD is detected in 14-25% of patients with 22q11.2dup while schizophrenia is less common in these patients than in the general population, suggesting that 22q11.2 duplication might be protective against schizophrenia.

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

Results: We formed a cohort of 8 patients with 22q11.2dup. The majority of patients in our cohort have microduplication of approximately 3Mb (80%). Also, the majority of them are familial cases and in 67% of cases, the 22q11.2 microduplication is inherited from the mother. Congenital heart defects were detected in 25% of our patients, while all tested patients have facial dysmorphism. iPSCs were generated from three patients with a familial form of 22q11.2dup and their mothers.

Conclusion: A cohort of patients with 22q11.2dup is formed and iPSCs were generated which can be used as a model system for studying NDDs.

Key words: 22q11.2 Duplication Syndrome; neurodevelopmental disorders; iPSCs; familial cases

Acknowledgements: This research was funded by the European Union`s Horizon Europe programme (Grant Agreement Number 101060201 (STREAMLINE)), the Ministry of Science, Technological Development and Innovation of the Republic of Serbia (grant number 451-03-47/2023-01/200042) and the Serbian Academy of Sciences and Arts (Grant number F-172).