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Content

22Q11.2 DELETION SYNDROME AS A TOOL FOR MODELLING AND RESEARCH OF NEURODEVELOPMENTAL DISORDERS

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Introduction: Neurodevelopmental disorders (NDDs) are a group of complex and heterogeneous disorders that include autism spectrum disorders, intellectual disability, schizophrenia and bipolar disorder. However, underlying pathophysiological mechanisms are mostly unknown. In order to get better understanding of the underlying mechanisms and to discover potential therapeutics we have focused our research on 22q11.2 Deletion Syndrome (22q11.2DS), caused by microdeletion of the region q11.2 of chromosome 22 and associated with a high risk for NDDs.

Methods: To study molecular mechanisms underlying intrafamilial phenotypic variability, we have identified families with the inherited form of 22q11.2DS with the aim of conducting the following analyses: whole genome sequencing in order to detect additional genetic variation(s) present in the affected child; generation of induced pluripotent stem cells (iPSCs) from peripheral blood mononuclear cells; analysis of the effects of 22q11.2 microdeletion on neural differentiation including organoids as 3D model system; transcriptome analysis of iPSC-derived neurons and astrocytes to determine differentially expressed gene sets and dysregulated pathways; and testing the metabolic changes and drug responsiveness of neurons and astrocytes by high-throughput cell-based assays.

Results: Peripheral blood mononuclear cells of the families with inherited form of 22q11.2DS were reprogrammed and established iPSCs were characterized. Generated iPSCs will be subjected to the further analyses.

Conclusion: Currently, most of the treatments of NDDs are symptom-based due to limited understanding of underlying pathophysiological mechanisms. It is expected that patient-derived iPSCs will enable a deeper understanding of unique disease mechanisms and may also provide a significant contribution in preclinical drug development.

Key words: iPSCs; transcriptome analysis; neural differentiation; organoids; drug responsiveness

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