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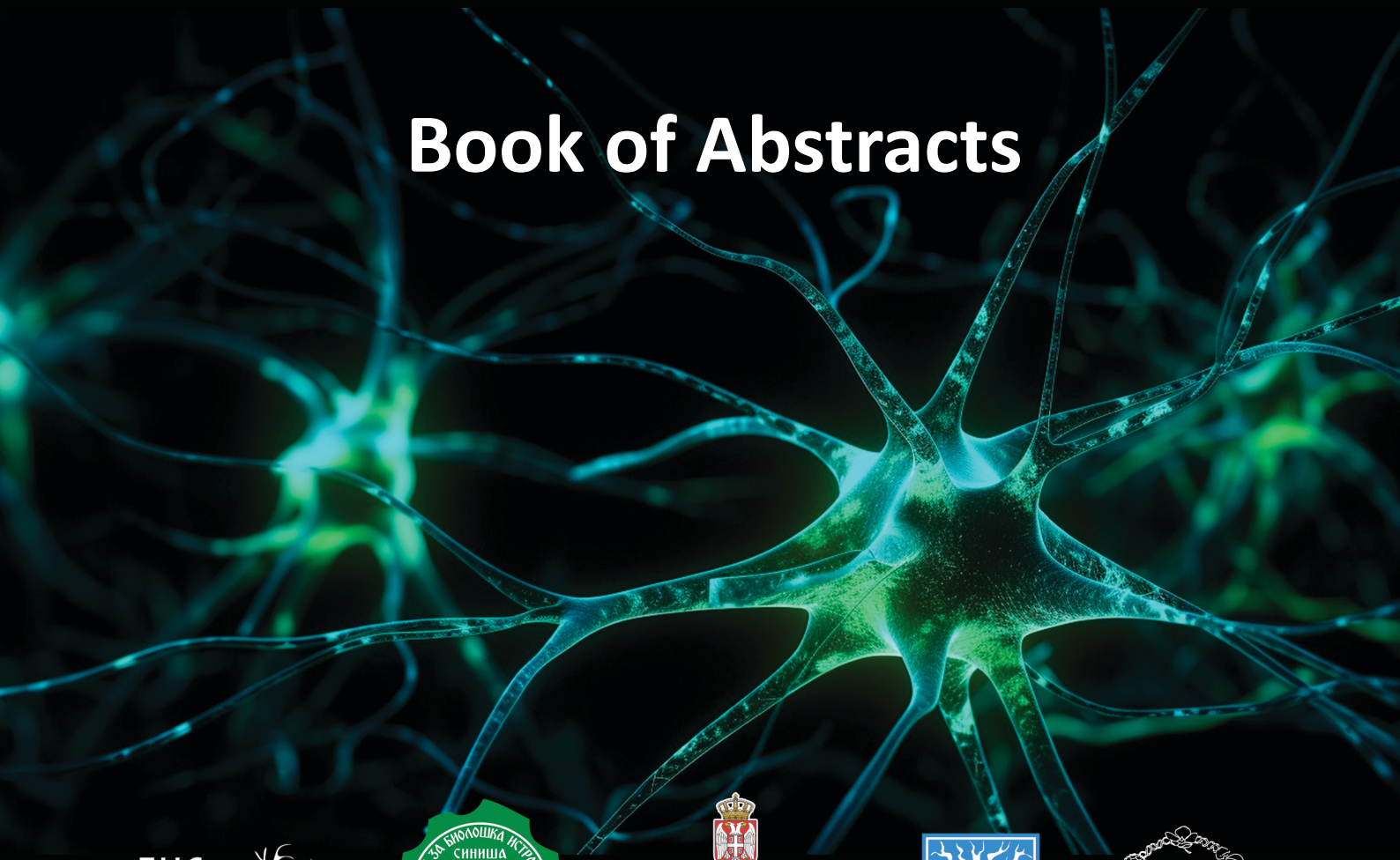

# Book of Abstracts

ДНС SNS

Институт за биолошка истраживања Синиша Станковић ИБИСС Универзитет у Београду

Република Србија Министарство науке, технолошког развоја и иновација

Универзитет у Београду Медицински факултет University of Belgrade Faculty of Medicine

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PROMEDIA Laboratory supply specialists

ZEISS

**8th CONGRESS OF SERBIAN NEUROSCIENCE SOCIETY with international participation**

**31 May – 2 June 2023. Belgrade, Serbia - BOOK OF ABSTRACTS**

**Published by:**
Serbian Neuroscience Society
Bulevar despota Stefana 142, 11060 Belgrade, Serbia

**Editors**
Selma Kanazir and Danijela Savić

**Assistant editors:**
Anica Živković
Željko Pavković

**Technical editor:**
Anđela Vukojević

**Graphic design:**
Olga Dubljević, Irina Veselinović



**ISBN: 978-86-917255-4-9**

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## Brain Disorders – From Genetics to Markers

## Neuroimmunoendocrine Interactions

# Genomic and clinical findings in patients with 22q11.2 duplication syndrome

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Neurodevelopmental disorders (NDDs), such as autism spectrum disorders (ASD), schizophrenia, and intellectual disability, are caused by disruption of early brain development. NDDs represent important public health challenge in modern societies with prevalence of about 10 to 15% of all births and the tendency of increasing worldwide. On the other side, treatments of NDDs are focused on symptoms due to 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 22q11.2dup might be protective against schizophrenia. We investigated genomic and clinical findings in cohort of 8 patients with 22q11.2dup. The majority of patients have 3Mb duplication. Five patients have 22q11.2 microduplication inherited from their parents. Other CNVs or SNVs are detected in 5 out of 8 patients. Common medical anomalies in our cohort of patients include developmental delay, facial dysmorphism, heart malformations, anomalies of the skeletal system, and anomalies affecting the eye. Characterization of a cohort of patients with 22q11.2dup is important since 22q11.2dup represents a powerful model to get insights into the molecular mechanisms underlying NDDs.

Acknowledgment: This research was funded by European Union`s Horizon Europe programme (Grant Agreement Number 101060201 (STREAMLINE)), 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).