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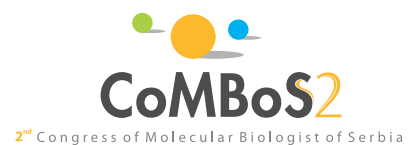
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Content

Welcome speech 4

Congress Organizers 5

MolBioS Award Winner 9

Plenary speakers 10

Session plenary speakers

- MOLECULAR BIOMEDICINE 11
- MOLECULAR BIOTECHNOLOGY 13
- MOLECULAR MECHANISMS OF CELL FUNCTIONS 16

Abstracts

- Session PLENARY LECTURES 20
- Session MOLECULAR BIOMEDICINE 25
 - PLENARY LECTURES 26
 - INVITED LECTURES 31
 - POSTERS 38
- Session MOLECULAR BIOTECHNOLOGY 100
 - PLENARY LECTURES 101
 - INVITED LECTURES 107
 - POSTERS 112
- Session MOLECULAR MECHANISMS OF CELL FUNCTIONS 126
 - PLENARY LECTURES 127
 - INVITED LECTURES 134
 - POSTERS 139
- MolBioS Student Session 157

Project Corner 182

Congress Friends 190

Sponsors 191

RARE METABOLIC DISEASES IN THE GENOMICS ERA

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Introduction: All inborn metabolic diseases are rare, having a prevalence less than 1:2000. Vast majority of them are monogenic and finding pathogenic genetic variants is needed to set the correct diagnosis, enable adequate treatment and provide genetic counseling to members of affected family. This study is an overview of genomic studies of rare metabolic diseases in Serbia.

Methods: Since 2005, more than 300 patients suspected to have a rare metabolic or neurometabolic disease have been analyzed using sanger sequencing, clinical-exome sequencing, whole-exome sequencing or whole-genome sequencing in order to find disease-causing or disease-modifying variants. Novel variants were characterized using *in silico* modelling or in *in vitro* eukaryotic assays (standard or CRISPR/Cas9 developed).

Results: Disease-causing variants were found in more than 60 different genes associated with a metabolic or neurometabolic disease. The most frequent disease was phenylketonuria (109 patients), followed by glycogen storage disease Ib (30 patients), while majority of diseases is seen only in a single patient. More than 40 new genetic variants were comprehensively characterized *in silico* or *in vitro*. For the first time, candidate modifiers (*SHANK* gene family) were identified in a group of phenylketonuria patients with an unusual phenotype.

Conclusion: In the genomics era, next-generation sequencing significantly shortens time to diagnosis and allows studying genetic modifiers of monogenic diseases and genotype-phenotype correlation. Furthermore, characterization of novel genetic targets boosts development of precision medicine.

Key words: rare diseases; next-generation sequencing; genomics; precision medicine

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