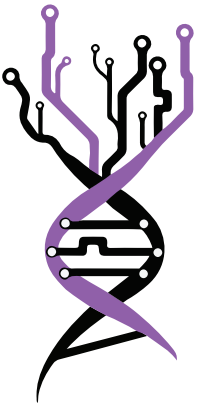
#BelBi2023 • Belgrade, Serbia

BOOK OF ABSTRACTS



4th Belgrade Bioinformatics Conference

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Dr. Ivana Morić
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FOREWORD

Dear colleagues and friends,

The 4th Belgrade Bioinformatics Conference - BelBi2023, where many high-quality scientific contributions were presented, has just ended. With great thanks to all participants, we now proudly present a book of abstracts that both reflects the scientific abundance and diversity of the conference and serves as a reminder of a memorable event.

Several research institutions, faculties, and scientific societies from Serbia joined forces in organizing this international conference, which covered numerous topics in computational biology, bioinformatics, and biomedical and health informatics. The main goal of BelBi2023 was to foster contact between scientists, both early stage career and senior researchers, allowing them to share experiences and latest advances in their fields. We sincerely hope that BelBi2023 has served as a platform for researchers from around the world to meet, initiate new collaborations, and expand professional contacts, and that all of you would become a part of the growing BelBi community.

We are grateful and proud to have welcomed more than 250 researchers from 21 countries. We have had 28 scientific sessions, consisting of more than 60 lectures (including eight Keynote talks), 47 presented posters, as well as three workshops and one satellite event – COST action. We have also organized seven industry lectures, including the NGS Challenge, two Meet the Expert Sessions, and one Business Coffee Break where ten start-up companies took part. And finally, the future BIO4 campus was presented and first panel on Serbia's resources for storage and analyses of genetic data was organized.

We would like to thank all the members of the International Advisory Board and the International Program Committee for their efforts and help in making this event a success. We are very grateful to the Ministry of Science, Technological Development and Innovation of the Republic of Serbia, SAIGE project, and UNDP-Serbia for their support. Finally, the Local Organizing Committee is very grateful to all the sponsors of the conference - BGI, Illumina & Elta'90MS, PacBio & East Diagnostics, ThermoFisher Scientific & Vivogen, Huawei, Labena, DSP Chromatography, RNIDS, Telekom Srbija, Alfa Genetics, Kefo and Superlab, hoping that they will stay with us for many years to come.

Looking forward to seeing you again at the 5th Belgrade Bioinformatics Conference.

Belgrade, July 2023

Dr. Valentina Đorđević
& Dr. Ivana Morić,
On behalf of BelBi2023
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POSTER PRESENTATION

Exploring the impact of rare Copy Number Variants on miRNA genes in CAKUT: Insights from integrated bioinformatic analysis and experimental validation

Ivan Jovanović[1]

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Rare copy number variants (CNVs) play a significant role in CAKUT development. However, the specific genetic drivers in certain CNVs associated with CAKUT remain unknown. To explore the genetic elements within CAKUT-associated CNVs, beyond the protein-coding genes, we leveraged the recently described comprehensive CNV landscape of CAKUT. MicroRNAs (miRNAs) are intriguing regulators of genomic networks and have the potential to be involved in CAKUT. Hereby, a pipeline for comprehensive analysis of miRNA genes affected by known, rare CNVs associated with CAKUT will be presented. The procedure is consisted of collection and synchronization of CNV regions specified in different hg assemblies with the hg19 assembly, mapping of the miRNA precursors, identification of the most frequently affected miRNAs and miRNA families by rare CNVs, bioinformatic interpretation of the top-rated miRNAs and prioritisation of key miRNAs for functional validation. Additionally, a method for estimation of the overall burden of rare CNVs on miRNA genes in CAKUT will be discussed. Remarkably, it was found that 80% of CAKUT patients with underlying rare CNV had at least one miRNA gene overlapping the identified CNV. Network analysis of the most frequently affected miRNAs has revealed the dominant regulation of the two miRNAs, hsa-miR-484 and hsa-miR-185-5p. Additionally, miR-548 family members have shown substantial enrichment in rare CNVs in CAKUT. The in vitro model which depicts the heterozygous deletion of the MIR484 has confirmed the study concept implying that rare CNVs affect the corresponding miRNA expression and subsequently dysregulatres miRNA target genes. The translational capacity of miRNA to be employed in therapeutic approaches is nowadays increasingly investigated. Therefore, the untangling of the mechanisms affected by dysregulated miRNAs could serve for future extension of genetic testing and the development of novel miRNA targeting strategies in CAKUT.

Keywords: CAKUT, CNV, miRNA.

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