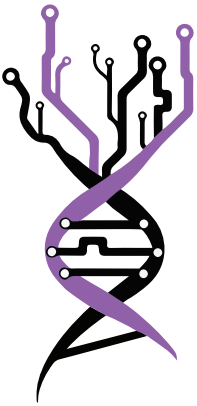
#BelBi2023 • Belgrade, Serbia

BOOK OF ABSTRACTS



4th Belgrade Bioinformatics Conference

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EDITORS
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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Decoding Cystic Fibrosis Phenotype

Aleksandra Divac Rankov*[1], Dušan Ušjak[1],
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Cystic fibrosis (CF) is a monogenic autosomal recessive disease caused by mutations in transmembrane conductance regulator (CFTR) gene. The golden standard for the diagnosis of CF is sweat chloride testing (>60 mmol/L) together with the identification of two CF-causing variants of CFTR gene. Nevertheless, about 0.01% of patients with elevated sweat chloride and high clinical suspicion of CF do not carry any CF-causing variants.
Here we present analysis of whole exome sequencing (WES) results for two patients with elevated sweat chloride levels and clinical presentation of CF in whom no CF-causing mutations were detected after CFTR gene whole coding region sequencing, and large insertion/deletion testing.
Genomic DNA was extracted from whole blood, subjected to library preparation using DNA nanoball technology from BGI and sequenced on DNBSEQ-G400 (MGI). Produced fastq files were mapped to hg38 reference genome using BWA/SAM tools. VCF files were generated using GATK (BaseRecalibrator, HaplotypeCaller) and annotated with InterVar and AnnoVar tools. Filtering of detected variants for disease relevance was done using the following criteria: QC Filter, GnomAD Allele Frequency, Functional consequences and phenotype-genotype relationship.
In both patients, similar number of variants predicted to impair protein function were detected (27 and 25). In two genes (CACNA1H and MUC5B) missense type variants were found in both patients and loss of function variants were found in 7 and 11 genes, respectively. Functional assessment of selected variants is underway.
Bioinformatics analyses are a valuable tool enabling identification of underlining genetic bases of disease phenotype, important in the context of optimal patient management and targeted therapies.

Keywords: whole exome sequencing (WES), cystic fibrosis, variant assessment

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