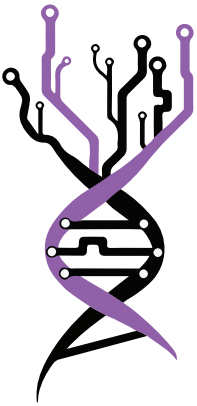
#BelBi2023 • Belgrade, Serbia

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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Potentially relevant variants of unknown significance in NGS-tested patients with suspected skeletal dysplasia

Marija Mijovic*[1], Goran Cuturilo[1,2], Jelena Ruml Stojanovic[1], Aleksandra Miletic[1], Brankica Bosankic[1], Hristina Petrovic[1], Bojana Vasic[1], and Nadja Vukasinovic[1]

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ACMG recognizes five different categories of sequence variants identified by next generation sequencing (pathogenic, likely pathogenic, variants of unknown significance, likely benign and benign). Sometimes, potentially relevant gene variants could be categorized as variants of unknown significance according to the level of available evidences. Because of that, detailed assessment of the phenotype-genotype correlation by the clinical geneticist in each individual case is crucially important. The interpretation and classification of a variant may change over time. Variant reinterpretation is defined as the practice of reevaluating all the evidence available about the pathogenicity of a genetic variant and taking into account any new evidence that is made available since the previous interpretation.

For the last seven years, we had 168 patients with clinically suspected locus heterogeneous skeletal dysplasia. Next generation sequencing (NGS) using clinical exome sequencing or whole exome sequencing was performed for all. All patients underwent detailed phenotype-genotype correlation investigation.

Molecular diagnosis by determining the pathogenic or likely pathogenic causative gene variant(s) was established for 102 out of 168 patients (60.71%). Additionally, in 10 patients (5.95%) variant of unknown significance (VUS) with good phenotype-genotype correlation was identified. These VUS variants could be potentially, and possibly are, causal, although there are no reliable evidences of their pathogenicity at the moment. In one of the positive patients in our study, the variant was initially classified as VUS, but with new evidence it was reclassified as likely pathogenic.

In the present study, a potentially relevant variant of unknown significance was detected in 5.95% of patients, which is a non-negligible proportion. For all these patients, we have organized clinical follow-up with periodic reinterpretation and reclassification of the detected variants.

Keywords: next generation sequencing, variant(s) of unknown significance, classification, reinterpretation, reclassification, skeletal dysplasia

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