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BOOK OF ABSTRACTS



4th Belgrade Bioinformatics Conference

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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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**A New Framework for the Use of Variant Interpretation Tools
in Clinical Practice**

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Current ACMG/AMP guidelines for the use of sequence variants for genetic diagnosis and treatment permit the use of *in silico* predictors as Supporting evidence (PP3 and BP4 criteria). These criteria, however, lack quantitative support and leave clinicians and scientists without standards for applying these criteria, leading to large interpretation variability. To address this challenge, our team built upon previous work and introduced a novel criterion that can be used to calibrate any computational model or any other continuous-scale evidence on any variant type. We used it to estimate score intervals corresponding to the four strengths of evidence for pathogenicity and benignity for fourteen missense variant interpretation tools on a carefully assembled data sets of known pathogenic and benign variants. We found that most tools achieved the Supporting evidence level for both pathogenic and benign classification using newly established data-driven thresholds. Importantly, at appropriate score thresholds, several *in silico* methods can also provide Moderate and Strong evidence levels for a limited number of variants. Based on these findings, we provided recommendations for quantitative revisions of the PP3 and BP4 criteria within ACMG/AMP guidelines and the future assessment of *in silico* methods for clinical interpretation.



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