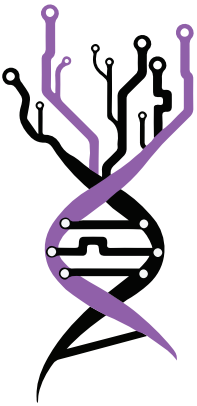
#BelBi2023 • Belgrade, Serbia

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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Profiling Pre-Replication Complex Mutations in Cancer

Jelena Kusic Tisma[1]*, Marija Orlic Milacic[2], Quang Trinh[2], Rhea Ahluwalia[2,3], Lincoln D. Stein[2,3]

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The pre-replication complex (preRC) consists of 15 proteins that mark DNA replication initiation sites and regulate replication timing. Deficiency in preRC proteins results in genomic instability (re-replication) and developmental defects (Meier-Gorlin syndrome). Our aim was to assess the scope of preRC gene aberrations in cancer. Variations in preRC genes were studied using CBio Portal software and TCGA PanCancer dataset. The functional impact of detected variants was evaluated in silico by three different prediction tools: SIFT (sequence and evolutionary conservation - based), PolyPhen2 (protein sequence and structure – based) and MutPred2 (supervised learning method based on neural networks).

No mutational hotspots were observed in any of the 15 preRC genes and no mutual exclusivity between mutations in preRC genes were detected. The highest alteration incidence in preRC genes was found in endometrial carcinoma and melanoma. The majority of the variations seen in preRC genes were non-synonymous. The functional assessment has shown that 253/1215 (21%) preRC gene mutations were predicted to be pathogenic with high confidence by 2/3 computational algorithms. None of the variants reached the high confidence pathogenicity score by all 3 prediction tool. In contrast, 49% of variants were predicted to be either benign by all three tools or benign by 2/3 or 1/3 tools, with the remaining 1/3 or 2/3, respectively, classifying them as low confidence pathogenic.

These finding suggest that mutations in preRC proteins might be passenger mutations and that cancer cells can tolerate them. The future step is to see whether incidence of coding vs. noncoding preRC mutations correlates with Tumor Mutation Burden (TMB) and Genome Instability Index (GII) of cancer.

Keywords: preRC, data mining, cBioPortal

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