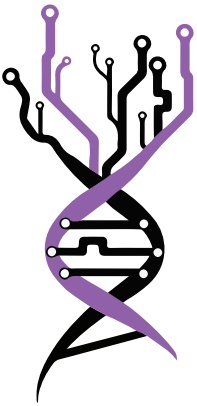
#BelBi2023 • Belgrade, Serbia

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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POSTER PRESENTATION

An integrated platform for genome assembly, comparative genomics and management of genomic variation databases

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The use of long read DNA sequencing technologies is producing an explosion of high-quality de-novo genome assemblies. The availability of these genomes represents a major step forward for evolution, population genomics, epidemiology, among other applications. A major bottleneck for many research groups continues to be the availability of tools to build and analyze the large datasets of genomes that can be produced using these technologies. In this talk, I summarize the functionalities developed by my research group in the version four of the Next Generation Sequencing Experience Platform (NGSEP) to perform a comprehensive analysis of long and short DNA sequencing reads. First, we designed new algorithms for assembly of haploid and diploid samples from long DNA sequencing reads. A minimizers table is constructed from the reads , using K-mer hash codes calculated from rankings relative to the mode of the k-mer counts distribution. Statistics collected during this process are used as features to build layout paths. For diploid samples, we integrated a reimplementation of the ReFHap algorithm to perform molecular phasing. Benchmark experiments using PacBio HiFi and Nanopore sequencing data for different species show that our solution has competitive contiguity and efficiency, as well as superior accuracy in some cases, compared to other currently used software. We also developed a functionality to perform ortholog identification and gene-based alignment of assembled genomes. Proteomes for each genome are extracted and homology relationships are efficiently predicted building indexes of aminoacid sequences by k-mer ocurrance. Then, genes are clustered in orthogroups based on the topology of the graph induced by the predicted relationships. Gene presence/absence matrices are derived from these orthogroups. If genome assemblies are provided as input, synteny relationships are identified for each pair of genomes. We also implemented algorithms to perform alignment of short and long reads to a reference genome. Based on aligned long reads, we improved the classical variants detector to detect long structural variants. Adding up these developments, NGSEP is a comprehensive tool to perform de-novo and reference-based analysis of DNA sequencing reads in a wide variety of experimental settings to solve different research goals.

Keywords: bioinformatics, algorithms, DNA sequencing, software, genome assembly

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