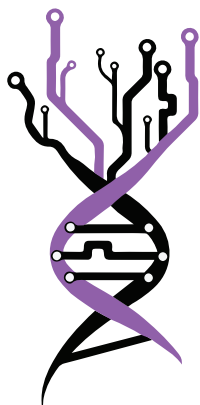


#BelBi2023 • Belgrade, Serbia

BOOK OF ABSTRACTS



4th Belgrade Bioinformatics Conference

HYBRID • 19 - 23 JUNE 2023

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ISBN: 978-86-82679-14-1

belbi.bg.ac.rs

Title	4 th Belgrade Bioinformatics Conference BOOK OF ABSTRACTS
Publisher	Institute of Molecular Genetics and Genetic Engineering, University of Belgrade Vojvode Stepe 444a, Belgrade, Serbia https://www.imgge.bg.ac.rs/
Editors	dr. Ivana Morić dr. Valentina Đorđević
Technical editor	Dušan Radojević
ISBN	978-86-82679-14-1
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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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Mapping of Disease Names to Disease Codes based on Natural Language Processing Techniques

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Information aggregation from various gen, disease, and gen-disease databases such as DisGeNet, COSMIC, HumsaVar, Orphanet, ClinVar, HPO, and Diseases into a unique database would enable researchers to analyze and compare valuable domain findings in a more convenient and systematic way. However, the aggregation poses numerous challenges due to non-uniform information annotation across the databases. In this work, we address the problem of mapping a disease name, when needed, into a standardized disease code (DOID) based on Natural Language Processing text representation techniques. We examine the benefits and limitations of using off-the-shelf embeddings such as Med2vec, and language models such as BioBERT, UmlsBERT, and PubMedBERT in retrieval scenarios with respect to standard full-text search. In addition to qualitative improvements, we elaborate on the technical requirements and computational complexities that come with the embracement of language models and semantic search.



ISBN: 978-86-82679-14-1