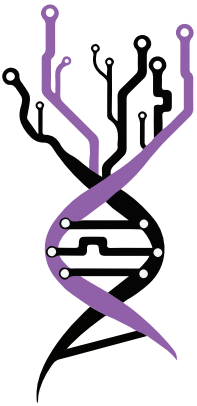
#BelBi2023 • Belgrade, Serbia

BOOK OF ABSTRACTS



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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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TABLE OF CONTENT

KEYNOTE LECTURES

Big Data in Biology: How EMBL delivers big data for biology, and some highlights of its application to human disease biology
Ewan Birney.....1

A tale of two stories: data-driven precision medicine and precision public health
Kristel Van Steen..... 2

Machine intelligence and network science for complex systems big data analysis
Carlo Vittorio Cannistraci..... 3

Targeting LLPS in disease: a new modality in drug development
Peter Tompa..... 4

Translating Bioinformatics Back To Healthcare: Facilitating the use of Artificial Intelligence at UW Medicine
Sean D. Mooney..... 5

Bioinformatics education course on gene networks reconstruction using online tools
Yuriy L. Orlov, Anastasia A. Anashkina..... 6

A New Framework for the Use of Variant Interpretation Tools in Clinical Practice
Predrag Radivojac..... 7

Privacy-preserving Systems Medicine
Jan Baumbach..... 8

INVITED LECTURES

Using AI/ML to transform molecular biology databases
Alex Bateman..... 9

Stereo-seq: Large Field of View-Spatially Resolved Transcriptomics at Nanoscale Resolution
Javier Batista Perez.....10

"What is life?'': Open quantum systems approach
Andrei Khrennikov.....11

Exploiting the linear organisation of omics network embedding spaces
Noël Malod-Dognin, Alexandros Xenos, Sergio Doria Belenguer, and Nataša Pržulj.....12

Inverting convolutional neural networks for super-resolution identification of regime changes in epidemiological time series
Jose M. G. Vilar.....13

Can we use biobanks to study infectious diseases?
Andrea Gelemanović.....14

An integrated platform for genome assembly, comparative genomics and management of genomic variation databases
Jorge Duitama..... 15

Bioinformatics and evolution of non-model organisms
Mikhail S. Gelfand..... 16

Computational bioengineering for heart disease
Nenad Filipovic, Themis Exarchos, and Djordje Jakovljevic..... 17

A Similarity-based Normative Framework for Bio-plausible Neural Nets
Anirvan Sengupta..... 18

Complexity driven evolution of Alternative splicing
Vladimir Babenko and Timophey Boltunov..... 19

Pangenomic Alignment: Strings plus Graphs
Travis Gagie..... 20

Circular Codes in the Genetic Information
Elena Fimmel and Lutz Strüngmann..... 21

Some Applications of Graph-Based Machine Learning Methods on Biological Data
Mladen Nikolić..... 22

From multifunctionality to polypathogenicity with intrinsic disorder
Vladimir N. Uversky..... 23

Exploring the impact of rare Copy Number Variants on miRNA genes in CAKUT: Insights from integrated bioinformatic analysis and experimental validation
Ivan Jovanović..... 24

Persistence of plasmids targeted by CRISPR interference in bacterial populations
Konstantin Severinov..... 25

Omics Data Fusion for Understanding Molecular Complexity Enabling Precision Medicine
Nataša Pržulj..... 26

An agnostic analysis of the human AlphaFold2 proteome using local protein conformations
Alexandre G. de Brevern..... 27

Uncovering resistance to microtubule targeting drugs
Mattia Pavani, Elena Chiroli, Paolo Bonaiuti, and Andrea Ciliberto..... 28

Computational tools and repositories for precision therapeutics in the post-genomic era
George P. Patrinos..... 29

Development of hybrid and optimized deep learning classifiers for speech recognition in tracheostomy patients: a case study
Themis Exarchos..... 30

Advancing Genomics with OrthoDB, BUSCO, and the LEM Framework
EV Kriventseva, M Manni, M Seppey, F Tegenfeldt, M Berkeley, D Kuznetsov, EM Zdobnov..... 31

Multiomics Integration by Non-Negative Tri-Matrix Factorization Reveals New Target Genes in Parkinson's Disease
Alexander Skupin.....32

Prediction of cell types using single-cell mRNA profiles
Vladimir Brusic..... 33

The complete solution and interpretation algorithms for large field-of-view and high-resolution spatial transcriptomics
Shuangsang Fang..... 34

To be folded, to be unfolded or to be aggregated with important functions: application of the directed coaggregation mechanism to combat bacterial communities
O.V. Galzitskaya, S.Yu. Grishin, A.V. Glyakina, M.V. Slizen, A.V. Panfilov, P.A. Domnin, A.P. Kochetov, A.A. Surin, S.V. Kravchenko, A.K. Surin, S.A. Ermolaeva..... 35

ORAL PRESENTATION

CADDIE - An online knowledge base for network-based mechanism exploration and drug repurposing in oncology
Michael Hartung, Elisa Anastasi, Zeinab M. Mamdouh, Cristian Nogales, Harald HHW Schmidt, Jan Baumbach, Olga Zolotareva, and Markus List..... 36

Mapping of Disease Names to Disease Codes based on Natural Language Processing Techniques
Anđelka Zečević, Jovana Kovačević, and Radoslav Davidović..... 37

Zero- and Few-Shot Machine Learning for Named Entity Recognition in Biomedical Texts
Miloš Košprdić, Nikola Prodanović, Adela Ljajić, Bojana Bašaragin, and Nikola Milošević..... 38

Clustering and classification of SARS-COV-2 isolates using RSCU
S. Malkov, M. Beljanski, G. Pavlović Lažetić, B. Stojanović, M. Maljković, A. Veljković, S. Kapunac, and N. Mitić..... 39

The use of Active Machine Learning for Protospacer-Adjacent Motif recovery in Class 2 CRISPR-Cas systems
Bogdan Kirillov, Aleksandra Vasileva, Oleg Fedorov, Maxim Panov, and Konstantin Severinov..... 40

Application of classification algorithms for hip implant surface topographies
Aleksandra Vulović, Tijana Geroski, and Nenad Filipović..... 41

Computational Modelling of Drug Effects on Cardiomyopathy and Analysis of Myocardial Work
Smiljana Tomasevic, Miljan Milosevic, Bogdan Milicevic, Vladimir Simic, Momcilo Prodanovic, Srboljub M. Mijailovich, Nenad Filipovic..... 42

Echocardiography-based Left Ventricle Cardiac Hypertrophy Simulations
Bogdan Milićević, Miljan Milošević, Vladimir Simić, Danijela Trifunović, Goran Stanković, Nenad Filipović, and Miloš Kojić..... 43

Decoding Cystic Fibrosis Phenotype
Aleksandra Divac Rankov, Dušan Ušjak, Martina Mia Mitić, and Jelena Kusic Tisma..... 44

Single cell 3' transcriptome profiling
Nevena Milivojević, Uršula Prosenc Zmrzljak, Biljana Ljujić, Valentina Đorđević, Marina Gazdić Janković, Marko Živanović, Feđa Puač, Miloš Ivanović, and Nenad Filipović..... 45

Modulating Horizontal Gene Transfer through Bistability in the Dynamics of Bacterial Restriction-Modification Systems
Marko Djordjevic, Lidija Zivkovic, and Magdalena Djordjevic..... 46

Cell-type-specific mechanistic drivers of progressive multiple sclerosis lesions
Elkjaer ML, Hartebrodt A, Oubounyt M, Weber A, Vitved L, Reynolds R, Thomassen M, Rottger R, Baumbach J, and Illes Z..... 47

AI-powered framework to predict the toxicity of microplastics
Junli Xu..... 48

Newest Advances on the FeatureCloud Platform for Federated Learning in Biomedicine
Niklas Probul, Mohammad Bakhtiari, Mohammad Kazemi Majdabadi, Balázs Orbán, Sándor Fejér, Supratim Das, Julian Klemm, Christina C Saak, Nina K Wenke, and Jan Baumbach..... 49

Deciphering key regulatory networks and drug repurposing candidates through scRNAseq data analysis using SCANet
Mhaned Oubounyt, Jan Baumbach, and Maria L. Elkjaer..... 50

From protein-protein to isoform-isoform interactions: the toolkit to map alternative splicing to interactome
Olga Tsoy, Zakaria Louadi, Chit Tong Lio, Jan Baumbach, Olga Kalinina, Alexander Gress, Tim Kacprowski, and Markus List..... 51

Drugst.One - A plug-and-play solution for online systems medicine and network-based drug repurposing
Andreas Maier, Michael Hartung, The Drugst.One Initiative, and Jan Baumbach..... 52

Fatty Acid Data Analysis Unravels Skeletal Site and Age-Specific Features of Human Bone Marrow Adiposity
Drenka Trivanović, Jovana Kovačević, Aleksandra Arsić, Marko Vujačić, Nikola Bogosavljević, Ivana Okić Djordjević, Milena Živanović, Slavko Mojsilović, Mirjana Maljković, and Aleksandra Jauković.... 54

Exploration of Pharmacogenomic Biomarkers in Chronic Immune Diseases Using Single-Cell RNA Sequencing
Mario Gorenjak, Larisa Goričan, Boris Gole, Uršula Prosenc, Erik Melén, Michael Kabesch, Anke H Maitland-van der Zee, Susanne Reinartz, Susanne J H Vijverberg, Uroš Potočnik and the PERMEABLE consortium..... 55

Dehydrins in the service of protecting the DNA helix from the aspect of molecular dynamics (MD)
Milan Senćanski, Ivana Prodić, Ana Pantelić, and Marija Vidović..... 57

Machine learning approach in inferring main population-level COVID-19 risk factors
Sofija Marković, Anđela Rodić, Ognjen Milićević, Igor Salom,
Magdalena Đorđević, and Marko Đorđević..... 58

Using whole exome sequencing to explore genetic basis of unicuspid aortic valve disease
Martina Mia Mitić, Dušan Ušjak, Maja Milošević, Marija Cumbo,
Sofija Dunjić Manevski, Branko Tomić, Ivana Petrović, Petar Otašević,
Slobodan Micović, Milovan Bojić, and Valentina Đorđević..... 59

Online *in silico* validation of disease and gene sets, clusterings or subnetworks with DIGEST
Klaudia Adamowicz, Andreas Maier, Jan Baumbach, and David B. Blumenthal..... 60

Alternative splicing impacts microRNA regulation within coding regions
Lena Maria Hackl, Amit Fenn, Zakaria Louadi, Jan Baumbach,
Tim Kacprowski, Markus List, and Olga Tsoy 61

Using AI to design antibodies
Goran Rakočević..... 62

Semantic unification and search of bioinformatics databases
A. Veljković, and N. Mitić..... 63

Beyond the Global Health Security Index: A Machine Learning Approach
to Analyzing the Official COVID-19 Deaths and Excess Deaths Data
Andjela Rodic, Sofija Markovic, Igor Salom, and Marko Djordjevic..... 64

Integration of differential transcriptomic and proteomic data in hydrated
and desiccated leaves of *Ramonda serbica* Panc.
Marija Vidović, Ilaria Battisti, Ana Pantelić, Dejana Milic,
Giorgio Arrigoni, Antonio Masi, and Sonja Veljović Jovanović..... 65

POSTER PRESENTATION

Possible role of estrogen metabolism and aldo-keto reductase activity in
chemoresistance of ovarian cancer
Nika Marolt, Andrew Walakira, Tadeja Režen, Damjana Rozman and Tea Lanišnik Rižner..... 66

Seven miRNAs potentially included in the chilling response of maize plants
in early stages of development
Manja Božić, Dragana Ignjatović-Micić, Nenad Delić, Marko Mladenović,
Jelena Vančetović, Bojana Banović Đeri, Ana Nikolić..... 67

Two contrasting late embryogenesis abounded protein family groups of
Ramonda serbica Panc.
Ana Pantelić, Strahinja Stevanović, Sonja Milić Komić, Nataša Kilibarda and Marija Vidović..... 68

De novo Genome Assembly of Sweet Chestnut (*Castanea sativa* Mill.)
Insights into the Molecular Basis of its Nutritional Properties
M. Aydin Akbudak and Ali Tevfik Uncu..... 69

Numerical and Biological Modeling Approach in the Analysis
of the Cancer Viability and Apoptosis
Katarina Virijević, Marko Živanović, Marina Gazdić Janković, Amra Ramović Hamzagić, Nevena Milivojević, Katarina Pecić, Dragana Šeklić, Milena Jovanović, Nikolina Kastratović, Ana Mirić, Tijana Đukić, Ivica Petrović, Vladimir Jurišić, Biljana Ljujić, Nenad Filipović..... 70

Root colonization ability of herbicide-resistant PGP bacteria
evaluated by 16S rRNA metabarcoding
Cristina Bez, Ivana Galic, Iris Bertani, Nada Stankovic, Vittorio Venturi..... 71

Genetic Complexity and Synteny Analysis of Castanea Genomes:
Unveiling the Significance of Chestnut Species in Ecological and Genomic Perspectives
Ali Tevfik Uncu and M. Aydin Akbudak..... 72

Elongation factor P (-like) protein and polyproline motifs
Marina Parr, Alina Sieber, Prof. Dr. Dmitrij Frishman and Dr. Jürgen Lassak..... 73

Comparative study of in silico protein design techniques
Ivan Tanasijević and Branka Rakić..... 74

Energy and information exchange between "donor" and "molecular bridge" structures:
non adiabatic polaron model
Dalibor Chevizovich, Vasilije Matic, and Zeljko Przulj..... 75

Profiling Pre-Replication Complex Mutations in Cancer
Jelena Kusic Tisma, Marija Orlic Milacic, Quang Trinh, Rhea Ahluwalia, Lincoln D. Stein..... 76

Combined experimental and theoretical study of Type-II toxin-antitoxin
system response to antibiotics
Bojana Ilic, Marko Đorđević, Hong-Yu Ou..... 77

Methodology, performance and retrainability survey of intrinsic disorder predictors
Nevena Ćirić and Jovana Kovačević..... 78

Evaluating ND1 and Cytb mitochondrial genes as markers for diversity analysis of
protected White-tailed eagle species from Serbia
*Slobodan Davidovic, Milica Stanković, Pavle Erić, Katarina Erić,
Aleksandra Patenković and Marija Tanasković*..... 79

Analysis of nucleotide sequence repeats in coronaviruses
S. Kapunac, S. Malkov, M. Beljanski, G. Pavlović Lažetić, B. Stojanović, M. Maljković, A. Veljković , N. Mitić..... 80

Genomic Surveillance and Phylogenetic Analysis of SARS-CoV-2 Variants in Serbia:
Insights into Evolutionary Dynamics and Genetic Diversity
Mirjana Novkovic, Bojana Banovic Djeri, Sasa Todorovic, and Valentina Djordjevic..... 81

Deciphering the reward-related impulsivity domains in rats:
The big data study of historical control
Jovana Aranđelović, Kristina Mirković, Jana Kojić, Miroslav Savić..... 82

Computer analysis of glioma gene network structure
Iarema P.O., Turkina V.A., Mayorova A.A., Orlov Y.L...... 83

Genome-wide association analysis for severe COVID-19 in Serbian population
Marko Zecevic, Nikola Kotur, Bojan Ristivojevic, Vladimir Gasic,
Branka Zukic, Sonja Pavlovic and Biljana Stankovic..... 84

Impact of different mapping tools on detection of small RNAs in bacterial outer membrane vesicles
Bojana Banović Đeri, Sofija Nešić, Ana Pantelić, Jelena Samardžić, Dragana Nikolić..... 85

In silico pre-selection of β-glucosidase gene for heterologous recombinant expression
Marija Atanaskovic, Ivana Moric, Milos Rokic, Lidija Senerovic..... 86

Supervised Machine Learning Approach for Prediction of
Occult Lymph Node Metastasis in T1-T2 Papillary Thyroid Carcinoma
Marina Popović Krneta, Nemanja Krajčinović, Zoran Bukumirić, and Miljana Tanić..... 87

Determinants of CRISPR array non-canonical adaptation mechanism
Marko Tumbas and Marko Đorđević..... 88

Data mining for long-non coding RNAs deregulated in colon cancer
through analysis of Gene Expression Omnibus database
Iva Pruner and Aleksandra Nikolic..... 89

Efficient bioinformatics workflow for *de novo* transcriptome assembly
of *Pelargonium zonale*
Dejana Milić, Ana Pantelić, Jelena Samardžić, Bojana Banović Đeri, Marija Vidović..... 90

Application of principal component analysis (PCA) and analytical hierarchy process (AHP) in analysis of articulatory characteristics of phonemes of children with 22q11.2 Deletion Syndrome
Danijela Drakulic, Marijana Rakonjac, Goran Cuturilo, Natasa Kovacevic-Grujicic,
Jelena Kusic-Tisma, Ivana Moric, Branka Zukic, and Milena Stevanovic..... 91

Integrated relational database of human protein-protein interactions
Bojana Jošić, Jovana Kovačević, Vladimir Perović, Nevena Veljković..... 92

Mining for the data about glycosylation in the bovines-
the analysis of the recently published studies
Anđelo Beletić, Ivana Duvnjak Orešković, Tea Pribić, and Gordan Lauc..... 93

Different approaches in microRNA analysis
Barbara Jenko Bizjan, Bine Stančič, Iva Sabolić,
Maja Štalekar and Uršula Prosenc Zmrzljak..... 94

In silico analysis and prediction of novel pharmacogenomic markers of pediatric ALL treatment
Vladimir Gašić, Nikola Kotur, Biljana Stanković, Đorđe Pavlović, Marina Jelovac, Jelena Perić, Bojan Ristivojević, Sonja Pavlović, and Branka Zukić..... 95

Exploring Changes in Diagnoses during the COVID-19 Era: Comparative Analysis
Despina Misheva, Marija Stojcheva, Hana Hasanicaj, Ana Mladenovska, Jovana Dobreva, Mary Lucas, Irena Vodenska, Lou Chitkushev, Dimitar Trajanov..... 96

Shotgun metagenomics reveals gut microbiota features associated with the efficacy of myeloid derived suppressor cells in the prevention of neuroinflammation
Marina Bekić, Nataša Ilić, Jelena Đokić, Dušan Radojević, Dragana Vučević, Saša Vasilev, and Sergej Tomić..... 97

Seeking an optimal variant calling pipeline for medical genetics
Yury A. Barbitoff, Alexandra Panteleeva, Alexander V. Predeus..... 98

Groundwater and soil as a reservoir for polyurethane-degrading bacteria
Milica Ciric, Brana Pantelic, Vladimir Šaraba, and Jasmina Nikodinovic-Runic..... 99

Developing bioinformatics pipeline for processing environmental DNA metabarcoding sequencing data
Iva Sabolić, Lucija Markulin, Teja Petra Muha, Barbara Jenko, Uršula Prosenc Zmrzljak..... 100

Evaluation of variant calling tools for detection of SNVs in BRCA1 and BRCA2 genes in patients from the Institute of Oncology and Radiology of Serbia
Isidora Pantović, Katarina Živić, Ivana Boljević, Milica Nedeljković, Radmila Janković, Miljana Tanić..... 101

Transcriptome analysis of *Pseudomonas aeruginosa* after MhqO dioxygenase treatment
Andjela Djokic, Ivana Moric, Lidija Senerovic and Lidija Djokic..... 102

PACSIN2 modifies miRNAs in extracellular vesicles, modulating thiopurine response
Alessia Norbedo, Marianna Lucafò, Carlotta Bidoli, Marco Gerdol, Metka Lenassi, Giuliana Decorti, Gabriele Stocco..... 103

Pathway analysis of $CD8^+$ T cell transcriptome in glioblastoma patients reveales multiple sclerosis signaling pathway as the top rated upregulated disease pathway in tumor infiltrating cells
Milan Stefanović, Ivan Jovanović, Aleksandra Stanković and Maja Živković..... 104

Transcriptomic profiling of white blood cells reveals new insights into the molecular mechanisms of thalidomide in children with inflammatory bowel disease
Marianna Lucafò, Letizia Pugnetti, Debora Curci, Carlotta Bidoli, Marco Gerdol, Fulvio Celsi, Sara Renzo, Monica Paci, Sara Lega, Paolo Lionetti, Alberto Pallavicini, Giuliana Decorti, Gabriele Stocco, Matteo Bramuzzo..... 105

The past, the present, and the future of RNA secondary structure prediction
Lazar Vasović..... 106

The use of tryptic food protein digests data in public proteomic repositories to assess the effects of chemical and post-translational modifications on digestion outcomes
Ivana Prodić, Teodora Đukić, Vesna Jovanović, Katarina Smiljanić..... 107

Machine learning-based data correlation between scanning electron microscopy images and energy-dispersive X-ray spectroscopy profiles
Ahmed Musa, Baeckkyoung Sung, Leon Abelmann..... 108

Protein structural differences in Cytochrome c oxidase subunit 1 of two Heterogynis species as a new approach for species delimitation
Marija Vidović, Vladislava Galović..... 109

Potentially relevant variants of unknown significance in NGS-tested patients with suspected skeletal dysplasia
Marija Mijovic, Goran Cuturilo, Jelena Ruml Stojanovic, Aleksandra Miletic, Brankica Bosankic, Hristina Petrovic, Bojana Vasic, and Nadja Vukasinovic..... 110

Analysis of Long COVID Phenotypes and their Impact on Mental Health and Daily Functioning: Insights from Twitter
Marko Markovikj, Jovana Dobreva, Mary Lucas, Irena Vodenska, Lou Chitkushev, Dimitar Trajanov..... 111

Metagenomic Analysis of Bacterial Community and Isolation of Representative Strains from Vranjska Banja Hot Spring, Serbia
Jovana Curcic, Danka Matijasevic, Nemanja Stanisavljevic, Srdjan Tasic, Milan Kojic, and Milka Malesevic..... 112

Using whole exome sequencing to explore genetic basis of unicuspid aortic valve disease

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Normal aortic valve consists of three cusps that develop in the embryonic stage. Unicuspid aortic valve (UAV) is a rare congenital anomaly resulting in only one cusp with estimated prevalence of 0.02% in general population. Aim of this study was to identify genetic variants possibly associated with development of UAV. The study included 17 subjects, namely 5 UAV patients and their healthy family members without UAV disorder. Total DNA was isolated from venous blood samples and whole exomes sequencing (WES) was performed using BGI's WES protocol. Adapter-trimmed and quality-filtered reads (fastp) were mapped to hg38 reference genome using BWA/SAMtools. VCF files were generated using GATK (BaseRecalibrator, HaplotypeCaller) and annotated with InterVar and AnnoVar tools. Rare heterozygous variants present in UAV patients were found in NOTCH1, TGFB2, MYH6, EGFR, FBN2, C1R, ROBO4 and TBX5, genes associated with development of aortic valves. Among these, most were missense mutations with damaging effects as predicted using *in silico* tools (SIFT and/or Polyphen). Only mutation in MYH6 p.Ala1130Ser was found in at least two different UAV patients. Also, rare homozygous missense mutation p.Gly577Ser with high damaging potential was found in ADAMTS5 gene. Besides, highly damaging heterozygous missense mutations were detected in gene interacting functional partners (STRING) of genes associated with development of aortic valves: DVL1, THBS1, NOTCH4, ADAMTS3, FBN1, NOTCH2, ADAM17, LRP5, WWTR1, C1S, ANKRD6 and TNNI1, as well as homozygous in ACAN and KNG1. Taken together, malfunctions in ADAMTS5, ACTA2, MYH6, FBN2, AXIN1, CELSR1 or TBX5 networks were found to be common in at least two UAV patients, suggesting existence of genetic basis in UAV disorder, possibly as a result of combined effects of multiple variants.

Keywords: unicuspid aortic valve, congenital heart disease, whole exome sequencing, genetic variants, valves

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