GENOMICS OF RARE DISEASES

SERBORDISINN & 2014 GOLDEN HELIX SYMPOSIUM

31. October – 1. November 2014.

Meeting venue: Hotel Tulip Inn Putnik, Belgrade, Serbia

(www.tulipinnputnikbelgrade.com)

ORGANIZING COMMITTEE

1. **Sonja Pavlovic, PhD,** (Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia)
2. **Maja Stojiljkovic, PhD,** (Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia)
3. **Prof. George P. Patrinos, PhD,** (University of Patras, Patras, Greece)
4. **Radoje Drmanac, PhD**, (Complete Genomics, Mountain View, CA, USA), honorable member of the committee

Co-organizers:

- SERBORDISinn project (EU-FP7-REGPOT), Institute of Molecular Genetics and Genetic Engineering, University of Belgrade
- Golden Helix Foundation

Communication sponsors:

Public Health Genomics

Orphanet

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Ministry of Education, Science and Technological Development RS (III41004)

Genomic Medicine Alliance

RD-Connect

SCIENTIFIC COMMITTEE

1. **Radoje Drmanac, PhD,** (Complete Genomics, Mountain View, CA, USA), honorable member of the committee
1. **Sonja Pavlovic, PhD,** (Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia)
2. **Prof. George P. Patrinos, PhD,** (University of Patras, Patras, Greece)
3. **Prof. Angela Brand MD PhD MPH,** (Institute for Public Health Genomics, Maastricht University, Maastricht, The Netherlands)
4. **Prof. Belen Perez, PhD,** (Centro de Biología Molecular, UAM, Madrid, Spain)
5. **Maja Stojiljkovic, PhD,** (Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia)
6. **Jelena Begovic, PhD,** (Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia)
7. **Prof. Dragana Janic, MD Phd,** (University Children's Hospital, Belgrade, Serbia)
8. **Maja Djordjevic, MD PhD,** (Mother and Child Healthcare Institute "Dr Vukan Cupic", School of Medicine, University of Belgrade, Belgrade, Serbia)

Friday, October 31th, 2014

08:00 – 17:00	**Registration for participants**
08:30 – 09:00	Welcome Addresses
KEYNOTE LECTURES I ***Chair: Sonja Pavlovic (Belgrade, Serbia)***	
09:00 – 09:30	**Radoje Drmanac** (Mountain View, USA; Shenzhen, China) *Whole genome sequencing for personal and population genomics: rare disease diagnosis and prevention*
09:30 – 10:00	**Hanns Lochmuller** (Newcastle upon Tyne, Great Britain) *RD-Connect and NeurOmics in rare disease research*
10:00 – 10:30	**Belen Perez** (Madrid, Spain) *Next generation sequencing applied to the diagnosis of inherited metabolic disorders*
10:30 – 11:00	COFFEE BREAK AND POSTER VIEWING
SESSION I: PUBLIC HEALTH OF RARE DISEASES ***Chair: Dragica Radojkovic (Belgrade, Serbia)***	
11:00 – 12:00	**Angela Brand** (Maastricht, The Netherlands) *Public Health Genomics - What's in for Rare Diseases?*
	Pilar Nicolas Jimenez (Bilbao, Spain) *Genomics of rare diseases within Europe. Legal issues.*
	Elisabeth Dequeker (Leuven, Belgium) *External quality assessment of genetic testing: a necessity for quality of care*
	Sonja Pavlovic (Belgrade, Serbia) *State of the art of rare disease activities in Serbia*
12:00 – 12:15	**CORPORATE LECTURE** **Szabolcs Kokeny** (Product Specialist EMEA, Illumina) *Gaining insight into genetic diseases with Illumina sequencing technology*

SESSION II: GENOMICS OF RARE DISEASES ***Chair: Milena Stevanovic (Belgrade, Serbia)***	
12:15 – 13:15	**Kostas Stamatopoulos** (Thessaloniki, Greece) *Immunogenetics in marginal-zone lymphomas*
	Beata Burzynska (Warsaw, Poland) *Genomics and new perspectives for rare red blood cell disorders*
	Istvan Balogh (Debrecen, Hungary) *Monogenic diabetes - lessons from a rare form of a common disease*
	Ayse Nazli Basak (Istanbul, Turkey) *The distinct genetics of ALS on Turkey*
13:15 – 14:00	**LUNCH**
KEYNOTE LECTURES II ***Chair: George Patrinos (Patras, Greece)***	
14:00 – 14:30	**Federico Innocenti** (North Carolina, USA) *Exceptional responders in cancer therapy*
14:30 – 15:00	**Michael Lee** (Yokohama, Japan) *Pharmacogenetic study of drug induced severe cutaneous disorders*
SESSION III: PHARMACOGENOMICS AND INDIVIDUALIZED THERAPY ***Chair: George Patrinos (Patras, Greece)***	
15:00 – 16:30	**Milan Macek Jr** (Praha, Czech Republic) *Pharmacogenomic studies in rare disease diagnostics and therapy: cystic fibrosis as a model*
	George P. Patrinos (Patras, Greece) *Personalized pharmacogenomics profiling*
	Alessio Squassina (Monserrato, Cagliari, Italy) *Cluster headache and bipolar disorder: is there a link?*
	Maria Puiu (Timișoara, Romania) *Pharmacogenetics - a promising perspective for tailoring the antipshychotic*

	treatment
	Lourdes Desviat (Madrid, Spain) *RNA-based therapies for rare diseases*
	Noora Ottman (Wageningen, The Netherlands) *Microbiota functionality and therapeutic possibilities*
16:30 – 16:40	**Best Abstract Award**
16:40 – 17:15	COFFEE BREAK AND POSTER VIEWING
	FREE AFTERNOON FOR PARTICIPANTS
17:00	**Organized transport from Tulip Inn Hotel to the IMGGE**
SERBORDISinn PROJECT – FUTURE PERSPECTIVES OF RESEARCH ON RARE DISEASES IN SERBIA Venue: IMGGE Institute, Vojvode Stepe 444a, Belgrade	
17:30 – 18:30	SERBORDISinn STEERING COMMITTEE MEETING (only for management team and members of SC)
18:30 – 19:30	SERBORDISinn PROJECT PARTNERS MEETING (open for all EU partners, Balkan partners and local partners)
19:30	**Organized transport from IMGGI to restaurant**
20:00	CONFERENCE DINNER for speakers **Organized transport from the restaurant to Tulip Inn Hotel**

Saturday, November 1st, 2014

SESSION IV: RARE DISEASES IN BALKAN COUNTRIES **(short presentations, discussions, initiatives)** ***Chair: Maja Stojiljkovic (Belgrade, Serbia)***	
10:00 – 10:10	**Zeljka Dukic** (Ministry of Education Science and Technological Development Republic of Serbia) *Balkan Region's participation in Horizon 2020*
10:10 – 11:40	**Maja Djordjevic** (Belgrade, Serbia) *Inborn errors of metabolism in Serbia*
	Irena Drmic Hofman (Split, Croatia) *Molecular diagnostics of rare cancers - time for regional networking?*
	Ksenija Fumic (Zagreb, Croatia) *Laboratory diagnosis of rare diseases in Croatia: present situation and trends*
	Snezana Kojic (Belgrade, Serbia) *Diagnosis of neuromuscular diseases in Serbia and launch of the Serbian Neuromuscular Disease Network NMD-SERBNET*
	Nina Maric (Banja Luka, Bosnia and Herzegovina) *Rare diseases detected in genetic counseling in Republic of Srpska*
	Ales Maver (Ljubljana, Slovenia) *Centre for Mendelian genomics: facilitating diagnostics of rare disorders*
	Olivera Miljanovic (Podgorica, Montenegro) *Approach to rare diseases in Montenegro*
	Borut Peterlin (Ljubljana, Slovenia) *Clinical exome sequencing improves diagnostics of rare diseases*
	Dijana Plaseska-Karanfilska (Skopje, Macedonia)
	Emil Polák (Bratislava, Slovakia) *Rare diseases in Slovakia: Diagnostics and future perspectives*
	Jelica Predojevic Samardzic (Banja Luka, Bosnia and Herzegovina) *Rare diseases in Republic of Srpska*

	Albena Todorova (Sofia, Bulgaria) *Molecular defects behind the syndromes in Bulgaria. The application of novel technologies in diagnostics of rare disease*
11:40 – 12:10	COFFEE BREAK AND POSTER VIEWING
12:10 – 13:40	**Goran Brajuskovic** (Belgrade, Serbia)
	Aleksandra Divac Rankov (Belgrade, Serbia) *Molecular diagnostics of rare pulmonary diseases in Serbia*
	Vita Dolzan (Ljubljana, Slovenia) *20 years of molecular genetic diagnostics in Slovenian CAH patients and families: CYP21A2 mutation spectrum and genotype-phenotype correlation*
	Danijela Drakulic (Belgrade, Serbia) *Microdeletion syndromes: our experience*
	Dragana Janic (Belgrade, Serbia) *Gaucher disease research in Serbia*
	Lejla Kapur Pojskic (Sarajevo, Bosnia and Herzegovina) *Inbreeding as contributing factor in prevalence of rare diseases*
	Ivana Novakovic (Belgrade, Serbia) *From linkage analysis to whole genome sequencing: discovering of the new gene responsible for Idiopathic Basal Ganglia Calcification*
	Jadranka Jovanovic-Privrodski (Novi Sad, Serbia)
	Jelena Roganovic (Rijeka, Croatia) *Fabry disease in children*
	Dusanka Savic – Pavicevic (Belgrade, Serbia)
	Zorica Sumarac (Belgrade, Serbia) *Gaucher Disease in Serbian Population: Enzymatic Laboratory Diagnostics*
	Marija Zaric (Banja Luka, Bosnia and Herzegovina)
13:40 – 14:00	**CLOSING REMARKS – FUTURE MEETINGS**
14:00 – 15:00	LUNCH