REGIONAL CONFERENCE ON RARE DISEASES

TOPIC IN FOCUS

Diagnostics in rare diseases

28 November 2024.

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PROGRAM

Hotel Sheraton, Novi Sad

09:00–9:30	**Registration & welcome coffee**
9:30–10:00	**Opening ceremony**

INTRODUCTION — Regulation

10:00–10:15	**„The importance of early diagnosis - the European trend "6 months“** Darija Julkovska, ERDERA-The European Joint Programme on Rare Diseases
10:15–10:30	**„The status of patients with rare diseases and their right to diagnosis from the point of view of medical law”** Hajrija Mujović, Institute of Social Sciences, Centre for Legal Research
10:30–10:45	**“The current situation in the diagnosis of rare diseases in Serbia”** Olivera Jovović, National Organization for Rare Diseases of Serbia, president
10:45–11:00	**"Diagnostics of rare diseases in the Republic of Srpska: challenges and advantages of a small population"** doc. Dr Nina Marić, Clinic for Children's Diseases UKC Republika Srpska, Center for Rare Diseases of the Republika Srpska
11:00–11:15	**"R69 Initiative - ICD-10 Code for "illness, unspecified"** Helen Hernandez, , Kal research Initiative - Initiative R69
11:15–11:30	**“Digital solutions for the diagnostic odyssey”** Vukašin Radulović, , Heliant, director
11:30–11:40	**Panel - questions**

OVERVIEW — Predictive diagnostics

11:40–12:00	**“Screening of carriers of genetic diseases in the age of genomic medicine”** Borislav Simović, Premium Genetics
12:00–12:20	**“Preimplantation genetic testing of monogenic diseases (PGT-M) for all couples at the risk of rare disease“** Mgr. Jakub Horak, Ph.D., Director of Preimplantation Genetic Testing (PGT), GENNET The Centre of Medical Genetics and Reproductive Medicine, Board Member of PGDIS-Preimplantation Genetic Diagnosis International Society, USA
12:20–12:40	**“Prenatal screening of hereditary disorders in Serbia”** Prim. dr sci. med. Aleksandra Novakov Mikić, Polyclinic Novakov
11:30–11:40	**Panel - questions**
12:50–13:30	**Lunch**

OVERVIEW — Targeted diagnostics

13:30–13:45	**Helene Cederroth, Wilhelm Foundation**, Undiagnosed network
13:45–14:00	**“Challenges of laboratory diagnostics of hereditary metabolic disorders in the era of new technologies”** Ksenija Fumić, PhD, Department of Medical Biochemistry and Hematology, Zagreb University Hospital
14:00–14:25	**„The Current and Prospective Applications of Next-Generation Sequencing in the Diagnosis of Rare Diseases“** Maja Stojiljković, PhD, Full Research Professor, Institute of Molecular Genetics and Genetic Engineering, University of Belgrade
14:25–14:40	**„Diagnostics and novel treatment options of severe short staturer“** Assoc. Prof. Primož Kotnik MD, PhD University Clinical Center Ljubljana, Pediatric Clinic, Clinical Department for Endocrinology, Diabetes and Metabolic Diseases, Ljubljana, Slovenia – Medical Faculty, University of Ljubljana
14:40–14:55	**„Challenges in the diagnosis of genetic diseases in Serbia today“** Prof. dr Goran Čuturilo, University Children's Hospital Belgrade
14:55–15:10	**Panel — questions**
15:10–15:25	**“Why timely diagnosis in rare diseases is important”** Dr Neena Nizar, Janssen Foundation, USA
15:25–15:40	**“Diagnostics of hereditary bone diseases”** doc. dr Adrijan Sarajlija, MD, PhD, Assoc. Professor of Pediatrics - University of Belgrade, Pediatrician, Clinical Genetics Consultant, Mother and Child Health Care Institute of Serbia "Dr. Vukan Cupic".
15:40–15:55	**„Challenges in the diagnosis of Williams syndrome”** PhD Sanda Huljev Frković, specialist in pediatrics, KBC Zagreb-Rebro, Croatia dr Aleksandra Perović, Associate Professor, University College London, Great Britain
15:55–16:10	**“Diagnosis of rare diseases in Vojvodina“** Prof. dr Ivana Kavečan Specialist in pediatrics, subspecialist in clinical genetics, Institute for Health Care of Children and Youth of Vojvodina
16:10–16:25	**„Experiences of the University Clinical Center of Serbia in the diagnosis of rare diseases and ways of improvement”** Prof. dr Zorica Šumarac, Center for Medical Biochemistry, Clinical center of Srbija, Belgrade
16:25–16:40	**“The importance of genetic testing in the diagnosis of rare neurological diseases of adulthood“** Dr Milena Janković, PhD, Full Research Professor, Laboratory for Molecular Genetic diagnosis of Neurological Diseases, Clinic for Neurology, Clinical Center of Serbia
16:40–16:55	**“Cegat - modern genetic diagnosis of rare diseases”** Dr med. Aleksandar Antović, medical director of Family Health
16:55–17:05	**Panel — questions**
PANEL DISCUSSION:	**Diagnosis of tuberous sclerosis from the point of view of the clinician and the patient** Moderator: Ivana Badnjarević
17:05–17:45	**Tamara Brtan,** , mother of a girl suffering from tuberous sclerosis **Dr Galina Stevanović,** Clinic for neurology and psychiatry for children and youth, Belgrade **Prim. dr sci. med. Aleksandra Novakov Mikić,** Polyclinic Novakov
17:45	**Q&A**

Conclusions and closing

Grad Novi Sad
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