# DAY 1 Thursday 9th of October 2025

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| **8:00-13:00** | **WS.01** |   **Workshop: Interpretation of NGS data** | **A. Maver** | |
| **13:00** | | **Lunch break** | | |
| **14:00-15:40** | **P1** | **Welcome address and plenary lectures *(B. Peterlin, D. Plaseska-Karanfilska)*** | | |
| | **W.01.P** 45 min | Using genomic medicine to make drug prescription safer and more effective | **W. Newman** | **United Kingdom** |
| | **W.02.P** 45 min | Gamechanger in rare disease research and diagnosis: the role of 3D facial gestalt scanning technology | **M. Macek** | **Czech Republic** |
| **15:40-16:50** | **S1** | **Session 1: Advances in rare disease diagnostics *(W. Newman, K. Writzl)*** | | |
| | **S1.01.I** 20 min | Long-read sequencing for rare disease research and diagnostics | **B. Van der Sanden** | **Netherlands** |
| | **S1.02.I** 20 min | ONT Sequencing | **S. Ossowski** | **Germany** |
| | **S1.03.I** 10 min | Structural variants in rare disease | **A. Kovanda** | **Slovenia** |
| | **S1.04.O** 10 min | Rapid Intraoperative Classification of Central Nervous System Tumours Using Real-Time Nanopore Sequencing | **S. Petrin** | **Slovenia** |
| | **S1.05.O** 10 min | The contribution of reanalysis of whole exome sequencing data to diagnosis rate | **H. Bas** | **Turkey** |
| **16:50** | | **Coffee break** | | |
| **17:20-18:50** | **S2** | **Session 2: What is new in Mendelian disorders *(M. Macek, L. Lovrečić)*** | | |
| | **S2.01.P** 20 min | Beyond Chromosomes: The Monogenic Causes of Early Pregnancy Loss | **D. Plaseska-Karanfilska** | **North Macedonia** |
| | **S2.02.I** 20 min | Chromatin remodeling disorders - challenging path to diagnosis and management | **L. Odak** | **Croatia** |
| | **S2.03.O** 10 min | Reporting Beyond the Primary Diagnosis: Actionable Additional Findings in WES/WGS | **B. Golob** | **Slovenia** |
| | **S2.04.O** 10 min | 3D Facial Gestalt Analysis of Individuals with Mutated PKD1 Genes in Polycystic Kidney Disease Patients | **M. Mihulová** | **Czech Republic** |
| | **S2.05.O** 10 min | Uncovering Dual Diagnoses through Trio-Based Whole Exome Sequencing (WES): Incidence and Clinical Implications | **F. Perino** | **Italy** |
| | **S2.06.O** 10 min | Resolving diagnostic challenges in skeletal dysplasia – a clinical overview of a ten years' experience of a single genetic center in Serbia | **M. Mijović** | **Serbia** |
| | **S2.07.O** 10 min | Genetic basis of female protective effect in neurodevelopmental disorders and beyond | **D. Perović** | **Serbia** |
| **18:50** | **S3-R** | **Roundtable: Genetic Medicine in Balkan countries: Challenges and solutions** | | |
| **18:50-20:00** | **S3.01.I** 10 min | Balkan Journal of Medical Genetics (BJMG): Gateway to publishing quality research | **D. Plaseska Karanfilska** | **North Macedonia** |
| | **S3.02.I** 10 min | The importance of European Reference Network in rare disease diagnosis and management: Slovenia's experience | **I. Babič Božović** | **Slovenia** |
| | **S3.03.I** 10 min | Competences of health professionals in genomic counselling | **N. Kregar Velikonja** | **Slovenia** |
| | **S3.04.P** | Maturity level of National Genomic Systems 10 min / Discussion 30 min | **B. Peterlin** | **Slovenia** |
| **20:00** | | **Welcome drink and get together** | | |

# DAY 2 - Morning Friday, 10th of October 2025

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| | **S4** | **Genetics of neurologic and neurodevelopmental disorders** *(O. Miljanović, M. Đurišić)* | | |
| 08:00-09:30 | **S4.01.P** 20 min | The contribution of genomic diagnostics to the elucidation of neurodevelopmental disorders | **O. Miljanović** | **Montenegro** |
| | **S4.02.O** 10 min | The FMR1 Premutation in Patients with Late-Onset Movement Disorders and Family Members of Fragile X Syndrome Patients: A Diagnostic Opportunity | **M. Pesić** | **Serbia** |
| | **S4.03.O** 10 min | Routine molecular genetic testing of GAA-FGF14-Related Ataxia and RFC1 CANVAS Spectrum Disorder | **H. Jaklič** | **Slovenia** |
| | **S4.04.O** 10 min | Monogenic Causes of Early-Onset Dementia: Evidence from Whole Exome Sequencing | **E. Shukarova Stefanovska** | **North Macedonia** |
| | **S4.05.O** 10 min | Expansion and contraction of CGG repeats in FMR1 gene within one family | **B. Pejović** | **Serbia** |
| | **S4.06.O** 10 min | ERBB4 exonic deletions in patients with intellectual disability and speech developmental delay | **M. Rasić** | **Serbia** |
| **09:10** | **S4.06.S** | **How to Combine Genomics, Deep Phenotyping, and AI to Diagnose Patients with Rare Diseases** | | **Genomize** |
| **09:30-10:30** | **S13** ARNOLD 1 | **Quality assurance in Balkan countries** ***(L. Lovrečić)*** (concurrent with P1 and P2 poster sessions)<br>**How to obtain regulatory compliance for in-house in vitro devices (IH-IVD's)?** *(H. Podgornik, UMCL, Slovenia)* 20 min<br>**The challenge of persistent poor performance in external quality control** *(W. Gutowska-Ding, EMQN, UK)* 20 min Virtual | | |
| **09:30-10:30** | **P1** ARNOLD 2 | **Concurrent Poster viewing session – Cancer** 5 min per poster | **P001-P010** ***(H. Butz, V. Šetrajčič Dragoš)*** | |
| | **P2** ROSA | **Concurrent Poster viewing session - Regional healthcare** 5 min per poster | **P011-P016** ***(A. Marjanovic, A. Maver)*** | |
| **09:30** | | **Coffee break** | | |
| | **S5** | **Advances in cancer diagnosis** *(V. Stegel, D. Primorac)* | | |
| **10:30-11:50** | **S5.01.P** 20 min | A New Era of AI and Whole Genome Sequencing (WGS) for Cancer Diagnosis and Treatment Strategies | **D. Primorac** | **Croatia** |
| | **S5.02.O** 10 min | Increased Identification of PARP Inhibitor-Eligible Patients in Epithelial Ovarian Cancer through HRD Testing at the Institute of Oncology Ljubljana | **V. Stegel** | **Slovenia** |
| | **S5.03.O** 10 min | Chaperone-Mediated Autophagy in Glioblastoma: A Multi-Omics Perspective | **Ö. Yildirim** | **Turkey** |
| | **S5.04.O** 10 min | Better diagnosis of liver cancer with the use of Xenium spatial transcriptomics | **U. Prosenc Zrmzljak** | **Slovenia** |
| | **S5.05.O** 10 min | Genes associated with higher mutational burden in tumors and improved response to checkpoint immunotherapy | **G. Kungulovski** | **North Macedonia** |
| | **S5.06.O** 10 min | Clinical verification of plasma cell immunoselection for FISH analysis in multiple myeloma | **H. Podgornik** | **Slovenia** |
| | **S5.07.O** 10 min | Computational Identification of Potential Therapeutic Agents Targeting the MUC16 Gene in Glioblastoma | **R. Kalkan** | **Northern Cyprus, Turkey** |
| | **S5.08.O** 10 min | Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome in a large exome dataset: Identification of novel variants in DNMT3B, ZBTB24, and CDCA7 genes | **F. Dereli Devrez** | **Turkey** |
| **12:00** | | **Lunch break** | | |

# DAY 2 Afternoon Friday 10th of October 2025

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| 13:00-14:30 | **S6** | **Concurrent session - Room Arnold 1 - Prenatal and preventive genomics** *(S. Hadjidekova, M. Xhetani)* | | |
| | **S6.01.P** 20 min | Recurrent Pregnancy Loss and Genetics: Exploring the Genetic Factors Behind Pregnancy Complications | **M. Xhetani** | **Albania** |
| | **S6.02.P** 20 min | Enhancing Prenatal Diagnosis of Fetal Congenital Anomalies Through Next-Generation Sequencing | **F. Burada** | **Romania** |
| | **S6.03.I** | Prenatal Genomic Testing - Current position and future directions 20 min | **L. Lovrečić** | **Slovenia** |
| | **S6.04.I** 20 min | Foetal radiation risk. Role of geneticists in biodosimetry service and genetic counselling | **J. Pajić** | **Serbia** |
| | **S6.05.O** 10 min | Familial Cases Show Higher Prevalence of Rare Predicted Pathogenic Variants in 56 Novel Genes Associated with Spontaneous Preterm Birth | **T. Mladenić** | **Croatia** |
| | **S6.06.O** 10 min | Preliminary serum metabolomics analysis highlighting tryptophan pathway alterations in spontaneous preterm birth | **S. Dević Pavlić** | **Croatia** |
| 13:00-14:30 | **S7** | **Concurrent session - Room Arnold 2 - Oncogenetics - the current state and challenges in the region** *(H. Podgornik, A. Patocs)* | | |
| | **S7.01.P** | Role of clinical and laboratory geneticists in precision cancer medicine 20 min | **A. Patocs** | **Hungary** |
| | **S7.02.I** 20 min | OncoOrigin: The Future of Precision Oncology through Integration of Machine Learning and Tumor Genomics for Identifying Primary Tumor Site | **P. Brlek** | **Croatia** |
| | **S7.03.I** 20 min | Challenging interpretation of germline variants in hereditary breast and ovarian cancer | **H. Butz** | **Hungary** |
| | **S7.04.O** 10 min | Spectrum of germline BRCA pathogenic variants in ovarian cancer patients from North Macedonia | **S. Kiprijanovska** | **North Macedonia** |
| | **S7.05.O** 10 min | Germline Screening for Hereditary Cancer Predisposition in the Bulgarian Population: Insights from 2024 | **N. Valcheva** | **Bulgaria** |
| | **S7.06.O** 10 min | Exploring attitudes towards nutritional advice amongst individuals affected by Lynch Syndrome in the UK | **I. Rennocks** | **United Kingdom** |
| | **S7.07.O** 10 min | Genetic Factors and Acute Kidney Injury: Influence on Cancer Treatment Strategies | **M. Imeraj** | **Albania** |
| 14:30-15:30 | **P03** Arnold 1<br>**P04** Arnold 2<br>**P05** Rosa | **Concurrent Poster viewing session – Case reports and Series** 5 min per poster | **P017-P026** *(A. Kovanda, K. Writzl)*<br>**P027-P036** *(J. Pajić, I. Babič Božović)*<br>**P037-P046** *(O. Antonova, F. Burada)* | |
| 15:30-16:30 | **S8** | **Screening and prevention programmes in the Balkan countries** *(T. Bahsi, F. Burada)* | | |
| | **S8.01.P** 20 min | PGT and hereditary breast cancer - can and should we break the chain? | **S. Hadjidekova** | **Bulgaria** |
| | **S8.02.P** 20 min | Combating Rare Diseases - Preconception and newborn screening programs in Turkey | **T. Bahsi** | **Turkey** |
| | **S8.03.O** 20 min | Facing Uncertainty in Prenatal Screening: Personal and Social Resources for Psychological Resilience | **J. Jakerlová** | **Czech republic** |
| 16:30-17:40 | **S9** | **Advanced treatments for genetic disorders and cancer** *(D. Biskup, P. Gasparini)* | | |
| | **S9.01.P** 30 min | On the way to personalized tumor vaccines | **D. Biskup** | **Germany** |
| | **S9.02.P** 30 min | Drug repurposing for inherited disease | **P. Gasparini** | **Italy** |
| | **S9.03.O** 10 min | The U-PGx project and PREPARE study in Slovenia: lessons learned on implementation of pharmacogenomics testing | **V. Dolžan** | **Slovenia** |
| 17:40 | **S9.04.S** | **PARPe Diem: Drawing Lessons from Experience to Shape Tomorrow** | **Astra Zeneca** | |
| 18:10-19:10 | **S10-R** | **Roundtable: International genomic forum: Strategy for Future Genomic Medicine** *(O. Miljanović, D. Primorac, P.Gasparini, B. Peterlin)* | | |
| 20:00 | | **Conference Dinner -** Grand Hotel Toplice | | |

# DAY 3 Saturday 11th of October 2025

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| 08:00-09:50 | **S11** | **Genetics of complex diseases and functional genomics** *(I. Babić Božović, V. Vidović)* | | |
| | **S11.01.I** 20 min | APOE, APP, and PSEN1 Mutation Screening in Alzheimer's Disease: A 15-Year Experience in Serbia | **A. Marjanović** | **Serbia** |
| | **S11.02.I** 20 min | Frequency of common variants predisposing to estrogen positive diseases in the Bulgarian population | **O. Antonova** | **Bolgaria** |
| | **S11.03.I** 20 min | Role of SIRT1 and SIRT3 Genetic Polymorphisms in the Risk of Acute Myocardial Infarction among Patients from the Republic of Srpska | **V. Vidović** | **Bosna and Herzegovina** |
| | **S11.04.O** 10 min | Genetic risk factors for anaphylaxis: Insights from somatic KIT p.D816V variant and Hereditary α-tryptasemia | **M. Rijavec** | **Slovenia** |
| | **S11.05.O** 10 min | Pediatric multiple sclerosis cases burdened with rare, predicted pathogenic variants in iron metabolism genes | **A. Turk** | **Slovenia** |
| | **S11.06.O** 10 min | Long-Read Sequencing in the Human Genome's Repetitive Landscape: Challenges and Clinical Opportunities | **T. Tesovnik** | **Slovenia** |
| | **S11.07.O** 10 min | Identifying patterns of differential methylation in multiple sclerosis by positional integration approach and their functional characterization | **N. Mele** | **Slovenia** |
| | **S11.08.O** 10 min | Connecting the Dots: Genetics, Diet, and Microbiome in Endometriosis (EM) | **A. Santin** | **Italy** |
| | **S11.09.O** 10 min | Severe Clinical Phenotype in Alport Syndrome Due to Two COL4A4 Exon Skipping Events | **A. Zupan** | **Slovenia** |
| **10:00** | **S11.10.S** | **NGS solutions for Human genetics** | | **Agilent** |
| **10:20-11:20** | **P06** ARNOLD 1 | **Concurrent Poster viewing session – complex and functional genomics** 5 min per poster | **P047-P056** *(L. Odak, A. Kovanda)* | |
| | **P07** ARNOLD 2 | **Concurrent Poster viewing session – diagnostics** 5 min per poster | **P058-P067** *(T. Pajič, M. Rijavec)* | |
| 11:20-13:00 | **S12** | **Expanding the genotype-phenotype landscape of genetic disorders in the Balkans** *(S. Bertok, A. Marjanović)* | | |
| | **S12.01.P** 20 min | Write according to the rules, read between the lines | **M. Djurišić** | **Serbia** |
| | **S12.02.O** 10 min | Genetics of porphyria. Efforts to associate specific mutations with clinical manifestations | **T. Todorov** | **Bulgaria** |
| | **S12.03.O** 10 min | Expanding the Genotypic Spectrum of Epidermolysis Bullosa: A Case Series | **E. H. Ceylan** | **Turkey** |
| | **S12.04.O** 10 min | Genotype-Phenotype Correlation in TTN Gene Variants | **M. B. Yilmaz** | **Turkey** |
| | **S12.05.O** 10 min | Challenges and Rewards in Diagnosing Diamond-Blackfan Anaemia: A Case Series from Cooperating Regional Tertiary Care Centres | **T. Pajič** | **Slovenia** |
| | **S12.06.O** 10 min | Body mass index is an overlooked confounding factor in clustering studies of 3D facial scans of children with autism spectrum disorder | **M. Schwarz** | **Czech republic** |
| | **S12.07.O** 10 min | Genetic Basis of Familial Hypercholesterolemia in Serbia: Detection of a Frequently Observed Mild-Phenotype LDLR Variant | **J. Komazec** | **Serbia** |
| | **S12.08.O** 10 min | Hereditary Myopathies in 45 Turkish Patients: Genetic Spectrum and Diagnostic Outcomes | **S. Demir** | **Turkey** |
| | **S12.09.O** 10 min | Biomolecular characterization of hereditary transthyretin amyloidosis in Bulgaria | **A. Todorova** | **Bulgaria** |
| **13:00-13:20** | | **Closing sessions** | | |
| | | Best poster and oral presentation award winners announcement<br>Closing remarks and thanks | | |
| **13:20** | | **Lunch break** | | |
| **14:00-16:00** | **WS.02** | **Workshop: Translational research on rare diseases** | **M. Stojiljkovic** | |