DAY 1 Thursday 9th of October 2025

8:00-13:00	**WS.01**	**Workshop: Interpretation of NGS data**	**A. Maver**	
13:00		**Lunch break**		
14:00-15:40	**P1**	**Welcome and plenary lectures *(B. Peterlin, D. Plaseska-Karanfilska)***		
	WA 10 min	Welcome address	**A. Maver**	**Slovenia**
	W.01.P 40 min	Using genomic medicine to make drug prescription safer and more effective	**W. Newman**	**United Kingdom**
	W.02.P 40 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	AI-Assisted Genome Diagnostics with Long Reads	**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.I 10 min	Establishment of the Slovenian genome database as a resource of population variability in the region	**A. Maver**	**Slovenia**
	S3.05.P	Maturity level of National Genomic Systems 10 min / Discussion 20 min	**B. Peterlin**	**Slovenia**
20:00		**Welcome drink and get together**		

DAY 2 - Morning Friday, 10th of October 2025

	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) **How to obtain regulatory compliance for in-house in vitro devices (IH-IVD's)?** *(H. Podgornik, UMCL, Slovenia)* 20 min **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, P067** ***(H. Butz, V. Šetrajčič Dragoš)***	
	P2 ROSA	**Concurrent Poster viewing session - Regional healthcare** 5 min per poster	**P011-P016, P068, P070-P072** ***(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

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 10 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 **P04** Arnold 2 **P05** Rosa	**Concurrent Poster viewing session – Case reports and Series** 5 min per poster	**P017-P026** *(A. Kovanda, K. Writzl)* **P027-P036** *(J. Pajić, I. Babič Božović)* **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

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	**P057-P067, P069** *(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 Closing remarks and thanks		
13:20		**Lunch break**		
14:00-16:00	**WS.02**	**Workshop: Translational research on rare diseases**	**M. Stojiljkovic**	