

DAY 2 - Morning

Friday, 10th of October 2025

09:30-10:30	P1	ARNOLD	Concurrent Poster viewing session – Cancer	P001-P010, P067 (H. Butz, V. Šetrajčič Dragoš)
	P001		Uncovering Immunogenic Gene Fusions in Glioblastoma for Personalized Immunotherapy	
	P002		Germline Mutations in Myeloid Malignancies in Slovenia	
	P003		Incidence of Multiple Malignancies in Patients with Triple-Negative Breast Cancer	
	P004		Overview of Referral Diagnoses for Tumor Genotyping by Next-Generation Sequencing at the Institute of Oncology Ljubljana	
	P005		Molecular and Cytogenetic Analyses of AML in Pediatric Patients in The Last Five Years- One Center Study	
	P006		<i>FGFR3</i> Expression and Tumour Size in Urothelial Carcinoma	
	P007		Assessment of Complete Cytogenetic Response in Chronic Myeloid Leukemia Patients Treated with Tyrosine Kinase Inhibitors in Southern and Eastern Serbia	
	P008		Association Between Genetic <i>HIF1A</i> Variants and the Efficacy of Cisplatin in Malignant Mesothelioma	
	P009		Xpert NPM1 Mutation Diagnostic Test Fort Monitoring of NPM1 mRNA Transcripts in Patients with Acute Myeloid Leukemia (AML)	
	P010		ALK Positive Mutation in a Female Diagnosed with Pulmonary Adenocarcinoma	
	P067		Gene Variants in Urothelial Bladder Cancer Analyzed by Sequencing	
	P2	ROSA	Concurrent Poster viewing session - Regional healthcare	P011-P016, P068, P070-P072 (A. Marjanovic, A. Maver)
	P011		Collaboration on Hereditary Angioedema with C1-Inhibitor Deficiency in South-Eastern Europe: Building a Regional Genetic Variant Landscape	
	P013		Assessing Complex Paediatric Cases via Chromosomal Microarray and Multidisciplinary Team Collaboration	
	P015		Genetic Background of Hereditary Erythrocytosis in Slovenian Population	
	P016		Trends in Breast Cancer Incidence and Histopathologic Patterns in a Regional Hospital Center: “Implications for <i>BRCA1/2</i> Genetic Screening”	
	P068		Variants in the <i>TP53</i> Gene in Patients with Urothelial Bladder Cancer Detected with Next-Generation Sequencing	
	P070		Polygenic Risk Score and Genetic Markers in Alcohol-Related Cirrhosis	
	P071		Pharmacogenomic Landscape of the Serbian Population	
	P072		Humanitarian Genetics: Reflections from a Pediatric Geneticist in Kabul, Afghanistan	

DAY 2 Afternoon Friday 10th of October 2025

14:30-15:30	P03 ARNOLD 1		Concurrent Poster viewing session – Case reports and Series	P017-P026 (A. Kovanda, K. Writzl)
	P017	Genetic Analysis of a Poirier–Bienvenu Neurodevelopmental Syndrome Caused by a de novo variant in <i>CSNK2B</i>		
	P018	De-novo, Novel Likely Pathogenic Variant in <i>SMC1A</i> Gene in a Patient with Intellectual Disability and Seizures History		
	P019	Case report: a Hemizygous DMD:c.958C>T Variant with Splicing Rescue and Attenuated Clinical Presentation		
	P020	Phenotypic Spectrum in Patients with Copy Number Variations		
	P021	Duplication of the <i>SOX3</i> Gene in an <i>SRY</i> -negative Boy with 46,XX Karyotype: A Case Report and a Literature Review		
	P022	Prevalence of <i>HLA-DQ2</i> and <i>HLA-DQ8/DR4</i> Haplotypes and Their Association with Clinical Manifestations in Patients from Bosnia and Herzegovina		
	P023	Mitochondrial Myopathy Caused by MT-ND5 Variant: Integrating WES and Mitochondrial DNA analysis		
	P024	The Role of <i>MTNR1A</i> and <i>MTNR1B</i> Variations in Chronic Insomnia		
	P025	Morbus Gaucher a Diagnostic Challenge		
	P026	Spectrum of NF1 Variants in a Patient Cohort from North-Eastern Slovenia		
	P04 ARNOLD 2		Concurrent Poster viewing session – Case reports and Series	P027-P035 (J. Pajić, I. Babić Božović)
	P027	A Multigenerational Nonsyndromic Hypopituitarism Family Associated with a Novel <i>GLI2</i> Variant		
	P028	Frequency of <i>ABCB1</i> C3435T and C1236T Polymorphisms in the Kosovo Population		
	P029	Expanding the Clinical and Molecular Landscape of Pseudoxanthoma Elasticum with a Novel <i>ABCC6</i> Variant		
	P030	Differential Diagnostic Obstacles for Harel-Yoon Syndrome		
	P031	A Case Report of a Patient with Neurodevelopmental Disorder with Hypotonia, Stereotypic Hand Movements, and Impaired Language – A Novel Variant In <i>MEF2C</i> Gene		
	P032	X-Linked Legacy: Multigenerational Expression of Aarskog-Scott Syndrome in a Single Family		
	P033	Rare Double Strike in Hypothalamic Signaling: A Case of Severe Pediatric Obesity Explained by <i>MC3R</i> and <i>NMUR2</i> Variants		
	P034	<i>MTNR1B</i> Polymorphisms and Circadian Phenotypes in Hashimoto's Disease		
	P035	The Fifth Known Case of <i>SASH3</i> -Associated Immunodeficiency: Novel Variant and Severe Inflammatory Phenotype		
	P05 ROSA		Concurrent Poster viewing session – Case reports and Series	P038-P046 (O. Antonova, F. Burada)
	P038	A Case Series on Suspected Bainbridge-Ropers Syndrome with a Two Novel Variation in <i>ASXL3</i> Gene		
	P040	A Case of Pallister-Killian Syndrome in a Newborn		
	P041	De Novo Ring Chromosome 20 in a Male Infant with Suspected Epilepsy		
	P042	Two Cases of Rare Pycnodysostosis Syndrome with <i>CTSK</i> Missense Variants		
	P043	Schuurs-Hoeijmakers Syndrome: Expanding the Phenotype of a Rare Disorder with a Recurrent <i>PACS1</i> Variant		
	P044	Clinical Exome Sequencing Identifies Pathogenic <i>SQSTM1</i> Variant in a Patient with Chorea and Gaze Palsy		
	P045	Analysis of the Association Between Collagen Gene Polymorphisms and Clinical Manifestations of Systemic Lupus Erythematosus		
	P046	Unravelling the Cause of Recurrent Venous Thrombosis in a Dabigatran-Treated Patient		

DAY 3

Saturday 11th of October 2025

10:20-11:20	P06 ARNOLD 1	Concurrent Poster viewing session – complex and functional genomics	P047-P056 (L. Odak, A. Kovanda)
	P047	Pulmonary <i>In Vitro</i> Model System Enables Exploration of Innovative Treatment Strategies for Rare Respiratory Diseases	
	P048	Establishing <i>In Vitro</i> Models for Glycogen Storage Disease Type Ib: A Platform for Therapeutic Investigations	
	P049	Functional Analysis of Novel <i>EGLN1</i> and <i>EPAS1</i> Variants in Hereditary Erythrocytosis	
	P050	Establishment of an <i>In Vitro</i> Insulin Resistance Model in HEPG2 Cells Through Glucose and Insulin Co-Treatment	
	P051	Methylation Levels and Genetic Variants of <i>MTHFR</i> Gene Are Not Risk Factors for Congenital Heart Defect in Down Syndrome	
	P052	Molecular diagnosis of sexually transmitted infections related to infertility: the efficiency of multiplex pcr panels	
	P053	Effects of <i>HLA-DRA</i> , <i>HLA-DQA1</i> , and <i>IL-6</i> Gene Variations to Multiple Sclerosis	
	P054	The Importance of HPV Genotyping in Albania: A First-of-its-kind Study at Geniuslab	
	P055	Detection of <i>Helicobacter pylori</i> in Stomach Cancer Patients Using dPCR	
	P056	miRNA and circRNA as Potencial Biomarkers for ALS	
	P07 ARNOLD 2	Concurrent Poster viewing session – diagnostics	P058-P066, P069 (T. Pajič, M. Rijavec)
	P057	Comprehensive Tryptase Genotyping and β III Frame-Shifted Allele Detection Employing Multiplex ddPCR	
	P058	Overcoming Diagnostic Challenges in <i>PKD1</i> Gene Analysis	
	P059	Diagnosis of Neurofibromatosis Type 1 Using CES, CMA and MLPA Methods	
	P060	Application of Quantitative PCR for <i>SMN1</i> Carrier Detection in Prenatal and Family Planning Settings	
	P061	Comparison of qPCR-HRM and Fragment Analysis Methods to Determine the Methylation Status of the <i>MLH1</i> Gene Promoter in Tumor Samples	
	P062	Haplotype Analysis of <i>MMP-9</i> Gene Polymorphisms and their Association with Hemorrhagic Risk Following Thrombolysis in Acute Ischemic Stroke Patients	
	P063	Implementation of <i>HLA-DQ2/DQ8</i> Genetic Testing for Coeliac Disease in a Clinical Laboratory	
	P064	Validation of the Automatic DNA Isolation from Various Human Tissues and of Bacterial DNA from Oral Mucosa Swabs by Method Based on Magnetic Beads	
	P065	Molecular Multitesting – Overview of Key Factors for High-Quality and Rapid Diagnostics	
	P066	Validation of Bioinformatic Tools for Pharmacogenomic Analysis on NGS Data	
	P069	A Retrospective Analysis of the Distribution and Positivity Rates of Molecular PCR Tests Performed in a Tertiary Care Center Throughout 2024	