



DRAFT OF THE AGENDA

DAY 1: Sunday, October 22, 2023

(All Local Time / GMT + 4)

09.30-10.00	WELCOME AND OPENING REMARKS	
10.00-12.30	SESSION A: DIAGNOSIS OF RARE AND UNDIAGNOSED DISEASES - COMMON CHALLENGES	
	The post-exome clinic: improving the impact of exome sequencing for developmental disorders	Sofia Douzgou Houge , Haukeland University Hospital, Norway
	The Diagnostic Implications of Pitfalls in Causal Variant Identification Based on 4,577 Molecularly Characterized Families	Lama Al-Abdi , King Faisal Specialist Hospital & Research Centre, Saudi Arabia
	How can we reach a diagnosis for more individuals with a rare condition, the Solve-RD perspective	Steven Laurie , The Barcelona Institute of Science and Technology, Spain
	DNA first strategies to reduce the diagnostic odyssey in rare disease patients	Wendy van Zelst-Stams , Radboudumc, The Netherlands
	Interpretation and classification of genetic variants	Gunnar Douzgos Houge , Haukeland University Hospital, Bergen, Norway
	TBC	Michael Brudno , University of Toronto, Canada
	TBC	Yasemin Alanay , Acibadem University, Turkey
12.30-13.30	LUNCH BREAK	
13.30-16.00	SESSION B: NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES	
	Therapy in GBA1 related Parkinson: the never-ending story	Ari Zimran , Gaucher Clinic at Shaare Zedek Medical Center, Israel
	Inherited (liver) metabolic diseases: Is there a therapeutic unmet need? Examples of preclinical data for gene therapy in Maple Syrup Urine Disease (MSUD)	Manuel Schiff , Hôpital Necker-Enfants Malades, France
	Dissecting clinical, genetic, and mechanistic heterogeneity of non-muscle actinopathies	Siddharth Banka , University of Manchester, UK
	Importance of whole genome sequencing for the early identification of rare genetic disorders	Arndt Rolfs , University of Rostock, Germany
	Novel mitochondrial augmentation therapy - from bench to bedside	Yair Anikster , Sheba-Tel HaShomer Hospital, Israel
	TBC	Stephen Meyn , University of Wisconsin-Medison, USA

	Oral EGFR inhibitor for severe Palmo-plantar keratoderma	Alain Hovnanian , Necker Enfants Malades Hospital, France
16.00-16.30	COFFEE BREAK	
16.30-18.45	SESSION C: RARE AND UNDIAGNOSED DISEASES WORLDWIDE	
	Undiagnosed rare diseases in the Czech Republic: activities on the National Coordination Centre for Rare Diseases and multidisciplinary nationwide collaboration.	Milan Macek , Charles University Hospital, Czech Republic
	Implementing GS-based diagnostics of rare diseases at the Karolinska	Anna Lindstrand , Karolinska University Hospital and Karolinska Institute, Sweden
	Rare Diseases in Georgia: achievements and challenges	Tinatn Tkemaladze , Tbilisi State Medical University; Oleg Kvlividze , Georgian Foundation for Genetic and Rare Diseases
	Diagnosing and managing rare diseases in limited resource settings: Pakistani experience	Salman Kirmani , Aga Khan University, Karachi, Pakistan
	TBC	Natalia Samonenko , National Children's Specialized Hospital "OKHMATDIT", Ukraine
	The landscape within the Indian Undiagnosed Diseases Program: insights & challenges	Ratna Puri , Institute of Medical Genetics & Genomics, India
	Rare and Undiagnosed Diseases: Turkish Perspectives	Uğur Özbek , Acibadem University, Turkey
18:45-19:30	POSTER SESSION	
20.30-23.00	WELCOME RECEPTION	