
## DRAFT OF THE AGENDA

**DAY 1: Sunday, October 22, 2023**

(All Local Time / GMT + 4)

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| **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 |
| | Capturing and Sharing Data on Rare Diseases Patients in the canadian Care4Rare Project | **Michael Brudno**, University of Toronto, Canada |
| | Rare Diseases in Diverse Populations: Clinical WGS Experience from Istanbul, an EMA Hub | **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 |
| | Beyond Exomes: New Opportunities for Undiagnosed Disease Programs | **Stephen Meyn,** University of Wisconsin-Medison, USA |

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| | 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 Insitute, Sweden |
| | Rare Diseases in Georgia: achievements and challenges | **Tinatin 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 |
| | Diagnosis and Treatment of Orhan Drugs in Ukraine | **Natalia Samonenko,** Center of Orphan Disease and Gene Therapy;l NSCH “OKHMATDYT”, 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** | |