DRAFT OF THE AGENDA

DAY 2: Monday, October 23, 2023

(All Local Time / GMT + 4)

09:00-10:00	**SESSION B/2: NOVEL DIAGNOSTICS AND THERAPEUTICS FOR RARE DISEASES** **Chair: David Adams**	
	(15 min total) Applications of Long-Read Sequencing for Rare Diseases	**Vorasuk Shotelersuk,** Chulalongkorn University, Thailand
	(10 min total) Solving undiagnosed disease beyond the exome: Functional enhancers with medical relevance identified by computational analysis and ChIP-STARR-seq in neural cell models enable prioritizing non-coding variants from patient whole genome sequencing studies	**Tahsin Stefan Barakat,** Department of Clinical Genetics, Erasmus MC University Medical Center, Rotterdam, The Netherlands
	(10 min total) Functional studies using *Drosophila* support clinical diagnosis and phenotypic expansion: *BMPR2* in neurodevelopmental disorders	**Shinya Yamamoto**, Baylor College of Medicine, Houston, Texas, USA
	(10 min total) Epigenetics in rare diseases: the role in Cystic Fibrosis	**Sandro Surmava,** Tbilisi State Medical University, Georgia
	(10 min total) Phenylalanine treatment in 8-month old girl with mitochondrial FARS2 deficiency	**Rachel Rock**, Metabolic Diseases Clinic, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel
10:00-11:00	**NEW DISEASES; SOLVED AND UNSOLVED CASES (6 MINUTES + 2 MINUTES FOR QUESTIONS)** **Chair: May Malicdan**	
	New Disease Discovered via UDNI Collaboration: Benign Adult Familial Myoclonic Epilepsy Type 8 (BAFME8)	**Vorasuk Shotelersuk,** Chulalongkorn University, Thailand
	PERCC1-associated congenital enteropathy: Delineating the natural history of a new disorder of enteroendocrine cell function	**Ben Pode-Shakked,** Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel
	Unraveling the genetic basis of rare hereditary neurological diseases in Mali.	**Guida Landoure,** Faculté de Médecine et d'Odontostomatologie, USTTB, Bamako, Mali
	The case of Rubinstein-Taybi syndrome - hunting for hidden mutation	**Anastasia Sukhiashvili,** Dept. Molecular & Medical Genetics, Tbilisi State Medical Univ, Tbilisi, Georgia
	Failure to thrive, ichthyosis, deafness, and endocrinopathies in an infant with a novel biallelic *AP1B1* mutation causing abnormal intracellular ATP7A trafficking	**Yehoshua (Joshua) Manor,** Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

	The undiagnosed disease masquerade: A family case of severe asthma successfully treated with mepolizumab	**Shmuel Prints,** Clalit Health Services, Beersheba, Israel
	Undiagnosed syndromic hyperinsulinemic hypoglycemia in an infant	**Roberto Giugliani,** Federal University, Rio Grande do Sul (UFRGS), Brazil
11:00-12:20	**SESSION E: INTERNATIONAL INITIATIVES ON RARE AND UNDIAGNOSED DISEASES** **Chair: Vorasuk Shotelersuk**	
	(12 min total) The International Rare Disease Research Consortium (IRDiRC): Making rare disease research efforts more efficient and collaborative, around the world	**David Pearce,** Sanford School of Medicine, University of South Dakota
	(12 min total) Consolidating the Rare Diseases Research Ecosystem: a Multistakeholder and Multidirectional approach	**Yanis Mimouni,** EJP RD, France
	(12 min total) CZI and Patient-Driven Research	**Tania Simoncelli,** Chan-Zuckerberg Initiative
	(12 min total) Challenges Unmasked: Navigating the complexities of adult rare and undiagnosed disease care	**Aung Min Saw,** Syndrome Without a Name (SWAN), Cardiff, Wales **Matthew Spencer,** Pediatrics, Cardiff
	(10 min total) Clinical utility of genetic diagnosis in adults with undiagnosed disease: An experience from Korean Adult Undiagnosed Disease Program	**Jaeso Cho,** Dept. Genomic Medicine, Seoul National University Hospital, Seoul, Korea
	(10 min total) A systematic approach for thousand severe unsolved pediatric conditions: results from the Telethon Undiagnosed Disease Program	**Vincenzo Nigro,** Telethon Institute of Genetics and Medicine, Pozzuoli, Italy
12:20-13:20	**LUNCH BREAK**	
13:20-16:30	**UDNI COMMITTEE & WORKING GROUP REPORTS**	**Chair: William Gahl**
	(10 min) Review of UDNI and Current Leadership	William Gahl
	(10 min) Membership; Trainee Category/Update	Eric Klee
	(10 min) Genetic Counseling Working Group	Janine Lewis, Stephanie Broley
	(10 min) Communications/Website	Marco Salvatore, Gianluca Ferrari, Domenica Taruscio
	(15 min) Developing Nations Working Group	Domenica Taruscio, Manuel Posada, Samuel Wiafe (Olaf Bodamer)
	(10 min) Education Working Group/UEMS/Medical Competence and Medical Specialty	Bela Melegh, Domenica Taruscio, Bruce Korf
	(15 min) Functional Research Working Group	Shinya Yamamoto
	(10 min) Patient Engagement Plus	Gulcin Gumus, Helene Cederroth Debbie Drell
	(10 min) Diagnostics Working Group: Linking to Hackathons and Emerging Technology	Ann Nordgren (Emma Palmer, Lorenzo Botto)
	(30 min) Hackathon Update and Followup; Video Summary of Stockholm Hackathon Impressions from Champions Future Hackathon (Nijmegen)	Mikk and Helene Cederroth Ann Nordgren Salman Kirmani, Samuel Wiafe, Aime Lumaka Wendy van Zelst-Stams
	(30 min) Data Sharing Committee: New Analytical Tools, Long Reads, etc. Calypso and iobio tools in the UDN	David Adams Alistair Ward, Univ of Utah
	(30 min) Champions Initiative Introduction (5 min) Ghana, Pakistan, DR Congo, Mali (5 min each) Intercontinental Exchange Discussion (15 min)	William Gahl, Helene and Mikk Cederroth, UDNI DNWG Samuel Wiafe, Salman Kirmani, Aime Lumaka, Guida Landoure
16:30-17:00	**COFFEE BREAK**	

17:00-19.00	**CURRENT ISSUES OF RARE AND UNDIAGNOSED DISEASES IN GEORGIA (IN GEORGIAN)**	
	Success and challenges in the treatment of SMA: Georgian experience	**Nana Tatishvili**; D. Tvildiani Medical University; Dept. of Neuroscience, M. Iashvili Children's Central Hospital
	Hope on the Horizon: Alagille Syndrome Treatment Advancements	**Mariam Ghughunishvili**; Givi Zhvania Pediatric Academic clinic
	Nonepileptic Paroxisms in Children	**Sophia Bakhtadze**, Dept. of Child Neurology, Tbilisi State Medical University; Givi Zhvania Pediatric Academic clinic
	Rare diseases outpatient monitoring program in Georgia - achivements, challenges and expectations	**Lali Margvelashvili**, Center of Medical Genetics and Laboratory Diagnostics
	Modern treatment of Duchenne muscular dystrophy	**Teona Shatirishvili**; D. Tvildiani Medical University; Dept. of Neuroscience, M. Iashvili Children's Central Hospital
	Cystic Fibrosis contemporary treatment issues: Georgian perspectives	**Dodo Agladze**, Petre Shotadze Tbilisi Medical Academy; Center of Medical Genetics and Laboratory Diagnostics
	Rare cases of sexual development disorders	**Jenara Kristesashvilil**; Faculty of Medicine, I. Javakhishvili State University; Reproductive Medical Center Universe; Tbilisi, Georgia
	Galactosemia: treatment recommendations	**Maia Kherkheulidze**; № 1 Dept. of Pediatrics, Tbilisi State Medical University; Givi Zhvania Pediatric Academic clinic
	Treatment aspects of Congenital Adrenal Hyperplasia	**Ekaterine Kvatatskhelia**; Ilia State University; Davit Metreveli Medical Center
17:00-18:00	**UDNI BUSINESS MEETING**	**Chair: W. Gahl**
	1. Board to approve new members 2. Votes: 2024 Meeting; RARE affiliation 3. 2024: Seoul, Sept. 5-7 (Jong-Hee Chae) 4. 2025: Call for proposals 5. BOD Turnover Discussion; Charter 6. RARE Journal affiliation 7. Issues Arising 8. Previous Topics for Discussion	
20:00-23.30	**GALA DINNER (kindly sponsored by the Wilhelm Foundation)**	