



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		
	(15 min) Applications of Long-Read Sequencing for Rare Diseases	Vorasuk Shotelersuk , Chulalongkorn University, Thailand
	(10 min) New Disease Discovered via UDNI Collaboration: Benign Adult Familial Myoclonic Epilepsy Type 8 (BAFME8)	Vorasuk Shotelersuk , Chulalongkorn University, Thailand
	TBC	TBC
	TBC	TBC
10:00-11:00 ABSTRACTS SESSION: SOLVED AND UNSOLVED CASES (6 MINUTES + 2 MINUTES FOR QUESTIONS)		
11:00-12:20 SESSION E: INTERNATIONAL INITIATIVES ON RARE AND UNDIAGNOSED DISEASES		
(~10-15 min each)	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
	CZI and Patient-Driven Research	Tania Simoncelli , Chan-Zuckerberg Initiative, USA
	EJP RD/RDP	Speaker TBC
	NIH Undiagnosed Diseases Program	Speaker TBC
	Brazil Rare Diseases	Roberto Giugliani , Federal University, Rio Grande do Sul (UFRGS), Brazil
	Cardiff, Wales SWAN Service	Aung Min Saw , SWAN clinic/NRU, Cardiff and Vale University, UK
12:20-13:20 LUNCH BREAK		
13:20-16:30 UDNI COMMITTEE & WORKING GROUP REPORTS		
	(10 min) Review of Current Leadership, Voting Results, Issues to Address	Chair: William Gahl , NIH, USA
	(10 min) Membership; Trainee Category/Update	William Gahl
	(10 min) Genetic Counseling Working Group	Eric Klee
	(10 min) Communications/Website	Janine Lewis, Stephanie Broley
	(10 min) Developing Nations Working Group	Marco Salvatore, Gianluca Ferrari, Domenica Taruscio
	(15 min) Education Working Group/UEMS/Medical Competence and Medical Specialty	Domenica Taruscio, Manuel Posada, Samuel Wiafe (Olaf Bodamer)
	(10 min) Functional Research Working Group	Bela Melegh, Domenica Taruscio
	(15 min) Patient Engagement Plus	Shinya Yamamoto
	(10 min) Diagnostics Working Group: Linking to	Gulcin Gumus, Helene Cederroth Debbie Drell
		Ann Nordgren (Emma Palmer,

	Hackathons and Emerging Technology	Lorenzo Botto)
	(30 min) Hackathon Update and Followup; Video Summary of Stockholm Hackathon Impressions from Champions	Mikk and Helene Cederroth Ann Nordgren Salman Kirmani, Samuel Wiafe, Aime Lumaka
	Future Hackathon (Nijmegen)	Wendy van Zelst-Stams
	(30 min) Data Sharing Committee: Update on New Analytical Tools, Long Reads, etc. IOBIO Team/Analysis	David Adams Alistair Ward, 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)	
	Presentation topics TBC	TBC (5-6 Speakers)
17:00-18:00	UDNI BUSINESS MEETING	
	<ol style="list-style-type: none"> 1. Confirm New Committee and Board Members; 2. Next Meeting: Seoul (Jong-Hee Chae); 3. Journal: RARE (Wendy van Zelst-Stams) 4. Sustainability/Future of UDNI; 5. Acknowledge UDNI in Papers; 6. Votes online after meeting; 7. Concluding remarks 	Chair: William Gahl, NIH, USA
20:00-23.30	GALA DINNER	