



11th International Conference on Rare and Undiagnosed Diseases

DAY 2: UDNI Committee and Working Group Reports

Tuesday, November 08, 2022

(all times are CET)

1200-1210	Welcome	Kaan Boztug, Georg Stary, Helene Cederroth, William Gahl
(1210-1450)	UDNI Committee & Working Group Reports	Chair: William Gahl
1210-1220	Review of Current Leadership, Voting Results, Issues to Address	William Gahl
1220-1230	Membership; Trainee Category/Update	Eric Klee
1230-1240	Genetic Counseling Working Group	Janine Lewis
1240-1255	Communications/Website	Domenica Taruscio, Marco Salvatore
1255-1310	Developing Nations Working Group	Domenica Taruscio, Manuel Posada, Samuel Wiafe, Olaf Bodamer
1310-1320	Education Working Group/UEMS/Medical Competence and Medical Specialty	Bela Melegh, Domenica Taruscio
1320-1330	Functional Research Working Group	Shinya Yamamoto
1330-1340	Patient Engagement Plus	Gulcin Gumus, Helene Cederroth, Debbie Drell
1340-1400	Diagnostics Working Group	Lorenzo Botto
1400-1450	Data Sharing Committee: Website Forum tour plus policy updates	David Adams, Alexa McCray
1450-1530	Break	
1530-1620	Champions Initiative Introduction (5 min) Ghana (10 min) Pakistan (10 min) DR Congo (10 min) Discussion (15 min)	William Gahl, Helene and Mikk Cederroth, UDNI DNWG Samuel Wiafe, Salman Kirmani, Aime Lumaka
(1620-1800)	International Updates and Abstracts	Chair: Paul Lasko
1620-1640	IRDIRC	David Pearce
1640-1655	Rare and Undiagnosed Disease Research in Saudi Arabia	Fowzan Alkuraya
1655-1710	New Phenotips Pedigree Analysis	Orion Buske
1710-1750	ABSTRACTS: New Diseases, Solved and Unsolved Cases (6 minutes + 2 minutes for questions)	
	1. Development of a web-based tool for the collection of structured phenotype and genotype data from patients submitted to the Undiagnosed Diseases Network International (UDNI)	Angelica Maria Delgado-Vega
	2. A novel immunodeficiency presenting with impaired CD8+ T-cell functions via perturbed	Sevgi Köstel Bal

	glycolysis amenable to metabolic reprogramming	
	3. Calcifying myopathy	Ana Mondadori dos Santos
	4. Progressive Scleroderma/scleroderma-like lesions, a new genetic entity?	Enrica Marchionni
1750-1840	UDNI Business Meeting	Chair: W. Gahl
	<ol style="list-style-type: none"> 1. Confirm New Committee and Board Members 2. Next Meeting Location (In person/virtual): 2023 Tbilisi 2024 Seoul (Jong Hee Chae) / Istanbul (Ugur Ozbek) 3. Sustainability/Future of UDNI 4. Acknowledge UDNI in Papers 5. Votes online after meeting 	
	Concluding remarks	Kaan Boztug, Georg Stary, Helene, Cederroth, William Gahl
1930	Dinner	