10th International Conference on Rare and Undiagnosed Diseases

Undiagnosed Diseases Network International
January 31-February 1, 2022
Torino, Italy

DAY 2 Tuesday, February 1, 2022 *(all times in USA Central Time)*

0700-0715	**Welcome and an Introduction to Torino**	Dario Roccatello, Domenica Taruscio, Helene Cederroth, William Gahl
(0715-0940)	**UDNI Committee & Working Group Reports**	Chair: William Gahl
0715-0730	Review of Current Leadership, Last Meeting Results, Issues to Address	William Gahl
0730-0740	Membership; Trainee Category	Eric Klee
0740-0750	Genetic Counseling Working Group	Janine Lewis; Stephanie Broley
0750-0810	Communications/Website	Domenica Taruscio Marco Salvatore
0810-0825	Developing Nations Working Group	Domenica Taruscio Manuel Posada Samuel Wiafe Olaf Bodamer
0825-0835	Education Working Group/UEMS/Medical Competence and Medical Specialty	Bela Melegh Domenica Taruscio
0835-0845	Functional Research Working Group	Shinya Yamamoto
0845-0900	Patient Engagement Plus	Gulcin Gumus, Helene Cederroth (Debbie Drell), Vanessa Boulanger
0900-0910	Data Sharing Committee	David Adams Alexa McCray
0910-0940	Diagnostics Working Group	Elizabeth Palmer
0940-1015	**Break**	
1015-1150	New Diseases, Solved and Unsolved Cases (6 minutes + 2 minutes for questions)	Chair: Roccatello Dario/Lisa Schimmenti
	1. EDNRB mosaicism in a patient with nonsyndromic deafness/ GJB2 and PTPRQ mutations in a family with nonsyndromic deafness	Darina Kachakova (Medical Faculty, Medical University – Sofia, Bulgari)
	2. Cardiac Arrhythmia Syndrome with ST-Segment Depression	Marchionni Enrica (Tor Vergata Hospital, University of Rome Tor Vergata, Rome, Italy)
	3. Adult-onset rapidly worsening progressive myoclonic epilepsy caused by a novel variant in DHDDS.	Jangsup Moon (Seoul National University Hospital)

	4. CNV positional effect suggests an enhancer-mediated SHH dysregulation in a boy with multiple congenital anomalies	Daniele Vitale (University of Naples Federico II, Napoli, Italy)
1150-1230	**Lunch**	
(1230-1410)	International Networks and Connections for Undiagnosed and Rare Diseases	Chair: Paul Lasko
1230-1240	IRDiRC: Rare Diseases Treatment Access Working Group	William Gahl Durhane Wong-Rieger Steve Groft
1240-1250	Global Commission to End the Diagnostic Odyssey for Children (Takeda, Eurordis, Microsoft Health)	Roberto Giugliani
1250-1300	ICORD: Central and South America and Caribbean Nations (ERCAL Initiative)	Steve Groft
1300-1310	UDN: Current and Future	Argenia Doss, NIH
1310-1325	Undiagnosed Diseases Network Foundation (UDNF)	Cristina Might
1325-1335	Solve RD – EU Horizon Program	Olaf Riess
1335-1410	New Diseases, Solved and Unsolved Cases **(6 minutes + 2 minutes for questions)**	Chair: Lorenzo Botto/ Bela Melegh
	1. Loss of seryl tRNA synthetase (SARS1) causes complex spastic paraplegia and cellular senescence	Edgard Verdura (Nostos Genomics, Barcelona)
	2. Biallelic copy number variations in both upstream & downstream enhancers of SHOX gene causes mesomelia and clubfoot without short stature	Bengisu Guner Yilmaz (Acibadem Mehmet Ali Aydinlar University, Istanbul, Turkey)
	3. Monoallelic and Biallelic Germline Mutations Affecting the Transcription Factor Helios Cause Pleiotropic Defects of Immunity	Daniel Mayr (St. Anna Children's Cancer Research Institute CCRI, Austria)
	4. Undiagnosed cutis laxa syndrome: two pairs of siblings with shared phenotype	Woojoong Kim (Seoul National University Children's Hospital)
1410-1500	**UDNI Business Meeting**	**Chair: William Gahl**
	1. Confirm New Committee and Board Members 2. Next Meeting Location (In person/virtual) 3. Sustainability/Future of UDNI 4. Acknowledge UDNI in Papers **Votes online after meeting**	
1500	**Concluding remarks**	Dario Roccatello & Savino Sciascia