10th International Conference on Rare and Undiagnosed Diseases

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

DAY 1 Monday, January 31, 2022 *(all times in USA Central Time)*

7:00-7:15	**Welcome and Introduction to Torino**	Dario Roccatello, Domenica Taruscio, William Gahl, Helene Cederroth
New Frontiers in rare diseases: from Diagnostics to classification challenges **Chairpersons:** Dario Roccatello & William Gahl		
7:15-7:45	Immunogenetics of the microcosm of the glomerular diseases characterized by focal segmental lesions of glomeruli	Fernando Fervenza (Mayo Clinic, Rochester USA)
7:45-8:15	The uncommon antigens of membranous nephropathy	Sanjeev Sethi (Mayo Clinic, Rochester USA)
8:15-8:35	The Piedmont Experience with Undiagnosed Diseases	Silvia Deaglio (University of Torino)
8:35-8:55	The Impact of undiagnosed renal diseases in the clinical practice	Roberta Fenoglio (University of Torino)
8:55-9:15	From diagnosis to characterization and clinical management: the DECODE project	Erica Daina (Mario Negri Institute)
9:15-9:25	**Espresso Break**	
9:25-10:00	Selected Abstracts (6 minutes + 2 minutes for questions)	
	1. Consistent count region-copy number variation (CCR-CNV): an expandable and robust tool for clinical diagnosis of copy number variation at the exon level using next-generation sequencing data	Man Jin Kim (Seoul National University Hospital)
	2. Condylo-mandibulo-dysplasia : A frequent unrecognized pathology.	Joel Ferri (University hospital of Lille, France)
	3. The FusX TALE Base Editor (FusXTBE) for rapid mitochondrial DNA programming of human cells	Ankit Sabharwal (Mayo Clinic, Rochester, MN)

	in vitro and zebrafish disease models in vivo.	
	4. Impaired SNAPC4function leads to global reduction of canonical splicing events and is associated with a disorder characterized by progressive spasticity, developmental delay, and speech dysarthria	May Malicdan (National Institutes of Health, Bethesda, MD)
10:00-10:40	**Break**	
Genome Engineering and Emerging experimental models and New Frontiers in –Omics Therapeutics **Chairpersons:** May Malicdan & Domenica Taruscio		
10:40-10:55	Understanding lysosomal dynamics in neurons through the dissection of the functional role of LYST	Jenny Serra-Vinardell (National Institutes of Health, Bethesda, MD)
10:55-11:10	The cross-section of lysosomal dysfunction and rare diseases: Investigation of a new mouse model for Salla disease	Marya Sabir (National Institutes of Health, Bethesda, MD)
11:10-11:40	Data mining and drug repurposing	Noel Southall (National Institutes of Health, Bethesda, MD)
11:40-12:00	Selected Abstracts (6 minutes + 2 minutes for questions)	
	1. Patient with acanthosis nigricans: Identification of germline EGFR likely pathogenic variant and treatment	Lauren Graham (University of Alabama at Birmingham Undiagnosed Diseases Program)
	2. Empiric Treatment for Persistent Fever from Suspected Autoinflammatory Disease	Jeffrey Z Shen (University of Alabama Birmingham)
12:00-12:10	**Espresso Break**	
From population Genomes to Precision Medicine - Implications for Rare Diseases in the European Networks **Chairpersons:** Savino Sciascia & Dario Roccatello		
12:10-12:40	An approach to undifferentiated autoinflammatory diseases	Marco Gattorno (RITA European Reference Network for Rare Diseases- Coordinator)
12:40-13:10	Diagnostic and therapeutic challenges in undefined rheumatic diseases	Marta Mosca (ReConnect-European Reference Network for Rare Diseases- Coordinator)

13:10-13:40	Diagnostic practice patterns in hereditary kidney diseases: Insights from the European Rare Kidney Disease Registry	Franz Schaefer (ERKnet-European Reference Network for Rare Diseases- Coordinator)
13:40-14:20	Lipoedema: a cluster of still undefined syndromes	Sandro Michelini (Past President of the European society of Lymphology)
14:20-14:40	**Break**	
Clinical tips and selected abstracts **Chairpersons:** Simone Baldovino & Elisa Menegatti Selected Abstracts (6 minutes + 2 minutes for questions)		
14:40-15:15	1. Particular food aversion and diet-conditioned phenotype	Miriam Rigoldi (Mario Negri Institute)
	2. PUS7 mutations resulting in increased protein translation and a possible link to autism	Ted Han (Glycosphingolipid Disorders Unit, National Institutes of Health, Bethesda)
	3. Novel Compound Heterozygous Variants of FARSA in a Patient with Fatal Systemic Disorder	Soo Yeon Kim (The Korean National Network of Rare Disease Centers)
	4. RhoG deficiency specifically abrogates human lymphocyte cytotoxicity and cause HLH	Artem Kalinichenko (1St. Anna Children's Cancer Research Institute, Vienna, Austria)
UDN in the world **Chairpersons:** David Adams & Domenica Taruscio		
15:15-15:25	UPD Mexico	Claudia Gonzaga-Jauregui (Baylor College of Medicine)
15:25-15:35	UPD in UK	Cristina Dias (Guy's & St. Thomas' NHS Foundation Trust)
15:35-15:45	UPD in Brazil	Francisco Nociti (University of Campinas – Piracicaba Dental School)
15:45-15:55	UPD in Austria	Vanja Nagy & Jörg Menche (CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Vienna, Austria)
15:55-16:05	**Espresso Break**	
16:05-17:00	Selected Abstracts Selected Abstracts (6 minutes + 2 minutes for questions)	
	1. Phase I Operation of The Korean Undiagnosed Diseases Program: expansion of nationwide network and development of infrastructures	Soo Yeon Kim (The Korean National Network of Rare Disease Centers)

	2. Curation and Expansion of Human Phenotype Ontology for Inborn Errors of Immunity	Julia Pazmandi (Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases, Vienna, Austria)
	3. Solve Unsolved: How to Improve Diagnosis for Rare Diseases	Annalaura Torella (TIGEM, Pozzuoli, Naples, Italy)
	4. The UAB Undiagnosed Diseases Program: A Fee-for-Service Clinical Program	Bruce Korf (University of Alabama at Birmingham Undiagnosed Diseases Program)
	5. Solving patients with rare diseases within Telethon Undiagnosed Disease Program through reanalysis of exome-phenome data	Manuela Morleo (TIGEM, Pozzuoli, Naples, Italy)
	6. Use of Exome, Genome and RNASeq sequencing to solve a patient of the Spanish Undiagnosed Rare Diseases Program (SpainUDP).	Beatriz Martinez-Delgado (Instituto de Investigación de Enfermedades Raras (IIER)
Undiagnosed to Rare Disease – from the Patient Perspective		
17:00-17:10	Helene Cederroth (The Wilhelm Foundation, Sweden)	
Final Remarks		
17:10-17:20	Dario Roccatello, Domenica Taruscio, William Gahl