



10TH INTERNATIONAL CONFERENCE ON UNDIAGNOSED DISEASES

A HYBRID EVENT

**Host: Dario Roccatello
University of Turin (Italy)**

Turin (Italy)

January 31st – February 1st, 2022

"La Centrale" Nuvola Lavazza

UDNI Program Committee

Dario Roccatello (Turin, Italy)

Lisa Schimmenti (Rochester, USA)

Eric Klee (Rochester, USA)

Wendy van Zelst-Stams (Nijmegen, Holland)

William Gahl (Bethesda, USA)

Bela Melegh (Pecs, Hungary)

Paul Lasko (Montreal, Canada)

Domenica Taruscio (Rome, Italy)

Helene Cederroth (Stockholm, Sweden)

Local Scientific Secretary (Turin, Italy)

Simone Baldovino

Roberta Fenoglio

Elisa Menegatti

Daniela Rossi

Savino Sciascia

Organizing Secretariat



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DAY I MONDAY, JANUARY 31ST, 2022

all times in USA Central Time (Central Europe Time +7)

07:00 Welcome and Introduction to Turin

Dario Roccatello, Domenica Taruscio, William Gahl, Helene Cederroth

■ NEW FRONTIERS IN RARE DISEASES: FROM DIAGNOSTICS TO CLASSIFICATION CHALLENGES

Chairpersons: Dario Roccatello, William Gahl

07:15 Immunogenetics of the microcosm of the glomerular diseases characterized by focal segmental lesions of glomeruli

Fernando Fervenza (Rochester, USA)

07:45 The uncommon antigens of membranous nephropathy

Sanjeev Sethi (Rochester, USA)

08:15 The Piedmont Experience with Undiagnosed Diseases

Silvia Deaglio (Turin, IT)

08:35 The Impact of undiagnosed renal diseases in the clinical practice

Roberta Fenoglio (Turin, IT)

08:55 From diagnosis to characterization and clinical management: the DECODE project

Erica Daina (Bergamo, IT)

09:15 Espresso Break

09:25 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, KR)

2. Condylomandibulo-dysplasia: A frequent and recognized pathology

Joël Ferri (Lille, FR)

3. The FusX TALE Base Editor (FusXTBE) for rapid mitochondrial DNA programming of human cells in vitro and zebrafish disease models in vivo

Ankit Sabharwal (Rochester, USA)

4. Impaired SNAPC4 function 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 (Bethesda, USA)

5. Evaluation on trisaccharide BM 652 as a potential biomarker in urine of enzyme replacement therapy naive patients with MPS I aged 6 months - 18 years

Bhawana Aggarwal (New Delhi, IND)

10:00 Break

■ **GENOME ENGINEERING AND EMERGING EXPERIMENTAL MODELS AND NEW FRONTIERS IN – OMICS THERAPEUTICS**

Chairpersons: May Malicdan, Domenica Taruscio

- 10:40 Understanding lysosomal dynamics in neurons through the dissection of the functional role of LYST

Jenny Serra-Vinardell (Bethesda, USA)

- 10:55 The cross-section of lysosomal dysfunction and rare diseases: Investigation of a new mouse model for Salla disease

Marya Sabir (Bethesda, USA)

- 11:10 Data mining and drug repurposing

Noel Southall (Bethesda, USA)

- 11:40 Selected Abstracts (6 minutes + 2 minutes for questions)

1. Patient with acanthosis nigricans: Identification of germline EGFR likely pathogenic variant and treatment

Lauren Graham (Birmingham, USA)

2. Empiric Treatment for Persistent Fever from Suspected Autoinflammatory Disease

Shen, Jeffrey Z (Birmingham, USA)

12:00 Espresso Break

■ **FROM POPULATION GENOMES TO PRECISION MEDICINE IMPLICATIONS FOR RARE DISEASES IN THE EUROPEAN NETWORKS**

Chairpersons: Savino Sciascia, Dario Roccatallo

- 12:10 An approach to undifferentiated autoinflammatory diseases

Marco Gattorno (Genoa, IT)

- 12:40 Diagnostic and therapeutic challenges in undefined rheumatic diseases

Marta Mosca (Pisa, IT)

13:10 Diagnostic practice patterns in hereditary kidney diseases: Insights from the European Rare Kidney Disease Registry
Franz Schaefer (Heidelberg, DE)

13:40 Lipoedema: a cluster of still undefined syndromes
Sandro Michelini (Rome, IT)

14:20 Break

■ **CLINICAL TIPS AND SELECTED ABSTRACTS**

Chairpersons: Simone Baldovino, Elisa Menegatti

14:40 Selected Abstracts (6 minutes + 2 minutes for questions)

1. Particular food aversion and diet- conditioned phenotype

Miriam Rigoldi (Bergamo, IT)

2. PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation

Ted Han (Bethesda, USA)

3. Novel Compound Heterozygous Variants of FARSA in a Patient with Fatal Systemic Disorder

Soo Yeon Kim (Seoul, KR)

4. RhoG deficiency specifically abrogates human lymphocyte cytotoxicity and cause HLH

Artem Kalinichenko (Vienna, AT)

■ **UDN IN THE WORLD**

Chairpersons: David Adams, Domenica Taruscio

15:15 UDP Mexico
Claudia Gonzaga-Jauregui (Mexico City, MEX)

15:25 UDP in UK
Cristina Dias (London, UK)

15:35 UDP in Brazil
Francisco Nociti (Sao Paolo, BRA)

15:45 UDP in Austria
Vanja Nagy (Vienna, AT)

15:55 Espresso Break

16:05 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 (Seoul, KR)
- 2. Curation and Expansion of Human Phenotype Ontology for Inborn Errors of Immunity**
Julia Pazmandi (Vienna, AT)
- 3. Solve Unsolved: How to Improve Diagnosis for Rare Diseases**
Annalaura Torella (Naples, IT)
- 4. The UAB Undiagnosed Diseases Program: A Fee-for-Service Clinical Program**
Bruce Korf (Birmingham, USA)
- 5. Solving patients with rare diseases within Telethon Undiagnosed Disease Program through reanalysis of exome- phenome data**
Manuela Morleo (Naples, IT)
- 6. Use of Exome, Genome and RNASeq sequencing to solve a patient of the Spanish Undiagnosed Rare Diseases Program (SpainUDP)**
Beatriz Martinez-Delgado (Madrid, ES)
- 7. Network analysis reveals rare disease signatures across multiple level of biological organization**
Jorg Menche (Vienna, AT)
- 8. Corean undiagnosed rare diseases program - adults: 2 years of experience**
Jong Hyeon Ahn (Seoul, KR)

■ UNDIAGNOSED TO RARE DISEASE – FROM THE PATIENT PERSPECTIVE

17:00 Helene Cederroth, Wilhelm Foundation

17:10 Final Remarks
Dario Roccotello, Domenico Taruscio, William Gahl

DAY 2 TUESDAY, FEBRUARY 1ST, 2022

all times in USA Central Time (Central Europe Time +7)

- 07:00** **Welcome and Introduction to Turin**
Dario Roccatello, Domenica Taruscio, Helene Cederroth, William Gahl
- **UDNI COMMITTEE & WORKING GROUP REPORTS**
Chairperson: William Gahl
- 07:15** **Review of Current Leadership, Last Meeting Results, Issues to Address**
William Gahl (Bethesda, USA)
- 07:30** **Membership; Trainee Category**
Eric Klee (Rochester, USA)
- 07:40** **Genetic Counseling Working Group**
Janine Lewis, Stephanie Broley (Bethesda, USA)
- 07:50** **Communications/Website**
Domenica Taruscio, Marco Salvatore (Rome, IT)
- 08:10** **Developing Nations Working Group**
Domenica Taruscio, Manuel Posada, Samuel Wiafe, Olaf Bodamer
- 08:25** **Education Working Group/UEMS/Medical Competence and Medical Specialty**
Bela Melegh, Domenica Taruscio
- 08:35** **Functional Research Working Group**
Shinya Yamamoto (Huston, USA)
- 08:45** **Patient Engagement Plus**
Gulcin Gumus, Helene Cederroth, (Debbie Drell), Vanessa Boulanger
- 09:00** **Data Sharing Committee**
David Adams, Alexa McCray
- 09:10** **Diagnostics Working Group**
Elizabeth Palmer (Sydeny, AUS)
- 09:40** **Break**
- 10:15** **New Diseases, Solved and Unsolved Cases** (6 minutes + 2 minutes for questions)
Chairpersons: Dario Roccatello, Lisa Schimmenti
- 1. EDNRB mosaicism in a patient with nonsyndromic deafness/ GJB2 and PTPRQ mutations in a family with nonsyndromic deafness**
Darina Kachakova (Sofia, BGR)

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2. Cardiac Arrhythmia Syndrome with ST- Segment Depression

Marchionni Enrica (Rome, IT)

3. Adult-onset rapidly worsening progressive myoclonic epilepsy caused by a novel variant in DHDDS

Jangsup Moon (Seoul, South Korea)

4. CNV positional effect suggests an enhancer- mediated SHH dysregulation in a boy with multiple congenital anomalies

Daniele Vitale (Naples, IT)

5. FIBCD1 is a conserved receptor for chondroitin sulphate proteoglycans of the brain extracellular matrix and a candidate gene for a complex neurodevelopmental disorder

Vanja Nagy (Vienna, AT)

12:00 Lunch

INTERNATIONAL NETWORKS AND CONNECTIONS FOR UNDIAGNOSED AND RARE DISEASES

Chairperson: Paul Lasko

12:30 IRDiRC: Rare Diseases Treatment Access Working Group

William Gahl, Durhane Wong-Rieger, Steve Groft

12:45 Approaching Undiagnosed Diseases in Brazil: The House of Rares Project

Roberto Giugliani

13:00 UDN: Current and Future

Argenia Doss

13:10 Undiagnosed Diseases Network Foundation (UDNF)

Cristina Might

13:25 Solve RD – EU Horizon Program

Olaf Riess

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13:35 **New Diseases, Solved and Unsolved Cases** (6 minutes + 2 minutes for questions)

Chairpersons: Lorenzo Botto, Bela Melegh

1. Loss of seryl tRNA synthetase (SARS1) causes complex spastic paraplegia and cellular senescence

Edgard Verdura (Barcelona, ES)

2. Biallelic copy number variations in both upstream & downstream enhancers of SHOX gene causes mesomelia and clubfoot without short stature

Bengisu Guner Yilmaz (Istanbul, TK)

3. Monoallelic and Biallelic Germline Mutations Affecting the Transcription Factor Helios Cause Pleiotropic Defects of Immunity

Daniel Mayr (Vienna, AT)

4. Undiagnosed cutis laxa syndrome: two pairs of siblings with shared phenotype

Woojoong Kim (Seoul, KR)

14:10 ■ **UDNI BUSINESS MEETING**

Chairperson: 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

5. Votes online after meeting

15:00 **Concluding remarks**

Dario Roccatello, Savino Sciascia

GENERAL INFORMATION

REGISTRATION

The conference will take place mainly in ONLINE mode.

In order to participate, you must send an email to: udni.torino2022@gmail.com, indicating NAME - SURNAME - PROFESSION.

By January 29th, the Organizing Secretariat will send the link for the connection.

