UDNI
Undiagnosed
Diseases Network
INTERNATIONAL

ISTITVTO SVPERIORE DI SANITÀ

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

selene
eventi & congressi

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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. Condylo-mandibulo-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 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 (Bethesda, USA)

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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 Roccatello

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)

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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)

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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 Roccatello, Domenica Taruscio, William Gahl

DAY 2 TUESDAY, FEBRUARY 1ST, 2022

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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 chondricitin 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.