



Scientific Program (updated 2025-08-12)

Tuesday, October 28, 2025

19:00 WELCOME SESSION

Wednesday, October 29, 2025

08:00 - 09:00 SESSION 1
OPENING SESSION
Chair: **Roberto Giugliani** (Brazil)
Co-chair: **Mikk Cederroth** (Sweden)

Welcome to the 14th UDNI
Roberto Giugliani (Brazil)

The origins of UDNI and lessons learned through the years
Helene Cederroth (Sweden)

From UDPs to Hackathons
William Gahl (USA)

The Undiagnosed Hackathon
Eric Klee (USA)

09:00 - 10:15 SESSION 2
PANEL: ONLINE TOOLS TO DIAGNOSE THE UNDIAGNOSED
Chair: **TBD**
Co-chair: **TBD**

Matchmaking Tools
Nara Sobreira (Brazil/USA)

Modern Clinical Phenotyping
Pawel Buczkowicz (Canada)

FaceMatch Consortium
Tracy Dudding-Byth (Australia)

Discussion and Wrap-Up

- 10:15 – 10:30 Break – Poster Viewing
- 10:30 - 12:00 SESSION 3
 PANEL: HIGHLIGHTS OF UDPS AND HACKATHONS WORLDWIDE
 Chair: **TBD** ()
 Co-chair: **TBD** ()
- Americas
Gabriela Repetto (Chile)
- Asia
Salman Kirmani (Pakistan) and **Ratna Puri** (India)
- Oceania
Gareth Baynam (Australia)
- UDNI LMIC WG: Aims, Structure and Results
Domenica Taruscio (Italy)
- Wrap-Up
- 12:15 – 13:15 Lunchtime Symposium / Poster Viewing
 (Spot Reserved for 1 x 60 min Sponsored Satellite Symposium)
- 13:30 - 14:30 SESSION 4
 SPOTLIGHT ON EMERGING RESEARCH
 (ORAL PRESENTATIONS OF 4 SELECTED ABSTRACTS)
 Chair: **TBD**
 Co-chair: **TBD**
- Abstract 1 - **TBD**
 Abstract 2 - **TBD**
 Abstract 3 - **TBD**
 Abstract 4 - **TBD**
- 14:30 - 16:00 SESSION 5
 PANEL: POLICY INNOVATIONS: STRATEGIES TO TACKLE THE UNDIAGNOSED
 Chair: **TBD**
 Co-chair: **TBD**
- WHO – Genomics and Global Policy
Anna Laura Ross (Switzerland)
- The Role of Rare Diseases International
Alexandra Heumber (Switzerland)
- The Brazilian MOH approach to rare diseases
Natan Monsores (Brazil)

The ERDERA Project
Oleg Kvividze (Georgia)

Wrap-Up

16:00 – 16:30 Break – Poster Viewing

16:30 - 18:00 SESSION 6
PANEL: INNOVATIVE APPROACHES FOR UNDIAGNOSED DISEASES
Chair: **TBD**
Co-chair: **TBD**

Computational Structural Genomics: A Transformational Approach
for Biology and Medicine
Michael Zimmermann (USA)

Innovative Tools for Diagnosis of Genetic Diseases
TBC

AI in Medical Genetics
Annick Rein (Israel)

The Challenge of Genetic and Genomic Newborn Screening:
The EU Screen4care Project
Alessandra Ferlini (Italy)

Wrap-Up

18:00 - 19:00 SESSION 7
CASE REPORTS: PRESENTATION AND DISCUSSION OF 4 SELECTED
CASES (2 SOLVED AND 2 UNSOLVED) – CASES TO BE SELECTED
FROM THE ABSTRACTS
Chair: **TBD**
Co-chair: **TBD**

Presenter 1: **TBD**
Presenter 2: **TBD**
Presenter 3: **TBD**
Presenter 4: **TBD**

Thursday, October 30, 2025

- 08:30 - 10:00 **SESSION 8**
PANEL: NETWORKS, TRAINING, AND EDUCATION ON UNDIAGNOSED DISEASES
Chair: **TBD**
Co-chair: **TBD**
- RIBERSER**
Manuel Posada (Spain)
- Proficiency Testing and Certification
Béla Melegh (Hungary)
- Education in Rare Undiagnosed Diseases
Rosa Andrea Pardo (Chile)
- Variant Interpretation Education
Andreas Laner (Germany)
- Wrap-Up
- 10:00 – 10:30 Break – Poster Viewing
- 10:30 - 12:00 **SESSION 9**
PANEL: GENOMIC STANDARDS AND POPULATION GENOMICS
Chair: **TBD**
Co-chair: **TBD**
- Genomic Standards, GA4GH
TBD
- Pangenome
TBD
- Genomics in Admixed Populations
Iscia Lopes-Cendes (Brazil)
- Genomic Data in Rare Diseases in Brazil
João Bosco (Brazil)
- Wrap-Up
- 10:00 - 12:00 **PROFICIENCY TESTING**
for registered applicants only
Chair: **Béla Melegh** (Hungary)
Co-chair: **Tinatin Tkemaladze** (Georgia)
- 12:15 – 13:15 Lunchtime symposium / Poster Viewing
(Spot Reserved for 2 x 30 min Sponsored Satellite Symposia)

- 13:30 - 14:30 **SESSION 10**
SPOTLIGHT ON EMERGING RESEARCH – ORAL PRESENTATIONS OF 4 SELECTED ABSTRACTS
Chair: **TBD**
Co-chair: **TBD**
- Abstract 5 - **TBD**
Abstract 6 - **TBD**
Abstract 7 - **TBD**
Abstract 8 - **TBD**
- 14:30 - 16:00 **SESSION 11**
PANEL: HOW TO CREATE, STRUCTURE, AND FUND AN INSTITUTIONAL UDP PROGRAM
Chair: **TBD**
Co-chair: **TBD**
- United States
Tammy McAllister (USA)
- Europe
Wendy van Zelst-Stams (Netherlands)
- Latin America
Thereza Loureiro (Brazil) and **Guilherme Baldo** (Brazil)
- Asia
Jong Hee Chae (South Korea)
- Wrap-Up
- 16:00 - 16:30 Break – Poster Viewing
- 16:30 - 18:30 **SESSION 12**
PANEL: FROM DIAGNOSES TO THERAPIES
Chair: **TBD**
Co-chair: **TBD**
- Methodology for rare disease trials
Maria Teresa Acosta (USA)
- Drug Repurposing and IRDiRC
Emilio Roldan (Argentina)
- RNA therapies and N=1 trials
TBC
- Therapeutic matching
David Adams (USA)

Building International Collaborative Efforts to Advance in Therapies
Fernando Goldsztein (Brazil)
Wrap-Up

18:30 – 19:00 SESSION 13
CLOSING SESSION
Chair: **W Gahl** (USA)
Co-chair: **Helene Cederroth** (Sweden)

Next-Gen Leaders: Voices of the Future in Rare Disease
Ömer Bahadır Kiliç & Ipek Balahan (Turkey)

Abstract Prize
TBD

Introduction to Next UDNI Conference
TBD

Closing
William Gahl (USA) and **Helene Cederroth** (Sweden)

Friday, October 31, 2025

08:00 - 14:30 UDNI BUSINESS AND COMMITTEE MEETINGS