



Scientific Program (updated 2025-07-04)

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 Coffee 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)

UDN1 LMIC WG: Aims, Structure and Results
Domenica Taruscio (Italy)

Wrap-Up

12:15 – 13:15 Lunch Symp / Poster Viewing
(Spot Reserved for 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
TBC

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 **Coffee 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 Zimmerman (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** **Coffee Break – Poster Viewing**
- 10:30 - 12:00** **SESSION 9**
PANEL: GENOMIC STANDARDS AND POPULATION GENOMICS
Chair: TBD
Co-chair: 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: T. Tkemaladze (Georgia)
- 12:15 – 13:15** **Lunch / Poster Viewing**
(Spot Reserved for Sponsored Satellite Symposium)

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

Therapeutic matching
TBC

Building International Collaborative Efforts to Advance in Therapies
Fernando Goldstein (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
Youth/ACIBADEM medical students

Abstract Prize
TBD

Introduction to Next UDNI Conference
TBD

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

19:00 **BUSES DEPART TO THE NETWORKING EVENING**

Friday, October 31, 2025

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