



## Scientific Program (updated 2025-09-16)

### Tuesday, October 28, 2025

19:00 Welcome Session (specific registration required)

### Wednesday, October 29, 2025

08:00 - 09:00 SESSION 1

OPENING SESSION

Co-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

Co-chair: **Ignacio Zarante** (Colombia)

Co-chair: **TBC**

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  
 Co-chair: **David Pearce** (USA)  
 Co-chair: **TBC**
- 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 Symposia / ULTRAGENYX and CENTOGENE  
 Poster Viewing
- 13:30 - 14:30 SESSION 4  
 SPOTLIGHT ON EMERGING RESEARCH  
 (ORAL PRESENTATIONS OF 4 SELECTED ABSTRACTS)  
 CO-chair: **Olaf Bodamer** (USA)  
 Co-chair: **TBC**
- 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  
 Co-chair: **Claudia Gonzaga-Jauregui** (México)  
 Co-chair: **TBC**
- 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  
Co-chair: **Salmo Raskin** (Brazil)  
Co-chair: **Tinatin Tkemaladze** (Georgia)

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

Unlocking the Power of Epigenetics through Data: EpiSign  
**Mary Jane Dickman** (Canada)

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  
Co-chair: **Dafne Horovitz** (Brazil)  
Co-chair: **Fulya Taylan** (Sweden)

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

## Thursday, October 30, 2025

- 08:00 - 10:00    SESSION 8  
PANEL: NETWORKS, TRAINING, AND EDUCATION ON  
UNDIAGNOSED DISEASES  
Co-chair: **Janine Lewis** (USA)  
Co-chair: **TBC**
- 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
- 09:30 – 10:00    Break – Poster Viewing
- 10:00 - 12:00    SESSION 9  
PANEL: GENOMIC STANDARDS AND POPULATION GENOMICS  
Co-chair: **May Christine Malicdan** (USA)  
Co-chair: **TBC**
- Genomics in Admixed Populations  
**Iscia Lopes-Cendes** (Brazil)
- Genomic Data in Rare Diseases in Brazil  
**João Bosco** (Brazil)
- Topic **TBC**  
Speaker **TBC**
- Wrap-Up
- 10:00 - 12:00    PROFICIENCY TESTING  
for registered applicants only  
Co-chair: **Béla Melegh** (Hungary)  
Co-chair: **Tinatin Tkemaladze** (Georgia)
- 12:15 – 13:15    Lunchtime Symposia / ASTRAZENECA  
Poster Viewing

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- 13:30 - 14:30    SESSION 10  
 SPOTLIGHT ON EMERGING RESEARCH – ORAL PRESENTATIONS OF  
 4 SELECTED ABSTRACTS  
 Co-chair: **Carolina Fischinger** (Brazil)  
 Co-chair: **TBC**
- 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  
 Co-chair: **Filippo Pinto e Vairo** (USA)  
 Co-chair: **TBC**
- 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  
 Co-chair: **Bibiana Oliveira** (Brazil)  
 Co-chair: **TBC**
- Methodology for rare disease trials  
**Maria Teresa Acosta** (USA)
- Drug Repurposing and IRDiRC  
**Emilio Roldan** (Argentina)
- 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  
Co-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 (invitation only)