TUESDAY, OCTOBER 28, 2025

19:00 — WELCOME SESSION (SPECIFIC REGISTRATION REQUIRED)

Location: Aimoré room, 2nd floor, Windsor Marapendi Hotel

WEDNESDAY, OCTOBER 29, 2025

08:00 - 09:00 — SESSION 1 - OPENING SESSION

Co-chair: ***Antoine Daher*** (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 - 10:00 — SESSION 2 - PANEL: ONLINE TOOLS TO DIAGNOSE THE UNDIAGNOSED

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

Co-chair: ***Filippo Pinto e Vairo*** (Brazil/USA)

Matchmaking Tools

Nara Sobreira (Brazil/USA)

Modern Clinical Phenotyping

Pawel Buczkowicz (Canada)

FaceMatch Consortium

Tracy Dudding-Byth (Australia)

10:00 - 10:30 — Coffee Break - Poster Viewing

Scientific Program

WEDNESDAY, OCTOBER 29, 2025

10:30 - 12:00 SESSION 3 - PANEL: HIGHLIGHTS OF UDPS AND HACKATHONS WORLDWIDE

Co-chair: ***David Pearce*** (USA)
Co-chair: ***Olaf Bodamer*** (USA)

Americas
Gabriela Repetto (Chile)

Asia
Salman Kirmani (Pakistan) & ***Ratna Puri*** (India)

Oceania - Recorded
Gareth Baynam (Australia)

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

Wrap-Up

12:00 Lunchtime Satellite Symposia (30 min each)

12:15 - 12:45 Genetic Variant to Clinical Manifestation: How Genotype Drives Phenotype and Guides the Diagnosis of an Intriguing Case
Carolina Fischinger (Brazil) - Sponsored by Ultragenyx

12:45 - 13:15 Overcoming Challenges in the Diagnostic of Rare Genetic Disorders
Carlos Leprevost (Brazil) - Sponsored by CentoGene

Poster Viewing

13:30 - 14:30 SESSION 4 - SPOTLIGHT ON EMERGING RESEARCH (ORAL PRESENTATIONS OF 4 SELECTED ABSTRACTS)

Co-chair: ***Eric Klee*** (USA)
Co-chair: ***Guilherme Baldo*** (Brazil)

n.05, p.30 Rare disease diagnostic platform megSAP/GSvar: our experience in processing 10.000 rare disease index short read genomes
Author: ***German Demidov***

n.24, p.37 The Role of RNA Sequencing to Resolve UDN Cases
Author: ***Pinar Bayrak-Toydemir***

n.02, p.29 Diagnosis Ofelia: AI-Based WhatsApp Screening for Rare Disease Risk Stratification in Colombia
Author: ***Ignacio Zarante***

n.81, p.56 Piloting a clinically-integrated undiagnosed disease program: evidence for clinical utility and clinician acceptability
Author: ***Lisa Ewans***

WEDNESDAY, OCTOBER 29, 2025

14:30 - 16:00 SESSION 5 - PANEL: POLICY INNOVATIONS: STRATEGIES TO TACKLE THE UNDIAGNOSED

Co-chair: ***Claudia Gonzaga-Jauregui*** (Mexico)
Co-chair: ***David Adams*** (USA)

- WHO – Genomics and Global Policy - Recorded
  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 Kvlividze (Georgia)
- Wrap-Up

16:00 - 16:30 Coffee 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 Dykeman (Canada)
- AI in Medical Genetics
  Annick Rein (Israel)
- The Challenge of Genetic and Genomic Newborn Screening: The EU Screen4care Project - Recorded
  Alessandra Ferlini (Italy)
- Wrap-Up

Scientific Program

WEDNESDAY, OCTOBER 29, 2025

18:00 - 19:00 SESSION 7 - CASE REPORTS: PRESENTATION AND DISCUSSION OF 4 SELECTED CASES (2 SOLVED AND 2 UNSOLVED)

Co-chair: ***Dafne Horovitz*** (Brazil)

Co-chair: ***Fulya Taylan*** (Sweden)

n.26, p.38 A familiar gene, an unfamiliar story: TCF4 and a new mechanism unearthed after a decade of research

Author: ***Laurence Faivre***

n.37, p.42 Whole genome sequencing in immunodeficiencies: case report of twins with Cartilage hair hypoplasia, Omenn syndrome

Author: ***Istvan Balogh***

n.72, p.53 Undiagnosed syndrome with cleft lip, cleft palate, atrioventricular septal defect, duplication of the hallux

Author: ***Eugenia Ribeiro Valadares***

n.76, p.54 Comprehensive Diagnostic Approach in a Pediatric Patient with Multiple Digestive, Urinary, and Neurological Symptoms.

Author: ***Bárbara Lawlor Sclavo***

THURSDAY, OCTOBER 30, 2025

08:30 - 10:00 SESSION 8 - PANEL: NETWORKS, TRAINING, AND EDUCATION ON UNDIAGNOSED DISEASES

Co-chair: ***TBC***

Co-chair: ***Alexandra Heumber*** (Switzerland)

RIBERSER

Manuel Posada (Spain)

Proficiency Testing and Certification

Béla Melegh (Hungary)

Education in Rare Undiagnosed Diseases

Rosa Andrea Pardo (Chile)

Variant Interpretation Education - Recorded

Andreas Laner (Germany)

Wrap-Up

10:00 - 10:30 Coffee Break – Poster Viewing

THURSDAY, OCTOBER 30, 2025

10:00 - 12:00 PROFICIENCY TEST (For registered applicants only)
Co-chair: ***Béla Melegh*** (Hungary)
Co-chair: ***Tinatin Tkemaladze*** (Georgia)
Location: Barás room, Ground floor, Windsor Marapendi Hotel

10:30 - 12:00 SESSION 9 - PANEL: GENOMIC STANDARDS AND POPULATION GENOMICS
Co-chair: ***May Christine Malicdan*** (USA)
Co-chair: ***Salman Kirmani*** (Pakistan)

Genomics in Admixed Populations
Iscia Lopes-Cendes (Brazil)

Genomic Data in Rare Diseases in Brazil
João Bosco (Brazil)

Building more representative reference datasets of human variation - Recorded
Daniel MacArthur (Australia)

Wrap-Up

12:15 - 13:15 Lunchtime Satellite Symposium (60 min)

Hypophosphatasia: an underrecognized disorder
Têmis Félix (Brazil) - Sponsored by **AstraZeneca**

Poster Viewing

13:30 - 14:30 SESSION 10 - SPOTLIGHT ON EMERGING RESEARCH – ORAL PRESENTATIONS OF 4 SELECTED ABSTRACTS
Co-chair: ***Carolina Fischinger*** (Brazil)
Co-chair: ***Olaf Bodamer*** (USA)

n.96, p.62 The role of ancestry-aware filtering in rare disease variant interpretation
Author: ***Zsolt Bánfai***

n.86, p.58 UDN Sweden: Integrating Short- and Long-Read WGS and RNA-seq to diagnose Undiagnosed Children with Rare Syndromes
Author: ***Fulya Taylan***

n.29, p.39 Germline Variants as Contributors to Focal Cortical Dysplasia: Insights from Whole-Exome Sequencing
Author: ***Flávia Paniza Marcone***

n.27, p.38 Biallelic PSTK variants cause generalized selenoprotein deficiency and progressive neurodegeneration
Author: ***May Christine Malicdan***

Scientific Program

THURSDAY, OCTOBER 30, 2025

14:30 - 16:00 SESSION 11 - PANEL: HOW TO CREATE, STRUCTURE, AND FUND AN INSTITUTIONAL UDP PROGRAM

Co-chair: ***Filippo Pinto e Vairo*** (Brazil/USA)
Co-chair: ***Ratna Puri*** (India)

United States - Recorded
Tammy McAllister (USA)

Europe
Wendy van Zelst-Stams (Netherlands)

Latin America
Thereza Cavalcanti (Brazil) and ***Guilherme Baldo*** (Brazil)

Asia
Jong Hee Chae (South Korea)

Wrap-Up

16:00 - 16:30 Coffee Break - Poster Viewing

16:30 - 18:00 SESSION 12 - PANEL: FROM DIAGNOSES TO THERAPIES

Co-chair: ***Bibiana Oliveira*** (Brazil)
Co-chair: ***David Pearce*** (USA)

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

THURSDAY, OCTOBER 30, 2025

18:00 - 19:00 SESSION 13 - CLOSING SESSION

Co-chair: ***William Gahl*** (USA)

Co-chair: ***Helene Cederroth*** (Sweden)

Next-Gen Leaders: Voices of the Future in Rare Disease

Ömer Bahadir Kiliç (Turkey) & ***Ipek Balaban*** (Turkey)

Abstract Prize

To be announced

Introduction to Next UDNI Conference

To be announced

Closing

William Gahl (USA)***, Helene Cederroth*** (Sweden) and ***Roberto Giugliani*** (Brazil)

19:30 Buses depart to Network Evening (specific registration required)

FRIDAY, OCTOBER 31, 2025

09:00 - 13:00 UDNI BUSINESS AND COMMITTEE MEETINGS (open to UDNI members and non-members)

Location: Aimoré II Room, 2nd floor, Windsor Marapendi Hotel