SCIENTIFIC PROGRAM (UPDATED 2025-10-11)

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: **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) 1

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** (USA)

Matchmaking Tools
Nara Sobreira (Brazil/USA)

Modern Clinical Phenotyping
Pawel Buczkowicz (Canada)

FaceMatch Consortium
Tracy Dudding-Byth (Australia)

10:00 – 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: **Olaf Bodamer** (USA)

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 Satellite Symposia (30 min each):

1) Genetic Variant to Clinical Manifestation: How Genotype Drives Phenotype and Guides the Diagnosis of an Intriguing Case
   Carolina Fischinger (Brazil) - Sponsored by Ultragenyx
2) 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)

1	Rare disease diagnostic platform megSAP/GSvar: our experience in processing 10.000 rare disease index short read genomes	German Demidov
2	The Role of RNA Sequencing to Resolve UDN Cases	Pinar Bayrak-Toydemir
3	Diagnosis Ofelia: AI-Based WhatsApp Screening for Rare Disease Risk Stratification in Colombia	Ignacio Zarante
4	Piloting a clinically-integrated undiagnosed disease program: evidence for clinical utility and clinician acceptability	Lisa Ewans

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

1	A familiar gene, an unfamiliar story: *TCF4* and a new mechanism unearthed after a decade of research	Laurence Faivre
2	Whole genome sequencing in immunodeficiencies: case report of twins with Cartilage hair hypoplasia, Omenn syndrome	Istvan Balogh
3	Undiagnosed syndrome with cleft lip, cleft palate, atrioventricular septal defect, duplication of the hallux	Eugenia Ribeiro Valadares
4	Comprehensive Diagnostic Approach in a Pediatric Patient with Multiple Digestive, Urinary, and Neurological Symptoms.	Bárbara Lawlor Sclavo

Thursday, October 30, 2025

08:30 - 10:00 SESSION 8
PANEL: NETWORKS, TRAINING, AND EDUCATION ON UNDIAGNOSED DISEASES
Co-chair: **Janine Lewis** (USA)
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
Andreas Laner (Germany)

Wrap-Up

10:00 – 10:30 Break – Poster Viewing

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
Daniel MacArthur (Australia)

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 Satellite Symposium (60 min):
From signs and symptoms to the diagnosis of Hypophosphatasia – How can the diagnostic process be performed earlier? What could a low alkaline phosphatase result indicate?
(presented by **Dr. Têmis Félix** and 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)

1	The role of ancestry-aware filtering in rare disease variant interpretation	Zsolt Bánfai
2	UDN Sweden: Integrating Short- and Long-Read WGS and RNA-seq to diagnose Undiagnosed Children with Rare Syndromes	Fulya Taylan
3	Germline Variants as Contributors to Focal Cortical Dysplasia: Insights from Whole-Exome Sequencing	Flávia Paniza Marcone
4	Biallelic *PSTK* variants cause generalized selenoprotein deficiency and progressive neurodegeneration	May Christine Malicdan

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: **Ratna Puri** (India)

United States
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 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

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 Balahan** (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 **Networking Evening**
(specific registration required)

Friday, October 31, 2025

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

Draft Business Agenda – UDNI Conference – Rio de Janeiro, Brazil

October 31, 2025

Oct 31	UDNI COMMITTEE & WORKING GROUP REPORTS	Chair: William Gahl
0900-0910	Welcome	William Gahl, Helene and Mikk Cederroth
0910-0920	Membership; Industry Category/Update	Eric Klee
0920-0930	Genetic Counseling Working Group	Janine Lewis (Stephanie Broley)
0930-0940	Communications/Website/ICORD	Marco Salvatore, Gianluca Ferrari, Domenica Taruscio
0940-0955	Low and Middle Income Countries Working Group	Domenica Taruscio, Manuel Posada, Samuel Wiafe, Olaf Bodamer
0955-1020	Champions Initiative Update	Salman Kirmani (5), Samuel Wiafe (5), Aime Lumaka, Guida Landoure, Ratna Puri (5)
1020-1030	Education Working Group/UEMS/Medical Competence and Medical Specialty	Bela Melegh, Domenica Taruscio, Bruce Korf
1030-1040	Functional Study Working Group	Shinya Yamamoto, May Malicdan, Stephen Pak
1040-1115	**Break**	
1115-1125	Patient Engagement Plus	Helene Cederroth
1125-1135	Diagnostics Working Group: Linking to Hackathons and Emerging Technology	Ann Nordgren, Emma Palmer, Lorenzo Botto, Fulya Taylan
1135-1145	Mayo Hackathon Update Future Hackathons	Eric Klee, Mikk and Helene Cederroth
1145-1200	Data Sharing and Technology Working Group	David Adams
	Other Business	William Gahl
1200-1300	Rare Cancers Working Group RARE Journal 2026 Meeting Issues Arising, Other Topics	Bela Melegh Wendy van Zelst-Stams (5) Bucharest (8) Jerusalem (8) Siena (8)