UDNI & Mayo Clinic Science Session
Virtual Livestream
All times listed in Central Standard Time (CST) and Greenwich Mean Time (GMT)

Friday, April 9th, 2021

6:50 CST/ 12:50 PM GMT  Welcome and Opening Remarks/Announcements

Alternative Omics Diagnostics

7:00 CST/13:00 GMT  Proteomics
Akhilesh Pandey, M.D., Ph.D., Mayo Clinic

7:30/13:30  Lyfes Languages and The Speed of Trust
Gareth Baynam, MBBS, Ph.D., The University of Western Australia

8:00/14:00  High Through-put Method of Genomic Interpretation in 3D
Raul Urrutia, M.D., Medical College of Wisconsin

8:30/14:30  Whole Methylome for Congenital Disorders
Linda Hasadsri, M.D., Ph.D., Mayo Clinic

Therapeutics

9:00/15:00  Gene Therapy for Primary Immunodeficiency and Metabolic Disease
R. Scott Mc Ivor, Ph.D., University of Minnesota

9:30/15:30  Break
Visit the international photo project The Undiagnosed online,
Joint venture Wilhelm Foundation and Rick Guidotti Positive Exposure

10:00/16:00  Propionic Acidemia and Gene Therapy
Michael Barry, Ph.D., Mayo Clinic

10:30/16:30  Congenital Disorders of Glycosylation; It’s Sugar Coded!
Eva Morava-Kozicz, M.D., Ph.D.

11:00/17:00  Brain on a Chip; Disease Modeling and Drug Screening in Mitochondrial Medicine
Tamas Kozicz, M.D., Ph.D., Mayo Clinic

11:30/17:30  Drug Repurposing Therapies
Ethan Perlstein, Ph.D., Perlara Laboratories and Maggie’s Pearl

12:00/18:00  Break
Visit the international photo project The Undiagnosed online,
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Genome Engineering

13:00 CST/19:00 GMT  CRISPR Brain Disease Modeling in Zebrafish  
Maura McGrail, Ph.D., Iowa State University

13:30/19:30  CRISPR engineering and transposon-mutagenesis for understanding peripheral nerve sheath tumorigenesis  
David Largaespada, Ph.D., Center for Genome Engineering, University of Minnesota

14:00/20:00  Functional Genomics  
Margot Cousin, Ph.D.

14:15/20:15  Phenotype identification in multiplexed RNA-sequencing with natural genetic variation  
Jin Zhang & Min Lou, Zhejiang University Medical Center

14:30/20:30  Break

Visit the international photo project The Undiagnosed online,  
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Novel Funding Models

15:00/21:00  Insurance & Philanthropy Support for UDN  
F. Sessions (Sesh) Cole, M.D., Washington School of Medicine in St. Louis

15:15/21:15  Antisense Therapeutics  
Stanley Crooke, M.D., Ph.D., n-Lorem Foundation

15:30/21:30  Cures Within Reach and a Donor-Directed Approach to Funding Repurposing Research  
Clare Thibodeaux, Ph.D., Cures Within Reach

15:45/21:45  Funding Model Example  
Annette Bakker, Ph.D., Children’s Tumor Foundation

16:00/22:00  Adjourn for the Day
UDNI & Mayo Clinic Science Session continued...

Virtual Livestream

All times listed in Central Standard Time (CST) and Greenwich Mean Time (GMT)

Saturday, April 10, 2021

6:50 CST/12:50 PM GMT Kick off the day

Emerging Science

7:00 CST / 13:00 GMT Genome Writers Guild
Shondra Miller, Ph.D., St. Jude Children’s Research Hospital

7:30/13:00 Mitochondrial Disorders: Towards a Therapy from Thin Air.”
Vamsi Mootha, M.D., Harvard Medical School, Harvard University

8:00/14:00 Protein Modeling
Thomas Caulfield, Ph.D., Mayo Clinic

8:30/14:30 How do we get to a definitive clinical result? The challenges of resolving complex structural variants.
Lisa Ewans, M.B.B.S., & Elizabeth Palmer, Ph.D., Sydney Children’s Hospital

8:45/14:45 Leber’s hereditary optic neuropathy (LHON)-associated mitochondrial tRNA mutation at position 73 displayed the pleiotropic effects on the processing, CCA adding activity, stability and aminoacylation of tRNA
Min-Xin Guan, Ph.D., Institute of Genetics, Zhejiang University School of Medicine

9:00/15:00 Break

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9:30/15:30 Genome Sequencing - Neonatal Rapid sequencing
Stephen Kingsmore, M.D., D.Sc., Rady Children’s Institute for Genomic Medicine

10:00/16:00 Updates in Prenatal Screening
Myra J. Wick, M.D., Ph.D., Mayo Clinic

10:30/16:30 Genomic Variations in South Asia and the Middle East
Sridhar Sivasubbu, Ph.D., IGIB in India

11:00/17:00 Break and Poster Viewing

Visit the international photo project The Undiagnosed online,
Joint venture Wilhelm Foundation and Rick Guidotti Positive Exposure
Moving from Undiagnosed to Rare Disease – from the Patient Perspective

12:00 CST/18:00 GMT  Keynote Session: On the Ethics of Value
Elliot Chaikof, M.D., Ph.D., Surgeon-in-Chief at the Beth Israel Deaconess Medical Center (BIDMC), & Johnson and Johnson Professor of Surgery, Harvard Medical School, Harvard University

13:00/19:00  Paige’s Story
Brendan Lanpher, M.D., Ph.D., Mayo Clinic

13:30/19:30  Chloe Barnes Advisory Council
Erica Barnes, Advisory Council on Rare Disease

13:45/19:45  Break
Visit the international photo project The Undiagnosed online, Joint venture Wilhelm Foundation and Rick Guidotti Positive Exposure

14:00/20:00  Precision Medicine: From n=1 to Everyone
Matt Might, Ph.D., University of Alabama at Birmingham

14:30/20:30  The Undiagnosed
Helene & Mikk Cederroth, Wilhelm Foundation

14:45/20:45  Introduction of UDNI Topics at Business Meeting
William A. Gahl, M.D., Ph.D., Undiagnosed Disease Network International

15:00/21:00  Adjourn

15:15/21:15  Join us for a casual post-session social hour via Zoom. Debrief with the program committee and attendees.
# 9th International Conference on Rare and Undiagnosed Diseases

Sunday, April 11, 2021 | Business Meeting

Livestreamed on behalf of Mayo Clinic, Rochester, Minnesota

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<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Chair/Leader</th>
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<tbody>
<tr>
<td>0700 CST</td>
<td>Welcome</td>
<td>Lisa Schimmenti, Eric Klee, Helene Cederroth, W. Gahl</td>
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<tr>
<td>07:15 CST</td>
<td><strong>UDNI Committee &amp; Working Group Reports</strong></td>
<td>Chair: W. Gahl</td>
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<tr>
<td>07:15-13:15</td>
<td>Review of Current Leadership, Last Meeting Results, Issues to Address</td>
<td>William Gahl</td>
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<td>07:30-13:30</td>
<td>Membership; Trainee Category</td>
<td>Eric Klee</td>
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<td>07:45-13:45</td>
<td>Genetic Counseling Working Group</td>
<td>Janine Lewis</td>
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<td>08:00-14:00</td>
<td>Communications/Website</td>
<td>Domenica Taruscio, Marco Salvatore</td>
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<td>08:15-14:15</td>
<td>Developing Nations Working Group</td>
<td>Domenica Taruscio, Manuel Posada, Samuel Wiafe</td>
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<td>08:30-14:30</td>
<td>Education Working Group/UEMS/Medical Competence and Medical Specialty</td>
<td>Bela Melegh, Domenica Taruscio</td>
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<td>08:40-14:40</td>
<td>Functional Research Working Group</td>
<td>Shinya Yamamoto</td>
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<td>08:50-14:40</td>
<td>Patient Engagement Plus</td>
<td>Gulcin Gumus</td>
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<td>09:05-15:05</td>
<td>Data Sharing Committee</td>
<td>David Adams, Alexa McCray</td>
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<td>09:20-15:20</td>
<td>Break</td>
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<td>09:50-15:00</td>
<td><strong>New Diseases, Solved and Unsolved Cases</strong></td>
<td>Chair: Ratna Puri, New Delhi</td>
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<td>09:50-15:00</td>
<td>(6 minutes + 2 minutes for questions)</td>
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<td>09:50-15:00</td>
<td>1. Deficiency of picornavirus host factor and PLAAT3 causes partial lipodystrophy</td>
<td>Nika Schuermans, Ghent U.</td>
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<td>09:50-15:00</td>
<td>2. PSMD12 haploinsufficiency, proteasome dysfunction, and autoinflammation</td>
<td>Kai Yan, Zhejiang U.</td>
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<tr>
<td>1. COX loss of function variants cause autosomal recessive sensory neuronopathy</td>
<td>Hai-Lin Dong, Zhejiang U.</td>
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<td>2. Genetics of ABCA4 retinopathy in a pseudo-dominant family with cone-rod dystrophy</td>
<td>Kunka Kamenarova, Bulgaria</td>
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<td>3. Chinese Hereditary Cancer Multigene Test Project: E107Nfs*3 mutation in CHEK2 gene</td>
<td>Jiawei Shou, Zhejiang U.</td>
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<td>4. Pediatric hereditary epidermolysis bullosa and ANCA-associated vasculitis</td>
<td>Guoping Huang, Zhejiang U.</td>
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<td>5. Recurrent pleuritis in a middle-aged woman</td>
<td>Karlijn van Vlerken, Erasmus</td>
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<td>6. Persistent heparan sulfate excretion in an undiagnosed patient with MPS phenotype</td>
<td>Roberto Giugliani, Brazil</td>
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<td>7. Short stature and microcephaly in two siblings</td>
<td>Swasti Pal, New Delhi</td>
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<td>8. An undiagnosed child with macrocephaly</td>
<td>Veronica Arora, New Delhi</td>
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<td>9. Biallelic P14KA variants cause hypomyelinating leukodystrophy</td>
<td>Aurora Pujol, Barcelona</td>
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**1120-1210**

17:20-18:20  **Lunch**

**12:10-14:30**
18:10-20:30  **International Networks and Connections for Undiagnosed and Rare Diseases**  
Chair: Paul Lasko, Montreal

**12:10**
18:10  IRDiRC: Rare Diseases Treatment Access Working Group  
William Gahl  
Durhane Wong-Rieger  
Steve Groft

**12:20**
18:20  WHO Collaborative Global Network for Rare Diseases  
Matt Bolz-Johnson, Rare Diseases International

**12:30**
18:30  Global Commission to End the Diagnostic Odyssey for Children (Takeda, Eurordis, Microsoft Health)  
Roberto Giugliani

**12:40**
18:40  iCORD: Central and South America and Caribbean Nations (ERCAL Initiative)  
Steve Groft

**12:50**
18:50  UDN: Current and Future  
Argenia Doss, NIH

**13:00**
19:00  G2MC/Regeneron  
Vajira Dissanayake

**13:10**
19:10  NORD Undiagnosed Rare Disease Patient Registry Natural History Program/Platform IAmRare  
Vanessa Boulanger

**13:20**
19:20  UDNi’s 5 unsolved cases for PhenomeCentral: Current Status, future steps  
Bela Melegh

**13:30**
19:30  Solve RD – EU Horizon Program  
Olaf Riess

**13:40-14:40**  
**Individual UDP Initiatives**  
*(6 minutes + 3 minutes for questions)*

1. Sydney, Australia: Gene2Care integration of genomics into clinical practice  
Elizabeth Palmer, U. New South Wales
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<th>2.</th>
<th>Ghent, Belgium: Belgian multidisciplinary program for Undiagnosed Rare Diseases</th>
<th>Nika Schuermans, Ghent U.</th>
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<tr>
<td>3.</td>
<td>Naples, Italy: Telethon Undiagnosed Diseases Program – 4-year pilot outcome</td>
<td>Morleo Manuela, TIGM, Naples</td>
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<td>4.</td>
<td>Dijon, France: 5-years and 199 novel cases of intellectual &amp; developmental disorders</td>
<td>Ange-Line Burel, Inserm-U. Burgundy-Franche Comte</td>
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<td>5.</td>
<td>Pusan, Korea: Whole exome sequencing using buccal swabs for the UDP in Korea</td>
<td>Chong Kun Cheon, Pusan U. Children’s Hospital</td>
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<td>6.</td>
<td>Western Australia: The psychology of rare diseases - Transformative Teamwork</td>
<td>Georgia Hay, Western Australia</td>
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<td>7.</td>
<td>Japan: Update on IRUD</td>
<td>Kenjiro Kosaki, Keio U.</td>
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**14:40**

**Break**

**15:10-16:35**

**UDNI Business Meeting**

**Chair: W. Gahl**

1. Confirm New Committee and Board Members
2. Next Meeting Location (In person/virtual)
3. Sustainability/Future of UDNI
4. Indigenous Nations WG (G. Baynam)
5. Unsolved Cases Website Curation WG: Role of Junior Members (Bela Melegh, Domenica Taruscio, Helene Cederroth)
6. UDNI Members Webpages
7. Acknowledge UDNI in Papers
8. UN-NGO Interactions
9. UDNI Distributing Patients: Referral center; Patient Case Postings (Domenica Taruscio)
10. Votes online after meeting

**16:35**

**Closing Comments**

**16:40**

**Adjourn**