

# UDNI & Mayo Clinic Science Session

## Virtual Livestream

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

### Friday, April 9<sup>th</sup>, 2021

6:50 CST/ 12:50 PM GMT      Welcome and Opening Remarks  
Drs. Klee & Babovic

#### 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  
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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, Center for Genome Engineering, University of Minnesota*
- 14:00/20:00 Speaker to be Announced
- 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,  
*Joint venture Wilhelm Foundation and Rick Guidotti Positive Exposure*

## 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 ASO Funding Source/Group  
*Stan 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, PhD, Director of Scientific Affairs, Cures Within Reach*
- 15:45/21:45 Funding Model Example  
*Annette Bakker, President, PhD, Children's Tumor Foundation*
- 16:00/22:00 Adjourn for the Day

## UDNI & Mayo Clinic Science Session continued...

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### 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 Title to be Confirmed  
*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, MBBS, Elizabeth Palmer, PhD, 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
- Visit the international photo project **The Undiagnosed** online,  
*Joint venture Wilhelm Foundation and Rick Guidotti Positive Exposure*
- 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 Title to be Confirmed  
*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  
*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 Title to be Confirmed  
*Matt Might, Ph.D., Hugh Kaul Precision Medicine Institute, 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

Registration 0630 CST/12:30 GMT

*(all times in United States Central Time and Greenwich Mean Time)*

0700 CST	<b>Welcome</b>	Lisa Schimmenti, Eric Klee, Helene Cederroth, W. Gahl
07:15 CST 13:15 GMT	<b>UDNI Committee &amp; Working Group Reports</b>	Chair: W. Gahl
07:15 13:15	Review of Current Leadership, Last Meeting Results, Issues to Address	William Gahl
07:30 13:30	Membership; Trainee Category	Eric Klee
07:45 13:45	Genetic Counseling Working Group	Janine Lewis
08:00 14:00	Communications/Website	Domenica Taruscio Marco Salvatore
08:15 14:15	Developing Nations Working Group	Domenica Taruscio Manuel Posada Samuel Wiafe
08:30 14:30	Education Working Group/UEMS/Medical Competence and Medical Specialty	Bela Melegh Domenica Taruscio
08:40 14:40	Functional Research Working Group	Shinya Yamamoto
08:50 14:40	Patient Engagement Plus	Gulcin Gumus
09:05 15:05	Data Sharing Committee	David Adams Alexa McCray
09:20 15:20	<b>Break</b>	
09:50-11:20 15:50-17:20	<b>New Diseases, Solved and Unsolved Cases</b> <i>(6 minutes + 2 minutes for questions)</i>	Chair: Ratna Puri, New Delhi
	1. Deficiency of picornavirus host factor and PLAAT3 causes partial lipodystrophy	Nika Schuermans, Ghent U.
	2. PSMD12 haploinsufficiency, proteasome dysfunction, and autoinflammation	Kai Yan, Zhejiang U.
	3. COX loss of function variants cause autosomal recessive sensory neuronopathy	Hai-Lin Dong, Zhejiang U.

	4. Genetics of ABCA4 retinopathy in a pseudo-dominant family with cone-rod dystrophy	Kunka Kamenarova, Bulgaria
	5. Chinese Hereditary Cancer Multigene Test Project: E107Nfs*3 mutation in <i>CHEK2</i> gene	Jiawei Shou, Zhejiang U.
	6. Pediatric hereditary epidermolysis bullosa and ANCA-associated vasculitis	Guoping Huang, Zhejiang U.
	7. Recurrent pleuritis in a middle-aged woman	Karlijn van Vlerken, Erasmus
	8. Persistent heparan sulfate excretion in an undiagnosed patient with MPS phenotype	Roberto Giugliani, Brazil
	9. Short stature and microcephaly in two siblings	Swasti Pal, New Delhi
	10. An undiagnosed child with macrocephaly	Veronica Arora, New Delhi
	11. Biallelic P14KA variants cause hypomyelinating leukodystrophy	Aurora Pujol, Barcelona
11:20-12:10 17:20-18:20	<b>Lunch</b>	
12:10-14:30 18:10-20:30	<b>International Networks and Connections for Undiagnosed and Rare Diseases</b>	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 19:40-20:40	<b>Individual UDP Initiatives</b> (6 minutes + 3 minutes for questions)	
	1. Sydney, Australia: Gene2Care integration of genomics into clinical practice	Elizabeth Palmer, U. New South Wales

	2. Ghent, Belgium: Belgian multidisciplinary program for Undiagnosed Rare Diseases	Nika Schuermans, Ghent U.
	3. Naples, Italy: Telethon Undiagnosed Diseases Program – 4-year pilot outcome	Morleo Manuela, TIGM, Naples
	4. Dijon, France: 5-years and 199 novel cases of intellectual & developmental disorders	Ange-Line Burel, Inserm-U. Burgundy-Franche Comte
	5. Pusan, Korea: Whole exome sequencing using buccal swabs for the UDP in Korea	Chong Kun Cheon, Pusan U. Children's Hospital
	6. Western Australia: The psychology of rare diseases - Transformative Teamwork	Georgia Hay, Western Australia
	7. Japan: Update on IRUD	Kenjiro Kosaki, Keio U.
14:40 20:40	<b>Break</b>	
15:10-16:35 21:10-22:35	<b>UDNI Business Meeting</b>	<b>Chair: W. Gahl</b>
	<ol style="list-style-type: none"> <li>1. Confirm New Committee and Board Members</li> <li>2. Next Meeting Location (In person/virtual)</li> <li>3. Sustainability/Future of UDNI</li> <li>4. Indigenous Nations WG (G. Baynam)</li> <li>5. Unsolved Cases Website Curation WG: Role of Junior Members (Bela Melegh, Domenica Taruscio, Helene Cederroth)</li> <li>6. UDNI Members Webpages</li> <li>7. Acknowledge UDNI in Papers</li> <li>8. UN-NGO Interactions</li> <li>9. UDNI Distributing Patients: Referral center; Patient Case Postings (Domenica Taruscio)</li> <li>10. Votes online after meeting</li> </ol>	
16:35 22:35	<b>Closing Comments</b>	
16:40 22:40	<b>Adjourn</b>	