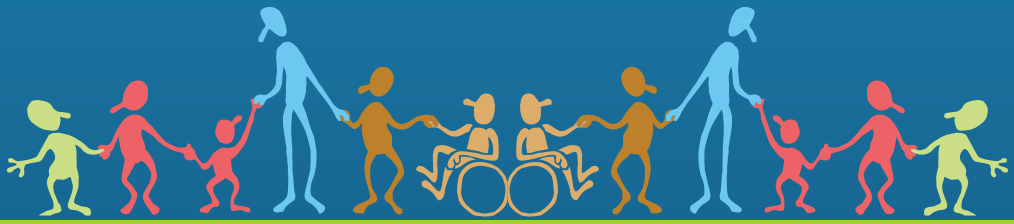
7th International Conference

Rare & Undiagnosed Diseases: Addressing Patient Needs for Rare Disorders in India

13th-15th April, 2019

The Leela Ambience Hotel, Gurugram, Delhi NCR | India

Scientific Program



Jointly Organized By

UDNI Undiagnosed Diseases Network INTERNATIONAL

Sir Ganga Ram Hospital Institute of Medical Genetics & Genomics

Wilhelm Foundation The Undiagnosed

ORD Organization for Rare Diseases India

IGIB Institute of Genomics & Integrative Biology Genomics Knowledge Partner

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Day 1, April 13th 2019		
0800 - 0900	**Registration**	
0900 hrs	**Welcome**	
Session 1: Diagnosis and Screening for Genetic Disorders Chairpersons: Thelma BK, Neerja Gupta, Sankar VH		
0910 - 0930	Genetic disorders in India - current status	Meenakshi Bhat, Bengaluru
0930 - 0950	Lessons learnt from newborn studies in India and way forward	Seema Kapoor, Delhi
0950 - 1010	Carrier screening – experience from India	Sheetal Sharda, Bengaluru
1010 - 1030	Roadmap for Management of Rare Disorders in India	Shubha R Phadke, Lucknow
1030 - 1100	**Tea/Coffee Break**	
Session 2: Rare Disorders Programs Chairpersons: Ishwar C Verma, Dwaipayan Bharadwaj, Prajnya Ranganath		
1100 - 1130	Population Genomics: The Alabama Genomic Health Initiative and the All of Us Research Program	Bruce R Korf, USA
1130 - 1200	Transforming Care for Children with Rare Disorders	Tamanna R Lal, USA
1200 - 1220	Rare diseases genomics in India – Guardian experience	Vinod Scaria, Delhi
1220 - 1240	Facioscapular muscular dystrophy – new insights	Meena Upadhyaya, UK
1240 - 1300	Artificial intelligence to drive innovation for rare disease treatments	Himani Sharma, USA
1300 - 1400	**Lunch and Posters**	
Session 3: Genomics of Rare Disorders Chairpersons: H Sharat Chandra, Mamta Muranjan, Sheela Nampoothiri		
1400 - 1430	Deep Phenotyping in the era of Genomics	Michael Brudno, Canada
1430 - 1500	Inferring variants in the era of Genomics	Madhuri Hegde, USA
1500 - 1520	Founder mutations in Agarwal Community	Ishwar C Verma, Delhi
1520 - 1550	Future of Technology for Rare disorders	Arndt Rolfs, Germany
1550 - 1605	Familial genetic disorders and lung cancer	Somasekar Seshagi, USA
1605 - 1620	Genomic Approaches to Tackle Undiagnosed Diseases	Shrikant Mane, USA
1620 - 1630	Discussion	
1630 - 1700	**Tea/Coffee Break and Posters**	
Session 4: Research Collaborations in Rare Diseases: Barriers and Opportunities		
1700 - 1750	Fostering Research Collaborations Panelists Alok Bhattacharya, Professor, Founder WWGM Ambrish Kapadia, Parent Project Muscular Dystrophy, Mumbai David Pearce, Sanford School of Medicine, South Dakota Anil Raina, GM (South Asia) & Head - India, Sanofi Genzyme	Moderator : Harsha K Rajasimha, USA
1750 - 1825	Role of Patient groups - Awareness & Education Panelists Anil Choube, IPWSA-Indian Prader Willi Syndrome Association Manjit Singh, LSDSS- Lysosomal Storage Disorder Support Society Samir Sethi, IRSF -Indian Rett Syndrome Foundation Archana Panda, Cure SMA Nicole Boice, Global Genes Vikas Bhatia, MERD India Rukshana, Primay Immune Deficiencies Parents Welfare Society	Moderator : Prasanna K Shirol, Bengaluru
1900 onwards	**Conference Banquet at Hotel The Leela Ambience**	

Day 2, April 14th 2019		
0800 – 0840	**Oral Abstracts Chairpersons:Bruce R Korf, Shubha R. Phadke, Inusha Panigrahi**	
0800 - 0806	Using structured data to streamline genomic diagnosis in the first Canadian rare disease research platform, Genomics4RD	O Buske, Genomics4RD, Canada
0806 - 0812	Utility of whole exome sequencing in diagnosis of mitochondrial disorders in pediatric cases	A Mishra, CSIR-IGIB, Delhi
0812 - 0818	An Artificial Intelligence Powered Care Continuum Platform for Inborn Errors of Metabolis	S Ulman, Philips Innovation Campus, Bengaluru
0818 - 0824	Looking beyond the scope: co-existence of multiple genetic etiologies in families unraveled by exome sequencing	D Gupta, SGRH, Delhi
0824 - 0830	Coexisting variants in complement regulatory genesIncreases sesceptibility to relapse in anti-factor H (FH) associated atypical hemolytic uremic syndrome (aHUS)-	P Khandelwal, AIIMS, Delhi

0830 - 0836	Utility of NGS Based Exome Testing as the first line of diagnosis for Congenital Disorders of Glycosylation	LS Matsa, Medgenome,Bengaluru
Session 5: Finding New Genes Chairpersons:Dr. Renu Saxena, Koumudi Godbole, Ashwin Dalal		
0840 - 0900	Genetic basis of three new recessive skeletal dysplasias	Girisha KM, Manipal
0900 - 0930	Intellectual disability - novel genes and mechanisms	May Malicdan, USA
0930 - 1000	Genomic approach to Undiagnosed Liver Disease	Silvia Vilarinho, Yale, USA
Session 6: Policy Needs, Current Status and Plans in Rare Disorders Chairpersons: Madhulika Kabra, Ashok Gupta, Vijay Chandru		
1000 - 1020	Rare disorders policy in India and its implementation	Himanshu Chauhan, India
1020 - 1040	International trends in Rare disorders policy & orphan drugs act	Stephen Groft, USA
1040 - 1100	Drug Discovery for Rare Disorders	Sam Santhosh, USA
1100 - 1130	**Tea/Coffee and Posters**	
Session 7: UDNI Inaugural Session: Welcome and Reports		
1130 - 1140	Lamp Lighting	
1140 - 1145	Welcome	Ishwar C Verma, Delhi
1145 - 1200	Roadmap for UDN-India	Ratna D Puri, Delhi
1200 - 1210	Address, Chief Guest	Dr Balram Bhargava Secretary, Department of Health Research, Director General, ICMR
1210 - 1220	Address, Guest of Honour	Dr S Venkatesh, Director General of Health Services
1220 - 1230	Vote of Thanks	Sunita Bijarnia- Mahay, Delhi
1230 - 1235	Welcome	Helene Cederroth, Wilhelm Foundation, Sweden
1235 - 1300	The UDNI: Vision and Way Forward	William A Gahl, National Institute of Health USA
1300 - 1400	**Lunch Break and Posters**	
1400 - 1415	**Meeting Goals**	William A Gahl, USA
	• List of Official UDNI Members, Board Members • Committee/Working Group Reports • Confirm Data Sharing Policies • Discuss Science and Cases • Address Future of UDNI • Vote on Charter, Appendices, BOD proposals • Voting Eligibility and Rules	
1415 - 1615	Committee & Working Group Reports	
1415 - 1430	Membership	Eric Klee, Mayo Clinic, USA
1430 - 1445	Program	Ratna D Puri, William A Gahl
1445 - 1450	Communications	Domenica Taruscio, Italy
1500 - 1530	• Functional Research • UW Infrastructure • Questionnaire • Website, UDN Genes, Model Match Maker • Rare Diseases Models & Mechanisms	Eric Klee, Mayo Clinic S.Meyn, U.Wisconsin, USA May Malicdan, NHGRI, USA S. Yamamoto, Baylor, USA Paul Lasko, Canada
1530 - 1545	Patient Engagement	V. Boulanger, NORD USA
1545 - 1615	Data Sharing (Current cases, # shared, email listserv, video on Phenome Central)	David Adams, USA Michael Brudno, Canada Tudor Groza, Australia
1615 - 1645	**Tea and Posters**	
1645 - 1730	Solved Cases; 8 minutes each (5 + 3)	
	TOP3A mitochondrial disease	D. Novacic
	Myofibrillar myopathy	B. Korf
	IGHMBP2 mutation and Charcot-Marie-Tooth 25	T.A. Cassini
	EXTL3-CDG and craniosynostosis	V. Arora
	IRF2BPL autistic spectrum; *DGAT1* & enteropathy	E. Lopez-Martin
	Robinow syndrome and Brachydactyly	R. Mishra
1830 onwards	**Dinner for UDNI members and Invitees (Wilhelm Foundation)**	

Day 3, April 15th 2019		
0800 - 0830	**Oral Abstracts** **Chairpersons: David Adams, Madhulika Kabra, Kuldeep Singh**	
0800 - 0806	The Initiation of Korean Undiagnosed Diseases Program: One-year experience	SY Kim, South Korea
0806 - 0812	Assessing the Clinical Utility of Exome Sequencing for Diagnosis of Inherited White Matter Disorders in a Cohort of Seventy-three Families from India	A Shukla, Manipal
0812 - 0818	Clinical genomics outcome of reanalyzed genomic datasets	S Kulshrestha, SGRH, Delhi
0818 - 0824	Genomics and other Omics tools for Enabling Medical Decisions (GOMED): Equitable access to Genomic Medicine	A Mathur, CSIR-IGIB, Delhi
0824 - 0830	A yet unsolved enigma of enchondromatosis- Report of three cases	R Kaur, AIIMS, Delhi
Session 8: UDNI Translational Research **Chairpersons: Radha Rama Devi, G Chandak, Sunita Bijarnia Mahay**		
0840 - 0910	New Therapies in Management of Gaucher Disease	Ari Zimran, Israel
0910 - 0930	Model Systems to Accelerate Drug Discovery	Sridhar Sivasubbu, India
0930 - 0945	National Newborn Screening Program for 40 rare metabolic diseases in Italy	Domenica Taruscio, Italy
0945 - 0955	WXNC and Genomics Medicine Ireland	C. Waters, WuXiNextCode
0955 - 1010	Mitochondrial augmentation therapy	Yair Anikster, Israel
1010 - 1020	Rare Disease Web Forum	Anil Raina, Sanofi Genzyme
1020 - 1050	**Tea/Coffee and Posters**	
	Cases, Reports	
1050 - 1210	Mystery Cases; 8 minutes each (5 + 3)	
	Suspected progeroid child	D. Hettiaracchchi
	Undiagnosed skin disorder	B. Korf
	Global delays, dysmorphisms	M. Lallar
	Short stature, microcephaly, delays, contractures	R. Puri
	Ataxia, vomiting after meals, seizures	V. Nagy
	Orbital neurofibromas, facial dysmorphism	D. Babovic-Vuksanovic
	Ectodermal dysplasia; spastic paraplegia & delays	G. Landoure
1210 - 1230	IRDiRC & Eur Joint Program on Rare Diseases	Daria Julkowska
1230 - 1240	Solve-RD/EURORDIS Community Engagement Task Force	Virginie Bros - Facer
1240 - 1250	DART: Diagnostic Annotation and Reporting Tool	Matthias Haimel, Vienna
1250 - 1305	Update from Italian Telethon UDP	G. Casari, V. Nigro
1305 - 1400	**Lunch Break**	
Session 9: UDNI: Going Forward		
1400 - 1500	**Business Discussion** -Ratify Charter, Appendices, Database -Confirm New Committee Members -Discuss Other Working Groups -Patient Recruitment -Next Meeting -ERN for Undiagnosed -Other Topics, Announcements	William A Gahl
1500 - 1545	**Reaching Out**	
1500 - 1515	Indigenous Populations	C. Poulton, Australia
1515 - 1530	Genetic Counseling	S. Broley, Australia J. Lewis, USA
1530 - 1545	Developing Countries	Madhulika Kabra, Delhi
1545 - 1615	**Tea/Coffee Break**	
1615 - 1700	Sustainability	William A Gahl
	Philanthropy	Paul Lasko, Canada
	COST Application, EJPRD, National Reps?	Audience
1700 - 1730	Other topics	
1730	Valedictory Program	
1830 onwards	**Dinner for UDNI members and invitees (Wilhelm Foundation)**	