Manipal Genetics Update VII

Cellular and Animal Models for Rare Genetic Diseases

January 18-20, 2024

Scientific Program

Day 1: January 18, 2024		
Time	**Topic**	**Speaker**
08:00AM-08:55AM	Registration	
08:55AM-09.00AM	Welcome note	Anju Shukla
Session I **Chairpersons**: Akhilesh Pandey, Gaurav Varshney, William Newman		
09:00AM-09.30AM	Rare disease research in India	Ashwin Dalal
09:30AM-10:00AM	Mechanisms underlying autosomal dominant leukodystrophy: Lessons from cell culture and models	Quasar S. Padiath
10:00AM-10:30AM	Progress for gene therapy in the leukodystrophies	Florian Eichler
10:30AM-11:00AM	Aicardi-Goutieres syndrome: Translational therapeutics	Adeline Vanderver
11:00AM-11:30AM	**Tea break**	
Session II **Chairpersons**: Prajnya Ranganath, Cecilia Lo, May Christine Malicdan		
11:30AM-12:00PM	Mass spectrometry in investigation of genetic disorders	Akhilesh Pandey
12:00PM-12.30PM	Inherited genetic-metabolic defects and epilepsy: Interfacing genes and biochemical pathways	Asuri N Prasad
12:30PM-01.00PM	*In silico, in vitro,* and *in vivo:* Combining multiple approaches to study candidate genes for monogenic disorders	Tess Holling
01.00PM-02:00PM	**Lunch break**	
Session III **Chairpersons:** Meenal Agarwal, Robert Taylor, Pavan Agrawal		
02:00PM-02:30PM	Modeling primary microcephaly with human brain organoids reveal fundamental roles of CIT kinase activity	Stephanie Bielas
02:30PM-03:00PM	Probing the mechanisms of chromatinopathies in brain development using human *in vitro* disease model	Bhavana Muralidharan
03:00PM-03:30PM	Optimized CRISPR tools for high-throughput functional genomics using zebrafish as a model	Gaurav Varshney
03:30PM-04:00PM	Patient-derived primary fibroblasts to study the pathophysiology of rare monogenic diseases: Possibilities and what we have learned	Frederike Leonie Harms
04:00PM-04:30PM	**Inauguration followed by conference photo**	
04:30PM-06:00PM	**Poster session I and tea break**	
07:00PM onwards	**Cultural program and gala dinner**	

Day 2: January 19, 2024		
Time	**Topic**	**Speaker**
Session IV **Chairpersons**: Kishore Parsa, Amita Moirangthem		
09:00AM-09:45AM	Advances in the molecular diagnosis of mitochondrial genetic diseases	Robert Taylor
09:45AM-10:30AM	Perrault syndrome: Still more to discover?	William Newman
10:30AM-11:00AM	**Tea break**	
Session V **Chairpersons**: S J Patil, Rakesh Mishra, Quasar S. Padiath		
11:00AM-11.30AM	Model organism screening in undiagnosed disease	Michael Francis Wangler
11:30AM-12:00PM	The road less travelled: Insights into the genetic landscape of congenital heart disease with mouse forward genetics	Cecilia Lo
12:00PM-12:30PM	Utilizing Murine Models to Unravel Pathophysiology and Therapeutics in Rare Disease Research	May Christine Malicdan
12:30PM-01:30PM	**Lunch break**	
Session VI **Chairpersons**: Stephanie Bielas, Michael Francis Wangler, Sheetal Sharda		
01:30PM-02:00PM	Molecular genetic analysis of a rare neurodevelopmental syndrome	Udai Bhan Pandey
02:00PM-02:30PM	Human-fly collaborative pipeline for diagnosis of rare genetic diseases in India	Manish Jaiswal
02:30PM-03:00PM	Modeling of rare haematological diseases using iPSCs	Shaji R V
03:00PM-04:30PM	**Poster Session II and Tea break**	
Session VII: Short talks **Chairpersons**: Udai Bhan Pandey, Girish Ratnaparkhi, Swadhin Chandra Jana		
04:30PM-04:50PM	Functional genomics of pediatric heart disease: Hypertrophic cardiomyopathy	Dhandapany Perundurai
04:50PM-05:10PM	Reproductive dysfunction in a zebrafish model of Fragile X syndrome	Aarti Sevilimedu
05:10PM-05:30PM	Role of *IER3IP1*, causative of microcephaly, epilepsy, and diabetes syndrome-1, in vesicular trafficking	Sonal Nagarkar Jaiswal
05:30PM-05:50PM	Nuclear mitochondria crosstalk associated with mitochondria DNA maintenance and biogenesis- *in vitro* and *in vivo* models	Sanjiban Chakrabarty
05:50PM-06:10PM	Overview of MPS II: Challenges and Management	Sheela Nampoothiri
07:30PM onwards	**Faculty dinner**	

Day 3: January 20, 2024		
Time	**Topic**	**Speaker**
Session VIII **Moderators**: Ashwin Dalal, Ratna D Puri, Aarti Sevilimedu		
09:00AM-10:00AM	Oral presentations	
10:00AM-10:30AM	From cutting edge science to policy: Navigating the landscape of rare genetic diseases in India	Rakesh Mishra
10:30AM-11AM	**Tea break**	
Session IX: Short talks **Chairpersons**: Shagun Aggarwal, Kiranam Chatti, Florian Eichler		
11:00AM-11:20AM	Investigating vesicular trafficking pathways in Griscelli syndrome	Ravi Manjithaya
11:20AM-11:40AM	Skeletal ciliopathies: Old and new players	Priyanka Upadhyai
11:40AM-12:00PM	Intriguing ribosomopathies: What have we learnt from zebrafish?	Anirban Chakraborty
12:00PM-12:20PM	Bi-allelic variants in *CSMD1* are implicated in a neurodevelopmental disorder with intellectual disability and variable cortical malformations	Anshika Srivastava
12:20PM-12:40PM	Modeling a spectrum of early-onset human developmental brain disorders: timing, mechanism, and therapy	Achira Roy
12:40PM-01:00PM	The VAPB social network in ALS: A versatile influencer at ER-membrane contact sites	Girish Ratnaparkhi
01:00PM-02:00PM	**Lunch Break**	
Session X: Sponsor talks **Chairperson**: Anupriya Kaur, Amita Moirangthem		
02:00PM-02:30PM	Current and innovative therapies in the management of plexiform neurofibromas	AstraZeneca
02:30PM-03:00PM	Sponsor talk	MedGenome
03:00PM onwards	**Valedictory**	