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**
08:30AM onwards	Registration	
08:55AM-09.00AM	Welcome note	Anju Shukla
	Session I	
09:00AM-09.30AM	Rare disease research in India	Shubha Phadke (SGPGIMS, India)
09:30AM-10:00AM	Mechanisms underlying autosomal dominant leukodystrophy: Lessons from cell culture and models	Quasar S. Padiath (University of Pittsburgh, USA)
10:00AM-10:30AM	Progress for gene therapy in the leukodystrophies	Florian Eichler (Massachusetts General Hospital, USA)
10:30AM-11:00AM	Aicardi-Goutieres syndrome: Translational therapeutics	Adeline Vanderver (Children's Hospital of Philadelphia, USA)
11:00AM-11:30AM	**Tea break**	
	Session II	
11:30AM-12:00PM	Mass spectrometry in investigation of genetic disorders	Akhilesh Pandey (Mayo Clinic, USA)
12:00PM-12.30PM	Inherited genetic-metabolic defects and epilepsy: Interfacing genes and biochemical pathways	Asuri N Prasad (Western University, Canada)
12:30PM-01.00PM	*In silico, in vitro,* and *in vivo:* combining multiple approaches to study candidate genes for monogenic disorders	Tess Holling (Institute of Human Genetics, Germany)
01.00PM-02:30PM	**Lunch and Poster Session I**	
	Session III	
02:30PM-03:00PM	Modeling primary microcephaly with human brain organoids reveal fundamental roles of CIT kinase activity	Stephanie Bielas (University of Michigan, USA)
03:00PM-03:30PM	Molecular genetic analysis of a rare neurodevelopmental syndrome	Udai Bhan Pandey (University of Pittsburgh, USA)
03:30PM-04:00PM	Optimized CRISPR tools for high-throughput functional genomics using zebrafish as a model	Gaurav Varshney (OMRF, USA)
04:00PM-04:30PM	Patient-derived primary fibroblasts to study the pathophysiology of rare monogenic diseases: Possibilities and what we have learned	Frederike Leonie Harms (Institute of Human Genetics, Germany)
06:00PM onwards	**Cultural program, inauguration, and dinner**	

Day 2: January 19, 2024		
Time	**Topic**	**Speaker**
	Session IV	
08:30AM-09:00AM	Advances in the molecular diagnosis of mitochondrial genetic diseases	Robert Taylor (Newcastle University, UK)
09:00AM-09:30AM	Ultra-rapid functional genomics for the diagnosis of mitochondrial disorders	David Arthur Stroud (University of Melbourne, Australia)
09:30AM-10:00AM	Solving medical mysteries with fruit flies: the role of mitochondria in rare pediatric neurodegeneration	Debdeep Dutta (Baylor College of Medicine, USA)
10:00AM-10:30AM	Perrault syndrome: still more to discover?	William Newman (University of Manchester, UK)
10:30AM-11:00AM	**Tea break**	
	Session V	
11:00AM-11.30AM	Model organism screening in undiagnosed disease	Michael Francis Wangler (Baylor College of Medicine, USA)
11:30AM-12:00PM	The road less travelled: insights into the genetic landscape of congenital heart disease with mouse forward genetics	Cecilia Lo (University of Pittsburgh, USA)
12:00PM-12:30PM	Utilizing Murine Models to Unravel Pathophysiology and Therapeutics in Rare Disease Research	May Christine Malicdan (National Institute of Health, USA)
12:30PM-02:00PM	**Lunch and Poster Session II**	
	Session VI	
02:00PM-02:30PM	Probing the mechanisms of chromatinopathies in brain development using human in vitro disease model	Bhavana Muralidharan (InStem, India)
02:30PM-03:00PM	Human-fly collaborative pipeline for diagnosis of rare genetic diseases in India	Manish Jaiswal (TIFR, India)
03:00PM-03:30PM	Modeling of rare haematological diseases using iPSCs	Shaji R V (CMC, India)
03:30PM-04:00PM	**Tea break**	
	Session VII: Short talks	
04:00PM-04:20PM	Functional genomics of pediatric heart disease: Hypertrophic cardiomyopathy	Dhandapany Perundurai (InStem, India)
04:20PM-04:40PM	Bi-allelic variants in *CCN2* cause a novel skeletal dysplasia	Aarti Sevilimedu (DRILS, India)
04:40PM-05:00PM	Role of *IER3IP1*, causative of microcephaly, epilepsy, and diabetes syndrome-1, in vesicular trafficking	Sonal Jaiswal (CCMB, India)
05:00PM-05:20PM	Nuclear mitochondria crosstalk associated with mitochondria DNA maintenance and biogenesis-in vitro and in vivo models	Sanjiban Chakrabarty (MSLS, India)
05:20PM-05:40PM	Sponsor talk# 1	Takeda
07:00PM onwards	**Faculty dinner**	

Day 3: January 20, 2024		
Time	**Topic**	**Speaker**
	Session VIII	
09:00AM-10:00AM	Oral presentations	Moderator: Ashwin Dalal (CDFD, India)
10:00AM-10:30AM	From cutting edge science to policy: Navigating the landscape of rare genetic diseases in India	Rakesh Mishra (TIGS, India)
10:30AM-11AM	**Tea break**	
	Session IX: Short talks	
11:00AM-11:20AM	Investigating vesicular trafficking pathways in Griscelli syndrome	Ravi Manjithaya (JNCASR, India)
11:20AM-11:40AM	Skeletal ciliopathies: Old and new players	Priyanka Upadhyai (KMC, Manipal, India)
11:40AM-12:00PM	Intriguing ribosomopathies: What have we learnt from zebrafish?	Anirban Chakraborty (NITTE University, India)
12:00PM-12:20PM	Bi-allelic variants in *CSMD1* are implicated in a neurodevelopmental disorder with intellectual disability and variable cortical malformations	Anshika Srivastava (SGPGIMS, India)
12:20PM-12:40PM	Modeling a spectrum of early-onset human developmental brain disorders: timing, mechanism, and therapy	Achira Roy (JNCASR, India)
12:40PM-01:00PM	The VAPB social network in ALS: A versatile influencer at ER-membrane contact sites	Girish Ratnaparkhi (IISER, India)
01:00PM-02:00PM	**Lunch**	
	Session X	
02:00PM-02:30PM	Current and Innovative Therapies in the management of NF1 with Plexiform Neurofibromas	AstraZeneca
02:30PM-03:00PM	Sponsor talk# 3	To be decided
03:00PM onwards	**Valedictory**	