



INDO-UK GENETIC EDUCATION FORUM

4<sup>th</sup> International Conference on Birth Defects (ICBD-2018)

&  
5<sup>th</sup> Annual Conference of the Society of  
Indian Academy of Medical Genetics

12<sup>th</sup> - 15<sup>th</sup> December, 2018

Scudder Auditorium, Christian Medical College, Vellore, Tamil Nadu (India)



## Conference Program

Day 1: 12 <sup>th</sup> December 2018 (Wednesday)		
Time	Topic	
17:30 - 19:00	Registration	
19:00 Onwards	Welcome/Social Evening with Delegates/ Faculty	
Day 2: 13 <sup>th</sup> December 2018 (Thursday)		
Time	Topic	Chairperson
08:30 Onwards	Registration	
09:00-10:00	<b>Welcome / Inauguration</b> Dr. Sumita Danda / Prof. Dhavendra Kumar	
<b>10:00-10:30</b>	<b>Tea/Coffee</b>	
10:30-11:30	<b>KEY NOTE LECTURE I:</b> <b>Global Perspectives of Congenital Malformations</b> Prof. Dian Donnai, Manchester, UK	Dr. Shubha Phadke Dr. M Naveed
11:30-13:30	<b>SESSION I: PHENOTYPES IN BIRTH DEFECTS</b> <b>Deciphering the Birth Defects Phenotypes</b> Dr. Helen Stewart, Oxford, UK <b>Neuromuscular Phenotypes in Clinical Neurogenetics</b> Dr. Mark Rogers, Cardiff, UK	Dr. Maya Thomas Dr. Siddharth Banka Dr. Prajnya Ranganath
	<b>Neuroimaging Phenotypes in Clinical Genetics/Genomics</b> Dr. Mohnish Suri, Nottingham, UK <b>Cardiovascular Phenotypes in Clinical Genetics/Genomics</b> Prof. Dhavendra Kumar, Cardiff, UK	Dr. Sridhar Gibikote Dr. Meenakshi Bhat Prof. Eamonn Maher
	<b>13:30-14:15</b>	<b>Lunch</b>
14:15-14:30	<b>SESSION II:</b> <b>Dr SS Agarwal Young Scientist Award</b> <b>Recipient:</b> Dr. Anju Shukla, Associate Professor, Department of Medical Genetics, Kasturba Medical College, Manipal	Prof. Dian Donnai Prof. Madhulika Kabra Dr. Helen Stewart Dr. Koumodi Godbole
14:30-15:20	<b>ORAL ABSTRACTS (4)</b> Mr Somashekar PH, Dr. Ranjana Mishra, Dr. Mallikarjun R Patil, Dr. Beena Suresh	
<b>15:20-16:00</b>	<b>Poster Session with Tea/Coffee</b>	
16:00-16:30	<b>SESSION III: GENOTYPES IN BIRTH DEFECTS</b> <b>One Genotype - Diverse Phenotypes</b> Lead: Dr. Siddharth Banka, Manchester, UK	Dr. Nicola Poplawski Dr. Ashutosh Halder Dr. Ashwin Dalal
16:30-17:30	<b>ORAL ABSTRACTS (5)</b> Ms Parneet Kaur, Dr, VS Vineeth, Dr, A Suneetha Susan, Dr, Stephanie Bielas, Dr Veronica Arora	
17:30-18:30	<b>KEYNOTE LECTURE II:</b> <b>Genomics of Birth defects: Congenital Heart Defects and mendelian disorders</b> Dr. Shrikant Mane, Yale University School of Medicine, USA	Dr Dhavendra Kumar Dr Neerja Gupta Dr Ratna Puri
<b>19:30 Onwards</b>	<b>Faculty Dinner, Big Bungalow, CMC Vellore</b>	

**Day 3: 14<sup>th</sup> December 2018 (Friday)**

<b>Time</b>	<b>Topic</b>	<b>Chairperson</b>
08:30-09:15	<b>KEY NOTE LECTURE:</b> <b>Molecular genetic investigation of foetal structural anomalies</b> Prof. Eamonn Maher, Cambridge	Dr. Shagun Aggarwal Dr. Sheela Nampoothiri Dr. Manisha Beck
09:15-10:00	<b>QUIZ on RARE Diseases:</b> Dr. Krati Shah & Dr. Beena Suresh, Dr. Divya P, Dr. Atanu K Dutta	
<b>GROUP PHOTO</b>		
10.00-11.00	<b>THE DR. SS AGARWAL ORATION</b> Prof. Dhavendra Kumar, Cardiff, UK	<b>Moderator:</b> Prof. Madulika Kabra, President SIAMG, India
<b>11.00-11.40</b>	<b>Poster Session with Tea/Coffee</b>	
11.40-12.45	<b>SESSION IV: EXPANDING PHENOTYPES OF MONOGENIC BIRTH DEFECTS; NOVEL FINDINGS FROM NGS (INDIAN SCENARIO)</b> [10mins presentations] 1. Skeletal Dysplasia - Dr. Neerja Gupta 2. Intellectual Disability - Dr. Ratna Puri 3. Dysmorphology - Dr. Prajnya Ranganath 4. Rasopathy - Dr. Sheetal Sharda 5. Fetal/prenatal - Dr. Shagun Aggarwal <b>Question Answer 15mins</b>	Dr. Mohnish Suri Dr. Kuldeep Singh Dr. Inusha Panigrahi Dr. Sankar V
12.45- 13.45	<b>Panel Discussion: Sustainable Access to Treatment : Changing Indian Scenario-Thalassemia / Haemophilia / NBS / LSD</b> Panellist from state wise centre 1. Uttar Pradesh - Prof. Shubha Phadke 2. New Delhi - Dr. Seema Kapur / Dr. Sunita Bijarnia 3. Kerala – Dr. Sankar V / Dr. Sheela Nampoothiri 4. Tamilnadu - Dr. S Danda / Dr. Srilakshmi R 5. Hyderabad – Dr. Radha Rama Devi 6. Karnataka – Dr. Meenakshi Bhat (5 mins presentation from each centre followed by comments of moderators and discussion)	<b>Moderators:</b> Prof. Madhulika Kabra Prof. Shubha Phadke
<b>13:45-14:30</b>	<b>Lunch</b>	
14:30-16:15	<b>SESSION V: NEOPLASIA IN CLINICAL DYSMORPHOLOGY</b> <b>Childhood Ocular Tumours - The Paradigm of Retinoblastoma</b> Dr. Helen Stewart, Oxford, UK <b>Neoplasia In Neurofibromatosis and Other Neuro-Cutaneous Disorders</b> Dr. Nicola Poplawski, Adelaide, Australia <b>Complexities Of Wilm's Tumour predisposition: The Beckwith-Wiedemann Syndrome spectrum</b> Prof. Eamonn Maher, Cambridge, UK	Dr. Shirley Hodgson Dr. Abraham Peedicayil Dr. Renu George Dr. Atanu K Dutta
<b>16:15-16:30</b>	<b>Tea/Coffee</b>	
16:30-17:30	<b>Dr. IC Verma Outstanding Researcher Award</b> <b>Recipient:</b> Dr. Radha Rama Devi	<b>Video conference</b> <b>Moderator:</b> Dr. Mohnish Suri
17:30-18:30	<b>SESSION VI: GENOMIC PERSPECTIVES OF GROWTH DISORDERS</b> <b>Phenotype-Genotype Correlation In Obesity Syndromes</b> Dr. Mieke van Haelst, Amsterdam, The Netherlands <b>Genetic Heterogeneity in Short Limbed Dysplasias</b> Dr. Charu Deshpande, London, UK <b>Monitoring of IUGR prenatally</b> Dr. Antoni Borell, Barcelona, Spain	Dr. Helen Stewart Dr. Anna Simon Dr. Sheela N Dr. Vrisha Madhuri Dr. Reeta Selvi Dr. S J Patil
18:30-19:15	<b>Industry in management of birth defects</b> Dr. Ramprasad VL, Mr. Anil Raina, Dr. Jayesh Sheth	Dr. Ashwin Dalal Dr. Kausik Mandal
<b>19:15-19:30</b>	<b>Valedictory Function</b>	
<b>19:30 Onwards</b>	<b>Conference Dinner - Scudder Auditorium</b>	

# Post Conference Workshop

Day 4: 15 <sup>th</sup> December 2018 (Saturday)				
PARALLEL CLINICAL WORKSHOPS: CLINICAL CASES & INTERACTIVE SESSIONS				
CANCER GENETICS WORKSHOP				
	Speaker	Topic	Time	Chairperson / Discussants
<b>Welcome and Introduction - Dr Shirley Hodgson</b>				
9.00 - 10.15	Dr Shirley Hodgson	Familial Cancer Overview	30 mins	Dr Abraham Peedicayil Dr Patricia Dr Jeba K
	Dr Anitha Thomas	Case on Lynch syndrome (Gynae Onco)	7 mins + 3min discussion	
	Dr Atanu Dutta	Case on Biallelic Mismatch Repair Deficiency (bMMRD)	15 mins	
	Dr Alka Ekbote	Breast cancer- Genetics	20 mins	
10.15 - 10.45	Dr Mark Rogers	Polyposis Syndrome	25 mins	Dr CE Eapen Dr Mark Ranjan
<b>10.45-11.15</b>	<b>Coffee Break</b>			
11.15 - 11.45	Dr. Eamonn Maher	Renal Cell Cancer	25 mins	Dr Nicola Poplawski Dr Atanu Dutta
11.45 - 12.30	Dr Nicola Poplawski	Cancer In Neurocutaneous Disorders	25 mins	Dr Susanne Pulimood Dr Alka Ekbote
		Case presentation (Dermatology)	15 mins	
<b>12.30 - 15.30</b>	<b>Lunch</b>			
13.30- 14.30	Dr Helen Stewart	Retinoblastoma/ Eye Tumours	25 mins	Dr Saban Horo Dr P Sundaresan
		Case presentation (Ophthalmology)	(7+ 3)mins	
	Dr P Sundaresan	Glioblastoma Gene Therapy	25 mins	Dr Deepa John Dr Rikki
14.30-15.00	Dr Humphrey Hodgson	Hepato-Biliary & Pancreatic Cancers	20 mins	Dr C E Eapen Dr Thomas Ram
		Case presentation (Gastroenterology)	(7+ 3)mins	
<b>15.00 - 15.20</b>	<b>Coffee Break</b>			
15.20-15.50	Panel discussion	Future of Cancer Tracking by minimal invasive methods. <b>Panellists:</b> Dr Nicola Poplawski Dr Anita Thomas Dr Patricia	30 mins	<b>Moderators:</b> Dr Shirley Hodgson Dr Abraham
15.50- 16.00	Dr Shirley Hodgson	Concluding Remarks and Wrap up	10 mins	

## CURRENT TRENDS IN CARDIO VASCULAR GENETICS

Time	Speaker	Topic	Chairpersons
<b>Welcome note and Introduction: Dr Dhavendra Kumar &amp; Dr Viji Samuel</b>			
9.00-9.30	Dr K Thangaraj	Genetic Heterogeneity of Cardiovascular diseases in India	Dr Dhavendra Kumar Dr Viji Samuel
9.30-10.00	Dr Charu Deshpande	Cardiovascular Developmental Disorders Aortic And Arterial Diseases	
<b>10.00 - 10.30</b>	<b>Coffee Break</b>		
<b>Cardiac Muscle Diseases- Cardiomyopathy</b>			
<b>10.30 -12.30</b>	Dr SJ Patil	Syndromic hypertrophic Cardiomyopathy	Dr Thangaraj Dr Tessa Homfray
	Dr Viji Samuel	Non syndromic HOCM	
	Dr Anoop G	Dilated /Restrictive cardiomyopathy	
	Mrs Saroja	Case presentation -NGS testing in Cardiomyopathy	
<b>12.30 - 13.30</b>	<b>Lunch</b>		
13.30 -14.00	Dr John Roshan	Cardiac Conduction And Rhythm Disorders – Inherited arrhythmias case presentation	Dr David Chase Dr Charu Deshpande
14.00- 14.30	Dr Tessa Homfray	NGS diagnostic/ Cascade Testing And Surveillance For Inherited cardiac diseases Prevention	Dr Sunil Chandy Dr SJ Patil
14.30 -15.30	Dr Dhavendra Kumar	Sudden Cardiac Death- Issues and Protocols	
<b>Coffee and depart</b>			

<b>WORKSHOP ON GENETIC COUNSELLING</b>			
<b>Time</b>	<b>Speaker</b>	<b>Topic</b>	<b>Chairpersons</b>
9.00 - 9.30	Ms Lori Dean	Elements of Psychosocial Counselling as applied in Clinical practice	Dr Radha Saraswathy Dr Smita Rao
9.30 - 10.00	Dr Antoni Borrell	Counselling for pregnancy mishaps: impact of new technologies	Dr Shubha Phadke Dr Lori Dean
10.00 - 10.30	Dr Shagun Aggarwal	NIPT and genetic counselling (pre- and post-test)	Ms Niby Jacob Dr Gautham AU
10.30 - 11.00	Dr Smita Rao	Informed Consent and Counselling in Whole Exome Sequencing	Dr Annie Hasan Dr Anju Shukla
<b>11.00 - 11.20</b>	<b>Coffee Break</b>		
<b>11.20 - 12.30</b>	<b>Interactive session-Cases</b>		
11.20 - 11.30	SGPGIMS	Fetal anomaly on USG	Dr Shubha Phadke Dr Antoni Borrell Dr Annie Hasan
11.30 - 11.40	KMC	Counseling for cancer	Dr Shagun Aggarwal Dr Smita Rao
11.40 - 11.50	NIMHANS	Late onset diseases eg. HD	Dr Gautham AU Ms Lori Dean
11.50 - 12.00	CMC	Carrier testing for DMD	Dr Sumita Danda Ms Niby Jacob Dr Radha Saraswathy
12.00 - 12.10	NIMS	Karyotype abnormality in RSA/Infertility	Dr Shagun Aggarwal Dr Shubha Phadke
12.10 - 12.45	<b>Question Answers</b>		
<b>12.45 - 13.30</b>	<b>Lunch</b>		
<b>CME on Rare Diseases Introduction and questionnaire</b>			
<b>Time</b>	<b>Speaker</b>	<b>Topic</b>	<b>Chairpersons</b>
14:00 - 14:10	Dr Sony M	Case- Lysosomal Storage Disorder (LSD)	Dr Kuldeep Singh Dr Reena Gulati Dr Sanjeeva G
14:10 - 14:40	Dr Jayesh Sheth	Laboratory Parameters and approach to LSD	
14.40 - 14.50	Dr Krati Shah	Treating LSD- Experience & Challenges	
14:50 - 15.10	Dr Sangeetha Y	Case based approach -Homocystinuria	Dr Maya Thomas Dr Radha Rama Devi
15.10 - 15:30	Dr Karthik M	Case based approach - Cerebral Creatine deficiency	
15:30 - 15:40	Mr Prasanna Shirol	Parents Perspective - Rare Disease Policy	
<b>15.40 - 16.00</b>	<b>Coffee Break</b>		
16.00 - 16.30	Dr Kausik Mandal	Case- Dymorpholgy. Working up of a novel dysmorphology syndrome	Dr Kalpana Gowrishankar Dr Beena Koshy
16:30 - 17.00	Dr Ruchika Goel	Case based approach - Immunodeficiency syndromes	Dr Krati Shah Dr Debashish Danda