



# **Third International Conference on Birth Defects (ICBD-2016) & Third National Conference of the Society for Indian Academy of Medical Genetics (SIAMG)**

**"Molecular and Metabolic basis of Disorders of Morphogenesis "**

**7<sup>th</sup>-10<sup>th</sup> December 2016, JAWAHARLAL AUDITORIUM, AIIMS, New Delhi, India**

## **Agenda**



**Jointly Organized by**

**INDO-UK GENETIC EDUCATION FORUM**

**SOCIETY OF THE INDIAN ACADEMY OF MEDICAL GENETICS (SIAMG)**

**18 HOURS CME/CPD APPROVAL BY THE FEDERATION OF THE UK ROYAL COLLEGES,  
THE ROYAL COLLEGE OF PHYSICIANS, LONDON, UK (REF. 107208)**

INAUGURAL CEREMONY-7 <sup>TH</sup> DECEMBER 2016		
5.30 pm- 6.30pm	Welcome and opening	Dr. V K Paul Dr. Madhulika Kabra
	Ganesh Vandana Lamp Lighting	Performance by MUSKAN All Dignitaries
	Address by President, SIAMG Address by Chair, Indo UK education Forum Words of Wisdom Address by Guest of Honor, Director, AIIMS	Dr. Shubha R Phadke Dr. Dhavendra Kumar Dr. I C Verma Dr. M C Misra
	Address by Chief Guest	Dr. Soumya Swaminathan (DG ICMR & Secretary DHR, GOI)
6:30-7.00pm	Theme lecture 'Our genes, genome and birth defects'	Dr. Jill Clayton-Smith, Manchester, UK
	Vote of Thanks	Dr. Neerja Gupta
<b>7:30 pm onwards</b> <b>Welcome Dinner</b>		

# CONFERENCE PROGRAM

DAY-1: 8 <sup>TH</sup> DECEMBER 2016			
Time	Event	Speaker	Chairpersons
08:00am - 09:00am	Registration		
09:00am-09:15am	Welcome & opening	Dr. Madhulika Kabra, India	
09:15am-09:45am (25+5)	Birth Defects: The changing scene in India	Dr. I C Verma, India	Dr. Dhavendra Kumar, UK Dr. Neena Raina, India
09:45am-10:15am (25+5)	Plenary Lecture 1 Bridging the gap between Genetic Practice & Research - Opportunities and Challenges		Dr. V. K. Paul, India Dr. Jill Clayton-Smith, UK
10:15am-10:45am (25+5)	Plenary Lecture 2 A Child with small head- Genes or Zika virus		
10:45am - 11:30am	Tea/Coffee and Group Photograph		
11:30am-01:30pm	Session 1: Clinical Dysmorphology		
11:30am-12:00 noon (25+5)	Clinical approach to a child with multiple malformations	Dr. Victoria Murday, UK	Dr. Madhulika Kabra, India Dr. Peter Turnpenny, UK Dr Seema Kapoor, India
12:00 Noon- 12:30pm (25+5)	Molecular diagnostics in clinical dysmorphology- lessons from DDD study	Dr. Jill Clayton-Smith, UK	
12:30pm-01:00pm (25+5)	Molecular diagnostics in clinical dysmorphology-Indian experience	Dr. Shubha Phadke, India	
01:00pm-01:30pm	Dysmorphology cases session I	Interesting known and unknown cases (NIMS, SGRH, SGPGI & AIIMS)	Discussants: Dr. Sujatha Jagdeesh, India Dr. Siddharth Banka, UK Dr. S J Patil, India
01:30pm - 02:30pm	LUNCHEON Poster Walk		Dr. Ratna Dua Puri, India Dr. Bertde Vries, NL
02:30pm-03:00pm (25+5)	Management and Therapy of Malformation Syndromes: What's New??	Dr. Peter Turnpenny, UK	Dr. Radha Rama Devi, India Dr. Sumita Danda, India
03:00pm - 06:00pm	Session II: Molecular Dysmorphology-1		
03:00pm-03:20pm (15+5)	Chromatin Modeling disorders	Dr. Siddhartha Banka, UK	Dr. Ratna Dua Puri, India Dr. Bert de Vries, NL
03:20pm-03:40pm (15+5)	Disorders of Nuclear Membrane and Lamins	Dr. Pradeep Vasudevan, UK	
03:40pm - 04:00pm (15+5)	Genetics of Obesity Related Disorders	Dr. Mieke van Halest, Netherlands (Web telecast)	Dr. Meenakshi Bhat, India Dr. Pradeep Vasudevan, UK
04:00pm - 04:20pm (15+5)	Bardet Biedl Syndrome: Mutation Profiling and Genetic Diagnostics	Dr. S Sripriya, India	
04:20pm-05:00pm (35+5)	Dr. SS Agarwal Memorial Oration of SIAMG "Epigenetics in Malformation Syndromes"	Dr. Jill Clayton-Smith, UK	Dr. Shubha Phadke, President,IAMG, India Dr. Peter Turnpenny, Past President, CGS, UK
05:00pm - 05:30pm	Tea/Coffee		
05:30pm-06:00pm	Dysmorphology case session II	Interesting known and unknown cases Cases from SGPGI, NIMS/ CDFD, SGRH, MAMC	Discussants: Dr. Daniela Pilz, UK Dr. Charu Desh Pande, UK Dr. Seema Thakur, India
07:30pm onwards	Gala dinner at Rose Garden, India International Centre		

**DAY-2: 9<sup>TH</sup> DECEMBER 2016**

Time	Event	Speaker	Chairpersons
08:00am - 09:30am	Free paper presentations & Dr SS Agarwal Young Scientist Award for 2016	Ms. Shalini S Nayak, India	Dr. Daniella Pilz, UK Dr. Sumita Danda, India Dr. Prakash Gambhir, India Dr. Siddharth Banka,UK
09:30am - 10:00am (25+5)	<b>Plenary Lecture 3</b> 'Unraveling mysteries of intellectual disability by Next generation genome sequencing'	Dr. Bert de Vries, NL	Dr. Neerja Gupta, India Dr. Ashwin Dalal, India
<b>10:00am - 11:30am</b>	<b>Session-III: Molecular Dysmorphology-2</b>		
10:00am - 10:20am (15+5)	Protein kinases- RAS/MAPK; P13-AK/ MTOR	Dr. Peter Turnpenny, UK	Dr. Girisha Katta, India Dr. Kaushik Mandal, India Dr. Kuldeep Singh, India
10:20am - 10:40am (15+5)	Disorders of Filamin and related genes	Dr. Daniela Pilz, UK	
10:40am - 11:00am (15+5)	RAS MAPK disorders - Experience from India	Dr. Meenakshi Bhat, India	
11:00am-11:30am (25+5)	Prof. IC Verma Research Award	Dr. Shubha R Phadke, India	Dr. IC Verma, India Dr. Jill Clayton Smith, UK Dr. Dhavendra Kumar, UK
<b>11:30am - 12:00 Noon</b>	<b>Tea/coffee</b>		
<b>12:00 noon - 01:30pm</b>	<b>Session IV: Metabolic basis of Malformations-1</b>		
12:00 noon - 12:20 pm (15+5)	Clinical heterogeneity in Cholesterol Biosynthesis - The Opitz syndromes	Dr. Pradeep Vasudevan, UK	Dr. Anil Jalan, India Dr. S J Patil, India Dr. Sunita Bijarnia, India
12:20pm - 12:40pm (15+5)	Metabolic dysmorphic syndromes- Diagnosis to Care	Dr. Charu Deshpande, UK	
12:40pm-01:00pm (15+5)	Incorporating NGS into diagnosis of IEMs	Dr. Siddharth Banka, UK	
01:00pm-01:30pm (25+5)	Dysmorphology case session III	Interesting known and unknown cases (SGPGI, CMC, Vellore, MAMC, NIMS,SGRH)	<b>Discussants:</b> Dr. Victoria Murday, UK Dr. Girisha Katta, India Dr. Sankar V H, India
<b>01:30pm - 02:30pm</b>	<b>LUNCHEON Poster Walk</b>		Dr. Kaushik Mandal, India Dr. Kuldeep Singh, India Dr. Peter Turnpenny, UK
<b>02:30pm - 04:30pm</b>	<b>Session V: Metabolic basis of Malformations- 2</b>		
02:30pm-02:50pm (15+5)	Cardiac phenotypes of Inherited Errors Metabolism	Dr. Dhavendra Kumar, UK	Dr. Seema Kapoor, India Dr. Sunita Bijarnia, India Dr. Ashwin Dalai, India
02:50pm-03:10pm (15+5)	Congenital disorders of glycosylation - Indian experience	Dr. Anil Jalan, India	
03:10pm-03:30pm (15+5)	Malformations in Mitochondrial genetic disorders	Dr. Charu Deshpande, UK	Dr. Radha Rama Devi, India Dr. Sheela Nampoothiri, India
03:30pm-03:50pm (15+5)	Vitamin responsive IEMs	Dr. Siddhartha Banka, UK	
03:50pm-04:10pm (15+5)	Coarse Facies-All is not MPS	Dr. Sujatha Jagdeesh, India	
<b>04:10pm-04:30pm</b>	<b>Tea/coffee</b>		
<b>04:30pm - 05:30pm</b>	<b>Session VI: Phenotyping-A mandatory tool</b>		
04:30pm - 04:50pm (15+5)	Getting Charged from CHARGE syndrome	Dr. Chetana Sachidanandan, India	Dr. Madhulika Kabra, India Dr. Sridhar Sivasubbu, India
04:50pm-05:10pm (15+5)	Solving the puzzle between face and bones	Dr. Sheela Nampoothiri, India	
05:10pm-05:30pm (15+5)	The value of phenotyping in the Era of Genomics	Dr. Bert de Vries, NL	
<b>05:30pm onwards</b>	<b>Valedictory Function</b>		