





Proudly present

Early Diagnosis of Biochemical Genetic Disorders Saves Children's Lives:

A Discussion on the Merits and Process to Successfully Implement Newborn Screening

> Wednesday 24 October 2018 9:00 a.m. – 3:30 p.m.

> > Vivanta-by-Taj, Goa, India

PROGRAM

Activities

Session Moderator:

Michael Bennett, PhD; Children's Hospital of Philadelphia and University of Pennsylvania; AACC Past President

8:00 – 9:00 a.m.	Registration
9:00 – 9:10 a.m. (10')	Welcome
Dr. Michael Bennett, PhD	AACC Past President
9:10 – 9:55 a.m. (45') Dr. Bennett	Global situation for newborn screening
9:55 – 10:40 a.m. (45')	Presentation to identify the size of the problem and the disease
Dr. Seema Kapoor / Prof.	patterns in the Indian population
Damodaran Vasudevan	
10:40 – 10:50 a.m. (10')	Break
10:50 – 11:20 a.m. (30')	Working to ensure accurate and reliable laboratory testing in
Dr. Carla Cuthbert, PhD	newborn screening: Activities of the U.S. Centers for Disease Control and Prevention in supporting Laboratory Quality.
11:20 – 11:50 a.m. (30')	Practical considerations for setting up a laboratory for newborn
Dr. Bennett	screening. Requirements to provide a complete service covering
	all newborns in the country. Quality control issues and reporting
44-50 40:00 m m (401)	requirements.
11:50 – 12:30 p.m. (40') Mr. Thomas Mookken	What we can learn from Goa State newborn screening program
12:30 – 1:30 p.m. (60')	Lunch
1:30 – 2:00 (30')	Case Reports Presentation
Dr. Kannan Vaidyanathan	 India Example: A family with two infants who succumbed to maternal hyperphenylalaninemia who were later diagnosed to have PKU
Dr. Vaidyanathan	 India Example: A case of a child with Maple Syrup Urine Disease (MSUD)
Dr. Bennett	 US Example: Excellent outcome for a patient diagnosed as a newborn with phenylketonuria (a good outcome as a result of screening
2:00 – 3:30 p.m. (90')	Panel Discussion
 Panelists: Dr. Bennett Dr. Cuthbert Dr. Saurabh Dani Dr. Kapoor Mr. Mookken Dr. Vaidyanathan Prof. Vasudevan 	(The goal of the roundtable is to come away with a plan for the future.)

BIOGRAPHIES

MICHAEL J. BENNETT, PHD, FRCPATH, DABCC, FACB



Dr. Michael J. Bennett is professor of Pathology and Laboratory Medicine at the University of Pennsylvania Perelman School of Medicine, chief of Laboratory Medicine and director of the Metabolic Disease laboratory at The Children's Hospital of Philadelphia. He holds the Evelyn Willing Bromley Endowed Chair in Clinical Laboratories and Pathology. He obtained his Ph.D. at the University of Sheffield, UK in the field of medical enzymology. Dr Bennett is past chair of the Pediatric and Maternal-Fetal and Proteomics and Metabolomics Divisions of AACC, He is past president of the National Academy of Clinical Biochemistry, past treasurer of AACC and immediate past-president of AACC. He is an associate editor for Clinical

Chemistry Journal, Annals of Clinical Biochemistry and sits on the editorial boards of Journal of Inherited Metabolic Diseases and Molecular Genetics and Metabolism. Dr Bennett's research activities include the use of mass spectrometry in the investigation of inborn errors of mitochondrial energy metabolism with a special emphasis on disorders of fatty acid metabolism. He has published over 290 original peer-reviewed articles and 50 book chapters in the field of pediatric clinical chemistry and inherited metabolic diseases. Dr. Bennett also carries out research on a rare untreatable neurodegenerative disorder of children called Batten disease.

CARLA D. CUTHBERT, PHD, FCCMG, FACMG

Dr. Carla D. Cuthbert is the chief of the Newborn Screening and Molecular Biology Branch (NSMBB) in the Division of Laboratory Sciences, National Center for Environmental Health, Centers for Disease Control and Prevention. She has held this position since December 2009. Dr. Cuthbert gives leadership and oversight to NSMBB, a branch that comprises several laboratories and services which support both domestic and international newborn screening programs by providing quality assurance materials, public health service and technical expertise, test development and translational research activities.



She is the ex-officio CDC representative on the U.S. Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. Dr. Cuthbert is also co-chair of the Interagency Coordinating Committee on Newborn and Child Screening which provides input to the U.S. Secretary of the Department of Health and Human Services on national newborn screening issues. Dr. Cuthbert is board-certified in Clinical Laboratory Biochemical Genetics from both the American College of Medical Genetics and Genomics and the Canadian College of Medical Geneticists.

DR. SAURABH DANI, FCPS, DGO, MBBS



Dr. Saurabh Dani is a practicing obstetrician and gynecologist with special interest in High Risk Pregnancy and Newborn Screening. Currently, he is practicing at Ankur Hospital, Cloudnine and Namaha Healthcare in Mumbai. He is the secretary of the Indian Society for Prenatal Diagnosis & Therapy (ISPAT), president-elect of the Association of Fellow Gynecologist of Mumbai. He has been the West-Zone coordinator of the Perinatology committee of FOGSI (Federation of Obstetric and Gynaecology Societies of India) for six years during which period he started promoting newborn screening amongst obstetricians. He was the co-convenor for the first stake-

holders meet on newborn screening, which saw participants from the National Neonatal Forum of India, NGOs working for newborn care, newborn screening equipment manufacturers and laboratories in India. He has participated in several local, national, and international conferences and given talks on various topics related to maternal and child health. He is the co-founder of Health N Wellness—a company with

a focus on preventive health. Under the banner of Health N Wellness, he has addressed in several national and multinational companies.

SEEMA KAPOOR, MD

Dr. Seema Kapoor is the director professor of Pediatrics and in-charge of the Division of Genetics & Metabolism at the Maulana Azad Medical College in New Delhi. She has authored 125 indexed publications in national and international journals and was awarded 25 gold medals during MBBS. She was also awarded the President's Medal for Best Lady Medico in 1982. Dr. Kapoor is currently coordinating a center for excellence in Biochemical Genetics. She is a member of multiple task force groups, including: the Task Force ICMR on inherited metabolic disorders, Task Force ICMR on Lysosomal storage Disorders, Task Force ICMR on Thalassemia, and DBT Task Force on Human Genetics. She is also a member



of the Research Cell of the Institute, the Disability Board, and Member Advisory to the Infertility Initiative in MAMC.

THOMAS MOOKKEN



Mr. Thomas Mookken is the founder and CEO of NeoGen Labs. He has a background in high technology and has worked primarily in the telecommunications industry in technical and marketing roles. He became interested in newborn screening when it was discovered, through a newborn screening test, that his son had G6PD Deficiency. Mr. Mookken received his MS in Electrical & Computer Engineering from the University of Texas at Austin and MBA from Carnegie Mellon University.

KANNAN VAIDYANATHAN, MD

Dr. Kannan Vaidyanathan is a professor in the Department of Biochemistry at Pushpagiri Institute of Medical Science and Research Center in Thiruvalla and deputy medical superintendent at Pushpagiri Medical College Hospital in-charge of laboratories.

Dr. Vaidyanathan earned his medical degree from the Trivandrum Medical College in 2003. From 2003 to 2006, he had a postdoctoral fellowship in the Department of

Microbiology & Cell Biology at the Indian Institute of Science in Bangalore. Thereafter, he worked at Amrita Institute of Medical Science, Kochi, heading the Metabolic Disorders Laboratory. He also worked at Medical College, Kottayam and Co-operative Medical College in Kochi, Kerala.

Dr. Vaidyanathan has won many awards, including the APFCB (Asia Pacific Federation of Clinical Biochemistry) Silver Jubilee scholarship in Beijing, China (2007) and KP Sinha – PS Krishnan award for best original research article in Indian Journal of Clinical Biochemistry (2011).

Other roles include managing editor of the Indian Journal of Clinical Biochemistry and the South Zone representative of the Association of Clinical Biochemists of India. Dr. Vaidyanathan has co-authored three textbooks in biochemistry and has more than 75 publications. He has delivered more than 60 lectures in national and international conferences in the US, Europe, Australia, South Africa, and China. He has guided six PhD students and more than 75 post-graduate students. He is the examiner for MBBS, BDS, PhD, and allied health sciences for all major universities in India. His areas of research include inborn errors of metabolism, clinical chemistry, and molecular biology of breast cancer.



D.M. VASUDEVAN, MD, FAMS, FACBI, FRCPATH



Dr. D.M. Vasudevan was the president of the Association of Clinical Biochemists of India (ACBI). Dr. Vasudevan has 50 years of teaching and research experience in various medical colleges. He has worked in the medical colleges of Calicut, Trivandrum, Kottayam, Trichur, Mangalore, Manipal, Gangtok (Sikkim), and Cochin. He has spent 30 years as a professor of Biochemistry. He is the founder dean of the Sikkim Manipal Institute of Medical Sciences in Gangtok, Sikkim and the founder principal and dean of Amrita Institute of Medical Sciences in Cochin.

In 1992, Dr. Vasudevan received a fellowship with the National Academy of Medical Sciences (FAMS), as well as the Dr. B.C. Roy Award for Eminent Medical Teacher (Indian Medical Council) from the President of India. Other awards include: Award of Best Doctor in the Medical Education Services, Government of Kerala and the KL Gupta Memorial Oration Award (ACBI) in 1995 and the Awadhesh Saran Memorial Oration Award of ACBI in 2016.

Dr. Vasudevan has authored the *Textbook of Biochemistry for Medical Students*, now in the 8th edition, which is widely used in all medical colleges in India and abroad; more than 150,000 copies sold (with a Spanish edition also available). In total, more than 240 of his papers have been published in national and international journals, some of which were cited in standard textbooks. More than 200 papers were presented in national and international conferences inside and outside India; and he gave invited speeches or theme talks in more than 100 conferences. He has guided 30 PhDs and is the recognised PhD guide of Calicut, Kerala, Manipal, and Amrita Universities.