

Atlanta Down Syndrome Research Conference

Saturday, March 10, 2012 -- Emory University School of Medicine

SPEAKERS' BRIEF BIOGRAPHIES

Omar Khwaja, MD, Ph.D. is Translational Medicine Leader for Down syndrome clinical trials at F. Hoffman – La Roche Ltd. Previously he was Instructor of Neurology at Harvard Medical School, and research staff and Director of the Rhett Syndrome program at Children's Hospital Boston. Dr. Khwaja received his undergraduate, his Master's degree (Developmental Biology and Neuroscience), his MD and a PhD in Human Molecular Genetics, from the University of Cambridge. He completed a fellowship in Clinical Genetics at the Royal Children's Hospital in Melbourne and a residency in Pediatrics at the Royal London Hospital in London, as well as residencies and fellowships in Neonatology at the Royal Children's Hospital in Melbourne and Great Ormond Street Hospital for Children in London. He trained as a resident in Child Neurology, as Chief Resident, and completed a fellowship in Fetal-Neonatal Neurology at Children's Hospital Boston--during which time he received an American Academy of Neurology Clinical Training Fellowship. Dr. Khwaja is an advocate for children with Down syndrome, Rett syndrome, autism and other neurologic disorders.

Stephanie Sherman, Ph.D. is Professor, Department of Human Genetics at Emory University. Her research program revolves around two syndromes (Down syndrome and Fragile X). She has used genetic epidemiological approaches to answer questions related to the underlying genetic mechanism leading to the syndrome and the resulting phenotype consequences. She has completed two large population-based studies of Down syndrome live births and their parents as well as collaborated with several sites in a research program to identify genes and/or environmental exposures that lead to the susceptibility of birth defects. Dr. Sherman has a B.S. from North Carolina State University, and a Ph.D. in Human Genetics from Indiana University Medical School.

Roger Reeves, Ph.D. is Professor, Department of Physiology, the Johns Hopkins University School of Medicine. His research goal is "to understand how gene dosage imbalance disrupts development in Down syndrome in order to develop and test potential therapies for Down syndrome features." His research interests focus on mechanisms of gene action in Down syndrome and he has used mouse models to identify and test hypotheses concerning Down syndrome "critical regions" on human chromosome 21. Among recent findings are the identification of genes that play a key role in tumor resistance; determination that a deficit in cranial neural crest is the (initial) basis for the hypomorphic craniofacial skeleton that produces the characteristic appearance of individuals with Down syndrome; and the discovery of the basis for and a "treatment" of a fundamental neuronal deficit in the trisomic brain. He has a B.S. from Bowling Green State University, a Ph.D. from the University of Maryland, and a Postdoc from The Johns Hopkins University.

Jamie Edgin, Ph.D. is Senior Research Associate, Department of Psychology, University of Arizona. She is a Developmental Psychologist specializing in the area of the Developmental Cognitive Neuroscience. Her area of expertise is cognitive development in neurodevelopmental disorders with hippocampal involvement, including Down syndrome, Williams syndrome, and extreme prematurity. Most recently, she has been studying the development of context-dependent memory in typical and atypical development. An important facet of her research program includes the support of clinical trials in Down syndrome through the validation of neuropsychological assessments and biomarkers (EEG and ERP), and she serves as a scientific advisor for several companies initiating these trials. In collaboration with Lynn Nadel, she co-founded the Down Syndrome Research Group.

Blythe Crissman, MS, CGC is a Certified Genetics Counselor and Clinical Coordinator of the Duke University Down Syndrome Clinic. The Duke Comprehensive Down syndrome clinic is a multidisciplinary clinic for the clinical care of approximately 700 children and young adults with Down syndrome. Ms. Crissman works with Priya Kishnani, MD, and others on the Duke Down syndrome research team, evaluating the effects of cholinesterase inhibitors on cognition and behavior in children and young adults with Down syndrome, including establishing a standard dosing regimen and developing a sensitive and specific test battery to detect changes in language and cognitive ability. They are currently involved in a multi-center Phase 2 pediatric trial using donepezil, a cholinesterase inhibitor. They are also exploring how a national Down syndrome registry program could be built and utilized to expand understanding of the condition.

George Capone, MD is Director, Down Syndrome Clinic, Kennedy Krieger Institute in Baltimore, MD. He is research scientist and associate professor of pediatrics at the Johns Hopkins University School of Medicine. Dr. Capone attended college at Wesleyan University and worked as a research assistant at the Dana Farber Cancer Institute in Boston before obtaining his medical degree from the University of Connecticut in 1983. After a residency and fellowship in pediatrics at the Children's Hospital Medical Center in Cincinnati, Dr. Capone came to Baltimore in 1988 to pursue a fellowship in neurobiology research at Johns Hopkins. He is the director of Kennedy Krieger Institute's Down Syndrome Clinic as well as attending physician on the institute's comprehensive rehabilitation unit. He and colleagues are exploring the neurobiologic basis of cognitive impairment and co-morbid neurobehavioral and psychiatric disorders associated with Down syndrome.

Jim Kucik, MPH is a health scientist with the Birth Defects Surveillance Team of the National Center on Birth Defects and Developmental Disabilities (NCBDDD), The Centers for Disease Control and Prevention. He received a master's degree in public health with a focus in epidemiology from Emory University in 2001. At NCBDDD, Jim has conducted research that investigates trends and causes of birth defects and has explored racial disparities in the occurrence of birth defects. Also, he contributes to the administration of CDC's MACDP birth defects' surveillance system.

Jeannie Visootsak, MD, FAAP is Medical Director of the Emory Down Syndrome Clinic and Fragile X Syndrome Clinic, and Assistant Professor, Department of Human Genetics and Pediatrics, Emory University School of Medicine. She has a B.S. from the University of Southern California, M.D. from the UCLA School of Medicine, and M.Sc. in Clinical Research from Emory University. Dr. Visootsak is Board Certified in Pediatrics and Developmental/ Behavioral Pediatrics. Her research interests include genotype-phenotype correlations in Down syndrome; Fragile X and sex chromosomal conditions; management of genetic intellectual disabilities syndromes; and developmental disabilities and access to care. She has conducted several studies on the impact of congenital heart defects on the development of young children with Down syndrome and is currently the Principal Investigator for the BP25543 Study to investigate a drug that is being developed to improve attention and memory in people with Down syndrome.