EINSTEIN
Albert Einstein College of Medicine
OF YESHIVA UNIVERSITY
Science at the heart of medicine

The Children's Hospital at Montefiore

The Rose F. Kennedy IDDRC and CERC
The Montefiore/Einstein Center
for Williams Syndrome
The Williams Syndrome Association

# 1st Annual Isabelle Rapin Conference on Communication Disorders

# WILLIAMS SYNDROME WORKSHOP AND ROUNDTABLE

**Thursday, December 6, 2012**
**9:00 am to 6:00 pm**
**Price Center /Block Research Pavilion**

williamssyndrome
ASSOCIATION

# *Schedule*

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| **8:30 – 9:00am** | **Continental Breakfast (available in Lower Lobby)** |

## Workshop in LeFrak Auditorium

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| **Introduction** | **Steven Walkley (Director of the Rose F. Kennedy IDDRC, Albert Einstein College of Medicine)** |
| **9:00 – 9:30am** | **Barbara Pober (Massachusetts General Hospital and Harvard Medical School)**<br>"Clinical Overview of Williams Syndrome and Initiatives to Promote Research" |
| **9:30 – 9:45am** | Questions/Discussion |
| **9:45 – 10:15am** | **Robert Marion and Joy Samanich (Albert Einstein College of Medicine and Montefiore Medical Center)**<br>"A Glimpse Inside the Einstein-Montefiore Williams Syndrome Center: A Treasure in Our Backyard" |
| **10:15 – 10:30am** | Questions/Discussion |
| **10:30 – 11:00am** | **Carolyn Mervis (University of Louisville)**<br>"Children with Williams Syndrome: Cognition, Language, and Psychopathology" |
| **11:00 – 11:15am** | Questions/Discussion |
| **11:15 – 11:45am** | **Tricia Thornton-Wells (Vanderbilt Kennedy Center)**<br>"From Sensation to Sociability in Williams Syndrome: Evidence from Structural and Functional MRI Studies" |
| **11:45 – 12 Noon** | Questions/Discussion |
| **12 Noon – 1:00pm** | **Informal Luncheon (available in Lower Lobby)** |
| **1:00 – 1:30pm** | **Brian Haas (University of Georgia)**<br>"Neuroimaging as a Tool to Unraveling the Williams Syndrome Social Phenotype" |
| **1:30 – 1:45pm** | Questions/Discussion |
| **1:45 – 2:15pm** | **Lucy Osborne (University of Toronto)**<br>"Of Mice and Men - New Insights into the Molecular Basis of Williams Syndrome" |
| **2:15 – 2:30pm** | Questions/Discussion |

## Roundtable Discussion in Room 451
### *(All speakers, Invited Discussants, Guests)*

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| **2:30 – 3:00pm** | **Refreshments** |
| **3:00 – 5:00pm** | **Moderated by Steven Walkley and Robert Marion (Albert Einstein College of Medicine)** |

## Reception in Lower Lobby

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| **5:00 – 6:00pm** | **Opportunity for family members to meet with the speakers and meeting participants** |

# *Abstracts*

**Speaker:** Barbara Pober, M.D.
Medical Geneticist
Department of Pediatrics, Genetics
Massachusetts General Hospital
Associate Professor of Pediatrics
Harvard Medical School

**Title:** Clinical Overview of Williams Syndrome and Initiatives to Promote Research
**Abstract:** Williams syndrome is a genetic disorder with a complex but characteristic phenotype. In addition to the occurrence of medical problems potentially impacting numerous organ systems, a distinctive neurodevelopmental phenotype is present. The cause of Williams syndrome is now known- it is a chromosomal microdeletion leading to the deletion of 26-28 genes. Clinically-based research efforts are underway on several fronts: natural history of Williams syndrome, genotype - phenotype correlations, and potential therapeutics. The Williams Syndrome Association is supporting development of the Williams Syndrome Clinical and Research Registry (www.registry.williams-syndrome.org) to accelerate the pace of work in these areas with the ultimate goal of improving the lives of persons with Williams syndrome.

**Speaker:** Robert Marion, M.D.
Director of Children's Evaluation and Research Center
Albert Einstein College of Medicine
Founder of Montefiore's Williams Syndrome Center

**Speaker:** Joy Samanich, M.D.
Pediatric Geneticist
Pediatrics
Montefiore Medical Center
The University Hospital for Albert Einstein College of Medicine

**Title:** A Glimpse Inside the Einstein-Montefiore Williams Syndrome Center: A Treasure in Our Backyard
**Abstract:** During this talk, the Einstein-Montefiore Williams Syndrome Center will be described. A true multidisciplinary clinic that provides complex medical and developmental services for individuals with Williams syndrome of all ages, the center is one of only a handful serving this population throughout the United States. Currently caring for approximately 75 families, the center meets monthly on the first Friday of every month. Coordinated by a genetic counselor, each clinic is attended by medical geneticists, a developmental behavioral pediatrician, a psychologist, speech therapists, a cardiologist, nephrologists, an endocrinologist, and a social worker, providing coverage of most medical issues. Family support activities are also available. This center provides a wealth of clinical material for translational research.

# *Abstracts*

**Speaker:** Carolyn B. Mervis, Ph.D.
Distinguished University Scholar and Professor
Co-Director, Experimental Psychology PhD Program
Director, Cognitive & Developmental Sciences Area
Department of Psychological & Brain Sciences
University of Louisville

**Title:** Children with Williams Syndrome: Cognition, Language, and Psychopathology

**Abstract:** This presentation will provide an overview of the cognitive, language, and psychopathology findings from my research program on children with Williams syndrome. Both large-sample cross-sectional results and medium-sample longitudinal findings will be described, focusing on the cognitive profile and the extent to which early development predicts later development for children with this syndrome. I will emphasize not only the similarities among children with Williams syndrome but also the variability.

**Speaker:** Tricia A. Thornton-Wells, Ph.D.
Assistant Professor of Molecular Physiology and Biophysics
Center for Human Genetics Research
Vanderbilt Institute of Imaging Science
Vanderbilt Kennedy Center for Research on Human Development

**Title:** From Sensation to Sociability in Williams Syndrome: Evidence from Structural and Functional MRI Studies

**Abstract:** Williams syndrome (WS) is a rare genetic neurodevelopmental disorder whose behavioral phenotype is characterized by increased non-social anxiety, sensitivity to sounds and hypersociability. By investigating the neural basis of WS, we aim to learn more about the relationship between genes, brain structure and brain function with respect to specific aspects of the WS phenotype. In the process, we gain insight into typical neurodevelopment and the basis of variability in sensory processing, anxiety and sociability in the general population. We present converging evidence from magnetic resonance imaging studies suggesting an altered pattern of brain structure and function in individuals with WS versus typically-developing controls, resulting in a spectral shift along a behavioral continuum. Brain regions implicated in socio-affective processes, including the prefrontal cortex, amygdala, insula, superior/middle temporal cortex and hippocampus, demonstrate differential patterns of cortical thickness and surface area, white matter structural connectivity, inter-network functional connectivity and task-related functional activation. These findings have important implications for understanding how interventions aimed at sensory processing might positively impact affect and behavior in WS, other developmental disorders and the neurotypical population.

# *Abstracts*

**Speaker:** Brian Haas, Ph.D.
Assistant Professor
Behavioral and Brain Sciences Program
University of Georgia

**Title:** Neuroimaging as a Tool to Unraveling the Williams Syndrome Social Phenotype

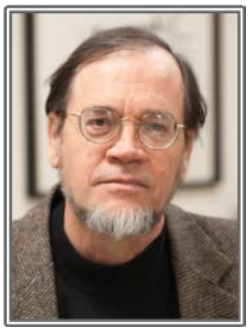
**Abstract:** Williams syndrome is often paired with a distinctive social-emotional phenotype. Recent advancements in psychological and neuroimaging research techniques have resulted in a more detailed understanding of the etiology of social behavior and emotion processing in this condition. In a series of studies, we have used neuroimaging as a tool to describe the biological basis of the Williams syndrome social phenotype. By using a functional magnetic resonance imaging (fMRI) approach, we demonstrated that individuals with Williams syndrome exhibit reduced amygdala response to fearful facial expressions and increased amygdala response to happy facial expressions, as compared to typically developing controls. In addition, we showed that the extent to which amygdala response to fearful facial expressions is attenuated is associated with the tendency to uninhibitedly approach strangers in Williams syndrome. Recently, we have used structural neuroimaging as a tool to demonstrate altered macro- and micro-structure of key brain regions involved in social cognition in WS. The results of these studies are an important step towards the design to syndrome and symptom specific treatment techniques for individuals with this condition.

**Speaker:** Lucy Osborne, Ph.D.
Associate Professor
Department of Medicine
Department of Molecular Genetics
Institute of Medical Science
University of Toronto

**Title:** Of Mice and Men - New Insights into the Molecular Basis of Williams Syndrome

**Abstract:** Although the genetic lesion responsible for Williams syndrome (WS) was identified 20 years ago, we are still striving to fully understand the role of individual genes in the unique and complex spectrum of symptoms that we see in people with WS. I will provide a brief introduction and overview of the genetics of WS as well as some of the mouse models that have been generated, followed by some examples of our own studies in humans and in mice that promise to shed new light on some of the biological processes that are altered in WS.

# *Roundtable Facilitators*



Steven Walkley, D.V.M., Ph.D.
Director, Rose F. Kennedy Intellectual and Developmental Disabilities Research Center
Professor, Dominick P. Purpura Department of Neuroscience
Professor, Department of Pathology
Professor, The Saul R. Korey Department of Neurology
Albert Einstein College of Medicine

Robert Marion, M.D.
Director of Children's Evaluation and Research Center
Albert Einstein College of Medicine
Founder of Montefiore's Williams Syndrome Center

# *Sponsors*

The Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC)
http://einstein.yu.edu/centers/iddrc/

The Rose F. Kennedy Children's Evaluation and Rehabilitation Center (CERC)
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The Montefiore/Einstein Center for Williams Syndrome
http://www.montekids.org/services/genetics/williams_syndrome/

The Williams Syndrome Association
http://williams-syndrome.org/

## *Notes:*