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# NEWS

## Rose F. Kennedy Intellectual and Developmental Disabilities Research Center

### Rare People and Rare Talents on a Rare Day

**F**ebruary 29 only occurs every four years. In recognition of this rare day, two Einstein centers — the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC), and the Children’s Evaluation and Rehabilitation Center (CERC), the clinical arm of the Rose F. Kennedy University Center for Excellence in Developmental Disabilities (RFK UCEDD) — co-sponsored a special concert, “Rare People and Rare Talents on a Rare Day.”

In offering opening remarks, Dr. Steven Walkley, director of the IDDRC noted, “Rare Disease Day started

*(continued on page 2)*





## MESSAGE FROM THE DIRECTOR

### STEVEN U. 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

While the Rose F. Kennedy Center has a 40-year history as an NIH-sponsored Intellectual and Developmental Disabilities Research Center, over the past year it has experienced a veritable renaissance in its programs and outreach to scientists and clinicians at Albert Einstein College of Medicine and its affiliated hospitals. For this center without walls, newly facilitated efforts by the Kennedy Center leadership are underway to bridge the disciplines of neuroscience and genetics and build productive collaborations between basic scientists and clinicians whose activities involve intellectual and developmental disabilities in children. New ties are also being forged between basic scientists in the Kennedy Center program and clinicians within Einstein's largest pediatric care center, known as CERC (Children's Evaluation and Rehabilitation Center), whose patient outreach encompasses the genetically diverse and socioeconomically compromised community of the Bronx.

Although the challenge of understanding and treating intellectual and developmental disabilities of children is substantial, the opportunities for advancement in understanding brain development and disease, and in developing new interventional therapies, have never been greater. The mandate of the Kennedy Center is for us to do all we can to foster these collaborations and to advance scientific discoveries and clinical efforts that will pave the way toward improvements in the lives of children with intellectual and developmental disorders.

## Rare People and Rare Talents on a Rare Day *(continued from page 1)*

four years ago on this date. Awareness has now reached a tipping point where people are becoming more familiar with this concept of 'rare disease'— which by definition at the National Institutes of Health is any disease in the U.S. affecting fewer than 200,000 people." He continued, "There are estimated to be 6,000 to 7,000 rare diseases, affecting about 25 million Americans in total. When we think about it in these terms, rare disease is not really very rare at all. Importantly, very few of these diseases – only about 250 – have approved therapies."

Dr. Robert Marion, director of CERC and the UCEDD, also welcomed those attending, adding, "Today's celebration is really about people. We're going to hear four very special people, individuals who have rare diseases but also rare talents." He then introduced the performers who also are current patients of his. They included:

**Zachary Cantor**, a singer with Williams syndrome, a developmental disorder characterized by mild to moderate intellectual disability and cardiovascular problems (during

his performance, Zach was accompanied by Charlotte Pharr, music therapist at the Williams Syndrome Center at Montefiore);

Singer **Ashley Appell** and saxophonist **Mervin Hernandez**, two individuals with Hermansky-Pudlak syndrome, which is characterized by blood clotting problems and a type of albinism of the skin, hair, and eyes that also affects vision; and

**Alena Galan**, a singer with Mucopolysaccharidosis Type VI, a lysosomal disorder which causes enlargement and inflammation of many tissues and organs, including severe skeletal abnormalities.

The festivities, which included a performance by a troupe of young girls performing Irish step dancing, were a great success. And the event achieved its dual goal of underscoring the difference that research can make in the lives of patients suffering from these conditions and of also inspiring people both with or without disabilities – as said by Alena Galan – "to do bigger things by keeping faith, courage, hope, and by never giving up."



### ON THE WEB

To learn more about the RFK IDDRC, please visit:  
[www.einstein.yu.edu/centers/IDDRC](http://www.einstein.yu.edu/centers/IDDRC)

## IDDRC



### Q: Why should scientists study rare diseases?

**A:** First and foremost, rare diseases represent important health conditions affecting both children and adults. Determining the underlying genetic causes of these conditions and the consequences that such defects have on cells and organs can lead the way to therapy development. Presently very few effective treatments are available for rare diseases.

Determining how rare diseases affect the brain and cause intellectual disability can provide important new insights into the mechanisms of learning and memory in all people.

Understanding of the causes and consequences of some rare disease can also lead to new ways of thinking about commoner diseases, like Alzheimer's and Huntington's, and novel ways to treat them.

# featured investigators

## Genome-wide DNA Methylation Profiles Associated with Abnormal Intrauterine Growth



**Dr. Francine Einstein**, an Associate Professor in the Department of Obstetrics & Gynecology and Women's Health, serves as a member of the Execu-

tive Committee of the RFK IDDC. Dr. Einstein is also the head of the epigenetics and brain development research cluster of the RFK IDDC. In this role, she fosters communication and collaboration between geneticists and neuroscientists to better understand the gene-environment interactions that affect neurodevelopment, cognitive functioning and plasticity of the brain. She organized the first of the RFK IDDC's cluster workshop series, entitled The Exposure and the Developing Brain in 2011, in which IDDC investigators and clinicians exchanged ideas, consolidated collaborations and learned of each other's work.



### ON THE WEB

For full article, please visit the website [www.einstein.yu.edu/centers/iddrc/investigators/](http://www.einstein.yu.edu/centers/iddrc/investigators/)

## Predicting Speech-language Deficits in Pre-lingual Infants



**Dr. Pierfilippo De Sanctis**, an Assistant Professor in the Department of Pediatrics, aims to establish himself as an independent translational researcher

in the study of developmental disabilities. His background is on studies probing the sensory-perceptual bases of intact sound and language processing in healthy adult populations, using cutting-edge neuroscience methodologies, including high-density electrophysiological recordings from the scalp and direct recordings from the cortical surface in epilepsy patients. His goal is to bring his expertise to bear on clinical pediatric and diagnostic issues, with an emphasis on the early detection of language impairment in children on the autism spectrum. Dr. De Sanctis is a current recipient of the pilot and feasibility award sponsored by the Rose F. Kennedy IDDC.

## Exploring the Hemichannel Properties of Cx26 Mutations: A Cause of Syndromic Deafness



The National Institute on Deafness and Other Communication Disorders reports that over 46 million people in the United States suffer from some form of com-

munication impairment or disorder. For over 20 years, biophysicist and channelologist, **Vytas K. Verselis**, Professor in the Dominick P. Purpura Department of Neuroscience, has been studying structure/function relationships and gating mechanisms of a class of channels called connexins which form an essential communication pathway between cells. Of particular interest to Dr. Verselis is connexin 26 (Cx26), a protein which is encoded by the GJB2 gene. Mutations in this gene are a major cause of pre-lingual, congenital deafness known as Keratitis-Ichthyosis-Deafness (KID) syndrome. The underlying basis of this syndrome appears to be aberrantly behaving Cx26 hemichannels, a relatively new mechanism of disease.

# discoveries

## New therapy for Niemann-Pick disease type C

Dr. Steven Walkley and his colleagues at the RFK IDDC, with assistance from its Cell and Molecular Imaging and Animal Behavior cores, discovered that the FDA-approved excipient, hydroxypropyl-beta-cyclodextrin (CD) effectively ameliorates brain and liver disease in the lysosomal disorder known as Niemann-Pick type C (NPC). Collaborative studies with Dr. Daniel Ory at the IDDC at Washington University in St. Louis led to identification of biomarkers for this

disease and their reduction following CD therapy. Studies in collaboration with Dr. Charles Vite at the IDDC at the University of Pennsylvania revealed the effectiveness of intrathecal CD therapy in a cat model of NPC disease. These highly collaborative efforts involving IDDC investigators (Walkley, Ory, Vite) at 3 institutions culminated in selection of CD and NPC disease by TRND (Therapeutics for Rare and Neglected Diseases) at NIH as a candidate compound and disease to move toward clinical trial development. The trial is expected to begin in early 2013.

## New gene linked to autism spectrum disorder (ASD)

RFK IDDC investigator, Dr. Noboru Hiroi, as recently cited in a news brief by the Simons Foundation Autism Research Initiative (SFARI), has discovered that deletion of one copy of T-box 1, or TBX1, a gene on the autism-linked chromosomal region 22q11.2, leads to social and communication deficits in mice. The results support the view that TBX1 could hold the key to autism-related features seen in individuals with a 22q11.2 deletion.



# our supporters

## NOTABLE GRANTS

**The Eunice Kennedy Shriver National Institute for Child Health and Human Development**, part of the National Institutes of Health, has awarded Einstein a five year, \$5.7 million grant to fund the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC). The grant supports Kennedy Center's ongoing efforts to improve the lives of children with intellectual and developmental disabilities (IDD) through

combined basic science research and clinical practice.

**The National Institutes of Health (NIH)** has awarded Einstein a five-year, \$2.8 million grant to study whether multisensory integration (the brain's processing of information from different senses) is impaired in people with autism. This research will continue at the Children's Evaluation and Rehabilitation Center (CERC) and could have important implications for

improving the quality of life of children with autism. In addition, the Cognitive Neurophysiology Laboratory (CNL) — the research arm for CERC — purchased several pieces of high-tech diagnostic and monitoring equipment, including advanced electroencephalogram (EEG) systems, stimulus delivery equipment and specialized video recording devices through a \$1 million capital grant from the New York State Senator **Jeffrey D. Klein**.

## UPDATES

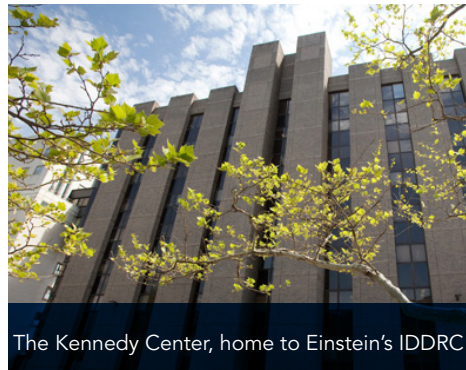
### New RFK IDDRC Members

Pierfilipo De Sanctis, Ph.D. (Pediatrics)  
Frederick J. Morrison, Ph.D. (Pediatrics)  
Harry Ostrer, M.D. (Pathology)  
Ji Ying Sze, Ph.D. (Molecular Pharmacology)

To become a member, please visit:  
<http://einstein.yu.edu/centers/iddrc/members/become-investigator.aspx>

### Congratulations!

On Saturday, May 5, 2012, Dr. Steven Walkley, director of RFK IDDRC, and Dr. Marc Patterson, a pediatric neurologist from the Mayo Clinic and a member of the RFK IDDRC's external advisory committee, were presented with certificates of special recognition by Senator Richard Blumenthal of Connecticut at the reception for the annual gala benefit and concert for Dana's Angels Research Trust (DART). *(more on the web)*



The Kennedy Center, home to Einstein's IDDRC



## UPCOMING EVENTS

The First Annual Isabelle Rapin Conference on Communication Disorders is scheduled for December 6, 2012 at the Price Center. The focus of this workshop and roundtable will be Williams Syndrome.

Please visit the website to view details of this and other upcoming events:  
<http://einstein.yu.edu/centers/iddrc/seminars-workshops/>

**To learn more about supporting the work of the Rose F. Kennedy IDDRC, please contact:**

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## ALBERT EINSTEIN ROSE F. KENNEDY IDDRC

**Our mission:** to improve the lives of children with intellectual and developmental disabilities through research and clinical outreach. The Center actively supports and encourages collaboration between bench scientists and clinicians.

### ADMINISTRATION

**Director**  
Steven U. Walkley, D.V.M, Ph.D.

**Associate Director**  
John J. Foxe, Ph.D.

**Administrator**  
Sukhi E. Lee, M.P.H.

### EXECUTIVE COMMITTEE MEMBERS

Joseph Arezzo, Ph.D.  
Francine Einstein, M.D.  
Robert Marion, M.D.  
Bernice Morrow, Ph.D.  
Elyse Sussman, Ph.D.  
Vytutas Verselis, Ph.D.