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

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.

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.

ON THE WEB
To learn more about the RFK IDDRC, please visit:
www.einstein.yu.edu/centers/IDDRC

Q&A

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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 Executive Committee of the RFK IDDRC. Dr. Einstein is also the head of the epigenetics and brain development research cluster of the RFK IDDRC. 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 IDDRC’s cluster workshop series, entitled The Expose and the Developing Brain in 2011, in which IDRC investigators and clinicians exchanged ideas, consolidated collaborations and learned of each other’s work.

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 IDDRC.

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 communication 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.

ON THE WEB
For full article, please visit the website www.einstein.yu.edu/centers/iddrc/investigators/

New therapy for Niemann-Pick disease type C

Dr. Steven Walkley and his colleagues at the RFK IDDRC, 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 IDDRC 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 IDDRC at the University of Pennsylvania revealed the effectiveness of intrathecal CD therapy in a cat model of NPC disease. These highly collaborative efforts involving IDDRC 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 IDDRC 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.
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)

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.

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