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NEWS

Rose F. Kennedy Intellectual and Developmental Disabilities Research Center



From left: Marc C. Patterson, M.D.; Steven U. Walkley, D.V.M., Ph.D.; John J. Foxe, Ph.D.; Vytautas Verselis, Ph.D.; Robert W. Marion, M.D.; John N. Constantino, M.D.; and BJ Casey, Ph.D.

Inaugural IDDRC Symposium Stimulates a Sense of Community

Newsletter for the Rose F. Kennedy
Intellectual and Developmental
Disabilities Research Center

ISSUE 3 • SPRING/SUMMER 2013

On April 30, 2013, the inaugural Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC) symposium was held in Einstein's Michael F. Price Center for Genetic and Translational Medicine/Harold and Muriel Block Research Pavilion. Founded more than 40 years ago, the IDDRC continues to serve as the core for Einstein's research into normal and abnormal brain development and brain function, and as a partner to Einstein's Children's Evaluation and Rehabilitation Center (CERC), which provides clinical care to patients with intellectual and developmental disabilities (IDDs).

The daylong symposium was designed to bring together IDDRC investigators at Einstein with clinical partners from Montefiore, the University Hospital and academic medical center for Einstein, where they could learn about one another's work and about collaboration opportunities. The Ethel and Samuel J. LeFrak Auditorium was filled to capacity, with guests including John N. Constantino, M.D. (Washington University School of Medicine in St. Louis), Marc C. Patterson, M.D. (Mayo Clinic) and BJ Casey, Ph.D. (Weill Medical

(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

In 1955, Saul R. Korey, M.D., became the founding chairman of Einstein's department of neurology, and before his untimely death eight years later created a program of science-based, translational neurology that persists to this day. As part of his innovative program, Dr. Korey in 1957 contacted the National Institute of Mental Health (NIMH) about establishing an interdisciplinary (or "ID") training program, one centered in neurology but with outreach to Einstein's other departments. NIMH funded Dr. Korey's ID program, which then flourished for three decades. Upon his death, its leadership passed to a series of distinguished faculty, including Dominick P. Purpura, M.D., who in the early 1970s established the department of neuroscience, one of the first in the country, in the recently completed Rose F. Kennedy Center for Research in Mental Retardation and Human Development. As a result, Dr. Korey's original ID program became the backbone of interdisciplinary postdoctoral training in the Kennedy Center and helped create a richly collaborative environment in basic and translational science, much of it focused on normal and abnormal brain development and function. That environment in turn influenced generations of students, postdocs and faculty, including yours truly. Such is the legacy of Saul Korey, whose brief years at Einstein shaped our academic community for decades thereafter. Now, in this 50th year since Dr. Korey's death, our goal is to ensure that it is indeed an enduring legacy, a constant guiding principle for our research in intellectual and developmental disabilities.

Inaugural IDDRC Symposium Stimulates a Sense of Community *(continued from page 1)*

College of Cornell University), who serve on the IDDRC external advisory board.

Four internationally recognized experts—in autism (Ralph-Axel Müller, Ph.D., San Diego State University), fragile X syndrome (Gary J. Bassell, Ph.D., Emory University School of Medicine), cochlear implants and cortical plasticity (Anu Sharma, Ph.D., University of Colorado at Boulder) and epigenetics and the brain (Janine M. LaSalle, Ph.D., University of California, Davis School of Medicine)—made presentations, as did selected speakers from Einstein. A poster session, featuring 30 abstract posters and six IDDRC scientific core posters, offered additional information on center members' work.

"The goal of this inaugural symposium was to build a sense of community among faculty



From left: Anu Sharma, Ph.D.; Gary J. Bassell, Ph.D.; Janine M. LaSalle, Ph.D.; and Ralph-Axel Müller, Ph.D.

by strengthening relationships within and between our various research groups," said Dr. Steven U. Walkley, director of Einstein's IDDRC. "By sharing our successes and featuring the various projects going on within the labs and clinics associated with the center, we hope to foster new collaborations that can augment current research and ultimately provide better care to individuals with intellectual disabilities."

in focus

Einstein's LEND program

Each year, trainees from 18 different disciplines come to Einstein to gain experience in working with individuals with intellectual disabilities and their families. This training is provided through our Leadership Education in Neurodevelopmental Disabilities (LEND) program. One of only 44 such programs in the United States, our LEND is the training arm of the Rose F. Kennedy University Center for Excellence in Developmental Disabilities (UCEDD), an IDDRC partner. Administered through the Health Resources and Services Administration's Maternal and Child Health Bureau at the U.S. Department of Health and Human Services, the LEND's mission is to train the next generation of leaders in the field of intellectual disabilities. For more than 40 years, through hands-on "in the trenches" clinical experiences at CERC, the UCEDD's clinical arm, our LEND has been carrying out this important work. Under the direction of Robert W. Marion, M.D., the program has trained thousands of professionals, including the directors of developmental programs at Cornell, Columbia, Mount Sinai and Einstein.



ON THE WEB

For full article, please visit

www.einstein.yu.edu/centers/childrens-evaluation-rehabilitation/training/

featured investigator

JUDY L. ASCHNER, M.D.



Professor of Pediatrics (Neonatology)
Professor of Obstetrics &
Gynecology and Women's Health
Michael I. Cohen, M.D., University Chair,
Department of Pediatrics
Albert Einstein College of Medicine
Physician-in-Chief
Chair of Pediatrics
The Children's Hospital at Montefiore

Dr. Aschner is the recently appointed physician-in-chief and chair of pediatrics at The Children's Hospital at Montefiore (CHAM), and professor and chair of pediatrics at Einstein. Previously, she was director of neonatology and the Julia Carell Stadler professor of pediatrics at the Monroe Carell Jr. Children's Hospital at Vanderbilt University School of Medicine.

Dr. Aschner is an internationally recognized leader in pediatrics, neonatology and perinatal biology, with clinical

interests in the prevention of chronic lung disease and prematurity, and in the treatment of infants with pulmonary hypertension. Her laboratory and translational research focus is on regulation of newborn pulmonary circulation, with much of her work identifying contributing factors to altered lung development and pulmonary hypertension in infants. Currently, Dr. Aschner is a principal investigator on a National Heart, Lung, and Blood Institute-funded multi-center study to identify novel predictive biomarkers for preterm infants at greatest risk for long-term respiratory morbidity.

Moving forward, Dr. Aschner is interested in fostering greater research at Einstein and CHAM and has already moved to further strengthen CERC at Einstein and its close ties to the IDDRC. Clearly, Dr. Aschner's commitment to our program is further evidence of the renaissance of IDD research at Einstein. Through her support and that of others, our newly configured Kennedy Center will continue to grow and prosper.



ON THE WEB

For full article, please visit www.einstein.yu.edu/centers/iddrc/legacy-series/

discoveries

2012 IDDRC Pilot Project Accomplishments



Genetic and Functional Characterization of ARHGAP11A—a Novel Autism Candidate



Brett S. Abrahams, Ph.D., assistant professor of genetics, and his graduate student, Rebecca Nebel, looked at the relationship between genetic variation and clinical presentation in a Bronx family using whole exome sequencing and DNA microarrays. This led to the discovery of a six-gene interval on chromosome 19, where deletions cause microcephaly and duplications cause macrocephaly (each overrepresented

in individuals with autism). All carriers of the duplication, the intellectually impaired proband as well as his typically developing family members, showed extreme macrocephaly. The researchers' findings are consistent with the idea that individual genetic variants can play an important role in aspects of disease without being associated with any clinical diagnosis.



Predicting Speech-Language Deficits in Pre-lingual Infants



Pierfilippo De Sanctis, Ph.D., research assistant professor of pediatrics, has adapted and optimized the Auditory Evoked Spread Spectrum Analysis (AESPA), originally developed to extract neural responses to speech in mature listeners (i.e. adults), for use in pre-lingual children. The AESPA measures patterns of electrical discharge from infants' language processing pathways when they are hearing language. Through the pilot project, Dr. De Sanctis determined age-appropriate stimulus material to tailor the experimental procedure to the infants. Preliminary results support the sensitivity of the AESPA method to map the development of receptive speech processing abilities across infancy and toddlerhood. Going forward, he will investigate the relationship between AESPA and standardize assessment tools such as the Bayley Scales of Infant and Toddler Development for cross-validation of the AESPA response.

A Novel Mechanism of Cross Correction for the Central Nervous System (CNS) in Lysosomal Disorders



Kostantin Dobrenis, Ph.D., assistant professor in the Dominick P. Purpura Department of Neuroscience, is investigating a novel mechanism by which microglia can deliver high amounts of lysosomal system proteins to neurons. Co-culture studies utilizing wild-type microglia and neurons from lysosomal storage disorders (Tay-Sachs and Sandhoff diseases) showed transfer of disease-deficient soluble lysosomal enzyme and suggested transfer of an integral membrane protein. Findings indicate a dependence on direct cell-cell interaction, show a transfer of independent membrane and lysosomal markers and point to a mechanism involving transfer of subcellular organelles. Defining underlying factors at play will contribute to our understanding of microglial-neuronal biology and could help delineate more effective modifications to hematopoietic stem cell replacement approaches that generate microglia to achieve cross-correction of CNS neurons.

IDDRC legacy series



Gary J. Bassell, Ph.D., is professor of cell biology and neurology at Emory University School of Medicine in Atlanta. Before moving to Emory in 2005, Dr. Bassell served on the faculty at Einstein for ten years, where his lab was first established in the department of anatomy and structural biology (1995-1998) and later relocated to the department of neuroscience and Kennedy Center (1998-2005). Dr. Bassell received his Ph.D. degree in cell biology from the University of Massachusetts Medical School, working in the laboratory of Robert H. Singer, Ph.D., who is the current chair of anatomy at Einstein. Dr. Bassell did postdoctoral work in the lab of Kenneth S. Kosik, M.D., at the Center for Neurological Diseases of Brigham and Women's Hospital and Harvard Medical School.

Dr. Bassell is the recipient of several awards, including the Basil O'Connor Scholar Award, the Irma Hirschl Career Scientist Award, the Autism Speaks Trailblazer Award and a Distinguished Investigator Award from the National Alliance for Research on Schizophrenia and Depression. His team investigates mechanisms of mRNA transport and local protein synthesis in neurons, and dysfunction of mRNA regulation in fragile X syndrome and other neurologic diseases.

For full article, please visit www.einstein.yu.edu/centers/iddrc/legacy-series/

UPCOMING EVENTS

The Second Annual Isabelle Rapin Conference on Communication Disorders is scheduled for **October 21, 2013**, at the Price Center. The focus of this workshop and roundtable will be dyslexia.

The annual CTSA symposium, Transforming Clinical and Translational Sciences, sponsored by the Harold and Muriel Block Institute for Clinical and Translational Research, will be held on **November 8, 2013**, at the Price Center. Invited guests include Emil D. Kakkis, M.D., Ph.D., president and founder of the EveryLife Foundation for Rare Diseases, and Christopher P. Austin, M.D., director of the National Center for Advancing Translational Sciences (NIH).

Please visit the website to view details of these and other upcoming events: www.einstein.yu.edu/centers/iddrc/seminars-workshops/

UPDATES

New RFK IDDRC Members

Judy L. Aschner, M.D. (Pediatrics)
John S. Butler, Ph.D. (Pediatrics)
Yonatan I. Fishman, Ph.D. (Neurology)

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

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

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NOTABLE GRANTS

Harry Shamon, M.D., on behalf of Einstein and Montefiore Medical Center, was awarded a renewal of the Clinical and Translational Science Award (CTSA), which supports the Harold and Muriel Block Institute for Clinical and Translational Research at Einstein and Montefiore.

1UL1TR001073-01, 1KL2TR001071-01 and 1TL1TR001072-01
9/1/2013-6/30/2018
Clinical and Translational Science Award

Other Recent Grants

Hannes E. Buelow, Ph.D. (PI)
1R21NS081505-01A1—NIH/NINDS
07/01/2013-06/30/2015

Establishing the role of a novel conserved gene in dendrite morphogenesis (originally supported by an IDDRC pilot grant)

1R01GM101313-01A1—NIH/NIGMS
01/01/2013-12/31/2016
Genetic analyses of heparan sulfate function in cell-cell interactions

Aleksandra Djukic, M.D., Ph.D. (PI)
Rett Syndrome Research Trust
05/2013-04/2014

Pharmacological treatment of Rett syndrome with glatiramer acetate (Copaxone)

Scott W. Emmons, Ph.D. (PI)
The G. Harold & Leila Y. Mathers Charitable Foundation
05/2013-04/2016
Connectomics of the nematode nervous system

Robert C. Kaplan, Ph.D. (PI)
HHSN268201300002I—NHLBI/NIH
06/01/2013-05/31/2020
Hispanic Community Health Study—Field Centers

Steven U. Walkley, D.V.M., Ph.D. (PI)
ML4 Foundation
09/1/2013-6/30/2014
Neurobiology and treatment of MLIV disease

Judith Wylie-Rosett, Ed.D., R.D. (PI)
1R01DK097096—NIDDK
09/01/2013-07/31/2017
Wellness Program Implementation: School & Student Toolkits

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
Lisa Guillory

EXECUTIVE COMMITTEE MEMBERS

Joseph C. Arezzo, Ph.D.
Francine H. Einstein, M.D.
Robert W. Marion, M.D.
Sophie Molholm, Ph.D.
Bernice E. Morrow, Ph.D.
Vytautas Verselis, Ph.D.