On December 6, 2012, the inaugural Isabelle Rapin Conference on Communication Disorders was held in Einstein’s Michael F. Price Center for Genetic and Translational Medicine/Harold and Muriel Block Research Pavilion. The event was established by Einstein’s Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (IDDRC) and the Children’s Evaluation and Rehabilitation Center (CERC), with additional sponsorship by the Williams Syndrome Center at The Children’s Hospital at Montefiore (CHAM) and the Williams Syndrome Association. Its goal was to bring together scientists and clinicians at Einstein and Montefiore, the University Hospital and academic medical center for Einstein, in order to raise awareness of communication disorders and to encourage collaboration toward identifying possible causes and treatments.

The conference was also established to honor the contributions Dr. Rapin has made—and continues to make—to the field of communication disorders during a career spanning more than half a century. “Dr. Rapin is a true pioneer in the field of child neurology, and it was fitting to dedicate this conference to her,” said Steven U. Walkley, D.V.M., Ph.D., director of the IDDRC, during introductory remarks. “Throughout her remarkable career at Einstein, she has expanded our knowledge and understanding of communication disorders while working with researchers, patients and their families. And she has mentored countless faculty and students.”

(continued on page 2)
First Isabelle Rapin Conference (continued from page 1)

The conference’s inaugural session detailed recent advances in research of Williams syndrome (WS), a rare genetic disorder that results in developmental problems that include delayed but remarkably fluent speech in the face of other cognitive deficits. Five major outside speakers were invited to give talks: Barbara Pober, M.D. (Massachusetts General Hospital and Harvard Medical School); Carolyn Mervis, Ph.D. (University of Louisville); Lucy Osborne, Ph.D. (University of Toronto); Tricia Thornton-Wells, Ph.D. (Vanderbilt University); and Brian Haas, Ph.D. (University of Georgia).

The Ethel and Samuel J. LeFrak Auditorium was filled to capacity for the event, requiring an overflow room with a video feed. Among those in attendance was the conference’s namesake, Isabelle Rapin, M.D., professor emerita of neurology and of pediatrics. She retired earlier in 2012. “I was truly heartened by the number of people who attended,” says Dr. Walkley. “It speaks to Dr. Rapin’s influence in the field and the esteem in which she is held.”

The seminar series was followed in the afternoon by interactive roundtable discussions among presenters, researchers and parents of individuals with WS. “Parents were able to obtain medical advice from clinicians and share stories with other families,” reports Michelle Disco, a genetic counselor at Montefiore Medical Center who served as one of the discussion moderators.

Establishment of the annual conference was inspired by a special event, “Rare People and Rare Talents on a Rare Day,” held in February 2012 and organized by Dr. Walkley along with Robert W. Marion, M.D., director of CERC, the Ruth L. Gottesman Chair in Developmental Pediatrics at Einstein and founder of the Williams Syndrome Center at CHAM. Among the featured performers that day was an individual with WS, recalls Dr. Walkley.

“During the event we were surprised to learn that people, including clinicians, were unfamiliar with the disorder—especially since there is a substantial clinic for Williams syndrome at Montefiore,” he says. Thus motivated, Drs. Walkley and Marion determined to initiate an annual conference that could educate clinicians and researchers about rare communication disorders. “Our goal to establish synergy between departments and across disciplines to drive research and treatment efforts was achieved, and we’re looking forward to planning next year’s conference,” concludes Dr. Walkley.

Rare People with Unique Talents

Einstein’s Rose F. Kennedy University Center for Excellence in Developmental Disabilities, the IDDRC and CERC hosted their second annual International Rare Disease Day on February 28. An art exhibit, “Rare People with Unique Talents on an Almost Rare Day,” was sponsored in collaboration with AHRC NYC and the Institutes of Applied Human Dynamics.

The artists who displayed their artwork in this exhibit are children and adults who participate in the programs named above. Works included various art media, including paintings, sculptures, paper mache and computer-generated artwork.

ON THE WEB
For full article, please visit www.einstein.yu.edu/centers/iddrc/highlights.aspx
Brain mechanisms of inattention in children with ADHD—an fMRI study

In a functional connectivity analysis using fMRI, Xiaobo Li, Ph.D., found that children with attention deficit hyperactivity disorder (ADHD) exhibited inappropriate development of the pulvinar nucleus. Such changes may lead to disrupted functional circuits for visual attention processing and thus contribute significantly to the pathophysiological mechanisms of inattentiveness symptoms in ADHD. This study was published in the Journal of the American Academy of Child and Adolescent Psychiatry (Volume 51, Number 11, pages 1197-1207.e4).

A forward genetic screen for genes involved in dendrite development

Hannes E. Buelow, Ph.D., identified a novel genetic pathway in C. elegans, the “meno-rin” pathway, that is required to shape dendrite branching patterns. He is now establishing whether this pathway also plays a critical role in mammalian dendritogenesis, and whether mutations in the menorin pathway may underlie defects in neuronal circuit formation.

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

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

Clinical trial update

NIH trial to test drug for Niemann-Pick Type C1

On January 23, the National Institutes of Health’s (NIH) National Center for Advancing Translational Sciences announced the launch of a Phase 1 clinical trial examining the safety of cyclodextrin for the treatment of Niemann-Pick Type C1 disease. Animal studies conducted by two groups of academic researchers, including Einstein’s Dr. Walkley and his graduate student, Cristin Davidson, were pivotal in establishing the efficacy of the compound for this fatal brain disorder. The first patient began receiving treatment on February 4 at the NIH Clinical Center.
NOTABLE GRANTS
The NIH has awarded researchers at Einstein a $3 million grant to investigate the short- and long-term consequences of soccer heading on the brain. The research is led by Michael L. Lipton, M.D., Ph.D., associate director of Einstein’s Gruss Magnetic Resonance Research Center and director of the IDDRC’s Translational Neuroimaging Core.

Other Recent Grants

Brett S. Abrahams, Ph.D. (PI)
The Beatrice & Samuel A. Seaver Foundation
1/6/2013-1/5/2014
Towards molecular convergence in autism via CNTNAP2

Brett S. Abrahams, Ph.D. (PI)
Autism Center of Excellence–University of California, Los Angeles
2/01/2013-1/31/2018
Autism genetics, phase II: increasing representation of human diversity

Maureen J. Charron, Ph.D. (PI)
Francine H. Einstein, M.D. (co-PI)
American Diabetes Association
1/1/2013-12/31/2015
Epigenetic effects of in utero BaP exposure

Alekandra Djukic, M.D., Ph.D. (PI)
International Rett Syndrome Foundation
10/1/2012-9/30/2013
Language abilities in girls with Rett syndrome: a pilot study of eye tracking

John J. Foxe, Ph.D. (PI)
Sophie Molholm, Ph.D. (co-PI)
National Science Foundation
9/1/2012-8/31/2013
The oscillatory control of selective attention: leveraging white matter microstructure and electrophysiology

Aristea S. Galanopoulou, M.D., Ph.D. (PI)
Solomon L. Moshe, M.D. (co-PI)
1R21NS078333-01A1—NIH/NINDS
9/30/2012-8/31/2014
Screening for new therapies for refractory infantile spasms

CURE, Citizens United for Research in Epilepsy
2/1/2013-1/31/2016
Identifying new therapies for infantile spasms

U.S. Department of Defense, PR121750
2/1/2013-1/31/2016
Identifying new therapies for infantile spasms

Jean M. Hébert, Ph.D. (PI)
Brain Research Foundation
1/1/2013-12/31/2014
How receptive is the adult neocortex to incorporating new projection neurons?

Noboru Hiroi, Ph.D. (PI)
Pablo E. Castillo, M.D., Ph.D. (co-PI)
1R01MH099660-01A1—NIH/NIMH
1/18/2013-12/31/2014
COMT and developmental memory capacity

Adam Kohn, Ph.D. (PI)
Hirschl-Weill-Caulier Career Scientist Award
1/2013-12/2017
Reading the mind: decoding neuronal population responses to predict perceptual judgments

Michael L. Lipton, M.D., Ph.D. (PI)
The Dana Foundation–David Mahoney Neuroimaging Program
Neuroimaging of cognitive dysfunction due to soccer heading-related brain injury

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

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UPCOMING EVENTS
The Inaugural Rose F. Kennedy Intellectual and Developmental Disabilities Research Center Symposium is scheduled for April 30, 2013, at the Price Center.
Please visit the website to view details of this and other upcoming events:
www.einstein.yu.edu/centers/iddrc/seminars-workshops/

EXECUTIVE COMMITTEE MEMBERS
Joseph C. Arezzo, Ph.D.
Francine H. Einstein, M.D.
Robert W. Marion, M.D.
Bernice E. Morrow, Ph.D.
Elyse S. Sussman, Ph.D.
Vytautas Verselis, Ph.D.