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Rose F. Kennedy Intellectual and Developmental Disabilities Research Center

First Isabelle Rapin
Conference on
Communication
Disorders:
Williams Syndrome
Workshop and
Roundtable



From left: Steven U. Walkley, D.V.M., Ph.D., director of the IDDRC; event honoree Isabelle Rapin, M.D.; and John J. Foxe, Ph.D., IDDRC associate director

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."



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

Synergy is appropriately defined as what happens when two individuals or groups working together produce results greater than the sum of what either could achieve alone. Now, in our second year as a newly funded center, the IDDRC and its affiliate programs have begun to show signs of synergy.

New collaborative interactions and their outcomes are evident not only between basic scientists in neuroscience and genetics, but also between these groups and the clinics that are part of CERC and CHAM.

In the past few months alone, two major areas of focus have been Rett and Williams syndromes and their respective clinics at CHAM. As detailed in this newsletter, the IDDRC sponsored an all-day workshop on Williams syndrome that brought together basic scientists and clinicians, as well as Williams syndrome families and representatives of the Williams Syndrome Foundation. For Rett syndrome, following a remarkable seminar by an IDDRC-invited speaker, Jonathan Kipnis, Ph.D. (University of Virginia), a Rett Syndrome Interest Group has been formed that meets monthly.

Through such efforts as these, we continue to do all we can to foster and synergize collaborations to advance scientific and clinical discoveries that lead to improvements in the lives of children with intellectual and developmental disabilities.

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



Aleksandra (Sasha) Djukic, M.D., Ph.D.
Associate Professor of Clinical Neurology
Saul R. Korey Department of Neurology
Associate Professor of Pediatrics
Albert Einstein College of Medicine
Director, Tri-State Rett Syndrome Center
The Children's Hospital at Montefiore

Dr. Djukic, a diplomate of the American Board of Psychiatry and of Neurology, specializes in the evaluation and treatment of children with cognitive impairment disabilities with a special expertise in girls with Rett syndrome. She directs the Tri-State Rett Syndrome Center at CHAM, one of the largest Rett syndrome clinics in the United States. According to Dr. Djukic, it was born in part from a science breakthrough a number of years ago showing that even severe symptoms of Rett syndrome in mouse models can be reversed. With support from division chair of neurology and of pediatric neurology Solomon L. Moshé, M.D., another IDDRC investigator, Dr. Djukic developed the clinically focused Rett center, with the hope that effective treatments could be developed for girls with the disorder. Research collaborations have begun to blossom, in parallel with basic science studies led by IDDRC investigator Aristeia S. Galanopoulou, M.D., Ph.D., associate professor of neurology and of neuroscience at Einstein and attending physician in the department of neurology at Montefiore.



ON THE WEB

For full article, please visit

www.einstein.yu.edu/centers/iddrc/investigators/



Michael Kraut, M.D., Ph.D.
Professor of Radiology
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Johns Hopkins University School of Medicine
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School of Behavioral and Brain Sciences
The University of Texas at Dallas

Dr. Kraut received a B.S. in Biomedical Engineering from the University of Southern California, followed by M.D. and Ph.D. degrees from Einstein. He carried out his doctoral research in the laboratories of Herbert G. Vaughan, Jr., M.D., and Joseph C. Arezzo, Ph.D., in the IDDRC. After graduation and a residency in diagnostic radiology at Beth Israel Hospital in Boston, he completed a fellowship in neuroradiology at Johns Hopkins Hospital, where he has remained on the faculty. Today, Dr. Kraut is a neuroradiologist and head of the Neuroimaging Core of the IDDRC at Johns Hopkins (The Kennedy-Krieger Institute). His ties to Einstein continue, however, as he currently serves as a member of its alumni board of governors.

Dr. Kraut's clinical interests span the field of neuroradiology and take their origins from his training at Einstein's IDDRC. His main research focus is in functional neuroimaging, especially as a tool to investigate cognitive operations related to semantic memory. A special interest, reflecting his background in neurophysiology developed while a doctoral student with Drs. Vaughan and Arezzo, is the use of functional imaging techniques in concert with electrophysiologic measures to elucidate the temporal sequence and spatial distribution of neural activation.



ON THE WEB

For full article, please visit

www.einstein.yu.edu/centers/iddrc/legacy-series/

discoveries

2012 IDDRC Pilot Project Accomplishments

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 "menorin" 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.

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.



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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)

Sophie Molholm, Ph.D. (co-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

Aleksandra 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. Moshé, 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

3/1/2013-2/28/2016

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/

UPDATES

New RFK IDDRC Members

Michael D. Brenowitz, Ph.D. (Biochemistry)
Chaim Putterman, M.D. (Medicine)
Julie Secombe, Ph.D. (Genetics)
Bridget Shafit-Zagardo, Ph.D. (Pathology)
Tao Wang, M.D., Ph.D. (Epidemiology & Population Health)

To become a member, please visit

www.einstein.yu.edu/centers/iddrc/members/become-investigator.aspx

CONGRATULATIONS!

2013 IDDRC Pilot and Feasibility Awards:

Epigenetic regulation by MeCP2 and its role in neuronal diseases

PI: Michael D. Brenowitz, Ph.D. (Biochemistry)

Synaptic connectivity using three mouse models of ASD

PI: Pablo E. Castillo, M.D., Ph.D. (Neuroscience)

Automated quantification of motor stereotypies in children with autism spectrum disorders

PI: Sylvie Goldman, Ph.D. (Neurology)

Exploring a role for AIDA-1 in schizophrenia and autism spectrum disorders

PI: Bryen A. Jordan, Ph.D. (Neuroscience)

The TWEAK/Fn14 pathway in the pathogenesis and treatment of neuropsychiatric systemic lupus erythematosus (NPSLE)

PI: Chaim Putterman, M.D. (Medicine)

Intellectual disability caused by mutations in KDM5C

PI: Julie Secombe, Ph.D. (Genetics)

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.

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Francine H. Einstein, M.D.

Robert W. Marion, M.D.

Bernice E. Morrow, Ph.D.

Elyse S. Sussman, Ph.D.

Vytautas Verselis, Ph.D.