

SPONSORED PROJECTS

Rubina A. Heptulla, M.B.B.S. (PI)
The influence of breakfast on hormone responses and cognitive performance, as assessed by CTET in obese adolescent males
 Sheryl and Daniel R. Tishman Foundation
 10/1/2016–9/30/2018

Anna Francesconi, Ph.D. (PI)
Metabotropic glutamate receptor functions in autophagy
 R01 MH108614
 7/1/2016–6/30/2021

Eliana Scemes, Ph.D. (PI)
Importance of pannexin to astrocyte and neuronal ATP signaling
 5R01 NS092786
 7/1/2016–6/30/2021

Steven U. Walkley, D.V.M., Ph.D. (PI)
Gene therapy studies in mucopolysaccharidosis IV disease
 ML4 Foundation
 6/1/2016–5/31/2017

Mark F. Mehler, M.D. (PI)
Aldrin Molero, M.D., Ph.D. co-investigator
Huntington's disease: A novel developmental oligodendroglialopathy
 1R01 NS096144
 4/1/2016–3/31/2021

Jose L. Peña, M.D., Ph.D. (PI)
Coding of auditory space in the avian brain
 2R01 DC007690
 12/1/2015–11/30/2020

E. Richard Stanley, Ph.D. (PI)
Analysis of a mouse model of adult-onset leukoencephalopathy with axonal spheroids and pigmented glia
 1R01 NS091519
 12/1/2015–11/30/2020

Steven U. Walkley, D.V.M., Ph.D. (PI)
Additional MPIs: Daniel Ory
 (Washington University School

of Medicine, St. Louis, MO) and **Fred Maxfield** (Weill Cornell Medical College, New York, NY)
Histone deacetylase inhibitors for treatment of Niemann-Pick C1 disease
 1R01 NS092653
 12/1/2015–11/30/2020

Elyse S. Sussman, Ph.D. (PI); Adam Kohn, Ph.D.; Odelia Schwartz, Ph.D.
Learning and updating internal visual models
 1R01 EY024858
 9/30/2015–6/30/2018

Peter D. Cole, M.D. (PI)
Markers of cognitive decline during treatment for childhood ALL
 1R21 CA187226
 9/17/2015–8/31/2017

Robert A. Coleman, Ph.D. (PI)
Tools for imaging the functional genome in living cells and tissues
Dr. Robert Singer, overall PI
 1U01 EB021236
 9/1/2015–8/31/2020

Caryn R. R. Rodgers, Ph.D. (PI)
Resilience among youth of color in impoverished urban communities
 6K23 HD074946
 9/1/2015–5/31/2018

R. Suzanne Zukin, Ph.D. (PI)
REST-activated program of gene expression in ischemia
 5R01 NS46742
 9/1/2015–3/31/2020

Eric Hollander, M.D. (PI)
An open-label extension study of CM-AT for the treatment of children with autism
 Curemark LLC
 8/1/2015–12/30/2016

Gary J. Schwartz, Ph.D. (PI)
Brainstem nutrient sensing in the integrative control of food intake
 6R01 DK105441
 4/15/2015–3/31/2019

2016 RFK IDDRC PILOT AWARDS

Meelad M. Dawlaty, Ph.D. (PI)
MECP2 and 5-hydroxymethylcytosine in epigenetic regulation of neuronal gene expression and etiology of Rett syndrome
 1/1/2016–12/31/2016

Pierfilippo De Sanctis, Ph.D. (PI)
The neurophysiological underpinnings of sensory-motor dysfunctions in autism spectrum disorder
 1/1/2016–12/31/2016

Kostantin Dobrenis, Ph.D. (PI)
Identification of novel pathogenic pathways and therapeutic targets in CNS of lysosomal storage disease
 1/1/2016–12/31/2016

Florence L. Marlow, Ph.D.; Alberto E. Pereda, M.D., Ph.D. (PIs)
Novel genetic models of MLIV
 1U01 EB021236
 1/1/2016–12/31/2016

Helmuth A. Sanchez Riquelme, Ph.D.; Cristin Davidson, Ph.D.; Vytautas Verselis, Ph.D. (PIs)
Cx26 and cochlear pathogenesis in deafness
 1/1/2016–12/31/2016

Deyou Zheng, Ph.D. (PI)
Uncovering the molecular link between CHD8 and ASD
 1/1/2016–12/31/2016

To become an RFK IDDRC member, please visit www.einstein.yu.edu/centers/iddrc/members/become-investigator.aspx.

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 among bench scientists and clinicians.

ADMINISTRATION

- Director**
Steven U. Walkley, D.V.M., Ph.D.
- Associate Director**
Sophie Molholm, Ph.D.
- Administrator**
Lisa Guillory, M.A.
- Administrative Assistant**
Frances Andrade

EXECUTIVE COMMITTEE MEMBERS

- Aleksandra Djukic, M.D., Ph.D.
- Craig Branch, Ph.D.
- Bernice E. Morrow, Ph.D.
- Vytautas Verselis, Ph.D.
- Melissa P. Wasserstein, M.D.

To support the work of the RFK IDDRC, contact:

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EVENTS

On May 24, 2016, the ICare4Autism National Conference, offered in conjunction with Montefiore and Albert Einstein College of Medicine, provided the opportunity to meet and network with leading professionals in the medical, academic, research, biopharma and healthcare industries. The all-day conference took place at Montefiore's Cherkasky Auditorium. See more at www.icare4autism.org.

On May 22, 2016, Einstein and Montefiore hosted the annual 22q at the Zoo event. See more at www.einstein.yu.edu/r/zoo2016. For more about 22q at Montefiore, please go to www.cham.org/programs-centers/22q11-2-deletion-syndrome.

IN THE MEDIA, 2015–16

John Greally, M.B., B.Ch., Ph.D., the RFK IDDRC NGEN director, was interviewed by *Nature* regarding his criticism of a *New Yorker* article (May 10, 2016) on epigenetics. See more at www.einstein.yu.edu/r/greally2016-nature.

The New York Times (December 3, 2015) interviewed **Dr. Greally** about a new study that suggests a father's experiences may influence the biology of his offspring. See more at www.einstein.yu.edu/r/greally2015-nytimes.

On November 4, 2015, as part of its New York City outreach program, **StoryCorps**, which has collected and archived more than 50,000 interviews with 100,000 participants, visited the Bronx for a day of interviews with parents whose children receive RFK IDDRC CERC services. See more at www.einstein.yu.edu/r/story2016.

The Telegraph (UK) (October 15, 2015) quotes **Scott W. Emmons, Ph.D.**, the Siegfried Ullmann Chair in Molecular Genetics, about his *Nature* study, which found that male nematode worms have neurons that allow them to prioritize mating. See more at www.einstein.yu.edu/r/emmons2016-telegraph.

UPCOMING EVENTS

To view details of upcoming RFK IDDRC-sponsored or IDD-relevant events, please visit www.einstein.yu.edu/centers/iddrc/seminars-workshops/.



Albert Einstein College of Medicine

Science at the heart of medicine

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NEWS

Rose F. Kennedy Intellectual and Developmental Disabilities Research Center

On Beauty: Celebrating International Rare Disease Day 2016

Einstein's Rose F. Kennedy Intellectual and Developmental Disabilities Research Center (RFK IDDRC) hosted its annual Rare Disease Day celebration on Monday, February 29, 2016—a rare "leap" day, which was chosen by EURORDIS, an alliance of rare-disease organizations in Europe, as a tribute to those who are genetically different.

In observance, Sophie Molholm, Ph.D., the new associate director of the RFK IDDRC, introduced the program and provided a brief recap of the history of the IDDRC at Einstein. She also underscored the importance of basic science research in translational medicine. Following her introductory remarks was a screening of the short documentary film *On Beauty* by Emmy-nominated filmmaker Joanna Rudnick. The film was shown to a packed and enthusiastic audience in the Ethel and Samuel J. LeFrak Auditorium in the Michael F. Price Center for Genetic and Translational Medicine/Harold and Muriel Block Research Pavilion. *On Beauty* features award-winning former fashion photographer Rick Guidotti, who challenges how beauty is defined by producing visually

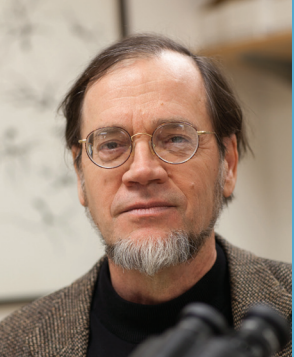


Rick Guidotti, director of Positive Exposure, with one of his photographs.

compelling images of people living with a wide range of genetic disorders. Through the nonprofit organization Positive Exposure, which Mr. Guidotti established to address and help people overcome myths and misperceptions about individuals born with genetic abnormalities, he aims to help those living with serious disorders change how they feel about themselves and how others perceive them. The film follows Mr. Guidotti during his travels

as he finds new photographic subjects, inspires hope and discusses his transition from the world of fashion to advocacy. Between the movie and presentations given by Mr. Guidotti and Ms. Rudnick, Alena Galan, an 18-year-old college-bound patient of Robert W. Marion, M.D., with Maroteaux-Lamy syndrome (mucopolysaccharidosis type VI), sang Miley Cyrus's "The Climb"—the song,

(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, Saul R. Korey Department of Neurology

The discovery of an intellectual or developmental disability (IDD), whether genetic or environmental, initiates a demanding yet critical array of responses from the medical community.

Whether such disease "discoveries" are more than a century old (Tay-Sachs disease) or very recent (Zika virus and microcephaly), the needs are similar: Disease causation, diagnostic tests and clinics specializing in patient care must be established, and in some cases natural history studies and patient registries are critical. Along the way, scientists, including both cognitive and bench neuroscientists, geneticists and cell biologists, are needed to provide insights into disease pathogenesis and therapy.

The journey from disease discovery to successful clinical care can be long. While no single academic medical center would necessarily be expected to focus on every step in this cycle of translational medicine, the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center, conjoined as it is with Einstein's basic science departments and the Children's Evaluation and Rehabilitation Center and the Children's Hospital at Montefiore, does indeed play a key role in fostering individual steps in this cycle of progress for a great variety of IDDs.

Rare Disease Day 2016 (continued from page 1)

she explained, that best summarizes her life and personal struggles.

Following the screening, performance and presentations, Ms. Rudnick and Mr. Guidotti answered questions before joining the audience of students, faculty and guests on Main Street in the Leo Forchheimer Medical Science Building for a meet-the-speakers reception, where members of the audience interacted with the presenters while enjoying an exhibit of Mr. Guidotti's photographs. See more at www.einstein.yu.edu/features/around-campus/440/rare-disease-day.



Speakers and presenters at Rare Disease Day included (from left): Joanna Rudnick, documentary filmmaker; Rick Guidotti; Alena Galan; Robert W. Marion, M.D., IDDRC Internal Advisory Committee member; Sophie Molholm, Ph.D., IDDRC associate director; and Howard M. Steinman, Ph.D., assistant dean for biomedical science education.

FOURTH ANNUAL ISABELLE RAPIN CONFERENCE ON COMMUNICATION DISORDERS: 22Q11.2DS (DIGEORGE SYNDROME/VCFS)

The RFK IDDRC hosted its fourth annual Isabelle Rapin Conference on December 3, 2015. Each year, the conference highlights research on a specific neurodevelopmental disorder, and this year the chosen focus was 22q11.2 deletion syndrome—also known as DiGeorge syndrome or velocardiofacial syndrome. The conference was organized by the assistant director of the IDDRC Neurogenomics Core (NGEN), Bernice E. Morrow, Ph.D., whose lab studies key genetic pathways involved in the development of 22q11.2 deletion syndrome. Guest speakers included Wendy R. Kates, Ph.D., of the State University of New York Upstate Medical University; Donna M. McDonald-McGinn, M.S., L.C.G.C., of the Children's Hospital of Philadelphia; Ann Swillen, Ph.D., of the University of Leuven, in Belgium; and Carrie E. Bearden, Ph.D., from the University of California–Los Angeles Center for the Assessment and Prevention of Prodromal States. Einstein faculty member Noboru Hiroi, Ph.D., a professor of psychiatry and behavioral sciences, gave a compelling talk on phenotypic variation and gene identification in DiGeorge syndrome, "22q11.2 Copy Number Variation: Identifying Individual Genes for Autism and Schizophrenia in Mouse Models." See more at www.einstein.yu.edu/r/rapin2015 and www.einstein.yu.edu/centers/iddrc/seminars-workshops/.



From left: Bernice E. Morrow, Ph.D.; Wendy R. Kates, Ph.D.; Donna M. McDonald-McGinn, M.S.; Ann Swillen, Ph.D.; Isabelle Rapin, M.D.; Carrie E. Bearden, Ph.D.; and Dr. Walkley.

featured investigators

MELISSA P. WASSERSTEIN, M.D.



Division Chief, Pediatric Genetic Medicine
Children's Hospital at Montefiore
Associate Professor of Pediatrics
Albert Einstein College of Medicine

On April 1, 2016, Melissa P. Wasserstein, M.D., was appointed division chief of pediatric genetic medicine at the Children's Hospital at Montefiore and associate professor of pediatrics at Einstein. She is also the new associate director of the Human Clinical Phenotyping Core (HCP) and will work with Dr. Sophie Molholm, associate director of the RFK IDDRC and director of the HCP. Before joining Einstein-Montefiore, Dr. Wasserstein was the medical director of the International Center for Types A and B Niemann-Pick at Mount Sinai Medical Center.

A biochemical geneticist and clinician-scientist, Dr. Wasserstein has focused during her career on the diagnosis and treatment of patients with inborn errors of metabolism, many of which have significant IDD-related manifestations. Over the

years, she has developed specific expertise in assessing how to better diagnose and treat rare metabolically related disorders.

Dr. Wasserstein is the principal investigator of a National Institute of Child Health and Human Development–funded multicenter pilot newborn screening program for four lysosomal storage diseases. The program assesses the validity of the screening assays, measures the incidence of disease in a diverse population and evaluates the ethics of adding these types of disorders to mandated screening panels. To date, this project has screened more than 41,000 babies in conjunction with the New York State Newborn Screening Laboratory, and works with the National Institutes of Health–funded Newborn Screening Translational Network to facilitate data sharing, using disease-specific data sets for long-term pediatric data repositories. Dr. Wasserstein has also been the principal investigator of many industry-sponsored clinical trials evaluating novel therapeutics for rare inborn errors of metabolism. She is a welcome addition to the RFK IDDRC.

KAMRAN KHODAKHAH, Ph.D.



Chair, Dominick P. Purpura Department of Neuroscience
Harold and Muriel Block Chair in Neuroscience
Albert Einstein College of Medicine

On May 10, 2016, Albert Einstein College of Medicine officially announced the appointment of Dr. Kamran Khodakhah as chair of the Dominick P. Purpura Department of Neuroscience. Dr. Khodakhah, an IDDRC member, joined the Einstein faculty in 2001 as an assistant professor. By 2007 he was a full professor and soon thereafter went on to receive secondary appointments in the Saul R. Korey Department of Neurology and in the department of psychiatry and behavioral sciences, where he is now vice chair of research.

Dr. Khodakhah's laboratory focuses on the role of the cerebellum and basal ganglia in motor coordination and in movement disorders such as ataxia (uncoordinated movement) and dystonia (involuntary muscle contraction). Over the years, his lab has maintained a large, continuously active portfolio of National Institutes of Health and foundation-related grants, and he has been an active member of the scientific research community. Further, he has served or is serving on a range of advisory boards, including those of the Simons Foundation, the Bachmann-Strauss Dystonia Foundation and the Gass Foundation. In 2007, Dr. Khodakhah received Einstein's LaDonne H. Schulman Excellence in Teaching Award. See more at www.einstein.yu.edu/r/khodakhah2016.

congratulations

We are pleased to announce that **Noboru Hiroi, Ph.D.**, RFK IDDRC member and professor of psychiatry and behavioral sciences, was awarded the 2016 Lilly Neuroscience Basic Research Award by the International College of Neuropsychopharmacology (CINP). Dr. Hiroi was honored at the opening ceremony of the CINP congress held in Seoul, Korea, on July 3–5, 2016. See more at www.einstein.yu.edu/r/hiroi2016.

Scott W. Emmons, Ph.D., the Siegfried Ullmann Chair in Molecular Genetics, received the Marshall S. Horwitz, M.D., Faculty Prize for Research on February 29, 2016, for his innovative research in the field of connectomics, which describes how the synapses of the nervous system are connected. See more at www.einstein.yu.edu/r/emmons2016.

On March 21, 2016, the Graduate Program in Biomedical Sciences held the 20th annual Julius Marmur Symposium, which honors excellence in graduate student research. This year two RFK IDDRC members had students in their labs recognized: **Phillip Campbell**, a graduate student in the laboratory of Florence L. Marlow, Ph.D., for his work on zebrafish development, and **Fanny Cazettes**, a computational neuroscientist in the laboratory of Jose Luis Peña, M.D., Ph.D., for her research on how the brain handles sensory uncertainty. See more at www.einstein.yu.edu/r/pena2016.