



**Mount  
Sinai**

*The Mindich  
Child Health and  
Development Institute*

# Annual Report 2017



## **The Mindich Child Health and Development Institute (MCHDI)**

is a translational research enterprise with the mission of advancing knowledge and therapies for diseases affecting infants, children, and adolescents. Led by Bruce D. Gelb, MD, the MCHDI provides an intellectually rich and supportive environment for fostering collaborative scientific investigation and Mount Sinai's "bench to bedside" philosophy, as well as training the next generation of scientific leaders in pediatric medicine.

Physician-scientists and scientists at the MCHDI work in a multidisciplinary manner with researchers and physicians in various departments and institutes at Mount Sinai. Together, we strive toward the objectives of developing robust paradigms for understanding the effects of genetics and environment on the health of infants, children and adolescents, and personalizing pediatric medicine through genetics and genomics.

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# Message from the Director

In 2017, the biomedical research community faced uncertainties about the future of federal support for scientific research, both quantitatively and qualitatively. Scientists made their voices heard through events such as the March for Science last spring. This past November, we hosted a workshop on communicating science, led by speaker James Rea from the Alan Alda Center at Stony Brook University, at our 5th Annual Retreat. We emphasized the importance of effectively communicating science, not only among researchers in different fields but to non-scientific audiences. As leaders in the field of child health research, it is crucial that we advocate for science and its significance in language that is accessible to parents, donors, and policy makers.

This year, the **Mindich Child Health and Development Center** (MCHDI) continued to recruit new faculty members to add to our core research areas: allergy & asthma, cardiovascular disease, neurodevelopmental disorders, obesity & diabetes and others. We currently total 61 faculty, including three extramural members (Minji Byun, PhD; Alan Groves, MBChB, MD; Shelley H. Liu, PhD) and three intramural members (Charlotte Cunningham-Rundles, MD, PhD; Ke Hao, ScD; Amy R. Kontorovich, MD, PhD), contributing to over 350 publications.

Furthermore, two major initiatives were announced as part of our efforts to address the long-term strategic aims that were set forth by our advisory boards and Leadership Council. In 2016, a Trainee Leadership Council (TLC) was formed to build a stronger community of young scientists focused on child health. The TLC hosted various workshops and events geared towards trainees. In 2017, they continued to host various social events and workshops, including a grant writing panel featuring trainees with funded grants. The TLC also worked with MCHDI staff to establish the first major program for trainees – a new trainee pilot grant program to fund early career scientists pursuing innovative basic, clinical, or translational research under the direction of MCHDI faculty. Two trainees with the most promising research ideas were awarded in the amount of \$10K for one year and presented their work during our 5th Annual Retreat.

Thanks to generous funding by the Genetic Disease Foundation, we were able to launch successfully the Mount Sinai Pediatric Precision Medicine Initiative, in collaboration with the **Departments of Pediatrics** and **Genetics and Genomic Sciences**, the **Icahn Institute for Genomics and Multiscale Biology**, **Charles Bronfman Institute for Personalized Medicine**, and the **Precision Immunology Institute** as well as other departments/institutes within Mount Sinai. The goal of this initiative is to perform rapid next generation sequencing for pediatric patients with rare undiagnosed diseases. The current standard is to utilize genetic testing as the last resort, often after a child and family have been subjected to months or years of a stressful and costly medical odyssey. This work will not only help children and families enrolled in the program, in some cases dramatically, but also help to establish pediatric precision medicine as the new standard of care at Mount Sinai and serve to educate pediatric trainees in the rapidly evolving practice of genomic medicine.



As we move forward into the new year, we hope to bridge the gap between research and practice, as well as science and policy. Now more than ever, is the time to unite the scientific community and advocate the need for increased funding to pursue quality research. The MCHDI will firmly support our diverse team of researchers and demonstrate how vital their work is in improving the lives of our children.

A handwritten signature in black ink, appearing to read "B. Gelb". The signature is fluid and cursive.

Bruce D. Gelb, Director

# Faculty Growth

In 2017, we welcomed three extramural and three intramural faculty members to our institute. Currently, we total 61 members consisting of scientists and physician-scientists across the disciplines of Allergy & Asthma, Cardiovascular Disease, Neurodevelopmental Disorders, Obesity & Diabetes, and more.



MCHDI faculty members from left to right: Nicole C. Dubois, PhD; Adolfo Garcia-Ocaña, PhD; Donald K. Scott, PhD; M. Cecilia Berin, PhD; Minji Byun, PhD; David Dunkin, MD; Dalila Pinto, PhD; Andrew J. Sharp, PhD; Dani Dumitriu, MD, PhD; Bruce D. Gelb, MD; Amy R. Kontorovich, MD, PhD; Martin J. Walsh, PhD; Jia Chen, ScD; Rupangi C. Vasavada, PhD

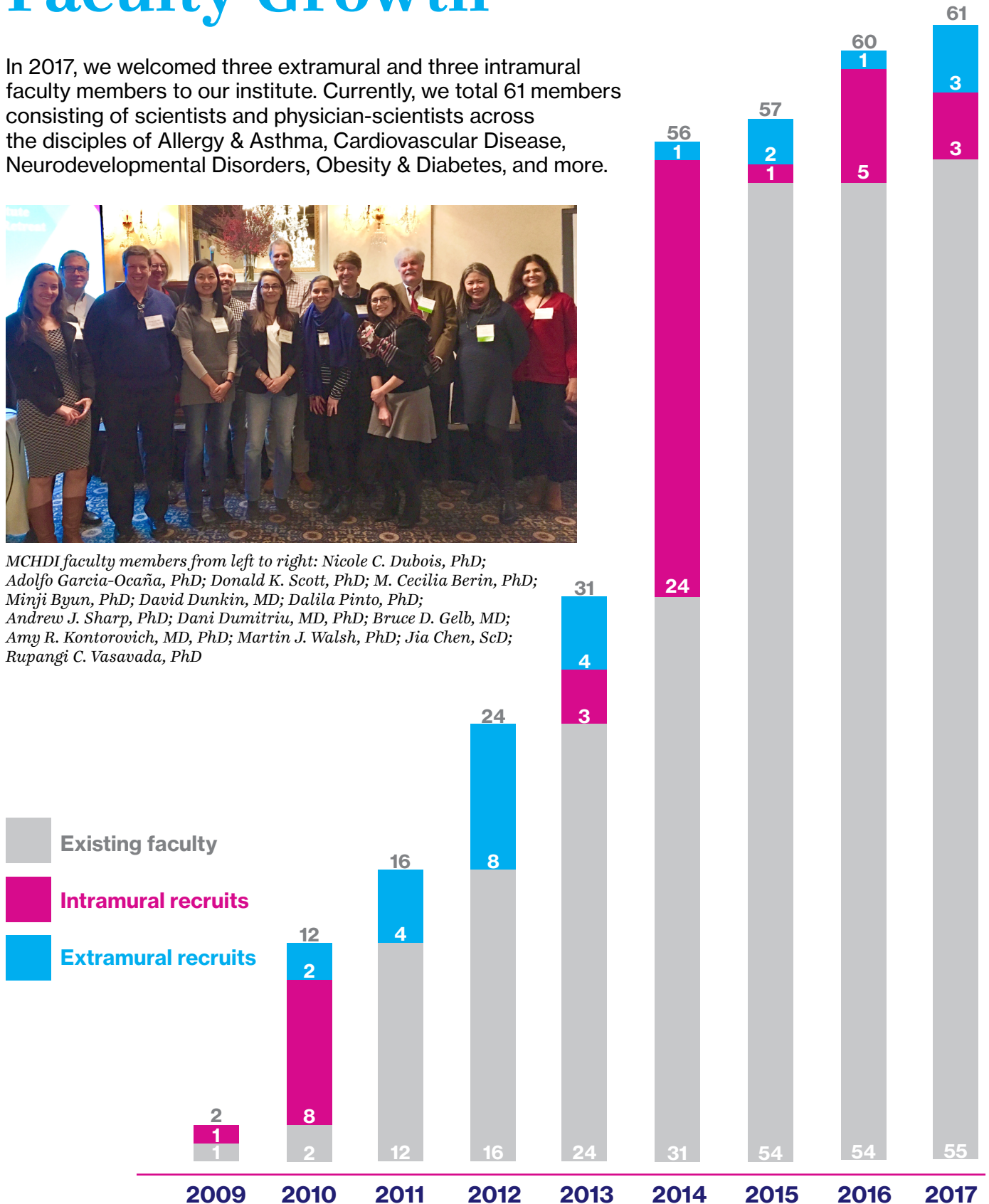


Chart of faculty recruits since our inception in 2009. In 2017, our institute recruited three new extramural and three intramural faculty members to our institute.



# New Faculty

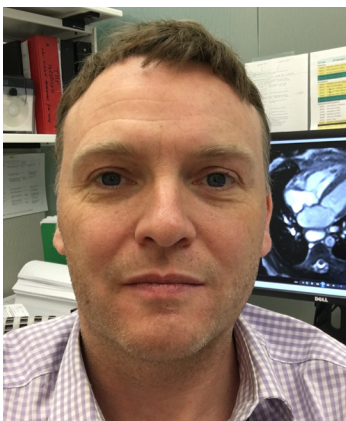
## New Extramural Faculty



*Minji Byun, PhD  
Assistant Professor, Medicine*

### ***Minji Byun, PhD***

Minji Byun, PhD is a tenure-track Assistant Professor in the Department of Medicine, Division of Clinical Immunology, and a member of the Precision Immunology Institute. She received her undergraduate degree in Life Science from POSTECH, South Korea, where she studied 3D structures of peptidoglycan recognition proteins by X-ray crystallography. She completed her PhD thesis at Washington University in St. Louis on poxvirus-encoded immune evasion mechanisms targeting the MHC class I antigen presentation pathway. She then performed her postdoctoral studies at The Rockefeller University, where she made the novel finding that pediatric Kaposi sarcoma is associated with primary immunodeficiency and mapped the disease cause down to the single gene level. She also ascertained the molecular mechanisms behind the immune defects caused by these mutations. In 2014, she joined as faculty at the Washington University in St. Louis, with a research focus on genetic and immunological mechanisms underlying susceptibility to rare immune disorders. In 2017, she was recruited to Icahn School of Medicine at Mount Sinai to join the Precision Immunology Institute. Her current research focus includes Kawasaki disease, an acute systemic vasculitis primarily affecting children, and idiopathic multicentric Castleman disease, a rare lymphoproliferative disorder affecting people of all ages. She hypothesizes that rare high-impact variants – inborn or acquired – underlie susceptibility to these disorders. Her laboratory uses various cutting-edge human genetics tools to identify candidate morbid variants, which are then investigated for their pathogenic roles in patient-derived cells as well as in vivo animal models.



*Alan Groves, MBChB, MD  
Associate Professor, Pediatrics*

### ***Alan Groves, MBChB, MD***

Alan Groves, MBChB, MD is an Associate Professor in the Division of Newborn Medicine within the Department of Pediatrics. He received his medical degree from the University of Edinburgh (1996) and undertook postgraduate pediatric training in the UK and New Zealand where he completed a research degree in neonatal echocardiography. Since completing his training, Dr. Groves has worked as an attending in Neonatal Medicine in London (Queen Charlotte's and Chelsea Hospital, St Thomas' Hospital) and New York (Weill Cornell). His primary research interest is in assessing heart function and growth using echocardiography and magnetic resonance imaging. He was awarded a Medical Research Council (UK) Clinician Scientist Fellowship to develop cardiac magnetic resonance imaging and advanced ultrasound techniques in newborns. These techniques have added significantly to understanding of the pathophysiology of the newborn circulation. Application of cardiac magnetic resonance imaging and atlas has demonstrated abnormal heart growth in infants born prematurely. He is currently funded by the American Heart Association to study a cohort of premature infants to determine the triggers and signaling

## *New Extramural Faculty* continued

### *Alan Groves, MBChB, MD continued*

mechanisms of altered heart development. His long-term research aim is to produce reductions in mortality and morbidity in newborn infants through improvements in circulatory monitoring and support.



*Shelley H. Liu, PhD  
Assistant Professor, Population  
Health Science and Policy*

### *Shelley H. Liu, PhD*

Shelley H. Liu, PhD is an Assistant Professor in the Center for Biostatistics within the Department of Population Health Science and Policy. She received her undergraduate degree in Biological Sciences, with a concentration in Physiology, and Statistics, from Northwestern University in 2011. She completed her PhD thesis in 2016 at Harvard University under advisor Brent Coull, where she developed Bayesian statistical methods for children's environmental health research. Specifically, she developed new models to study how time-varying exposures to chemical mixtures affect neurodevelopment, and identified critical time windows of exposure. She also studied the interaction and effect modification of co-exposures. During her PhD, she also developed a method to account for missing data in HIV viral genetic linkage analysis. Since joining the Icahn School of Medicine at Mount Sinai, she is interested in children's health research and environmental epidemiology.

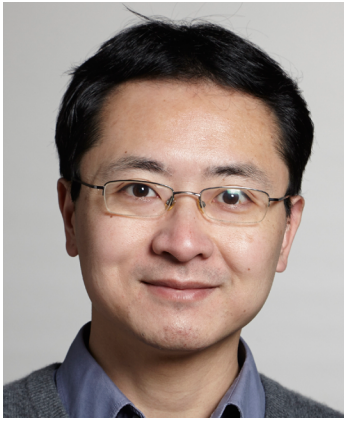
## *New Intramural Faculty*



*Charlotte Cunningham-  
Rundles, MD, PhD  
Professor, Medicine  
Professor, Pediatrics*

### *Charlotte Cunningham-Rundles, MD, PhD*

Charlotte Cunningham-Rundles, MD, PhD, the David S. Gottesman Professor of Medicine/Immunology, and Pediatrics, is acting head of the Division of Clinical Immunology, Department of Medicine. She is a graduate of Duke University, and received her MD from Columbia College of Physicians and Surgeons, and her PhD in Immunology from New York University School of Medicine. She has served on a number of training grant review committees as well as editorial boards and national organizations. She is the PI of an interactive PPG on primary immunodeficiency, with Drs. Andrea Cerutti, PJ Maglione, Sergio Lira, and Adrian Ting at Mount Sinai; Eric Meffre at Yale University, Dr. Jean Laurent Casanova at Rockefeller University. She is the PI of a NIH sponsored U24 Cooperative Grant on Primary Immune Deficiency (USIDNet), and is an investigator in a new Clinical Sequencing Exploratory Research (CSER) Consortium with colleagues from Mount Sinai and Montefiore. Her overall area of investigation is human immunodeficiency diseases and immuno-reconstitution. Her research focus has been on the pathogenesis and treatment of B cell defects leading to hypogammaglobulinemia. However, due to the complex nature of B cell activation, the genetic causes are multiple and include molecular defects in the bone marrow, lymph node and cellular compartments. With her colleagues at Rockefeller, Yale and the NIH, they have been using whole genomic sequencing to elucidate causes of B cell failure. Their studies show that they can identify the defective gene in about 30% of cases, leading also to better treatment options.



*Ke Hao, ScD  
Associate Professor, Genetics  
and Genomic Sciences*

***Ke Hao, ScD***

Ke Hao, ScD is a tenured Associate Professor in the Department of Genetics and Genomic Sciences, and a member of the Icahn Institute for Genomics and Multiscale Biology. He received his undergraduate degree in Biological Sciences and Technology at Tsinghua University, China. He graduated with a Doctor of Science degree at Harvard University in Environmental Health. He then performed his postdoctoral studies in the Department of Biostatistics at Harvard University, focusing on algorithm development and analysis of genetics and genomics data. In 2012, he was recruited to Icahn School of Medicine at Mount Sinai. His current research activities include: (1) environmental health, (2) genetics of human diseases, and (3) computational biology algorithm development. He invented an integrated system, Bio3Air, to measure the amount of ambient PM2.5 inhaled over a long period of time with very high spatial and time resolution. The system is now applied in environment epidemiology studies in China led by Dr. Hao. He was also involved in a large number of genetics studies, including linkage scans, genome-wide association studies (GWAS) and whole-exome/whole genome studies (NGS) on many disease areas (e.g COPD and IBD). Dr. Hao led projects to systematically characterize genetic control of transcriptome and proteome (i.e., eQTLs and pQTLs) in human tissues and also construct gene networks. The xQTLs were integrated with GWAS or NGS to provide insight on the molecular mechanism underlying disease genetic predisposition.



*Amy R. Kontorovich, MD, PhD  
Assistant Professor, Medicine  
Medical Director, Adult  
Cardiovascular Genetics*

***Amy R. Kontorovich, MD, PhD***

Amy R. Kontorovich, MD, PhD is an Assistant Professor of Medicine in Cardiology and is the Medical Director of Adult Cardiovascular Genetics in the Zena and Michael A. Wiener Cardiovascular Institute. Dr. Kontorovich received a BSE in Bioengineering from the University of Pennsylvania where she studied biomechanical effects on endothelial cells in atherogenesis. She fulfilled the MD and PhD degrees from Stony Brook University. Her doctoral work with Dr. Ira Cohen involved developing a novel quantum-dot nanoparticle-based method for tracking stem cells after delivery to the heart. Dr. Kontorovich completed both the Internal Medicine residency and fellowship in Cardiology at the Icahn School of Medicine at Mount Sinai. Her research is focused on myocarditis, an inflammatory condition of the heart that can lead to sudden cardiac death and cardiomyopathy. She studies genetic factors that mediate this rare and potentially fatal outcome of infection by common viruses.



# Faculty Research Areas

## *Asthma and Allergy*

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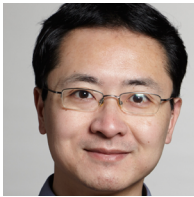
**M. Cecilia Berin, PhD** (Associate Professor, Pediatrics)  
**Research Areas:** Immune mechanisms of food allergy and regulation of immune tolerance  
**Faculty Interactions:** Rosalind J. Wright, David Dunkin, Hugh A. Sampson, Scott H. Sicherer, Anna Nowak-Wegrzyn, Supinda Bunyavanich

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**Supinda Bunyavanich, MD, MPH** (Associate Professor, Pediatrics & Genetics and Genomic Sciences)  
**Research Areas:** Integrative genomics of asthma and allergic diseases  
**Faculty Interactions:** Hugh A. Sampson, Andrew J. Sharp, Scott H. Sicherer, Xiu-Min Li, M. Cecilia Berin, Anna Nowak-Wegrzyn

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**Ke Hao, ScD** (Associate Professor, Genetics and Genomic Sciences)  
**Research Areas:** Genetic Pleiotropy, mendelian randomization, inflammatory bowel disease, placenta biology, ambient air particulate matter exposure  
**Faculty Interactions:** Jia Chen, Luca Lambertini

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**Xiu-Min Li, MD** (Professor, Pediatrics)  
**Research Areas:** Integrative medicine for induction of immune tolerance for food allergy, asthma and Inflammatory Bowel Disease  
**Faculty Interactions:** Hugh A. Sampson, Julie Wang, Scott H. Sicherer, Anna Nowak-Wegrzyn, Jia Chen, Martin J. Walsh, David Dunkin, Supinda Bunyavanich

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**Anna Nowak-Wegrzyn, MD** (Associate Professor, Pediatrics)  
**Research Areas:** Food allergy, FPIES, oral immunotherapy, anaphylaxis, milk and egg allergy  
**Faculty Interactions:** M. Cecilia Berin, Supinda Bunyavanich, Hugh A. Sampson, Julie Wang, Xiu-Min Li, Scott H. Sicherer

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**Hugh A. Sampson, MD** (Kurt Hirschhorn Professor, Pediatrics)  
**Research Areas:** Immunopathogenesis of food allergy and anaphylaxis  
**Faculty Interactions:** M. Cecilia Berin, Supinda Bunyavanich, David Dunkin, Scott H. Sicherer, Xiu-Min Li, Julie Wang, Anna Nowak-Wegrzyn, Rosalind J. Wright

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**Scott H. Sicherer, MD** (Director, Jaffe Food Allergy Institute; Division Chief, Pediatric Allergy; Elliot Roslyn Jaffe Professor, Pediatrics)  
**Research Areas:** Food allergy epidemiology, treatments, natural course, quality of life  
**Faculty Interactions:** Eyal Shemesh, Supinda Bunyavanich, M. Cecilia Berin, Hugh A. Sampson, Julie Wang, Xiu-Min Li, Anna Nowak-Wegrzyn

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**Julie Wang, MD** (Associate Professor, Pediatrics)  
**Research Areas:** Novel therapeutics for food allergy, epidemiology and management of food allergy in minority, urban populations  
**Faculty Interactions:** Hugh A. Sampson, Scott H. Sicherer, Xiu-Min Li, Anna Nowak-Wegrzyn, Shelley H. Liu

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## Asthma and Allergy continued



**Karen M. Wilson, MD, MPH** (Debra and Leon Black Division Chief, General Pediatrics; Vice-Chair for Clinical and Translational Research, Pediatrics; Professor, Pediatrics)

**Research Areas:** Secondhand tobacco smoke, secondhand marijuana smoke, inpatient respiratory illness

**Faculty Interactions:** Rosalind J. Wright, Shelley H. Liu



**Rosalind J. Wright, MD, MPH** (Dean, Translational Biomedical Research; Director, Clinical and Translational Science Award; Horace W. Goldsmith Professor, Children's Health Research; Professor, Pediatrics & Environmental Medicine & Public Health)

**Research Areas:** Developmental epidemiology, environmental and chemical exposures, social and psychological influences in neurodevelopmental diseases, health disparities

**Faculty Interactions:** Robert O. Wright, Manish Arora, Allan C. Just, Chris Gennings, Annemarie Stroustrup, M. Cecilia Berin, Jia Chen, Karen M. Wilson, Hugh A. Sampson, Susan Teitelbaum

## Cardiovascular Disease



**Harold S. Bernstein, MD, PhD** (Adjunct Professor, Pediatrics)

**Research Areas:** Drug development (target validation through clinical proof of concept), heart failure, metabolic syndrome, diabetes, thrombosis, chronic kidney disease

**Faculty Interactions:** Bruce D. Gelb



**Chen-Leng Cai, PhD** (Associate Professor, Cell, Developmental & Regenerative Biology)

**Research Areas:** Heart development and regeneration

**Faculty Interactions:** Bruce D. Gelb, Nicole C. Dubois, Anne Moon, Martin J. Walsh, Jianlong Wang



**Nicole C. Dubois, PhD** (Assistant Professor, Cell, Developmental & Regenerative Biology)

**Research Areas:** Heart development, stem cell differentiation, disease modeling

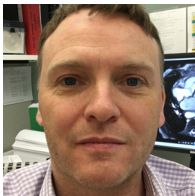
**Faculty Interactions:** Bruce D. Gelb, Chen-Leng Cai, Robert S. Krauss, James J. Bieker, Andrew J. Sharp, Martin J. Walsh, Michael Rendl, Jianlong Wang



**Bruce D. Gelb, MD** (Gogel Family Professor and Director, Mindich Child Health and Development Institute; Professor, Pediatrics & Genetics and Genomic Sciences)

**Research Areas:** Genetics of cardiovascular diseases, stem cell research

**Faculty Interactions:** Ross L. Cagan, Nicole C. Dubois, Alex Kolevzon, Andrew J. Sharp, Dusan Bogunovic, Chen-Leng Cai, Megan K. Horton, Harold S. Bernstein, Anne Moon, Annemarie Stroustrup, Charlotte Cunningham-Rundles, Amy R. Kontorovich



**Alan Groves, MBChB, MD** (Associate Professor, Pediatrics)

**Research Areas:** Hemodynamics, cardiac function, echocardiography, magnetic resonance imaging

**Faculty Interactions:** Annemarie Stroustrup



**Anne Moon, MD, PhD** (Adjunct Professor, Pediatrics)

**Research Areas:** Developmental biology of congenital heart disease and limb defects, functions of Tbx and Fibroblast Growth Factors

**Faculty Interactions:** Bruce D. Gelb, Chen-Leng Cai

## Cardiovascular Disease continued



**Amy R. Kontorovich, MD, PhD** (Medical Director, Adult Cardiovascular Genetics; Assistant Professor, Medicine)

**Research Areas:** Myocarditis, genetics of cardiovascular diseases, stem cell research

**Faculty Interactions:** Bruce D. Gelb

## Neurodevelopmental Disorders



**Manish Arora, PhD, BDS, MPH** (Professor, Environmental Medicine & Public Health & Dentistry)

**Research Areas:** Environmental epidemiology and exposure biology

**Faculty Interactions:** Robert O. Wright, Rosalind J. Wright, Hirofumi Morishita, Megan K. Horton, Chris Gennings, Jia Chen, Allan C. Just, Lisa M. Satlin, Annemarie Stroustrup, Shanna H. Swan, Philip J. Landrigan, Jeffrey M. Saland, Avi Reichenberg, Susan Teitelbaum, Shelley H. Liu



**Joseph D. Buxbaum, PhD** (Deputy Chair, Department of Psychiatry; Director, Seaver Autism Center for Research and Treatment; Professor, Psychiatry, Neuroscience, & Genetic and Genomic Sciences)

**Research Areas:** Autism spectrum disorder, neurodevelopmental disorders, gene discovery, functional genetics, molecular and cellular neuroscience, cell and animal model systems

**Faculty Interactions:** Alex Kolevzon, Hirofumi Morishita, Coro Paisan-Ruiz, Andrew J. Sharp, Shanna H. Swan, Dorothy E. Grice, Avi Reichenberg, Ross L. Cagan, Dalila Pinto, Eyal Shemesh



**Jia Chen, ScD** (Professor, Pediatrics, Environmental Medicine & Public Health, Medicine, & Oncological Sciences)

**Research Areas:** Environmental epigenetics, molecular epidemiology

**Faculty Interactions:** Robert O. Wright, Manish Arora, Rosalind J. Wright, Luca Lambertini, Chris Gennings, Ke Hao, Andrew J. Sharp, Xiu-Min Li, Susan Teitelbaum



**Dani Dumitriu, MD, PhD** (Assistant Professor, Neuroscience)

**Research Areas:** Functional and structural connectomics underlying resilience to mouse social defeat

**Faculty Interactions:** Megan K. Horton, Hirofumi Morishita, Vilma Gabbay, Lisa M. Satlin, Robert O. Wright



**Lisa Eiland, MD** (Assistant Professor, Pediatrics)

**Research Areas:** Stress and neurodevelopment

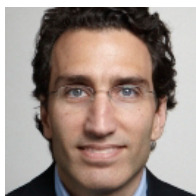
**Faculty Interactions:** Annemarie Stroustrup



**Megan K. Horton, PhD, MPH** (Assistant Professor, Environmental Medicine & Public Health)

**Research Areas:** Children's environmental health, exposure assessment, pediatric neuroimaging

**Faculty Interactions:** Bruce D. Gelb, Robert O. Wright, Chris Gennings, Annemarie Stroustrup, Allan C. Just, Shanna H. Swan, Manish Arora, Dani Dumitriu



**Alex Kolevzon, MD** (Director, Child and Adolescent Psychiatry; Professor, Psychiatry & Pediatrics)

**Research Areas:** Autism spectrum and other neurodevelopmental disorders

**Faculty Interactions:** Joseph D. Buxbaum, Bruce D. Gelb, Avi Reichenberg, Dalila Pinto, Jeffrey M. Saland, Eyal Shemesh, Shanna H. Swan

## Neurodevelopmental Disorders *continued*

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**Robert S. Krauss, PhD** (Professor, Cell, Developmental & Regenerative Biology & Oncological Sciences)

**Research Areas:** Hedgehog signaling and birth defects, muscle stem cells and regeneration

**Faculty Interactions:** Nicole C. Dubois, Michael Rendl, James J. Bieker, Marek Mlodzik

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**Luca Lambertini, PhD, MPH** (Assistant Professor, Environmental Medicine & Public Health & Obstetrics, Gynecology and Reproductive Science)

**Research Areas:** Placental biomarkers of altered fetal and child development

**Faculty Interactions:** Dalila Pinto, Eyal Shemesh, Jia Chen, Andrew J. Sharp, Susan Teitelbaum, Philip J. Landrigan, Donald K. Scott, Adolfo Garcia-Ocaña, Ke Hao

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**Philip J. Landrigan, MD, MSc, DIH** (Professor, Environmental Medicine & Public Health & Pediatrics)

**Research Areas:** Global environmental health, children's environmental health, occupational medicine

**Faculty Interactions:** Robert O. Wright, Susan Teitelbaum, Manish Arora, Luca Lambertini

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**Marek Mlodzik, PhD** (Professor and Chair, Cell, Developmental & Regenerative Biology; Professor, Ophthalmology and Oncological Sciences)

**Research Areas:** Genetics and cell biology of planar cell polarity establishment, cell biology of Wnt-signaling and Notch-signaling

**Faculty Interactions:** Ross L. Cagan, Robert S. Krauss

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**Hirofumi Morishita, MD, PhD** (Associate Professor, Psychiatry, Ophthalmology & Neuroscience)

**Research Areas:** Mechanisms of perceptual and cognitive development, drug repurposing for neurodevelopmental disorders

**Faculty Interactions:** Manish Arora, Joseph D. Buxbaum, Dani Dumitriu

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**Coro Paisán-Ruiz, PhD** (Assistant Professor, Neurology, Psychiatry, & Genetics and Genomic Sciences)

**Research Areas:** Genetics of movement disorders, zebrafish models of neurodegeneration

**Faculty Interactions:** Joseph D. Buxbaum

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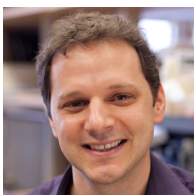


**Dalila Pinto, PhD** (Assistant Professor, Psychiatry & Genetics and Genomic Sciences)

**Research Areas:** Genetics and genomics of neurodevelopmental disorders (such as autism, epilepsy, schizophrenia), structural variation, transcriptomics, gene regulation, non-coding RNA

**Faculty Interactions:** Alex Kolevzon, Martin J. Walsh, Joseph D. Buxbaum, Eyal Shemesh, Luca Lambertini

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**Andrew J. Sharp, PhD** (Associate Professor, Genetics and Genomic Sciences)

**Research Areas:** Epigenomics, transcriptomics, genome function, structural variation, imprinting, congenital disorders

**Faculty Interactions:** Bruce D. Gelb, Nicole C. Dubois, Supinda Bunyavanich, Joseph D. Buxbaum, Jia Chen, Luca Lambertini

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## Neurodevelopmental Disorders *continued*



**Annemarie Stroustrup, MD, MPH** (Associate Professor, Pediatrics, Obstetrics, Gynecology and Reproductive Science, & Environmental Medicine & Public Health)

**Research Areas:** Neurodevelopment, perinatal environmental exposures

**Faculty Interactions:** Robert O. Wright, Rosalind J. Wright, Susan Teitelbaum, Chris Gennings, Manish Arora, Megan K. Horton, Shanna H. Swan, Allan C. Just, Lisa M. Satlin, Lisa Eiland, Bruce D. Gelb, Alan Groves



**Shanna H. Swan, PhD** (Professor, Environmental Medicine & Public Health)

**Research Areas:** Prenatal exposures, sexually dimorphic development, phthalates, stress, anogenital distance, neurodevelopment, analgesics

**Faculty Interactions:** Joseph D. Buxbaum, Robert O. Wright, Manish Arora, Dorothy E. Grice, Alex Kolevzon, Avi Reichenberg, Chris Gennings, Megan K. Horton, Allan C. Just, Annemarie Stroustrup, Susan Teitelbaum



**Robert O. Wright, MD, MPH** (Ethel H. Wise Professor and Department Chair, Environmental Medicine & Public Health; Director, Lautenberg Laboratory for Environmental Health; Director Institute for Exposomics)

**Research Areas:** Environmental causes of neurodevelopmental disorders and child obesity

**Faculty Interactions:** Rosalind J. Wright, Manish Arora, Chris Gennings, Allan C. Just, Megan K. Horton, Avi Reichenberg, Annemarie Stroustrup, Lisa M. Satlin, Jia Chen, Dani Dumitriu, Susan Teitelbaum, Shanna H. Swan, Shelley H. Liu

## Obesity and Diabetes



**Brian D. Brown, PhD** (Associate Professor, Genetics and Genomic Sciences)

**Research Areas:** Immunology and immunotherapy, autoimmune disease, microRNA regulation, and biotechnology

**Faculty Interactions:** TBA



**Ross L. Cagan, PhD** (Director, Center for Personalized Cancer Therapeutics; Professor, Cell, Developmental & Regenerative Biology, Oncological Sciences & Ophthalmology)

**Research Areas:** Drosophila as a tool to develop therapeutics for cancer, diabetes, and rare mendelian diseases

**Faculty Interactions:** Bruce D. Gelb, Joseph D. Buxbaum, Marek Mlodzik



**Adolfo García-Ocaña, PhD** (Professor, Medicine)

**Research Areas:** Diabetes, pancreatic beta cell biology

**Faculty Interactions:** Rupangi C. Vasavada, Donald K. Scott, Martin J. Walsh, Luca Lambertini



**Allan C. Just, PhD** (Assistant Professor, Environmental Medicine & Public Health)

**Research Areas:** Epigenomics, environmental exposures, endocrine disruptors, air pollution, obesity, birth outcomes

**Faculty Interactions:** Robert O. Wright, Rosalind J. Wright, Manish Arora, Chris Gennings, Annemarie Stroustrup, Shanna H. Swan, Megan K. Horton, Susan Teitelbaum, Avi Reichenberg



**Ruth J.F. Loos, PhD** (Professor, Environmental Medicine & Public Health)

**Research Areas:** Genetics of obesity and related cardiometabolic traits, genetic epidemiology, epidemiology

**Faculty Interactions:** Susan Teitelbaum, Martin J. Walsh



## Obesity and Diabetes continued



**Donald K. Scott, PhD** (Professor, Medicine)

**Research Areas:** Obesity and diabetes

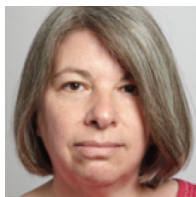
**Faculty Interactions:** Adolfo García-Ocaña, Rupangi C. Vasavada, Martin J. Walsh, Jaime Chu, Luca Lambertini



**Rupangi C. Vasavada, PhD** (Associate Professor, Medicine)

**Research Areas:** Diabetes, pancreatic beta cell biology, growth factors, cell signaling

**Faculty Interactions:** Adolfo García-Ocaña, Donald K. Scott, Jaime Chu



**Susan Teitelbaum, PhD** (Professor, Environmental Medicine & Public Health)

**Research Areas:** Environmental chemical exposure assessment, pubertal development, physical growth and development

**Faculty Interactions:** Jia Chen, Manish Arora, Philip J. Landrigan, Luca Lambertini, Annemarie Stroustrup, Shanna H. Swan, Robert O. Wright, Rosalind J. Wright, Allan C. Just, Ruth J.F. Loos, Chris Gennings



**Martin J. Walsh, PhD** (Professor, Pharmacological Sciences, Genetics and Genomic Sciences & Pediatrics)

**Research Areas:** Chromatin biology, RNA biology and Gene transcription in cancer, early development and metabolism

**Faculty Interactions:** Nicole C. Dubois, Dalila Pinto, Donald K. Scott, Ruth J.F. Loos, Chen-Leng Cai, Adolfo García-Ocaña, Xiu-Min Li, Jianlong Wang

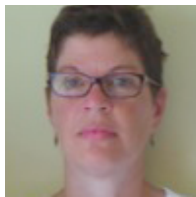
## Psychiatric Disorders



**Vilma Gabbay, MD** (Associate Professor, Psychiatry & Neuroscience)

**Research Areas:** Pediatric mood disorders, neuroimaging

**Faculty Interactions:** Eyal Shemesh, Dani Dumitriu



**Dorothy E. Grice, MD** (Professor, Psychiatry)

**Research Areas:** Genetic and epidemiological studies of OCD, Tourette disorder, autism and related childhood-onset neuropsychiatric disorders, prenatal exposures, including smoking, functional analysis of identified risk genes

**Faculty Interactions:** Avi Reichenberg, Joseph D. Buxbaum, Shanna H. Swan



**Avi Reichenberg, PhD** (Professor, Psychiatry & Environmental Medicine & Public Health)

**Research Areas:** Autism, schizophrenia, other psychiatric disorders

**Faculty Interactions:** Joseph D. Buxbaum, Alex Kolevzon, Dorothy E. Grice, Robert O. Wright, Manish Arora, Chris Gennings, Shanna H. Swan, Allan C. Just



**Eyal Shemesh, MD** (Professor, Pediatrics; Senior Faculty, Psychiatry)

**Research Areas:** Measurement and biological correlates of self-care behaviors

**Faculty Interactions:** Joseph D. Buxbaum, Alex Kolevzon, Vilma Gabbay, Scott H. Sicherer, Jeffrey M. Saland, Luca Lambertini, Dalila Pinto

## Other Research Focuses

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**James J. Bieker, PhD** (Professor, Cell, Developmental & Regenerative Biology)  
**Research Areas:** Transcriptional regulation of gene expression in erythroid cells, derivation of marked stem cells  
**Faculty Interactions:** Robert S. Krauss, Nicole C. Dubois

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**Dusan Bogunovic, PhD** (Assistant Professor, Microbiology & Pediatrics)  
**Research Areas:** Genetics of infectious and inflammatory diseases, Type I Interferons, Pseudo-TORCH syndrome, Neurolisteriosis  
**Faculty Interactions:** Bruce D. Gelb, Charlotte Cunningham-Rundles

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**Minji Byun, PhD** (Assistant Professor, Medicine)  
**Research Areas:** Genetics of immune disorders, primary immunodeficiency, immune dysregulation  
**Faculty Interactions:** Charlotte Cunningham-Rundles

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**Jaime Chu, MD** (Assistant Professor, Pediatrics)  
**Research Areas:** Disorders of glycosylation, cancer metabolism, liver fibrosis  
**Faculty Interactions:** Donald K. Scott, Rupangi C. Vasavada

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**Charlotte Cunningham-Rundles, MD, PhD** (David S. Gottesman Professor, Medicine; Professor, Pediatrics)  
**Research Areas:** Primary Immune Deficiency, B cells, antibody, B cell memory, hypogammaglobulinemia, immune reconstitution  
**Faculty Interactions:** Bruce D. Gelb, Dusan Bogunovic, Minji Byun, Birte Wistinghausen

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**David Dunkin, MD** (Assistant Professor, Pediatrics)  
**Research Areas:** Tolerance induction and therapeutics in Inflammatory Bowel Disease  
**Faculty Interactions:** Hugh A. Sampson, M. Cecilia Berin, Xiu-Min Li

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**Chris Gennings, PhD** (Professor, Environmental Medicine & Public Health & Population Health Science and Policy)  
**Research Areas:** Biostatistical methods development for environmental health  
**Faculty Interactions:** Robert O. Wright, Rosalind J. Wright, Megan K. Horton, Shanna H. Swan, Allan C. Just, Manish Arora, Annemarie Stroustrup, Avi Reichenberg, Jia Chen, Susan Teitelbaum, Shelley H. Liu

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**Shelley H. Liu, PhD** (Assistant Professor, Population Health Science and Policy)  
**Research Areas:** Biostatistics, environmental mixtures, public health  
**Faculty interactions:** Robert O. Wright, Manish Arora, Chris Gennings, Karen M. Wilson, Julie Wang

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**Michael Rendl, MD** (Associate Professor, Cell, Developmental & Regenerative Biology & Dermatology)  
**Research Areas:** Stem cells, hair regeneration, morphogenesis  
**Faculty Interactions:** Robert S. Krauss, Nicole C. Dubois

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## Other Research Focuses *continued*

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**Jeffrey M. Saland, MD** (Associate Professor, Pediatrics)

**Research Areas:** Kidney disease in children, lipoprotein metabolism in children with CKD, hemolytic uremic syndrome.

**Faculty Interactions:** Manish Arora, Eyal Shemesh, Alex Kolevzon, Lisa M. Satlin

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**Lisa M. Satlin, MD** (Herbert H. Lehman Professor and System Chair, Pediatrics)

**Research Areas:** Ontogeny and mechanoregulation of epithelial ion channels in secretory epithelia

**Faculty Interactions:** Annemarie Stroustrup, Robert O. Wright, Manish Arora, Jeffrey M. Saland, Dani Dumitriu

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**Jianlong Wang, PhD** (Associate Professor, Cell, Developmental & Regenerative Biology)

**Research Areas:** Stem cell pluripotency, somatic cell reprogramming, epigenetic control, transcriptional regulation

**Faculty Interactions:** Martin J. Walsh, Chen-Leng Cai, Nicole C. Dubois

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**Birte Wistinghausen, MD** (Associate Professor, Pediatrics)

**Research Areas:** Pediatric and adolescent Non-Hodgkin-Lymphoma, immunodeficiency associated lymphomas, post-transplant lymphoproliferative syndrome

**Faculty Interactions:** Charlotte Cunningham-Rundles

# Awards and Publications

## *Awards/Honors/Patents*

**Supinda Bunyavanich, MD, MPH**, American Academy of Allergy, Asthma, and Immunology 2017 Annual Meeting, American Academy of Pediatrics Section on Allergy and Immunology Outstanding Pediatric Abstract Award

**Jaime Chu, MD**, Molly Markoff Foundation, Art in Giving/Rachel Molly Markoff Foundation Research Award

**Charlotte Cunningham-Rundles, MD, PhD**, Clinical Immunology Society, President's Award 2017

**Bruce D. Gelb, MD**, Mount Sinai, Jacobi Medallion Award

**Robert S. Krauss, PhD**, Keynote Speaker, 38th Annual 2017 David W. Smith Workshop on Malformations and Morphogenesis, Stowe, VT

**Philip J. Landrigan, MD, MSc, DIH**, National Council for Science and the Environment, Lifetime Achievement Award, 2017

**Shelley H. Liu, PhD**, Eastern North America Region of the International Biometric Society, Distinguished Student Paper Award

**Shelley H. Liu, PhD**, American Statistical Association, Biometrics Section Student Paper Award

**Shelley H. Liu, PhD**, American Statistical Association, Section in Epidemiology Travel Award

**Shelley H. Liu, PhD**, International Society of Environmental Epidemiology, New Researcher Best Abstract Award 2017

**Ruth J.F. Loos, PhD**, Clarivate Analytics, Highly Cited Researcher 2017

**Donald K. Scott, PhD**, American Diabetes Association, Basic Science Innovation Award, Targeting the ChREBP-Nrf2 axis to expand functional beta cell mass

**Scott H. Sicherer, MD**, Clarivate Analytics, Highly Cited Researcher 2017

**Annamarie Stroustrup, MD, MPH**, Perinatal Research Society, Full Member

**Rupangi Vasavada, PhD**, U.S. Utility Patent, "Use of Osteoprotegerin (OPG) to increase human pancreatic beta cell survival and proliferation"

## *Publications*

Curtin P, Curtin A, Austin C, **Gennings C**, Tammimies K, Bölte S, **Arora M**. **Recurrence quantification analysis to characterize cyclical components of environmental elemental exposures during fetal and postnatal development.** *PLoS One*. 2017 Nov 7;12(11):e0187049.

**Arora M, Reichenberg A**, Willfors C, Austin C, Gennings C, Berggren S, ... Bolte S. **Fetal and postnatal metal dysregulation in autism.** *Nat Commun*. 2017 Jun 01;8:15493.

Goswami R, Blazquez AB, Kosoy R, Rahman A, **Nowak-Wegrzyn A, Berin MC**. **Systemic innate immune activation in food protein-induced enterocolitis syndrome.** *J Allergy Clin Immunol*. 2017 Jun;139(6):1885-96.e9.

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Sims R, van der Lee SJ, Naj AC, Bellenguez C, Badarinarayan N, Jakobsdottir J, ... **Buxbaum JD**, ... Schellenberg GD. **Rare coding variants in plcg2, abi3, and trem2 implicate microglial-mediated innate immunity in alzheimer's disease.** *Nat Genet*. 2017 Sep;49(9):1373-84.

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# Grants

## Agency

	Funding from New Grants (\$)	Funding from Existing & New Grants (\$)
National Human Genome Research Institute/NIH/DHHS	\$3,352,780	\$4,000,768
National Institute Of Mental Health/NIH/DHHS	2,662,166	4,689,036
National Institute Of Diabetes And Digestive And Kidney Diseases/NIH/DHHS	1,454,134	5,074,844
Department Of The Army	1,235,655	1,235,655
National Institute Of Allergy And Infectious Diseases/NIH/DHHS	1,167,250	5,899,923
Johns Hopkins University	621,823	621,823
Columbia University	528,094	540,940
National Heart, Lung, And Blood Institute/NIH/DHHS	516,610	2,003,058
Sean Parker Foundation	500,000	500,000
Broad Institute	447,907	447,907
New York Community Trust	410,531	410,531
National Institute Of Environmental Health Sciences/NIH/DHHS	304,249	5,907,746
National Institute Of Neurological Disorders And Stroke/NIH/DHHS	251,737	622,518
National Eye Institute/NIH/DHHS	248,690	963,173
Juvenile Diabetes Research Foundation	150,001	150,001
American Diabetes Association, Inc.	115,000	115,000
DBV Technologies	112,509	112,509
Benaroya Research Institute At Virginia	100,869	419,541
Henry Ford Health System	98,527	98,527
Aimmune Therapeutics, Inc.	69,399	69,399
Gilead Sciences	65,000	65,000
Vanderbilt University	60,522	60,522
PX Global Advisors, LLC	51,300	51,300
Children's Cardiomyopathy Foundation	50,000	50,000
Promentis Pharmaceuticals, Inc.	49,951	49,951
Burroughs Wellcome Fund	30,000	30,000
Phadia	10,419	10,419
National Institute Of Health Office Of The Director/NIH/DHHS		10,751,590
Seaver Foundation		1,250,001
National Institute Of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS		1,140,728
National Institute Of General Medical Sciences/NIH/DHHS		1,095,324
Albert Einstein College Of Medicine		753,891
National Institute Of Child Health And Human Development/NIH/DHHS		662,650
Emory University		508,729
New York State Office Of Science, Technology, And Academic Research		500,000
National Institute Of Dental And Craniofacial Research/NIH/DHHS		416,275
Drexel University		299,065
Boston Children's Hospital		251,858
Harvard University		179,830
National Institute Of Biomedical Imaging And Bioengineering/NIH/DHHS		162,433
American Academy Of Pediatrics		155,192
University Of Pittsburgh		135,600
Food Allergy Research & Education		120,000
Hirschl/Weill-Caulier Trust		105,000
March Of Dimes		102,694
Blacksmith Institute		100,003
New York University, Washington Square		98,812
Washington University		86,337
University Of Washington		81,963
Human Frontier Science Program Organization		77,500
Immune Deficiency Foundation		77,085
European Commission		75,625
American Heart Association - Founders Affiliate		66,000
Cincinnati Children's Hospital		51,600
University Of Chicago		43,960
St. Jude Children's Research Hospital		33,068
Seattle Children's Research Institute		20,079
Weill (Joan And Sanford I.) Medical College Of Cornell University		19,952
Children's Oncology Group		9,334
<b>Total</b>	<b>\$14,665,123</b>	<b>\$53,632,269</b>

# Material Transfer Agreements

## Research Focus

	Outgoing Material Transfer Agreements (#)	Technology Licenses (#)
Neurodevelopmental disorders	4	5
Cardiovascular disease	5	4
Diabetes and Obesity	6	5
Allergy and Asthma	0	6
Others	1	1
<b>Total</b>	<b>16</b>	<b>21</b>

# Licenses

## Licenses

	Total Number
Antigens/Antibodies	10
Reagents/Methods/Cell Lines	3
Genes/Adapters/Vectors/Oligonucleotides	6
Gene Testing/Therapeutics	2
<b>Total</b>	<b>21</b>



# Pilot Projects Funded for 2017-2018

## *Trainee Pilot Awards*

The inaugural trainee pilot program was established during the summer of 2017. Two new trainee pilot projects were awarded in the amount of \$10K in the predoctoral and postdoctoral divisions. The purpose of the program is to support postdoctoral fellows or PhD/MD-PhD students in pursuing an independently funded research project that is separate from their ongoing projects under their current Principal Investigator/mentor. Successful applications were required to (a) demonstrate that they can achieve independence and will generate preliminary data that could lead to career development or other grants and (b) be relevant to children's health.

### ▲ Postdoctoral Division

**Investigator:** Michael S. Breen, PhD, Postdoctoral fellow, Seaver Autism Center for Research and Treatment, Department of Psychiatry and Genetics and Genomic Sciences

**Project Title:** Cell-type Specific Transcriptome Profiling in Umbilical Cord Blood

**Primary Mentor:** Joseph D. Buxbaum, PhD, Department of Psychiatry and Genetics and Genomic Sciences

**Secondary Mentors:** Dan J. Stein, PhD (University of Cape Town) and Meaghan Jones, PhD (University of British Columbia)

### ▲ Predoctoral Division

**Trainee Investigator:** Oscar Rodriguez, PhD Candidate, Department of Genetics and Genomic Sciences

**Project Title:** Using Long-Reads to Identify Repeat Expansions in Undiagnosed Dominantly-Inherited Ataxias

**Primary Mentor:** Andrew J. Sharp, PhD, Department of Genetics and Genomic Sciences

**Secondary Mentor:** Ali Bashir, PhD, Department of Genetics and Genomic Sciences



*Michael S. Breen, PhD  
Postdoctoral Fellow,  
Department of Psychiatry and  
Genetics and Genomic Sciences*



*Oscar Rodriguez,  
PhD Candidate, Department of  
Genetics and Genomic Sciences*

# Pilot Projects Funded for 2017-2018 continued

## *Faculty Pilot Awards*

Three pilot projects were selected for \$70K in institutional funding for a one year period starting January 1, 2017. The purpose of the pilot program is to provide MCHDI faculty with funding for initial stages of research projects, with the goal of generating sufficient data to apply for larger, external grants. Projects are encouraged that are likely to (a) improve children's health, (b) promote collaboration within the MCHDI, and (c) leverage additional extramural funding for the Principal Investigators (PIs).

**Principal Investigators:** Nicole C. Dubois, PhD; Andrew J. Sharp, PhD

**Project Title:** Generating Cardiac Purkinje Cells from Human Pluripotent Stem Cells

**Principal Investigators:** Allan C. Just, PhD; Shanna H. Swan, PhD; Avi Reichenberg, PhD

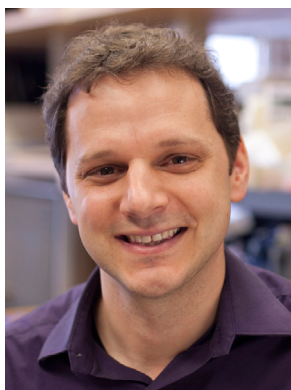
**Project Title:** Novel Biomarkers of Intrauterine Brain Development

**Principal Investigators:** Minji Byun, PhD; Paul J. Maglione, MD, PhD

**Project Title:** Investigation of Germline Mutations in PIK3CG in Primary Immunodeficiency



*Nicole C. Dubois, PhD  
Assistant Professor,  
Cell, Developmental &  
Regenerative Biology*



*Andrew J. Sharp, PhD  
Associate Professor,  
Genetics and Genomic  
Sciences*



*Allan C. Just, PhD  
Assistant Professor,  
Environmental Medicine &  
Public Health*



*Shanna H. Swan, PhD  
Professor, Environmental  
Medicine & Public Health*



*Avi Reichenberg, PhD  
Professor, Psychiatry  
Professor, Environmental  
Medicine & Public Health*



*Minji Byun, PhD  
Assistant Professor,  
Medicine*



*Paul J. Maglione, MD, PhD  
Assistant Professor,  
Medicine*



# Annual Retreat

The 5th Annual MCHDI Retreat was held at the Harmonie Club on November 28, 2017. Over 100 faculty members, trainees, and guests were in attendance. The retreat planning committee members: Martin J. Walsh, M. Cecilia Berin, and Jia Chen introduced two new segments, including an interactive workshop on communicating science and group activity during lunch. James Rea was invited from Stony Brook University's Alan Alda Center to lead the workshop exercise on effectively describing diverse areas of research to a broad audience. There were short talks from trainee pilot grant recipients (Michael S. Breen, PhD; PI: Joseph D. Buxbaum and Oscar Rodriguez; PI: Andrew J. Sharp) and faculty pilot awardees (Nicole C. Dubois, PhD; Allan C. Just, PhD; and Minji Byun, PhD) throughout the day. During lunch, group leaders facilitated discussion to get feedback on suggested topics/speakers for next year as well as brainstorm new ideas for the MCHDI to develop. The afternoon session resumed with short talks by the Young Investigators Competition finalists. Ryan Walker, PhD (PI: Ruth J.F. Loos) and Rosemary Li (PI: Rupangi C. Vasavada) were the final winners of their respective postdoctoral and predoctoral divisions. The parent's perspective segment was moderated by Bruce D. Gelb and focused on the story of 4-year-old Markella Giammalva, who was diagnosed with Dravet Syndrome. Her mother, Dara, and grandfather, Charles Gitlitz, described the trials and tribulations to discovering the cause of Markella's seizures and how their family is coping with her condition. Our faculty and trainees continue to be touched and inspired by first-hand accounts of families caring for patients with childhood diseases and work actively to improve treatment options through their research.



# Communications

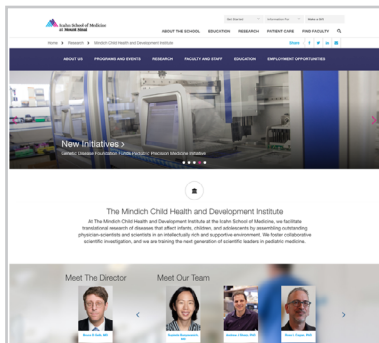
MCHDI delivers the latest updates on research advancements, events and news, both internally and externally via various communication channels. Below is information about the MCHDI website, newsletter and social media platforms:

**Website** ▲ Our website includes detailed information about our signature programs, shared resources, trainee education, and employment opportunities. You can also find our complete list of faculty and links to their research websites as well as the latest press releases featuring our faculty. Our annual reports and MCHDI newsletters are also accessible via our website. Please visit our website at [www.mountsinai.org/mindich](http://www.mountsinai.org/mindich)

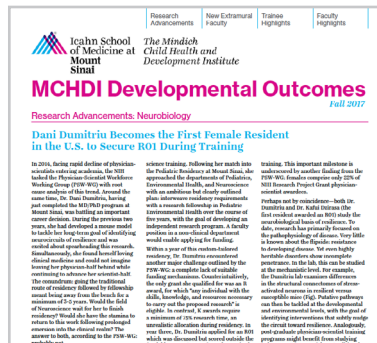
**Newsletter** ▲ The MCHDI Developmental Outcomes is a bi-annual newsletter distributed internally to faculty, trainees, and other institute affiliates to highlight important research breakthroughs, publications, awards, and events within MCHDI. View our latest newsletters featured on [icahn.mssm.edu/research/mindich/about/newsletters](http://icahn.mssm.edu/research/mindich/about/newsletters)

**Facebook** ▲ Our official MCHDI Facebook page was launched in 2014 and currently has 500+ likes and followers. Our team posts almost daily to share updates on faculty research, seminars, and events, and other information relevant to children's health. Please like and follow our page at [www.facebook.com/mindichchdi](http://www.facebook.com/mindichchdi)

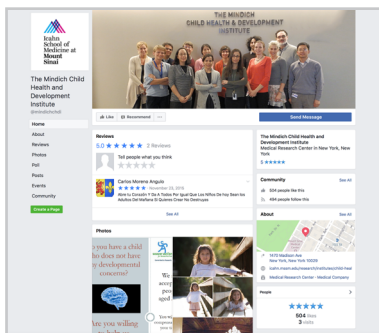
**Twitter** ▲ Our tweets are streaming on our website in real time. Follow or tweet to us @MindichCHDI or on our website at [www.mountsinai.org/mchdi](http://www.mountsinai.org/mchdi)



MCHDI website



MCHDI newsletter



MCHDI Facebook



MCHDI Twitter



# Shared Resources

## *Grant Forward*

Grant Forward is a pre-award funding database with a comprehensive list of federal, foundation, and other funding sources. It offers a user-friendly search interface, automated e-mail alerts, and tailored grant recommendations. Grant Forward subscriptions for MCHDI faculty and trainees are covered by our Institute. To sign up please visit: [www.grantforward.com](http://www.grantforward.com)



## *BioMe Biobank*

The BioMe Biobank contains the largest collection of DNA and plasma samples at Mount Sinai, enabling high-throughput disease genotyping and phenotyping while maintaining patient confidentiality through the Epic electronic medical record (EMR). The goal is to integrate patient clinical care information and research data. Observational epidemiologic studies of children have expanded in the past decade in response to the rising prevalence of childhood diseases including obesity, autism, and asthma and of environmental risk factors such as lead and pesticides, and the ability to genotype DNA has enabled further inquiry into the genetic basis of childhood diseases. MCHDI, in collaboration with the Charles R. Bronfman Institute for Personalized Medicine, is funding the collection of DNA samples from pediatric patients with allergies, and since February 2012 the Jaffe Food Allergy Institute has recruited >1000 enrollees. The pediatric cohort is comprised of samples from diverse racial and ethnic groups.

For more information please visit: [www.ica hn.mssm.edu/research/institutes/institute-for-personalized-medicine/innovation-and-technology/biome-platform](http://www.ica hn.mssm.edu/research/institutes/institute-for-personalized-medicine/innovation-and-technology/biome-platform)

## *Biorepository CORE Shared Resource Facility*

The biorepository CORE facility provides basic histology services such as processing and embedding section fixed and frozen tissues from animal or human sources. In addition, services include DNA/RNA/miRNA extractions, preparing and analyzing tissue microarrays, and supporting functions for tissue procurement, both from consented and anonymized collections. For a full list of their services visit their website at: [ica hn.mssm.edu/research/resources/shared-resource-facilities/histology](http://ica hn.mssm.edu/research/resources/shared-resource-facilities/histology)

# New Initiatives

## *Pediatric Precision Medicine Initiative*

During the summer of 2017, the MCHDI announced the launch of the Mount Sinai Pediatric Precision Medicine Initiative, made possible by the generous support of the **Genetic Disease Foundation** (GDF). At Mount Sinai, the strong commitment to developing scientific infrastructure for genomic technology and precision medicine has positioned our institution to lead the nation in applying state-of-the-art genomic research methodology to advancing patient care. This new initiative will bring together Mount Sinai care providers and investigators with diverse expertise throughout the **Departments of Pediatrics and Genetics and Genomic Sciences**, as well as the **Icahn Institute for Genomics and Multiscale Biology**, the **Charles Bronfman Institute for Personalized Medicine**, and the **Precision Immunology Institute**.



Precision medicine (PM) uses individualized patient data to accurately and rapidly diagnose disease, better predict the outcomes of medical issues, and treat illnesses more precisely and effectively. Currently, medical problems with strong genetic underpinnings such as birth defects, neurodevelopmental delays, and inborn errors of immunity are ones that typically manifest during infancy, childhood and/or adolescence, and where a PM approach can be transformative. Moreover, these types of conditions can lead to diagnostic odysseys, during which young patients are subjected to extensive medical testing for months or years, families wait anxiously for definitive answers, and effective therapies, when available, are delayed. Through the MCHDI's exciting new Pediatric Precision Medicine Initiative, we will improve outcomes by applying state-of-the-art genomic technologies as early as possible in the course of a child's disorder.

Recent advances in genomic medicine have enabled the PM approach that we will undertake. Using just a few drops of blood from the child, we are able to perform high-capacity DNA sequencing to examine the genes that provide instructions for all of the body's proteins. Especially when compared to similar sequencing of the patient's parents in order to identify the rare differences, our ability to pinpoint disease-causing DNA mutations is unparalleled in medical history. To date, our experiences have proven that this approach can solve medical mysteries, identifying known disease genes presenting in unexpected ways as well as allowing us to pinpoint novel ones.

Through the generous support of the **GDF**, we are offering this PM approach to infants, children, and teenagers with some of the most complex and difficult-to-diagnose medical issues. In addition to accelerating and improving their care, this program will advance medical education by preparing the young physicians in pediatric training at Mount Sinai to use genomic medicine effectively, allowing them to better serve the community in their future practices.



**Mount Sinai** *The Mindich  
Child Health and  
Development Institute*

# Leadership and Staff

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**Program Manager** Elena Lum, PhD

**Administrative Manager** Risa Slaughter

**Administrative Coordinator** Monica Nunez-Taveras

**Senior Executive Director of Development** Monica Sohn

**Director of Development** Caryn R. Karo

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Birte Wistinghausen, MD

Robert O. Wright, MD, MPH

Rosalind J. Wright, MD, MPH

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please visit our website at [www.mssm.edu/mchdi](http://www.mssm.edu/mchdi)