



**Mount
Sinai**

*The Mindich
Child Health and
Development Institute*

Annual Report 2018



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 2018, we have taken steps to implement major strategic and fundraising goals. The **Mindich Child Health and Development Center** (MCHDI) family has grown to 65 faculty members who specialize in allergy & asthma, cardiovascular disease, neurodevelopmental disorders, obesity & diabetes, and others. These members include three new faculty recruited extramurally (John Bucuvalas, MD, Isabelle Meyts, MD, PhD, and Rebecca Trachtman, MD) and seven internal Mount Sinai faculty (Silvia De Rubeis, PhD, Hala Harony-Nicolas, PhD, Laura Huckins, PhD, Alison P. Sanders, PhD, Paige M. Siper, PhD, Bryn D. Webb, MD, and Anusha Yeshokumar, MD). Our members have been more active than ever, contributing to over 380 publications this year.

In this past year, the MCHDI made significant progress in its initiative focused on pediatric precision medicine. The Undiagnosed Diseases Program, which was initiated with generous support from the **Genetics Disease Foundation**, has continued to enroll infants, children, and adolescents with a wide range of disorders that appear to be genetic but are unexplained. In this past year, Dr. Bryn Webb took the reins of the program and a new coordinator, Priyal Agrawal, was hired. The Program has continued to be successful in solving cases, of which some have proven to be actionable. The NHGRI-funded project for the Clinical Sequencing Evidence-Gathering Research Consortium called NYCKidSeq (Mount Sinai's Principal Investigators are Carol Horowitz, Eimear Kenny and me) has progressed and, at year's end, was poised to recruit its first subjects who will undergo genome sequencing and select gene resequencing. This project, in collaboration with Montefiore Hospital and the New York Genome Center, is focused on our diverse populations of New York City, who tend to be underserved in genomic medicine, to test the efficacy of a novel method for communicating genetic testing results. Finally, Dr. Annemarie Stroustrup is the site Principal Investigator on a project called GEMINI, a subaward from Tufts for an NCATS-funded project that will compare a new gene resequencing panel to rapid genome sequencing for newborns and young infants with apparent genetic disorders. Efforts are underway as part of Mount Sinai's Genomic Health Initiative, led by Dean Charney, to identify novel opportunities to further expand pediatric precision medicine.

Another outgrowth of the MCHDI strategic plan was a joint venture with the Precision Immunology Institute, led by Miriam Merad, MD, PhD, creating the Center for Inborn Errors of Immunity. Children affected with inborn errors of immunity or primary immunodeficiencies tend to suffer severe complications from the relevant infections, and there are often therapeutic options for reducing morbidities (see p. 30). Dr. Merad and I were successful in recruiting Dr. Isabelle Meyts, a leader in research on primary immunodeficiencies. Dr. Meyts, who is currently part-time at Mount Sinai and will become full-time in the fall, is planning for this Center's direction and initiatives. Joining her in the Center from its outset will be three other MCHDI members—Drs. Dusan Bogunovic, Minji Byun and Charlotte Cunningham-Rundles.



As we move forward into the new year, we are focusing on expanding our efforts in the parts of the MCHDI strategic plan that have been initiated but also initiating efforts in pediatric clinical trials and newborn medicine/prematurity. We hope to expand the scope of clinical trials being undertaken with children in the Mount Sinai Health System while enjoying economies of scale that a centralized unit is expected to provide. We also remain fully committed to helping all of MCHDI's faculty members and their trainees through our programming and infrastructure.

A handwritten signature in black ink, which appears to read "B. Gelb".

Bruce D. Gelb, Director

Faculty Growth

In 2018, we welcomed three new external faculty and seven internal faculty members to our institute. Currently, we total 65 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 (no longer at Mount Sinai), PhD; Bruce D. Gelb, MD; Amy R. Kontorovich, MD, PhD; Martin J. Walsh, PhD; Jia Chen, ScD; Rupangi C. Vasavada, PhD (no longer at Mount Sinai)

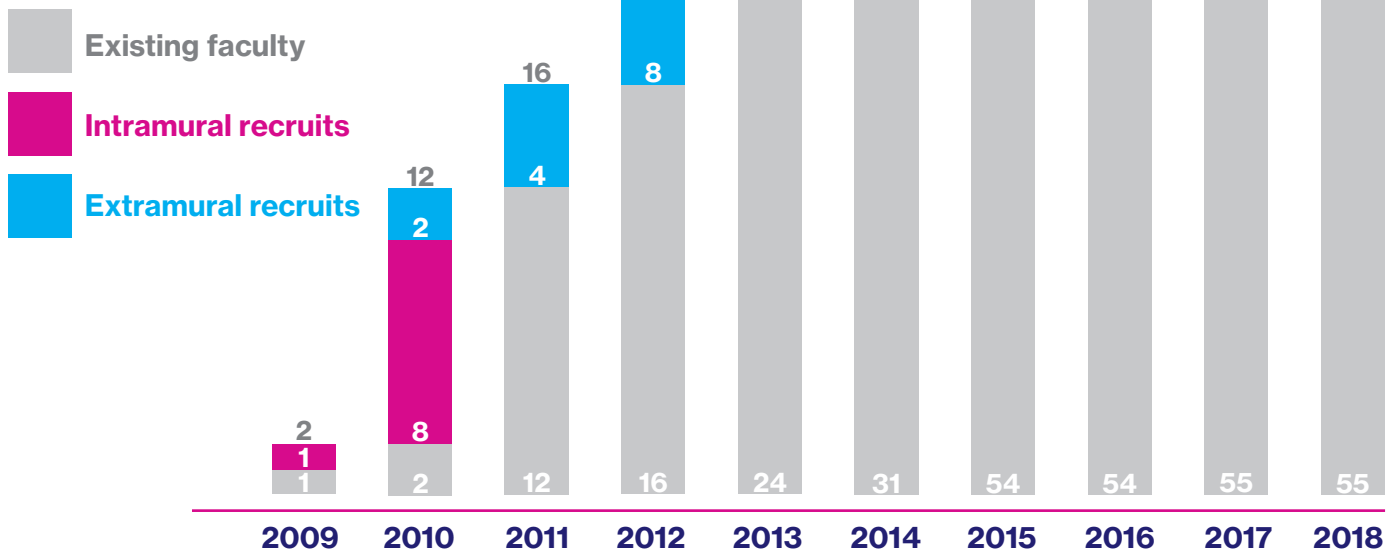


Chart of faculty recruits since our inception in 2009. In 2018, our institute recruited three new external and seven internal faculty members to our institute.

New Faculty

New Extramural Faculty



*John Bucuvalas, MD
Professor, Pediatrics
Chief, Division of Hepatology
Vice Chair, Faculty Affairs,
Pediatrics*

John Bucuvalas, MD

John Bucuvalas, MD, is the Chief of the Hepatology and Vice Chair of Faculty Affairs in the Jack and Lucy Clark Department of Pediatrics at the Icahn School of Medicine at Mount Sinai and the Kravis Children's Hospital at Mount Sinai. He also serves as the Director of Solid Organ Transplant Outreach for the Recanati-Miller Transplant Institute. He graduated from Harvard College, *magna cum laude*, in biology and then obtained his medical degree at Harvard Medical School. He completed his pediatric residency including a year as Chief Resident before his gastroenterology fellowship at Cincinnati Children's Hospital. He is board certified in pediatric gastroenterology with a certificate of added qualification in transplant hepatology. He advanced to Professor of Pediatrics at the University of Cincinnati and served as Director of the Integrated Solid Organ Transplant Program before coming to Mount Sinai. The overall goal of Dr. Bucuvalas's research is to give child and adolescent liver transplant candidates and recipients the promise of full and meaningful lives by delivering reliable state-of-the-art care integrated across disciplines, ensuring that we acquire and apply new knowledge and improve processes in the constantly evolving delivery system. His primary research efforts, funded by NIH, focus on clinical and translational studies to define predictors of operational tolerance, to predict risk and determine the mechanism of long-term structural liver allograft injury and to define strategies to mitigate nonadherence in transplant recipients.



*Isabelle Meyts, MD, PhD
Professor, Pediatrics
Director, Center for Inborn
Errors of Immunity*

Isabelle Meyts, MD, PhD

Isabelle Meyts obtained her medical degree at the Katholieke Universiteit Leuven, Belgium, in 1999. In 2007 she obtained her license in Pediatrics. She obtained her PhD degree in 2007 with papers studying the role of Th subsets in allergic airway inflammation. At the University Hospitals Leuven, Belgium, in the Department of Pediatrics, she built the Pediatric Primary Immunodeficiency Unit and is responsible for the diagnosis and treatment of children affected by primary immunodeficiency. She has built expertise in hematopoietic stem cell transplantation for primary immunodeficiency. Her efforts led to the recognition of the Department of Primary Immunodeficiencies as a Jeffrey Modell Foundation Diagnostics and Research Center in 2011. Her research centers on the genetic and functional unraveling of inborn errors of immunity. Recent focus has been on DADA2 and on the various innate immune system disorders. In 2018 she became President of the European Society for Immunodeficiencies.

New Extramural Faculty continued



Rebecca Trachtman, MD
Assistant Professor, Pediatrics

Rebecca Trachtman, MD

Rebecca Trachtman, MD, MS, is an Assistant Professor in the Department of Pediatrics at the Icahn School of Medicine at Mount Sinai. Dr. Trachtman is a pediatric rheumatologist, interested in outcomes and treatment in systemic autoimmune and autoinflammatory disease. She received her undergraduate degree from Barnard College and her medical degree from Albert Einstein College of Medicine. Dr. Trachtman then completed her pediatric residency training at NYU Langone Medical Center, followed by her pediatric rheumatology fellowship training at Hospital for Special Surgery/Weill Cornell Medicine, where she was the recipient of the Charles L. Christian Award for excellence in musculoskeletal research. During her fellowship training, Rebecca also attained a master's degree in Clinical and Translational Investigation through the Weill Cornell Medicine Clinical and Translational Science Center. Dr. Trachtman's areas of clinical focus include juvenile idiopathic arthritis, systemic lupus erythematosus, vasculitis and periodic fever syndromes. Her research has a clinical and translational focus, specifically evaluating biomarkers and patient-reported outcomes in juvenile idiopathic arthritis, in order to improve treatment. Dr. Trachtman's main area of focus right now is biomarkers for distinction of disease flare and infection in systemic juvenile idiopathic arthritis.

New Intramural Faculty



Silvia De Rubeis, PhD
Assistant Professor, Psychiatry

Silvia De Rubeis, PhD

Silvia De Rubeis, PhD, is an Assistant Professor at the Seaver Autism Center for Research and Treatment, Department of Psychiatry, and Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai. Dr. De Rubeis is a molecular neuroscientist and geneticist interested in understanding the genetic, molecular, and cellular mechanisms underlying autism spectrum disorder (ASD) and intellectual disability (ID). Dr. De Rubeis completed her PhD in Cellular and Molecular Biology at the University of Rome "Tor Vergata" in Italy. During her first postdoctoral training with Dr. Claudia Bagni at the Katholieke Universiteit Leuven and Vlaams Instituut voor Biotechnologie (VIB) in Leuven, Belgium, she studied how the regulation of mRNA translation shapes the synaptic development in the context of Fragile X syndrome, a leading monogenic cause of ASD and the most common inherited form of ID. She then joined ISMMS for a second postdoctoral training in Genetics and Genomics in the lab of Dr. Joseph Buxbaum. While there, she studied the role of rare genetic variation in ASD through large-scale exome sequencing analyses and discovered novel genes and loci conferring risk. Her lab studies developmental defects resulting from disruptive mutations in novel high-risk genes identified from genomic studies in ASD and ID. Her research takes a genetics-first approach for functional analyses in cellular and mouse models and strives to take into account clinically relevant aspects that emerge from patient-based research.



*Hala Harony-Nicolas, PhD
Assistant Professor, Psychiatry*

Hala Harony-Nicolas, PhD

Dr. Hala Harony-Nicolas is an Assistant Professor at the Department of Psychiatry and a member of the Seaver Autism Center, the Friedman Brain Institute and the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai (ISMMS). Dr. Harony-Nicolas is a molecular and behavioral neuroscientist. She received her PhD in Molecular Biology at the Technion Institute, Israel, and completed her first postdoctoral training in Molecular Neurobiology at University of Haifa, Israel, where she studied the epigenetic regulation of the mouse oxytocin receptor gene. She joined Dr. Joseph Buxbaum's laboratory and the Seaver Autism Center at the Icahn School of Medicine at Mount Sinai, where she completed her second postdoctoral training and was thereafter promoted to the level of Instructor. During her training, she completed the validation and characterization of a new transgenic rat model for autism; the Shank3-deficient rat, and demonstrated the ameliorative effect of the oxytocin peptide on synaptic plasticity, social memory, and attentional deficits in this model. Studies in Dr. Harony-Nicolas's laboratory are focused on understanding the mechanisms by which autism-associated mutations lead to the manifestation of behavioral deficits by asking how they affect (1) social brain circuits, (2) the oxytocin system, known to modulate social behavior, and (3) the interaction between both in health and disease. Her laboratory applies behavioral and cutting-edge molecular neuroscience approaches, with the ultimate goal of identifying molecular targets for treatment and uncovering altered brain circuits that can be manipulated with circuit-specific noninvasive interventions.



*Laura Huckins, PhD
Assistant Professor, Genetics
and Genomic Sciences.*

Laura Huckins, PhD

Laura Huckins, PhD, is an Assistant Professor of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai. Psychiatric disorders are complex, impairing, and highly stigmatized conditions, conferring significant morbidity and mortality, and imposing significant emotional, social, and financial burdens on sufferers and their families. Substantial progress in understanding the genetic basis of these disorders has been made through global collaborative projects, and a number of significantly associated variants have been identified. Despite this, little is known about the biological mechanisms underlying these disorders, and few effective treatments exist. In particular, no effective medications exist for AN, which is understudied and underfunded compared to schizophrenia or bipolar disorder. Similarly, PTSD is underfunded compared to other psychiatric disorders, and existing funding largely focuses on military and combat-based PTSD, rather than interpersonal violence. The Huckins lab will focus on understanding the multi-omic aetiopathology of psychiatric disorders and trajectories, with an emphasis on understudied disorders and vulnerable groups. They will leverage statistical and analytical expertise to develop and apply machine-learning algorithms, in order to provide novel insights into understudied questions. To this end, her lab will have three synergistic foci: (1) development of statistical algorithms; (2) generation of new cohorts using clinical recruitment and mining of electronic medical records (EMRs); and (3) identification of novel associations. Dr. Huckins intends that these goals will complement and strengthen existing expertise in the Pamela Sklar Division of Psychiatric Genomics.



*Alison P. Sanders, PhD
Assistant Professor, Pediatrics
and Environmental Medicine &
Public Health*

Alison P. Sanders, PhD

Dr. Sanders is an Assistant Professor of Environmental Medicine & Public Health and Pediatrics at the Icahn School of Medicine at Mount Sinai. She is an environmental health engineer and molecular epidemiologist with a unique skill set in geospatial statistics and environmental toxicology, having graduate and postgraduate training in engineering, toxicology, and perinatal epidemiology. Her research involves developing novel approaches to tackle complex research questions in kidney health and development, integrating research in chemical mixtures, the microbiome, epigenetics, and perinatal health. She directs the Laboratory for Environmental Nephrotoxicology and is developing novel methods to use kidney organoids in environmental health science. Dr. Sanders's work is funded by an NIEHS K99/R00 Pathway to Independence Award. She is the Director of Postdoctoral Professional Development within the Department of Environmental Medicine & Public Health, a new program she designed to facilitate early-career scientists building successful careers in and out of academia. Dr. Sanders received her undergraduate degree from the University of Virginia, her master's from the University of Wisconsin and her doctoral degree in Environmental Sciences & Engineering from the University of North Carolina.



*Paige M. Siper, PhD
Assistant Professor, Psychiatry*

Paige M. Siper, PhD

Paige M. Siper, PhD, is a licensed clinical psychologist, Chief Psychologist of the Seaver Autism Center for Research and Treatment, and an Assistant Professor in the Department of Psychiatry. She has expertise in the diagnosis, neuropsychological assessment, and treatment of individuals with a variety of neurodevelopmental disorders, including autism spectrum disorder (ASD) and intellectual disability. Dr. Siper's research focuses on biomarker discovery and sensory processing using electrophysiological and behavioral approaches. Dr. Siper is the co-developer of the Sensory Assessment for Neurodevelopmental Disorders (SAND), which is the first clinician-administered observation and corresponding caregiver interview to quantify sensory reactivity according to DSM-5 criteria for ASD. Dr. Siper is currently using a type of EEG known as a visual evoked potential, along with the SAND and comprehensive clinical characterization of patients, to identify biological and bio-behavioral markers of idiopathic and single-gene forms of ASD. Her long-term research goals involve the integration of neural and neuropsychological findings to identify subtypes, monitor disease trajectory, and objectively measure treatment response. Dr. Siper has a strong interest in ensuring inclusion of severely affected individuals in research studies. Dr. Siper graduated with honors and distinction from Cornell University with a Bachelor of Science. She earned her PhD in Clinical Psychology from Yeshiva University and completed her clinical psychology internship at the University of Miami Miller School of Medicine. Dr. Siper was the recipient of an Autism Speaks Meixner Translational Research Postdoctoral Fellowship, which she completed at the Seaver Autism Center at Mount Sinai. She joined the Mount Sinai faculty in 2014.



*Bryn D. Webb, MD
Assistant Professor, Genetics &
Genomic Sciences
Assistant Professor, Pediatrics*

Bryn D. Webb, MD

Bryn D. Webb, MD, is a physician-scientist and Assistant Professor in the Departments of Genetics & Genomic Sciences and Pediatrics, with expertise in pediatrics, clinical and molecular genetics, and genomics. She serves as Co-Director of the Mitochondrial Medicine Program in the Division of Medical Genetics at Mount Sinai and Co-Director of the multidisciplinary Cleft and Craniofacial Program at Mount Sinai. Her research has focused on gene discovery for rare congenital anomalies and patients with undiagnosed disease, and her prior research accomplishments have included the following: identification of pathogenic variants in HOXB1 in a subset of patients with congenital facial paralysis and strabismus (Webb et al, Am J Hum Genet, 2012); identification of a novel mitochondrial disorder caused by recessive single nucleotide variants in MARS2 (Webb et al, Hum Mutat, 2015); identification of an additional causative disease gene (DACT1) for Townes Brocks syndrome (Webb et al, Hum Mutat, 2017); and identification of a novel mitochondrial disorder caused by recessive variants in MRPS34 (Lake NJ*, Webb BD*, et al, Am J Hum Genet, 2017; *= co-first author). She currently has an NIH K08 Career Development Award to further study mitochondrial aminoacyl tRNA synthetase disorders, including MARS2 deficiency, by creating cellular models of these diseases with CRISPR/Cas9 technology and by employing RNA-seq and network analysis. She is also an investigator in the Pediatric Precision Medicine initiative, which aims to diagnose and improve care for pediatric patients with undiagnosed disease at Mount Sinai.



*Anusha Yeshokumar, MD
Assistant Professor, Neurology
and Pediatrics*

Anusha Yeshokumar, MD

Anusha Yeshokumar, MD, is an Assistant Professor in the Departments of Pediatrics and Neurology at the Icahn School of Medicine at Mount Sinai. As an autoimmune neurologist, her research focuses on understanding outcomes after autoimmune encephalitis and identifying clinical and biologic factors associated with these outcomes. This work involves numerous national and international collaborations. Clinically, Dr. Yeshokumar cares for children and adults with autoimmune encephalitis, multiple sclerosis, neuromyelitis optica, neurosarcoidosis, and other autoimmune diseases that affect the central nervous system. Dr. Yeshokumar joined the Icahn School of Medicine at Mount Sinai in 2017. She received her undergraduate degree from the Johns Hopkins University in Baltimore, where she majored in Neuroscience. She obtained her MD degree from Jefferson Medical College in Philadelphia. She completed her pediatrics residency at the Westchester Medical Center followed by pediatric neurology residency at Johns Hopkins Hospital, where she was awarded the Jay Slotkin Award for Excellence in Research. She then pursued a combined adult and pediatric fellowship in the Multiple Sclerosis and Neuroimmune Fellowship Program at the Children's Hospital of Philadelphia and Hospital of the University of Pennsylvania, funded by the National Multiple Sclerosis Society.

Faculty Research Areas

Asthma and Allergy



M. Cecilia Berin, PhD (Professor, Pediatrics)

Research Areas: Immune mechanisms of food allergy and regulation of immune tolerance



Supinda Bunyavanich, MD, MPH (Associate Professor, Pediatrics, & Genetics and Genomic Sciences)

Research Areas: Integrative genomics of asthma and allergic diseases



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



Anna Nowak-Wegrzyn, MD (Professor, Pediatrics)

Research Areas: Food allergy, FPIES, oral immunotherapy, anaphylaxis, milk and egg allergy



Hugh A. Sampson, MD (Kurt Hirschhorn Professor, Pediatrics)

Research Areas: Immunopathogenesis of food allergy and anaphylaxis



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



Julie Wang, MD (Professor, Pediatrics)

Research Areas: Novel therapeutics for food allergy, epidemiology and management of food allergy in minority, urban populations



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



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

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



Nicole C. Dubois, PhD (Assistant Professor, Cell, Developmental & Regenerative Biology)
Research Areas: Heart development, stem cell differentiation, disease modeling



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



Alan Groves, MBChB, MD (Associate Professor, Pediatrics)
Research Areas: Hemodynamics, cardiac function, echocardiography, magnetic resonance imaging



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



Amy R. Kontorovich, MD, PhD (Medical Director, Adult Cardiovascular Genetics; Assistant Professor, Medicine)
Research Areas: Myocarditis, genetics of cardiovascular diseases, stem cell research



Alison P. Sanders, PhD (Assistant Professor, Pediatrics, and Environmental Medicine & Public Health)
Research Areas: Environment, toxic metals, kidney development, hypertension, cardiovascular disease, biomarkers

Neurodevelopmental Disorders



Manish Arora, PhD, BDS, MPH (Professor, Environmental Medicine & Public Health, & Dentistry)
Research Areas: Environmental epidemiology and exposure biology



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



Jia Chen, ScD (Professor, Pediatrics, Environmental Medicine & Public Health, Medicine, & Oncological Sciences)
Research Areas: Environmental epigenetics, molecular epidemiology



Silvia De Rubeis, PhD (Assistant Professor, Psychiatry)
Research Areas: Intellectual disability, autism spectrum disorder, functional genetics, cell and animal model systems, brain development



Lisa Eiland, MD (Assistant Professor, Pediatrics)
Research Areas: Stress and neurodevelopment



Hala Harony-Nicolas, PhD (Assistant Professor, Psychiatry)
Research Areas: Brain circuits of social behavior, mechanisms of action of the oxytocin hypothalamic system, animal models for autism spectrum disorder



Megan K. Horton, PhD, MPH (Assistant Professor, Environmental Medicine & Public Health)
Research Areas: Children's environmental health, exposure assessment, pediatric neuroimaging



Laura Huckins, PhD (Assistant Professor, Genetics and Genomic Sciences)
Research Areas: Psychiatric Genetics, specializing in understudied disorders and disorders affecting vulnerable populations. Particular focus on Anorexia Nervosa, PTSD, Sexual Assault, OCD. Secondary focus on machine learning algorithms; transcriptomic imputation; multi-omic eQTL-based methodologies.

Neurodevelopmental Disorders *continued*



Alex Kolevzon, MD (Director, Child and Adolescent Psychiatry; Professor, Psychiatry, & Pediatrics)
Research Areas: Autism spectrum and other neurodevelopmental disorders



Robert S. Krauss, PhD (Professor, Cell, Developmental & Regenerative Biology, & Oncological Sciences)
Research Areas: Hedgehog signaling and birth defects, muscle stem cells and regeneration



Paige M. Siper, PhD (Assistant Professor, Psychiatry)
Research Areas: Autism, intellectual disability, biomarker discovery, sensory processing



Luca Lambertini, PhD (Assistant Professor, Environmental Medicine & Public Health, & Obstetrics, Gynecology and Reproductive Science)
Research Areas: Placental biomarkers of altered fetal and child development



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



Hirofumi Morishita, MD, PhD (Associate Professor, Psychiatry, Ophthalmology, & Neuroscience)
Research Areas: Mechanisms of perceptual and cognitive development, drug repurposing for neurodevelopmental disorders



Coro Paisán-Ruiz, PhD (Associate Professor, Neurology, Psychiatry, & Genetics and Genomic Sciences)
Research Areas: Genetics of neurological and neurodevelopmental diseases, disease modeling in zebrafish



Dalila Pinto, PhD (Assistant Professor, Psychiatry, & Genetics and Genomic Sciences)
Research Areas: Genetics and genomics of neurodevelopmental disorders (particular focus on autism, epilepsy, schizophrenia, OCD), structural variation, transcriptomics, gene regulation, non-coding RNA

Neurodevelopmental Disorders continued



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

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



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

Research Areas: Neurodevelopment, perinatal environmental exposures



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

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



Bryn D. Webb, MD (Assistant Professor, Genetics and Genomic Sciences)

Research Areas: Identifying genetic etiologies of congenital anomalies, mitochondrial disorders, undiagnosed disease



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



Anusha Yeshokumar, MD (Assistant Professor, Neurology and Pediatrics)

Research Areas: Autoimmune Encephalitis, outcomes research, inflammatory biomarkers, cognition, behavior

Obesity and Diabetes



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



Adolfo García-Ocaña, PhD (Professor, Medicine)

Research Areas: Diabetes, pancreatic beta cell biology

Obesity and Diabetes continued



Allan C. Just, PhD (Assistant Professor, Environmental Medicine & Public Health)
Research Areas: Epigenomics, environmental exposures, endocrine disruptors, air pollution, obesity, birth outcomes



Ruth J.F. Loos, PhD (Professor, Environmental Medicine & Public Health)
Research Areas: Genetics of obesity and related cardiometabolic traits, genetic epidemiology, epidemiology



Donald K. Scott, PhD (Professor, Medicine)
Research Areas: Obesity and diabetes



Susan Teitelbaum, PhD (Professor, Environmental Medicine & Public Health)
Research Areas: Environmental chemical exposure assessment, pubertal development, physical growth and development



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

Psychiatric Disorders



Vilma Gabbay, MD (Associate Professor, Psychiatry, & Neuroscience)
Research Areas: Pediatric mood disorders, neuroimaging



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



Avi Reichenberg, PhD (Professor, Psychiatry, & Environmental Medicine & Public Health)
Research Areas: Autism, schizophrenia, other psychiatric disorders

Psychiatric Disorders *continued*



Eyal Shemesh, MD (Professor, Pediatrics; Senior Faculty, Psychiatry)
Research Areas: Measurement and biological correlates of self-care behaviors

Other Research Focuses



James J. Bieker, PhD (Professor, Cell, Developmental & Regenerative Biology)
Research Areas: Transcriptional regulation of gene expression in erythroid cells, derivation of marked stem cells



Dusan Bogunovic, PhD (Assistant Professor, Microbiology, & Pediatrics)
Research Areas: Genetics of infectious and inflammatory diseases, Type I Interferons, Pseudo-TORCH syndrome, Neurolisteriosis



Brian D. Brown, PhD (Professor, Genetics and Genomic Sciences)
Research Areas: Immunology and immunotherapy, autoimmune disease, microRNA regulation, and biotechnology



John Bucuvalas, MD (Professor, Pediatrics)
Research Areas: Outcomes after liver transplantation, allograft injury in pediatric liver transplant recipients



Minji Byun, PhD (Assistant Professor, Medicine)
Research Areas: Genetics of immune disorders, primary immunodeficiency, immune dysregulation



Jaime Chu, MD (Assistant Professor, Pediatrics)
Research Areas: Disorders of glycosylation, cancer metabolism, liver fibrosis



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

Other Research Focuses continued



David Dunkin, MD (Assistant Professor, Pediatrics)

Research Areas: Tolerance induction and therapeutics in Inflammatory Bowel Disease, mechanisms of inflammatory diseases of the gastrointestinal tract



Chris Gennings, PhD (Professor, Environmental Medicine & Public Health, & Population Health Science and Policy)

Research Areas: Biostatistical methods development for environmental health



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

Research Areas: Biostatistics, environmental mixtures, public health



Isabelle Meyts, MD, PhD (Professor, Pediatrics)

Research Areas: Adenosine deaminase 2 deficiency, innate immunity, malignancy and primary immunodeficiency



Michael Rendl, MD (Associate Professor, Cell, Developmental & Regenerative Biology, & Dermatology)

Research Areas: Stem cells, hair regeneration, morphogenesis



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

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



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

Research Areas: Ontogeny and mechanoregulation of epithelial ion channels in secretory epithelia, 3D bioprinting of kidney tubules



Rebecca Trachtman, MD (Assistant Professor, Pediatrics)

Research Areas: Biomarkers, patient-reported outcomes in juvenile idiopathic arthritis



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

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

Faculty Research Interactions

Vignette Highlight: Dusan Bogunovic, PhD



Bruce D. Gelb, MD

Gogel Family Professor and Director, MCHDI Professor, Pediatrics & Genetics and Genomic Sciences

Collaborating on PPM and characterization of innate immune defects in children



Charlotte Cunningham-Rundles, MD, PhD

David S. Gottesman Professor, Medicine Professor, Pediatrics

Collaborating on childhood inflammatory disorders



Dusan Bogunovic, PhD

Assistant Professor, Microbiology & Pediatrics

Collaborating on genetics of immune mediated gastrointestinal defects

Collaborating on human genetics of inborn errors of childhood diseases via PPM

Collaborating on characterization of congenital defects leading to renal disease



David Dunkin, MD

Assistant Professor, Pediatrics



Jeffrey M. Saland, MD

Associate Professor, Pediatrics



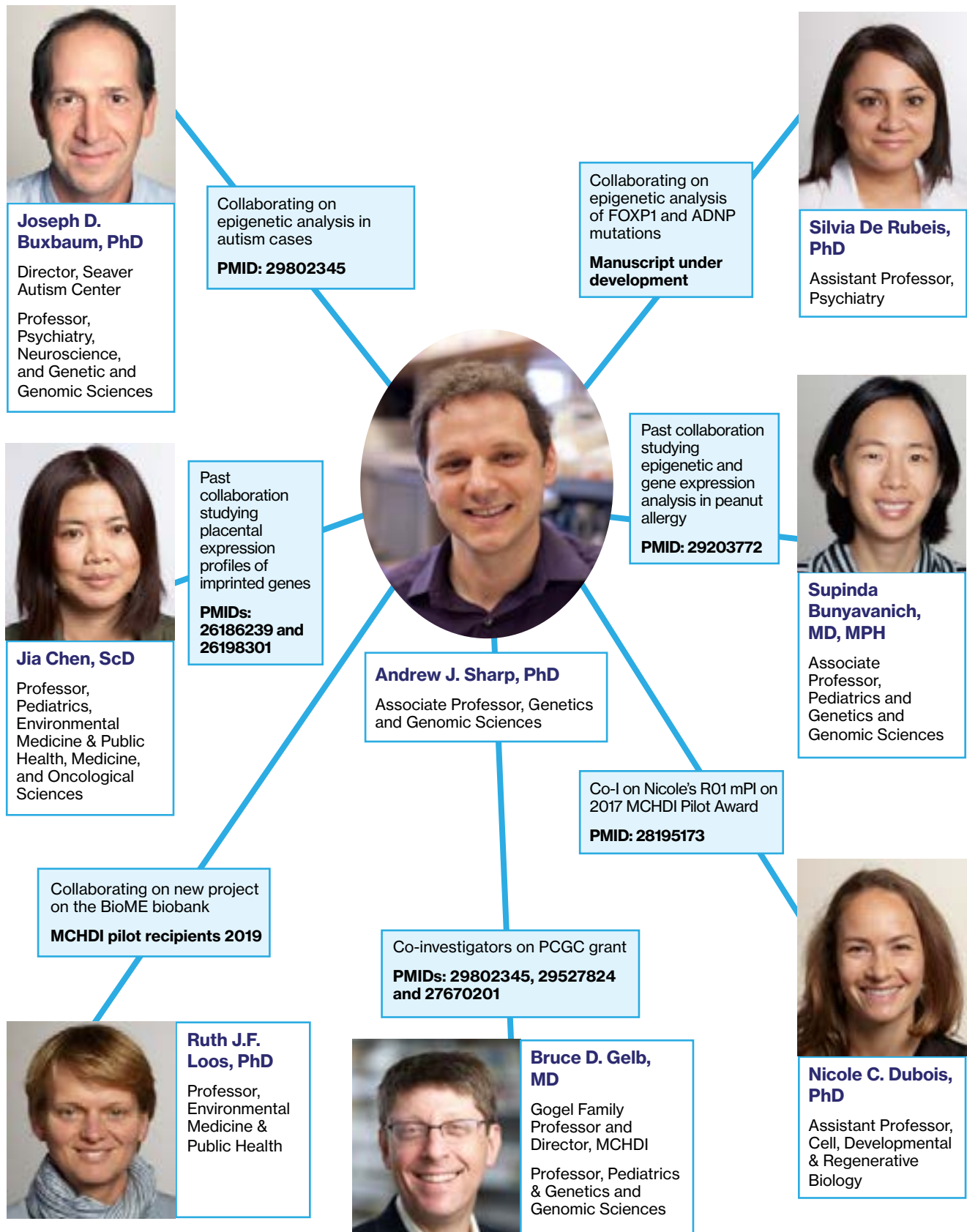
Bryn D. Webb, MD

Assistant Professor, Genetics & Genomic Sciences

Assistant Professor, Pediatrics

Faculty Research Interactions

Vignette Highlight: Andrew J. Sharp, PhD



Awards and Publications

Awards/Honors/Patents

James J. Bieker, PhD, Guest Speaker, Scientific Program on Red Cell Biology, American Society of Hematology, San Diego, CA 2018

Bruce D. Gelb, MD, Icahn School of Medicine at Mount Sinai, Jacobi Medallion Award 2018

Lisa M. Satlin, MD, University of Alabama at Birmingham, The 2nd Annual James A. Schafer Lectureship, "Cell-specific Function and Regulation of Mechanosensitive Ion Channels in the Distal Nephron"

Lisa M. Satlin, MD, Experimental Biology Annual Meeting in San Diego, 2018 Carl W. Gottschalk Award of the American Society of Physiology (Renal Section)

Lisa M. Satlin, MD, American Physiological Society (Renal Section), 2018 Carl W. Gottschalk Distinguished Lectureship, "In the Flow: Cell-specific expression and regulation of BK channels in the distal nephron"

Lisa M. Satlin, MD, University of Buffalo, Suk-Ki Hong Memorial Lecture, Departments of Medicine and Physiology and Biophysics, "Bedside to bench: a journey of discovery into the ontogeny and renal regulation of potassium homeostasis"

Eyal Shemesh, MD, Texas Children's Hospital / Baylor College of Medicine. Alpard Visiting Professorship and Alpard 7th Annual Lecture: "Failure to Follow Medical Recommendations – Making Sense of an Important but Hidden Cause of Morbidity"

Anusha Yeshokumar, MD, Icahn School of Medicine at Mount Sinai, Department of Neurology's Excellence in Teaching Award 2018

Trainee Honors/Awards

Giovanna Collu, PhD, PI: Marek Mlodzik, PhD & **Kathryn Bambino, PhD**, PI: Jaime Chu, MD, Genetics Society of America, Career Development Symposium Award

Maya Deysenroth, PhD, PI: Jia Chen, ScD, Teratology Society Meeting, Travel Award 2018

Maya Deysenroth, PhD, PI: Jia Chen, ScD, NIEHS Extramural Paper of the Month, "Intrauterine multi-metal exposure is associated with reduced fetal growth through modulation of the placental gene network"

Gleicy Hair, PhD, PI: Alison P. Sanders, PhD, FutureTox IV Progress to Maturity: Predictive Toxicology for Healthy Children, SOT, Travel Award 2018

Hsi-en John Ho, MD, PI: Supinda Bunyavanich, MD, MPH, American College of Allergy Asthma and Immunology, Travel Award 2018

Felix Richter, MD/PhD candidate, PI: Bruce D. Gelb, MD, American Society of Human Genetics, Epstein Trainee Award Semi-Finalist and Oral Presentation

Publications

Smith TM, Austin C, Green DR, Joannes-Boyau R, Bailey S, Dumitriu D, ... **Arora M. Wintertime stress, nursing, and lead exposure in Neanderthal children.** *Sci Adv.* 2018 Oct 31;4(10):eaau9483.

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Grants

Agency	Funding from New Grants (\$)	Funding from Existing & New Grants (\$)
National Institute of Diabetes and Digestive and Kidney Diseases/NIH/DHHS	2,561,657	8,939,894
National Institute of Allergy and Infectious Diseases/NIH/DHHS	1,524,998	6,496,285
National Institute of Neurological Disorders and Stroke/NIH/DHHS	1,289,818	1,499,182
National Institute of Child Health and Human Development/NIH/DHHS	1,207,682	1,809,601
National Institute of Environmental Health Sciences/NIH/DHHS	1,164,283	7,428,631
National Heart, Lung, and Blood Institute/NIH/DHHS	1,035,326	2,946,998
New York State Stem Cell Board	887,830	887,830
National Institute of General Medical Sciences/NIH/DHHS	823,755	1,417,305
Icahn School of Medicine at Mount Sinai	732,418	732,418
Highline Therapeutics, Inc.	430,985	430,985
National Cancer Institute/NIH/DHHS	421,587	421,587
Emory University	300,762	300,762
National Institute on Drug Abuse/NIH/DHHS	254,250	254,250
Rutgers	193,266	193,266
University of Southern California	150,000	150,000
Northwestern University	117,375	117,375
University of North Carolina	85,055	149,495
Alnylam Pharmaceuticals, Inc.	81,074	81,074
Moebius Syndrome Foundation	80,000	80,000
Rho Federal Systems	76,177	76,177
Art in Giving/The Rachel Molly Markoff Foundation	75,000	75,000
Minerva Neurosciences, Inc.	52,053	52,053
American Foundation for Suicide Prevention	49,998	49,998
Albireo AB	43,566	43,566
Castleman Disease Collaborative Network	42,508	42,508
University of California, San Francisco	38,902	38,902
Sanofi US Services Inc.	38,000	38,000
Brain and Behavior Research Foundation	35,000	35,000
Binational Science Foundation	30,000	30,000
Aimmune Therapeutics, Inc.	8,000	8,000
Children's Hospital of Philadelphia		4,787
Seattle Children's Research Institute		20,207
University of Pittsburgh		50,850
New England Research Institute		63,051
Gilead Sciences		65,000
Vanderbilt University		69,516
Hirschl/Weill-Caulier Trust		70,000
American Heart Association		72,630
European Commission		75,625
Immune Deficiency Foundation		76,785
Duke University		79,648
University of Washington		80,194
New York University, Washington Square		83,582
Henry Ford Health System		98,526
Queens College of CUNY		99,768
Johns Hopkins University		101,893
Broad Institute		114,016
American Diabetes Association, Inc.		114,997
Food Allergy Research & Education		120,000
Harvard University		132,404
University of Colorado		143,307
American Academy of Pediatrics		183,880
Drexel University		239,451
Cincinnati Children's Hospital		453,446
Boston Children's Hospital		655,887
National Institute of Dental and Craniofacial Research/NIH/DHHS		497,855
New York State Office of Science, Technology, and Academic Research		500,000
Columbia University		552,501
National Eye Institute/NIH/DHHS		1,008,706
National Institute of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS		816,157
Albert Einstein College of Medicine		924,593
Seaver Foundation		1,483,141
Benaroya Research Institute at Virginia		2,051,627
National Human Genome Research Institute/NIH/DHHS		4,777,104
National Institute of Mental Health/NIH/DHHS		5,123,518
National Institutes of Health Office of the Director/NIH/DHHS		10,881,143
Total	\$13,831,325	\$66,711,937

Material Transfer Agreements

Research Focus	Outgoing Material Transfer Agreements (#)	Technology Licenses (#)
Neurodevelopmental disorders	8	6
Cardiovascular disease	0	4
Diabetes and Obesity	8	5
Allergy and Asthma	0	10
Others	3	1
Total	19	26

Licenses

Licenses	Total Number
Antigens/Antibodies	11
Reagents/Methods/Cell Lines	7
Genes/Adapters/Vectors/Oligonucleotides	6
Gene Testing/Therapeutics	2
Total	26

Pilot Projects Funded for 2018-2019

Trainee Pilot Awards

Two new trainee awardees were selected for the second annual trainee pilot program in the amount of \$10K over a one-year period. The purpose of the program is to support postdoctoral/clinical 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 applicants 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: Hsi-en John Ho, MD, Clinical Fellow, Department of Pediatrics

Project Title: The Oral Microbiome and Metabolic Alterations in Food Allergy

Primary Mentor: Supinda Bunyavanich, MD, MPH, Department of Pediatrics and Genetics and Genomic Sciences

Secondary Mentor: Scott H. Sicherer, MD, Chief, Department of Pediatrics and Director, Jaffe Food Allergy Institute

Secondary Mentor: Alexander V. Grishin, PhD, Department of Pediatrics

▲ Predoctoral Division

Investigator: Milo R. Smith, PhD, Postdoctoral Fellow, Department of Neuroscience, Genetics and Genomic Sciences, Psychiatry, and Ophthalmology

Project Title: A cell-type specific in vitro model to rapidly screen modulators of neurodevelopmental plasticity

Primary Mentor: Hirofumi Morishita, MD, PhD, Department of Neuroscience, Genetics and Genomic Sciences, Psychiatry, and Ophthalmology and Joel T. Dudley, PhD, Department of Genetics and Genomic Sciences

Secondary Mentor: Nan Yang, PhD, Department of Neuroscience



*Hsi-en John Ho, MD,
Clinical Fellow, Department of
Pediatrics*



*Milo R. Smith, PhD
Postdoctoral Fellow,
Department of Neuroscience,
Genetics and Genomic Sciences,
Psychiatry, and Ophthalmology*

Pilot Projects Funded for 2018-2019 continued

Faculty Pilot Awards

Three pilot projects were selected for \$70K in institutional funding for a one-year period starting January 31, 2019. 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: M. Cecilia Berin, PhD, and David Dunkin, MD

Project Title: Food-specific T cells in Eosinophilic Esophagitis

Principal Investigators: Hirofumi Morishita, MD, PhD, and Yasmin Hurd, PhD

Project Title: Neurodevelopmental Basis of Genetic Vulnerability for Cannabis Use Disorder

Principal Investigators: Andrew J. Sharp, PhD, and Ruth J.F. Loos, PhD

Project Title: Association studies of multicopy genes in the Mount Sinai BioMe exome sequencing cohort



*M. Cecilia Berin, PhD
Professor, Pediatrics*



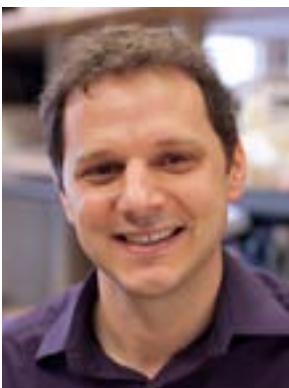
*David Dunkin, MD
Assistant Professor,
Pediatrics*



*Hirofumi Morishita, MD,
PhD
Associate Professor,
Psychiatry, Ophthalmology
and Neuroscience*



*Yasmin Hurd, PhD
Director, Addiction Institute
Professor, Pharmacological
Sciences, Neuroscience and
Psychiatry*



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



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

Annual Retreat

The Sixth Annual MCHDI Retreat was held at the Harmonie Club on November 27, 2018. Over 100 faculty members, trainees, and guests were in attendance. The retreat planning committee members: Amy R. Kontorovich, Dalila Pinto, Avi Reichenberg and Maya Deysenroth introduced new elements to the program this year, including select faculty talks and the poster board game. There were talks from trainee/faculty pilot grant recipients and select talks from faculty throughout the day. Dr. Bruce D. Gelb moderated a panel on conventional and nonconventional sources of funding with panelists from the Grants and Contracts Office and Mount Sinai Innovation Partners. The Young Investigators Competition participants Erik De Water, PhD (PI: Ruth J.F. Loos) and Felix Richter, MD/PhD candidate (PI: Bruce D. Gelb) were awarded in the postdoctoral and predoctoral divisions, respectively. For the parent's perspective segment, we invited a family with a child affected by cystic fibrosis. The audience enjoyed an engaging segment with Dr. Gelb as the moderator. The day rounded off with the poster session and poster board game hosted by the Trainee Leadership Committee: Jennie Altman, Maya Deysenroth, and Felix Richter. Participants were asked to match hints on the game card with the correct poster, and three winners who completed the most hints were awarded a fun gift bag of prizes.



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/mchdi

Newsletter ▲ The MCHDI Developmental Outcomes is a biannual 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

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, events, and other information relevant to children's health. Please like and follow our page at 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



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 email 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



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 more than 1000 enrollees. The pediatric cohort is comprised of samples from diverse racial and ethnic groups.

For more information, please visit: www.icahn.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: icahn.mssm.edu/research/resources/shared-resource-facilities/histology

Strategic Plan Implementation

Inborn Errors of Immunity Program

Inborn errors of immunity comprise over 330 different monogenic conditions in which one or more components of the human immune system are affected. These conditions are expressed as a combination of various phenotypes: infection (life-threatening or recurrent), malignancy, autoinflammation, autoimmunity or severe allergy. Most of these conditions manifest in childhood – some can first manifest in adulthood. Inborn errors of immunity carry important morbidity and mortality and are a significant burden to health economics systems. Moreover, uncovering the genetic diagnosis in an early stage of the disease is crucial for designing the optimal treatment, be it a drastic measure (hematopoietic stem cell transplantation or gene therapy), a therapy targeted to the cellular pathway that is defective, or a generic treatment with antibiotics/immunoglobulins to prevent irreversible end-organ damage.

As such, this program has found an excellent niche within the Mindich Child Health and Development Institute: the program is per se a “bed to bench and back” program. Hence, the program aims to embrace all physicians, physician-scientists, and scientists working on the immune system and the organ systems affected by a defect in the immune system. This broad research effort encompassing genomic strategies as well as classic molecular immunology aims to unravel the pathophysiology of known but poorly described inborn errors of immunity and to decipher new inborn errors of immunity. Moreover, through specific pathway analysis, we aim to develop new tools for targeted therapy in children and adults affected by inborn errors of immunity.

Pediatric Precision Medicine Program

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 Program, 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 **Genetic Disease Foundation** 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.





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