



Icahn School
of Medicine at
Mount
Sinai

*The Mindich
Child Health and
Development Institute*

2022 ANNUAL REPORT



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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Director's Message

Bruce D. Gelb, MD, Director

What a difference a year makes! As I wrote this message a year ago, in early 2022, we were in the midst of the COVID-19 wave from the Omicron variant. Our hopes then were that this wave would wane fast and enable us to move to a manageable endemicity. Indeed, looking back on the year as it unfolded, January 2022 was the worst of it by far and the summer wave was quite modest. Thus far, no significant COVID-19 wave has occurred this winter. Recognizing this, the U.S. Government will end its national and public health emergency declarations in mid-May. This aligns with our increasing return to normality at Mount Sinai—dispensing with masks in our research areas and resuming many of our in-person activities. While SAR-CoV-2 vaccinations are poised to become a routine part of our health maintenance, much like influenza vaccinations, we should celebrate the scientific triumphs that have liberated us from this scourge, saving countless lives.

Before becoming too triumphalist, let us acknowledge that research will be needed for many years to come in order to understand the true impact of COVID-19, particularly about today's children and adolescents. How does seeing only one's immediate family, at least without masks, and rarely interacting with peers for extended periods impact the growth and devel-

opment of young children? What are the effects from isolation on maturation for adolescents, particularly ones with developmental or social skills challenges, and can those be overcome as society reopens? I think about the impact of the Dutch famine during the winter of 1944–45, which led to epigenetic alterations that continue to contribute to certain health risks in subsequent generations to the present day. Not to equate nutritional and social deprivations, but one can imagine the impact of the past three years echoing for quite some time. The opportunity and obligation to study such phenomena lie in front of us and our successors.

Our School announced that it was undertaking a bold new biobanking project, the Mount Sinai Million Health Discoveries Program. This program's goal is to recruit a cohort of one million diverse individuals through the Mount Sinai Health System, whose DNA will undergo exome sequencing. Those sequencing data will then be analyzed using the rich clinical data available through our electronic medical records. For the MCHDI, the added excitement is the institutional commitment that 10%, or 100,000, of recruited individuals will be infants, children and adolescents, which our institute will oversee. A notable gap in most of the existing large biobanks with exome or genome sequencing, for example, the United Kingdom Biobank, All of Us (to date), the Million Veterans Project, has been the absence of pediatric participants. This has limited the ability of child health researchers to undertake large-scale genomic medicine studies that enable new discoveries for diseases of childhood. It has also limited our ability to understand the impact of putatively damaging genetic variation on children's health on a population basis. Recruiting this size cohort at Mount Sinai will clearly be a major lift, but the potential benefits to child health research are substantial.

Finally, the MCHDI is working with our partners in the Department of Pediatrics and Mount Sinai Children's to initiate some major new research endeavors. We hope that 2023 will see their successful launches. Stay tuned for more about those initiatives as they come to fruition.

FACULTY GROWTH

MCHDI ANNUAL FACULTY GROWTH 2022

In 2022

We welcomed five new external faculty and four internal faculty members to our institute.

85 Members

In 2022, we welcomed five new external faculty and four internal faculty members to our institute. Currently, we total 85 members consisting of scientists and physician-scientists across the disciplines of Allergy & Asthma, Cardiovascular Disease, Neurodevelopmental Disorders, Obesity & Diabetes, and more.

Existing faculty
Internal recruits
External recruits

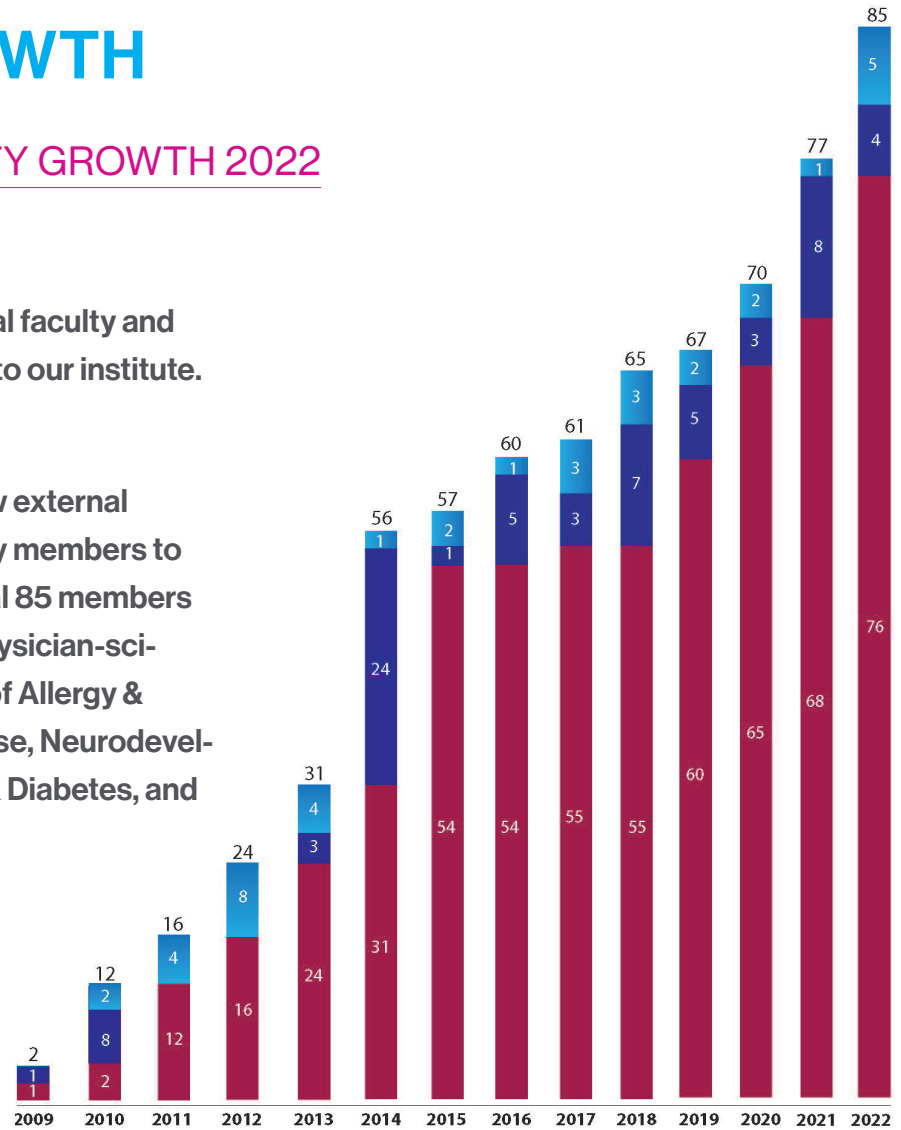


Chart of faculty recruits since our inception in 2009. In 2022, our institute recruited five new external and four new internal faculty members to our institute.

MCHDI FACULTY MEMBERS

MCHDI faculty members from left to right: Nicole C. Dubois, PhD; Adolfo Garcia-Ocaña, PhD; Donald K. Scott, PhD; M. Cecilia Berin, PhD (no longer at Mount Sinai); Minji Byun, PhD (no longer at Mount Sinai); David Dunkin, MD; Dalila Pinto, PhD; Andrew J. Sharp, PhD; Dani Dumitriu, MD, PhD (no longer at Mount Sinai); 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)






Icahn
School of
Medicine at
Mount
Sinai

NEW FACULTY

NEW EXTRAMURAL FACULTY



Nathalie Chami, PhD

Nathalie Chami, PhD, is an instructor of Environmental Medicine & Public Health in the lab of Dr. Ruth Loos at the Icahn School of Medicine. The focus of her research has been on gene discovery in monogenic and polygenic traits and disorders, including blood cell traits, familial dilated cardiomyopathies, obesity, and related metabolic traits using genotype and sequencing data.

She is currently leading one of the largest studies aimed at identifying novel genes associated with BMI and obesity using whole exome and whole genome sequencing analysis in TOPMed and the UK Biobank, two of the largest datasets available to the research community. She is also leveraging these datasets to study the penetrance of variants that cause monogenic disease in order to improve interpretation of rare variants. In addition, she is interested in identifying genetic factors that increase the risk of obesity but otherwise decrease the risk of cardiometabolic traits such as cholesterol and blood pressure and vice

versa with the purpose of elucidating the mechanisms that uncouple weight gain from susceptibility to cardiovascular outcomes.

Dr. Chami received a BSc in biomathematics and an MSc in human genetics from McGill University. She then pursued her doctoral studies at the Montreal Heart Institute. Dr. Chami received a postdoctoral grant from the Canadian Institute of Health Research from 2017-2020 and completed her postdoctoral training at the Icahn School of Medicine at Mount Sinai.



Sarah Duncan-Park, PhD

Sarah Duncan-Park, PhD, is an Assistant Professor in the Department of Pediatrics, Division of Developmental-Behavioral Pediatrics at the Icahn School of Medicine at Mount Sinai. Dr. Duncan-Park received her undergraduate degree summa cum laude from Providence College and attended Teachers College, Columbia University, where she received her Master of Arts in clinical psychology.

She then earned her doctorate in clinical psychology from Fordham University, where she received an F31 Ruth L. Kirschstein Predoctoral Individual National Research Service Award through the NICHD to investigate patterns of medication adherence in pediatric solid organ transplant recipients. Dr. Duncan-Park completed her predoctoral internship training with the NYU-Bellevue Clinical Psychology Internship Program – Child and Adolescent Track, as well as a part-time postdoctoral fellowship through NYU's

Integrated Behavioral Health program, during which time she also served as a study manager at Mount Sinai. Her areas of expertise include youth and caregiver coping with pediatric chronic illness, understanding and supporting medication adherence, and intervention development in this population.

NEW EXTRAMURAL FACULTY - CONTINUED



Son Duong, MD

Son Duong, MD, received his medical doctorate at the University of Virginia School of Medicine. He completed his residency training in pediatrics at UPMC Children's Hospital of Pittsburgh and completed a Master of Science in clinical research at the University of Pittsburgh.

He then completed his Pediatric Cardiology Fellowship at Lucile Packard Children's Hospital at Stanford, and an advanced noninvasive imaging fellowship at Icahn Mount Sinai and Mount Sinai Kravis Children's Hospital. Dr. Duong's research is focused on application of advanced data analysis techniques to large-scale data sources for better

prediction of outcomes in patients with congenital heart disease. He is currently a faculty member of the Artificial Intelligence in Medical Science (AIMS) Lab. As a specialist in cardiac imaging, he is developing artificial intelligence-assisted prediction tools for cardiac structure and function from multimodality data.



Joan Han, MD

Joan Han, MD, is a Professor of Pediatrics and Chief of the Division of Pediatric Endocrinology and Diabetes in the Jack and Lucy Clark Department of Pediatrics at the Icahn School of Medicine at Mount Sinai and Mount Sinai Kravis Children's Hospital. She earned her undergraduate and medical degrees from Harvard University.

She completed her residency in pediatrics at Boston Children's Hospital and Boston Medical Center, and pursued further advanced training in a clinical research fellowship at Nemours Children's Clinic in Jacksonville, Florida, and a pediatric endocrinology fellowship at the National Institutes of Health in Bethesda, Maryland. Prior to joining Mount Sinai, she was Associate Professor of Pediatrics and Director of the Pediatric Obesity Program at the University of Tennessee Health Science Center and Le Bonheur Children's Hospital in Memphis, Tennessee. She is board certified in general pediatrics, pediatric endocrinology, and obesity medicine and has published broadly in these fields. Dr.

Han's primary research efforts focus on the neuroendocrine regulation of energy balance and cognitive functioning as well as the genetic, environmental, and behavioral determinants of metabolic health in the general population and in patients with rare genetic disorders associated with obesity and type 2 diabetes. She served as Pediatric Associate Editor for the *International Journal of Obesity* from 2015-2021. She is a member of the American Pediatric Society, the Society for Pediatric Research, the American Pediatric Society, the Pediatric Endocrine Society, the Endocrine Society, and The Obesity Society, and is a fellow of the American Academy of Pediatrics.



Elvin Wagenblast, PhD

Elvin Wagenblast, PhD, is an Assistant Professor of Oncological Sciences and Pediatrics at the Icahn School of Medicine at Mount Sinai. Dr. Wagenblast earned his PhD in biological sciences from Cold Spring Harbor Laboratory.

He most recently was a Human Frontier Science Program Fellow and Banting Fellow in the laboratory of John Dick, PhD, FRS, at Princess Margaret Cancer Centre at the University of Toronto. There, he developed methodologies for genome editing technologies in human primary blood stem cells. In the summer of 2022, Dr. Wagenblast joined

Mount Sinai to study childhood leukemia. With the support of a Damon Runyon-Rachleff Innovation Award, the central question of his laboratory is to understand how a normal blood stem cell can become cancerous and how this process is different in children versus adults.

INTRAMURAL FACULTY



Sharon Baumel-Alterzon, PhD

Sharon Baumel-Alterzon, PhD, is an instructor in the Diabetes, Obesity and Metabolism Institute at the Icahn School of Medicine at Mount Sinai. Dr. Baumel-Alterzon received her PhD in 2014 from the Technion-Israel Institute of Technology, where she studied parasitic diseases.

In 2018, Dr. Baumel-Alterzon joined Dr. Donald Scott's lab at Mount Sinai to study the underlying mechanisms that regulate the expansion of pancreatic β -cells with the idea of finding therapeutic targets that can regenerate β -cells in diabetes. Specifically, her work focuses on the role of Nrf2 transcription factor, a master regulator of anti-oxidative response, on adaptive β -cell expansion, survival, and identity. Dr. Baumel-Alterzon has recently shown that Nrf2 is required for adaptive β -cell expansion under situations of overnutrition and that activation of the Nrf2 pathway using pharmacological agents increases human β -cell proliferation, highlighting the promising therapeutic potential of compounds that modulate Nrf2 to induce β -cell regenera-

tion for diabetes. Since defects in maternal β -cell adaptive expansion can lead to gestational diabetes mellitus, Dr. Baumel-Alterzon has been recently awarded with a NIH/NIDDK KO1 award to study the role of Nrf2 in β -cell turnover during pregnancy. Additionally, due to the sharp increase in the annual incidence of both type 1 (1.8%) and type 2 (4.8%) diabetes in the pediatric population in the last decades, as part of MCHDI trainee pilot award, Dr. Baumel-Alterzon began to explore the role of Nrf2 in regulating β -cell expansion at early stages of life. Dr. Sharon Baumel-Alterzon's final goal is to identify mechanistic targets for therapeutic intervention in diabetes.



Megan Januska, MD

Megan Januska, MD, is an Assistant Professor in the Division of Pediatric Pulmonology in the Jack and Lucy Clark Department of Pediatrics and in the Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai and Mount Sinai Kravis Children's Hospital.

After receiving her undergraduate degree from Grinnell College and her medical degree from the Geisel School of Medicine at Dartmouth, Dr. Januska completed her pediatric residency and pediatric pulmonology fellowship training at the Icahn School of Medicine at Mount Sinai.

Supported by a Cystic Fibrosis Foundation Clinical Fellowship Award, Dr. Januska developed a research project focusing on the cellular and molecular mechanisms that define the pediatric cystic fibrosis airway through the application

of single-cell technologies to minimally invasive respiratory specimens obtained during flexible bronchoscopy. Leveraging the developed workflow, Dr. Januska now aims to create a single-cell atlas of the normal pediatric airway along with a corresponding model system through the generation of patient-derived airway organoids with the support of a KL2 Scholars Award. Ultimately, Dr. Januska intends to apply the workflow and generated dataset to develop novel and minimally invasive methods to investigate and diagnose rare and severe pediatric respiratory disorders.

INTRAMURAL FACULTY - CONTINUED



Behrang Mahjani, PhD

Behrang Mahjani, PhD, has a unique background in analyzing complex biological data using advanced statistical models. He completed his BSc at K.N.Toosi University of Technology in 2004 and his first MSc in complex adaptive systems with a specialization in population genetics at the Chalmers University of Technology, Sweden, in 2008.

He continued at the Chalmers University of Technology and received his second MSc in mathematical statistics in 2011. He then completed his PhD at Uppsala University, Sweden, in statistical computing in 2016. His doctoral dissertation was focused on the development of new analytical methods for the genetic mapping of complex traits.

Dr. Mahjani spent one year as a postdoctoral fellow at the Department of Biostatistics and Epidemiology at Karolinska

Institutet, where he received training in epidemiology and statistical methods for register-based research. Then, he was a postdoctoral fellow at the Department of Psychiatry at the Icahn School of Medicine at Mount Sinai under the mentorship of Drs. Joseph Buxbaum and Dorothy Grice. Dr. Mahjani's primary interest is to better understand the developmental mechanisms and trajectories of childhood neuropsychiatric disorders, from the prenatal period through adolescence.



Sarah Stanley, PhD

Sarah A. Stanley, PhD, is an Assistant Professor the Icahn School of Medicine at Mount Sinai in the Diabetes, Obesity and Metabolism Institute, and Neuroscience. Her research focuses on developing and optimizing tools to image and modulate neural circuits and applying these to understand neural control of metabolism.

After receiving her undergraduate and medical degrees from Cambridge University, Dr. Stanley completed her endocrinology training and Ph.D. at Imperial College London. Supported by a Medical Research Council fellowship, Dr. Stanley moved to Rockefeller University for postdoctoral training, focusing on developing novel neuromodulatory tools to study the neural circuits regulating glucose metabolism. Since joining the Diabetes Obesity and Metabolism

Institute at Mount Sinai, Dr. Stanley's lab has continued to develop and optimize novel imaging and neuromodulatory tools to examine the roles of central and peripheral neural circuits in the regulation of glucose metabolism and determine how these circuits are disrupted in metabolic disease. Ultimately, the aim of these studies is to identify new methods to prevent and treat diseases such as diabetes.



FACULTY RESEARCH AREAS

ASTHMA AND ALLERGY



M. Cecilia Berin, PhD

(Adjunct Professor, Pediatrics)

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



Supinda Bunyavanich, MD, MPH

(Professor, Pediatrics, and Genetics and Genomic Sciences)

Research Areas: Integrative genomics of asthma and allergic diseases



Maria Curotto de Lafaille, PhD

(Associate Professor, Pediatrics)

Research Areas: Immunology of allergic diseases, B lymphocyte responses



Ke Hao, ScD

(Professor, Genetics and Genomic Sciences)

Research Areas: Genetic pleiotropy, mendelian randomization, inflammatory bowel disease, placenta biology, ambient air particulate matter exposure



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 and anaphylaxis



Karen M. Wilson, MD, MPH

(Adjunct Professor, Pediatrics)

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

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

(Associate Professor, Cell, Developmental & Regenerative Biology)

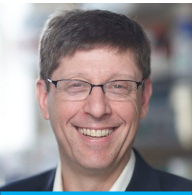
Research Areas: Heart development, stem cell differentiation, disease modeling



Son Duong, MD

(Assistant Professor, Pediatrics)

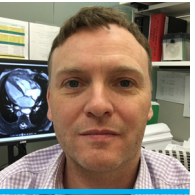
Research Areas: Artificial intelligence in cardiac imaging, pediatric cardiology



Bruce D. Gelb, MD

(Dean of Child Health Research, Gogel Family Professor and Director, Mindich Child Health and Development Institute; Professor, Pediatrics, and Genetics and Genomic Sciences)

Research Areas: Genetics of cardiovascular diseases, stem cell research, pediatric precision medicine



Alan Groves, MBChB, MD

(Adjunct Professor, Diagnostic, Molecular and Interventional Radiology)

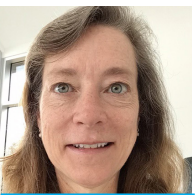
Research Areas: Hemodynamics, cardiac function, echocardiography, magnetic resonance imaging



Yuval Itan, PhD

(Associate Professor, Genetics and Genomic Sciences)

Research Areas: Human disease genomics, computational biology, and bioinformatics



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; Associate Professor, Medicine)

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

NEURODEVELOPMENTAL DISORDERS



Mafalda Barbosa, MD, PhD

(Assistant Professor, Genetics and Genomic Sciences)

Research Areas: Genetics of neurodevelopmental disorders, precision medicine, rare diseases



Jennifer Bragg, MD

(Associate Professor, Pediatrics)

Research Areas: Neurodevelopmental disorders, sensory processing disorders, impact of parental stress on neurodevelopment, developmental programming, neurodevelopment in children with congenital heart disease, whole genome sequencing in infants and neonates



Michael S. Breen, PhD

(Assistant Professor, Psychiatry, and Genetics and Genomic Sciences)

Research Areas: Functional genomics of neurodevelopmental and neuropsychiatric disorders, transcriptomics, single-cell RNA-sequencing, stem cells, RNA editing and biology



Joseph D. Buxbaum, PhD

(Deputy Chair, Department of Psychiatry; Director, Seaver Autism Center for Research and Treatment; Professor, Psychiatry, Neuroscience, and 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, and Oncological Sciences)

Research Areas: Environmental epigenetics, molecular epidemiology



Tirtha K. Das, PhD

(Assistant Professor, Cell, Developmental & Regenerative Biology)

Research Areas: Integrating fly plus vertebrate disease models, cancer, rare mendelian diseases, therapeutics development



Silvia De Rubeis, PhD

(Associate Professor, Psychiatry)

Research Areas: Intellectual disability, autism spectrum disorder, functional genetics, cell and animal model systems, brain development



Jennifer Foss-Feig, PhD

(Associate Professor, Psychiatry)

Research Areas: Autism spectrum and related neurodevelopmental disorders, neuroimaging, interactive social neuroscience, biomarker discovery, sensory processing

NEURODEVELOPMENTAL DISORDERS - CONTINUED



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



Lisa Eiland, MD

(Associate Professor, Pediatrics)

Research Areas: Stress and neurodevelopment



Hala Harony-Nicolas, PhD

(Associate Professor, Psychiatry, and Neuroscience)

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

(Associate Professor, Environmental Medicine & Public Health)

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



Laura Huckins, PhD

(Assistant Professor, Genetics and Genomic Sciences)

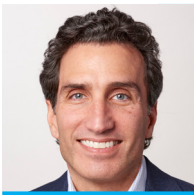
Research Areas: Psychiatric genetics, specializing in understudied disorders and disorders affecting vulnerable populations. Particular focus on anorexia nervosa, PTSD, sexual assault, OCD. Secondary focus on machine learning algorithms, transcriptomic imputation, multi-omic eQTL-based methodologies



Magdalena U. Janecka, PhD

(Assistant Professor, Psychiatry)

Research Areas: Neurodevelopmental disorders; epidemiology, epigenetics, environmental risk factors



Alex Kolevzon, MD

(Director, Child and Adolescent Psychiatry; Professor, Psychiatry, and Pediatrics)

Research Areas: Autism spectrum and other neurodevelopmental disorders



Robert S. Krauss, PhD

(Professor, Cell, Developmental & Regenerative Biology, and Oncological Sciences)

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

NEURODEVELOPMENTAL DISORDERS - CONTINUED



Paige M. Siper, PhD

(Adjunct Assistant Professor, Psychiatry)

Research Areas: Autism, intellectual disability, biomarker discovery, sensory processing



Luca Lambertini, PhD

(Assistant Professor, Obstetrics, Gynecology and Reproductive Science)

Research Areas: Placental biomarkers of altered fetal and child development



Behrang Mahjani, PhD

(Assistant Professor, Psychiatry, Genetics and Genomic Sciences, and Artificial Intelligence and Human Health)

Research Areas: Genetics of neurodevelopmental disorders



Florence Marlow, PhD

(Associate Professor, Cell, Developmental & Regenerative Biology)

Research Areas: Genetics of early patterning and germline, neurodevelopment



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

(Professor, Psychiatry, Ophthalmology, and Neuroscience)

Research Areas: Mechanisms of perceptual, cognitive, and social development relevant to neurodevelopmental disorders



Dalila Pinto, PhD

(Assistant Professor, Psychiatry, and Genetics and Genomic Sciences)

Research Areas: Genetics and genomics of neurodevelopmental disorders (particular focus on autism, epilepsy, schizophrenia, OCD), structural variation, transcriptomics, gene regulation, noncoding RNA

NEURODEVELOPMENTAL DISORDERS - CONTINUED



Avi Reichenberg, PhD

(Professor, Psychiatry, and Environmental Medicine & Public Health)

Research Areas: Autism, schizophrenia, other psychiatric disorders



Andrew J. Sharp, PhD

(Professor, Genetics and Genomic Sciences)

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



Annemarie Stroustrup, MD, MPH

(Adjunct Associate Professor, Pediatrics and Environmental Medicine & Public Health)

Research Areas: Neurodevelopment, perinatal environmental exposures, identifying genetic etiologies of congenital disease



Shanna H. Swan, PhD

(Professor, Environmental Medicine & Public Health)

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



Pilar Trelles, MD

(Adjunct Assistant Professor, Psychiatry)

Research Areas: Autism spectrum disorder, neurodevelopmental disorders, health disparities



Bryn D. Webb, MD

(Adjunct Assistant Professor, Genetics and Genomic Sciences)

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



Anusha Yeshokumar, MD

(Adjunct Assistant Clinical Professor, Pediatrics, and Neurology)

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

OBESITY AND DIABETES



Sharon Baemel-Alterzon, PhD

(Instructor, Medicine)

Research Areas: Beta-cell biology, oxidative stress, cell proliferation, cell cycle, cell division, apoptosis/cell death, diabetes, obesity, gene expressions, gene regulation, knockout mice, molecular biology, transcription factors



Ross L. Cagan, PhD

(Adjunct Professor, Cell, Developmental & Regenerative Biology)

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



Nathalie Chami, PhD

(Instructor, Environmental Medicine & Public Health)

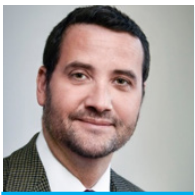
Research Areas: Genetics of complex traits, monogenic disease, genetics of obesity and cardiometabolic outcomes



Lauryn Choleva, MD

(Instructor, Pediatrics)

Research Areas: Type 2 diabetes, type 1 diabetes, hypoglycemia



Fernando Ferrer, MD, FACS, FAAP

(Professor, Urology)

Research Areas: Cancer, bioactive lipids, renal injury, biomarkers, renal obstruction



Adolfo García-Ocaña, PhD

(Professor, Medicine)

Research Areas: Diabetes, pancreatic beta cell biology



Allan C. Just, PhD

(Assistant Professor, Environmental Medicine & Public Health)

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



Joan Han, MD

(Chief, Division of Pediatric Endocrinology and Diabetes; Professor, Pediatrics)

Research Areas: Pediatric obesity, neuroendocrine regulation of energy balance, genetic obesity syndromes and disorders of the leptin pathway

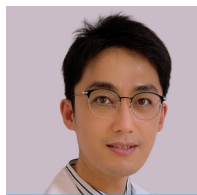
OBESITY AND DIABETES - CONTINUED



Ruth J.F. Loos, PhD

(Professor, Environmental Medicine & Public Health; Charles Bronfman Professor in Personalized Medicine)

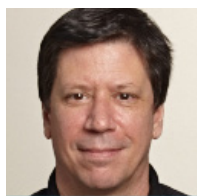
Research Areas: Genetics of obesity and related cardiometabolic traits, genetic epidemiology, epidemiology



Geming Lu, MD

(Instructor, Medicine)

Research Areas: Type 1 diabetes, type 2 diabetes, autoimmune disorders (IBD and MS), immunometabolism, beta cell regeneration, multiomic data analysis



Donald K. Scott, PhD

(Professor, Medicine)

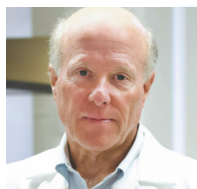
Research Areas: Metabolic regulation of transcription, beta cell regeneration and preservation, diabetes



Sarah Stanley, PhD

(Associate Professor, Medicine, and Neuroscience)

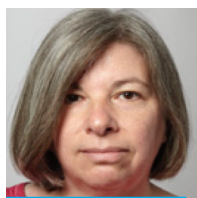
Research Areas: Neural control of metabolism



Andrew F. Stewart, MD

(Professor, Diabetes, Obesity and Metabolism Institute, Irene and Dr. Arthur M. Fishberg Professor, Medicine)

Research Areas: Type 1 diabetes, type 2 diabetes, beta cell regeneration, drug discovery



Susan Teitelbaum, PhD

(Professor, Environmental Medicine & Public Health)

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

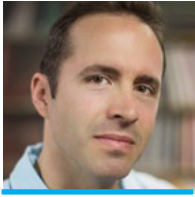


Nita Vangeepuram, MD, MPH

(Assistant Professor, Pediatrics, Environmental Medicine & Public Health, Population and Health Science and Policy)

Research Areas: Youth diabetes prevention, community-based participatory research, health equity research

OBESITY AND DIABETES - CONTINUED



Ryan W. Walker, PhD

(Assistant Professor, Environmental Medicine & Public Health)

Research Areas: Clinical microbiome, obesity, nutrition, environmental exposures

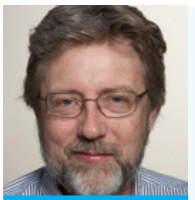


Martin J. Walsh, PhD

(Professor, Pharmacological Sciences, Genetics and Genomic Sciences, and Pediatrics)

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

OTHER RESEARCH FOCUSES



James J. Bieker, PhD

(Professor, Cell, Developmental & Regenerative Biology)

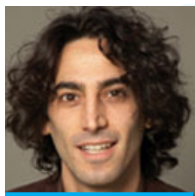
Research Areas: Transcriptional regulation of gene expression in erythroid cells



Dusan Bogunovic, PhD

(Professor, Microbiology, Oncological Sciences, and Pediatrics)

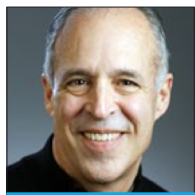
Research Areas: Genetics of infectious and inflammatory diseases, type I interferons, Pseudo-TORCH syndrome, neurolisterosis



Brian D. Brown, PhD

(Professor, Genetics and Genomic Sciences)

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



John Bucuvalas, MD

(Professor, Pediatrics)

Research Areas: Outcomes after liver transplantation, allograft injury in pediatric liver transplant recipients

OTHER RESEARCH FOCUSES - CONTINUED



Minji Byun, PhD

(Adjunct Assistant Professor, Medicine)

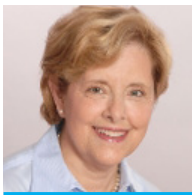
Research Areas: Genetics of immune disorders, clonal hematopoiesis, immune dysregulation



Jaime Chu, MD

(Assistant Professor, Pediatrics)

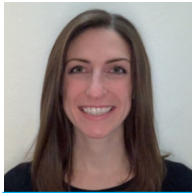
Research Areas: Disorders of glycosylation, cancer metabolism, liver fibrosis, environmental toxicants in liver disease



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



Sarah Duncan-Park, PhD

(Assistant Professor, Pediatrics)

Research Areas: Behavioral health intervention development, psychosocial adjustment to pediatric chronic illness



David Dunkin, MD

(Associate 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, and Population Health Science and Policy)

Research Areas: Biostatistical methods development for environmental health



Katherine Guttman, MD, MBE

(Assistant Professor, Pediatrics)

Research Areas: Palliative care, family-centered care, parent-physician communication, research ethics



Shelley H. Liu, PhD

(Assistant Professor, Population Health Science and Policy)

Research Areas: Biostatistics, environmental mixtures, public health

OTHER RESEARCH FOCUSES - CONTINUED



Megan Januska, MD

(Assistant Professor, Pediatrics, and Genetics and Genomic Sciences)

Research Areas: Integrative genomics of pediatric lung development and disease states, including cystic fibrosis



Kaustav Mukherjee, PhD

(Instructor, Cell, Developmental & Regenerative Biology)

Research Areas: Hematopoietic transcription regulation, genomics and epigenetics, single-cell technologies, erythroid disorders



Praveen Raju, MD, PhD

(Associate Professor, Neurology, and Pediatrics)

Research Areas: Pediatric brain tumors, developmental neurobiology, BBB drug delivery



Michael Rendl, MD

(Professor, Cell, Developmental & Regenerative Biology, and Dermatology)

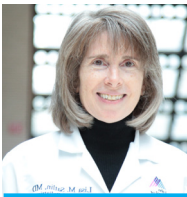
Research Areas: Stem cells, hair regeneration, morphogenesis



Jeffrey M. Saland, MD

(Chief, Pediatric Nephrology, and Hypertension; Professor, Pediatrics)

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



Lisa M. Satlin, MD

(Herbert H. Lehman Professor and Chair, Pediatrics)

Research Areas: Ontogeny and mechanoregulation of epithelial ion channels in secretory epithelia, generation and characterization of functional bioengineered kidneys



Eyal Shemesh, MD

(Professor, Pediatrics, and Psychiatry)

Research Areas: Remote intervention paradigms, biological correlates of non-adherent behaviors, multisite and multidisciplinary clinical trials

OTHER RESEARCH FOCUSES - CONTINUED



Christopher Sturgeon, PhD

(Associate Professor, Cell, Developmental & Regenerative Biology, and Medicine)

Research Areas: Hematopoiesis, development, pluripotent stem cells, adoptive immunotherapy



Rebecca Trachtman, MD

(Assistant Professor, Pediatrics)

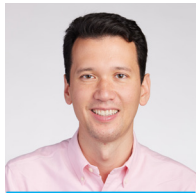
Research Areas: Biomarkers, patient-reported outcomes in juvenile idiopathic arthritis



Ernest Turro, PhD

(Associate Professor, Genetics and Genomic Sciences)

Research Areas: Biostatistics, statistical genetics, functional genomics, Bayesian modeling, rare diseases, inherited blood disorders, primary immunodeficiencies, mitochondrial genetics



Elvin Wagenblast, PhD

(Assistant Professor, Oncological Sciences, and Pediatrics)

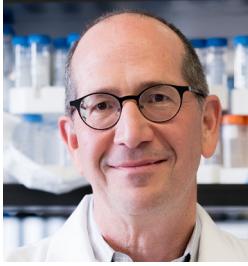
Research Areas: Blood stem cells, leukemia



FACULTY RESEARCH INTERACTIONS



Faculty Highlight: Avi Reichenberg, PhD



Joseph D. Buxbaum, PhD



Alex Kolevzon, MD



Dorothy E. Grice, MD



Magdalena U. Janecka, PhD



Paige M. Siper, PhD



Shanna H. Swan, PhD



Megan K. Horton, PhD, MPH



Anna-Sophie Rommel, PhD



Chris Gennings, PhD

Joseph D. Buxbaum, PhD

- Collaborator on environmental autism research
- Collaborator on Autism Risk and Maternal Cardiometabolic Health study
- Collaborator on Population-Based Autism Genetics and Environment study

Alex Kolevzon, MD

- Collaborator on environmental autism research
- Collaborator on Autism Risk and Maternal Cardiometabolic Health study
- Collaborator on Autism and Prenatal Endocrine Disruptors study

Dorothy E. Grice, MD

- Collaborator on Epidemiology of OCD and related disorders

Magdalena U. Janecka, PhD

- Mentor
- Collaborator on Maternal health in pregnancy and autism risk – genetic and non-genetic mechanisms
- Collaborator on Prenatal medication exposure in autism, birth complications, and developmental disabilities

Paige M. Siper, PhD

- Collaborator on environmental autism research

Shanna H. Swan, PhD

- Collaborator on Autism Risk and Maternal Cardiometabolic Health study
- Collaborator on Autism and Prenatal Endocrine Disruptors study

Megan K. Horton, PhD, MPH

- Collaborator on Metal mixtures, exposure windows, and neurodevelopmental trajectories from adolescence to adulthood
- Collaborator on Early life exposure to metal mixtures and neuroimaging of internalizing behaviors in childhood

Anna-Sophie Rommel, PhD

- Collaborator on The longer-term impact of prenatal exposure to SARS-CoV-2 infection and the COVID-19 vaccine on behavior, cognition, and brain functioning in the child

Chris Gennings, PhD

- Collaborator on Autism and Prenatal Endocrine Disruptors study



Faculty Highlight: Nicole Dubois, PhD



Supinda Bunyavanich, MD, MPH



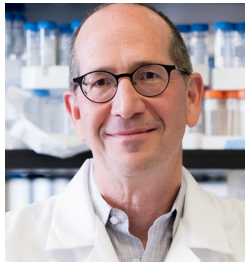
Bruce D. Gelb, MD



Magdalena U. Janecka, PhD



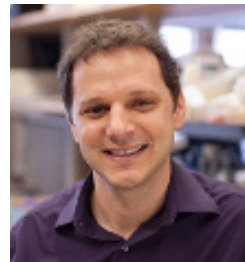
Silvia De Rubeis, PhD



Joseph D. Buxbaum, PhD



Robert S. Krauss, PhD



Andrew J. Sharp, PhD



David Dunkin, MD



Marek Mlodzik, PhD

Supinda Bunyavanich, MD, MPH

- Co-member of MCHDI Internal Advisory Board

Bruce D. Gelb, MD

- Mentor
- Co-sponsor for trainee fellowships David Gonzalez (F31) and Tasneem Ebrahim (AHA)
- Collaborator on mechanisms of de novo sarcomere formation

Magdalena U. Janecka, PhD

- Co-chair for MCHDI Incubator Faculty Meetings

Silvia De Rubeis, PhD

- Collaborator on "The role of RNA-binding protein DDX3X in neurodevelopmental disorders and congenital heart disease"

Joseph D. Buxbaum, PhD

- Collaborator on "The role of RNA-binding protein DDX3X in neurodevelopmental disorders and congenital heart disease"
- Co-member of MCHDI Internal Advisory Board

Robert S. Krauss, PhD

- Mentor

Andrew J. Sharp, PhD

- Collaborator, co-investigator on "Early mechanisms of cardiac specification" R01 HL134956
- Co-member of MCHDI Internal Advisory Board
- Collaborator, co-investigator on MCHDI pilot award "Generating Cardiac Purkinje Cells from Human Pluripotent Stem Cells" (past)
- Co-author on "Foxa2 identifies a cardiac progenitor population with ventricular differentiation potential" by Bardot et al., *Nat Comm* 2017

David Dunkin, MD

- Co-chair for MCHDI Incubator Faculty Meetings (past)

Marek Mlodzik, PhD

- Mentor

Praveen Raju, MD, PhD

- Co-member of MSTP admissions committee

Christopher Sturgeon, PhD

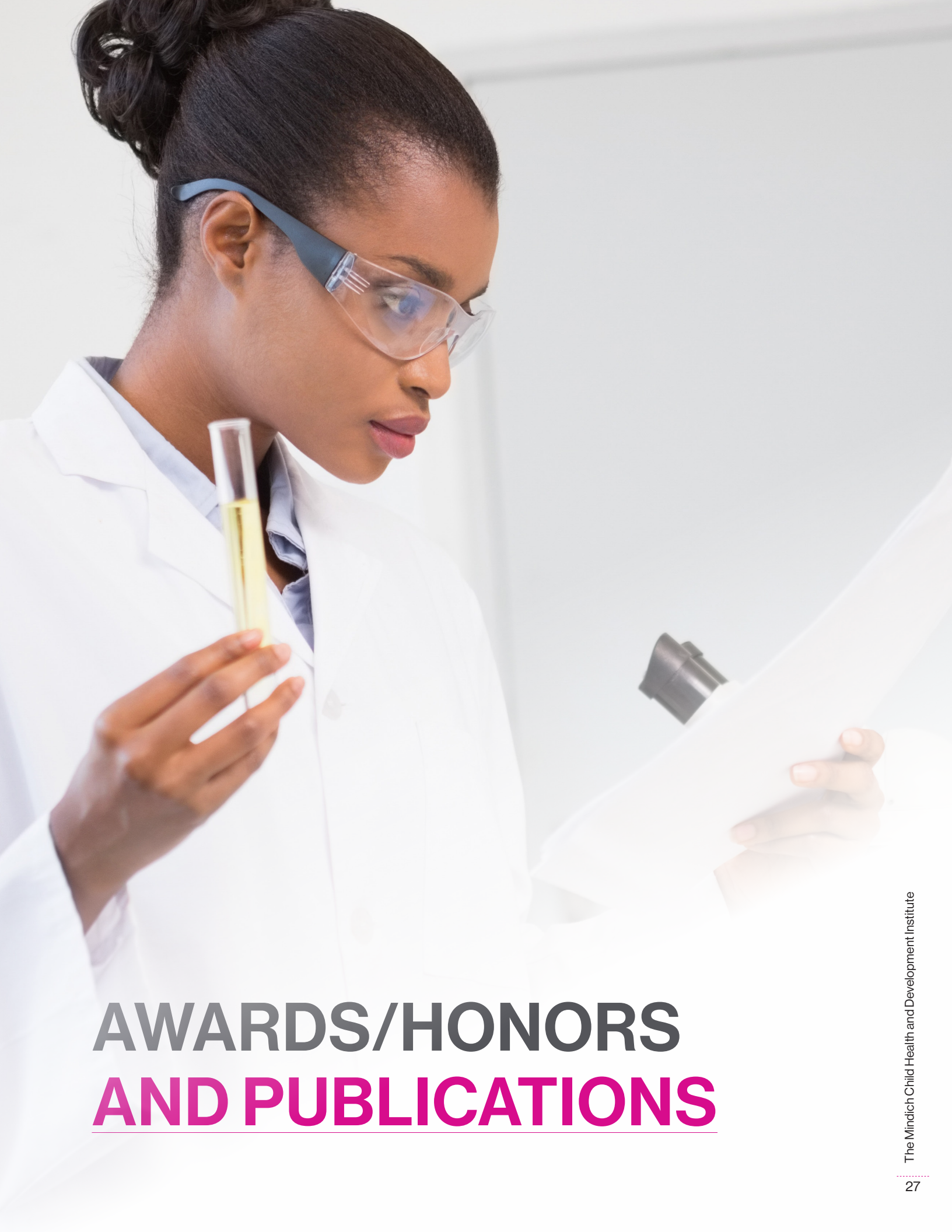
- Collaborator on "Mechanisms of macrophage-myocyte interactions"



Praveen Raju, MD, PhD



Christopher Sturgeon, PhD



AWARDS/HONORS AND PUBLICATIONS

FACULTY HONORS/AWARDS

Tirtha K. Das, PhD, Co-Chair and Speaker, 63rd Annual Drosophila Conference, “Human Disease Modeling” oral platform presentation section and “Flies on Drugs” workshop

David Dunkin, MD, American Gastroenterological Association, AGA Fellow

Bruce D. Gelb, MD, Promotion to Dean of Child Health at the Icahn School of Medicine at Mount Sinai

Yuval Itan, PhD, Elected as a member to the Henry Kunkel Society (HKS)

Amy R. Kontorovich, MD, PhD, Promotion to Director of the New Mount Sinai Center for Inherited Cardiovascular Diseases

Dalila Pinto, PhD, Chair, Symposium “Autism Spectrum Disorders,” 24th Biennial Meeting of the International Society for Developmental Neuroscience (ISDN), Vancouver, May 8, 2022

FACULTY PUBLICATIONS

Fu JM, Satterstrom FK, Peng M, Brand H, Collins RL, ... **Barbosa M**, ... **De Rubeis S**, **Buxbaum JD**, Daly MJ, Devlin B, Roeder K, Sanders SJ, Talkowski ME. **Rare coding variation provides insight into the genetic architecture and phenotypic context of autism.** *Nat Genet.* 2022 Sep;54(9):1320-31.

Baumel-Alterzon S, Katz LS, Brill G, Jean-Pierre C, Li Y, Tse I, Biswal S, **Garcia-Ocaña A**, **Scott DK**. **Nrf2 regulates β -cell mass by suppressing β -cell death and promoting β -cell proliferation.** *Diabetes.* 2022 May 1;71(5):989-1011.

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Berin MC, Agashe C, Burks AW, Chiang D, Davidson WF, Dawson P, ... **Sicherer SH**, ... **Sampson HA**. Allergen-specific t cells and clinical features of food allergy: Lessons from cofar immunotherapy cohorts. *J Allergy Clin Immunol.* 2022 Apr;149(4):1373-82.e12.

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Cuddlestone WH, Li J, Fan X, Kozenkov A, Lalli M, Khaliq S, ... **Breen MS**. **Cellular and genetic drivers of rna editing variation in the human brain.** *Nat Commun.* 2022 May 30;13(1):2997.

Svensson-Arvelund J, Cuadrado-Castano S, Pantsulaia G, Kim K, Aleynick M, Hammerich L, ... **Brown BD**, ... Brody **JD**. **Expanding cross-presenting dendritic cells enhances oncolytic virotherapy and is critical for long-term anti-tumor immunity**. *Nat Commun*. 2022 Nov 22;13(1):7149.

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GRANTS

AGENCY NAME	Funding from New Grants (\$)	Funding from New & Existing Grants (\$)
National Institute of Allergy and Infectious Diseases/NIH/DHHS	2,907,000	9,354,985
National Institute of Mental Health/NIH/DHHS	2,196,686	7,775,335
National Institute of Child Health and Human Development/NIH/DHHS	1,290,846	2,763,785
National Institute of Environmental Health Sciences/NIH/DHHS	1,259,559	5,475,318
National Institute of Diabetes and Digestive and Kidney Diseases/NIH/DHHS	722,990	9,289,212
National Center for Advancing Translational Sciences/NIH/DHHS	338,000	338,000
Additional Ventures Foundation	333,847	333,847
ChadTough Defeat DIPG Foundation	200,000	200,000
Biomarin Pharmaceutical	171,186	171,186
National Institute on Aging/NIH/DHHS	166,240	335,240
Leducq Foundation (Fondation Leducq)	154,935	154,935
University of Colorado	154,194	154,194
Simons Foundation	150,000	400,000
International OCD Foundation	100,000	100,000
Rhythm Pharmaceuticals, Inc.	76,369	76,369
American Society Of Hematology	62,500	62,500
FRAXA Research Foundation	50,000	50,000
Celgene International II Sarl	41,408	41,408
Benaroya Research Institute at Virginia	27,755	2,469,900
Solta Therapeutics	1,255	1,255
National Heart, Lung, and Blood Institute/NIH/DHHS	0	3,779,037
National Institute of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS	0	2,666,316
National Institute of Neurological Disorders And Stroke/NIH/DHHS	0	1,300,321
Johns Hopkins University Medical School	0	1,124,096
National Institute of General Medical Sciences/NIH/DHHS	0	1,098,774
National Eye Institute/NIH/DHHS	0	940,159
Albert Einstein College of Medicine	0	870,771
National Center for Complementary and Integrative Health/NIH/DHHS	0	707,878
National Institute of Dental and Craniofacial Research/NIH/DHHS	0	567,838
University of Wisconsin-Madison	0	390,167
New York State Stem Cell Board	0	363,550
Rutgers	0	336,207
Emory University	0	312,409
Montefiore Hospital	0	243,068
Columbia University	0	224,306
Johns Hopkins University	0	200,000
President and Fellows of Harvard College	0	183,846
Children's Hospital Pittsburgh	0	124,796
Massachusetts General Hospital	0	107,509
Food Allergy Research & Education	0	100,000
University of Washington	0	100,000
Cystic Fibrosis Foundation	0	99,979
Van Andel Institute	0	83,013
Vanderbilt University Medical Center	0	77,540
Broad Institute	0	66,478
The Johns Hopkins University	0	52,069
University of Cambridge	0	51,952
Jounce Therapeutics	0	50,850
University of Pittsburgh	0	50,850
Children's Hospital Med. Center, Ohio	0	50,000
Alex's Lemonade Stand Foundation for Childhood Cancer	0	50,000
Tufts University	0	43,739
Ann & Robert H. Lurie Children's Hospital of Chicago	0	42,968
Washington University School of Medicine in St. Louis	0	42,721
Washington University	0	41,951
Us-Israel Binational Science Foundation	0	40,000
Brain and Behavior Research Foundation	0	35,000
Hirsch/Well-Caulier Trust	0	35,000
European Commission	0	32,185
Virginia Commonwealth University	0	31,002
Leukemia and Lymphoma Society	0	27,824
Cincinnati Children's Hospital	0	9,000
Total	10,404,770	56,302,639

MATERIAL TRANSFER AGREEMENTS/LICENSES

Research Focus	Outgoing Material Transfer Agreements (#)	Licenses (#)
Neurodevelopmental disorders	3	18
Cardiovascular disease	1	1
Diabetes and Obesity	1	8
Allergy and Asthma	0	0
Others	1	2
Total	6	29

Licenses	Total Number
Antigens/Antibodies	6
Reagents/Methods/Cell Lines	9
Genes/Adapters/Vectors/Oligonucleotides	7
Gene Testing/Therapeutics	4
Mouse and Cell Models	3
Total	29



PILOT PROJECTS FUNDED FOR 2021-2022

TRAINEE PILOT AWARDS

Two new trainee awardees were selected for the 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 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.



Bhavana Shewale

PhD Candidate, Cell, Developmental & Regenerative Biology

Project Title: Investigating the Role of Rho-Signaling in Cardiac Actin Nucleation During De Novo Sarcomerogenesis

Primary Mentor: Nicole Dubois, PhD, Associate Professor, Cell, Developmental & Regenerative Biology, Mindich Child Health and Development Institute

Secondary Mentors: Robert Krauss, PhD, Professor & Program Director, Cell, Developmental & Regenerative Biology

Marek Mlodzik, PhD, Professor & Chair, Cell, Developmental & Regenerative Biology

FACULTY PILOT AWARDS

Three pilot projects were selected for \$75K in institutional funding for a one-year period starting March 1, 2022. 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).

Project Title: Genotype-Phenotype Correlations in Cardiac and Skeletal Muscle of Fatty Acid Oxidation Deficiencies

Principal Investigators: Nicole C. Dubois, PhD (Communicating PI), Sander Houten, PhD (Co-PI), George Diaz, MD, PhD (Co-I), Nenad Bursac, PhD (Co-I), Justin Cross, PhD (Co-I)



Nicole C. Dubois, PhD (Communicating PI)

*Associate Professor, Cell,
Developmental & Regenerative Biology
Mindich Child Health and
Development Institute
Black Family Stem Cell
Institute
Cardiovascular Research
Institute*



Sander Houten, PhD (Co-PI)

*Associate Professor, Genetics
and Genomic Sciences*

FACULTY PILOT AWARDS - CONTINUED



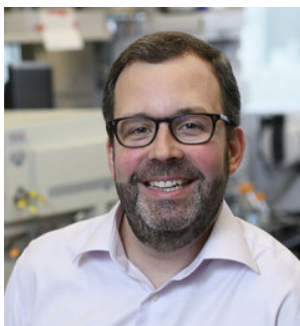
George Diaz, MD, PhD (Co-I)

Professor, Genetics and Genomic Sciences and Pediatrics



Nenad Bursac, PhD (Co-I)

*Professor, Biomedical Engineering and Cell Biology
Duke Cancer Institute
Director of Duke Regeneration Center
Duke University, Durham*



Justin Cross, PhD (Co-I)

*Director, Donald B. and Catherine C. Marron Cancer Metabolism Center
Memorial Sloan Kettering Cancer Center, New York*

Project Title: Immunophenotypic Comparison of Systemic Juvenile Idiopathic Arthritis and Multisystem Inflammatory Syndrome in Children

Principal Investigators: Rebecca Trachtman, MD (Communicating PI), and Dusan Bogunovic, PhD (Co-PI)



Rebecca Trachtman, MD (Communicating PI)

*Assistant Professor, Pediatrics
Mindich Child Health and Development Institute*



Dusan Bogunovic, PhD (Co-PI)

*Professor, Pediatrics, Microbiology, and Oncological Sciences
Mindich Child Health and Development Institute
Director, Center for Inborn Errors of Immunity
Precision Immunology Institute*

FACULTY PILOT AWARDS - CONTINUED

Project Title: Whole-Genome Sequencing of Neonatal Fatalities

Principal Investigators: Ernest Turro, PhD (Communicating PI), Felix Richter, MD, PhD (Co-I), Katherine Guttman, MD, MBE (Co-PI), Mafalda Barbosa, MD, PhD (Co-PI)



**Ernest Turro, PhD
(Communicating PI)**
*Associate Professor,
Genetics and Genomic
Sciences
Mindich Child Health and
Development Institute
Charles Bronfman Institute
for Personalized Medicine*



Felix Richter, MD, PhD (Co-I)
*Pediatrics Resident Physician
Mindich Child Health and
Development Institute*



**Katherine Guttman,
MD, MBE (Co-PI)**
*Assistant Professor,
Pediatrics
Mindich Child Health and
Development Institute*



**Mafalda Barbosa, MD, PhD
(Co-PI)**
*Assistant Professor, Genetics
and Genomic Sciences
Mindich Child Health and
Development Institute*

ANNUAL RETREAT

Our 10th Annual Retreat was hosted at the Harmonie Club on November 30, 2022, with ~120 faculty, trainees, staff, and volunteers at the event. It was a long-awaited opportunity to reconnect with colleagues in person in the midst of a pandemic that has devastated many, and affected us all. The retreat planning committee was composed of our MCHDI Director, Bruce Gelb, MD, Committee Chair, Florence Marlow, PhD, MCHDI faculty members, Ernest Turro, PhD, and Jennifer Foss-Feig, PhD, and trainee leadership committee member, Vahe Khachadourian, PhD. Our keynote speaker and panelist was Lilianna Solnica-Krezel, PhD (Alan A. and Edith L. Wolff Distinguished Professor and Head Department of Developmental Biology, Co-Director, Center of Regenerative Medicine, Washington University School of Medicine) who presented on the topic “Forward and Reverse in Zebrafish to Understand Development and Disease.” Our panel on “Beating Burnout, Work/Life Balance in Academia and Mental Health Resources” were also led

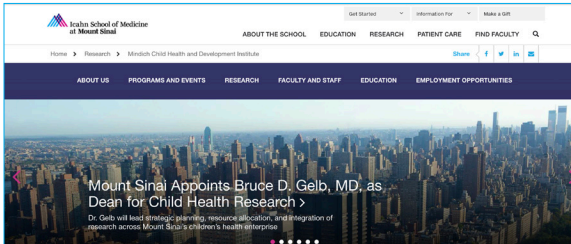
by Ashley Doukas, PhD (Associate Clinical Director, The Center for Stress, Resilience, and Personal Growth), Acanthus Fairley, LCSW (Workforce Retention Coordinator), Employee Assistance Program, and Basil Hanss, PhD (Senior Associate Dean of Postdoctoral and Student Affairs) who shared their personal experience and provided mental health resources for our audience.

We congratulated our Young Investigators Competition (YIC) winners in the postdoctoral division, Daniel Greene, PhD (PI: Ernest Turro, PhD) and predoctoral winners, Michael Espino Horesh (PI: Dusan Bogunovic, PhD) and Rollie Hampton (PIs: Sarah Stanley, PhD, and Adolfo Garcia-Ocaña, PhD). The awardees for best posters selected by judges were Clifford Liu, MS (PI: Bruce Gelb, MD), Lauren Dierdoff (PI: Silvia De Rubeis, PhD), Christos Sazeides, MS (PI: Dusan Bogunovic, PhD) and the crowd favorite voted by attendees, Mariana Waldetario, PhD (PI: Maria Lafaille, PhD).

COMMUNICATIONS

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

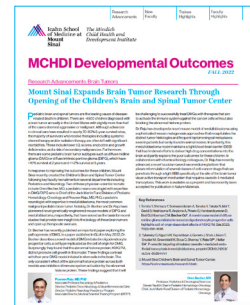
Website



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 at <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.

Twitter

Our tweets are streaming on our website in real time. Follow or tweet to us @MindichCHDI or visit our website at www.mountsinai.org/mchdi.





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-mails alert, and tailored grant recommendations. Grant Forward subscriptions for MCHDI faculty and trainees are covered by our Institute.

To sign up, please visit: <https://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 composed 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

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:

<http://ica hn.mssm.edu/research/resources/shared-resource-facilities/histology>

STRATEGIC PLAN IMPLEMENTATION

PEDIATRIC CLINICAL TRIALS OFFICE

INTRODUCTION

The Pediatric Clinical Trials Office (PCTO) within the Mindich Child Health and Development Institute (MCHDI) serves the needs of researchers within the Mount Sinai Health System who wish to conduct clinical trials with pediatric participants. The program became active after the pandemic surge in Fall 2020. We are partnered with the Clinical Trials Office (CTO) in Internal Medicine to manage existing and “in development” trials, whether they involve investigator-initiated, industry-funded, or federally funded efforts. Of note, in addition to the range of services offered by the CTO, we support “extension” trials (adult studies extending into pediatric populations) and NIH-funded clinical research.

PCTO STAFF

In 2022, the PCTO workforce doubled in size to provide support to our growing portfolio.

Current staff

[Michele Cohen, MS, CCRC](#) – Co-Director / [Eyal Shemesh, MD](#) – Co-Director

[Yair Bitton, MPH, MBA, CCRP](#) – Program Manager

[Catherine Swarts, MS](#) – Clinical Research Coordinator II

[Navjot Kaur](#) – Financial Analyst

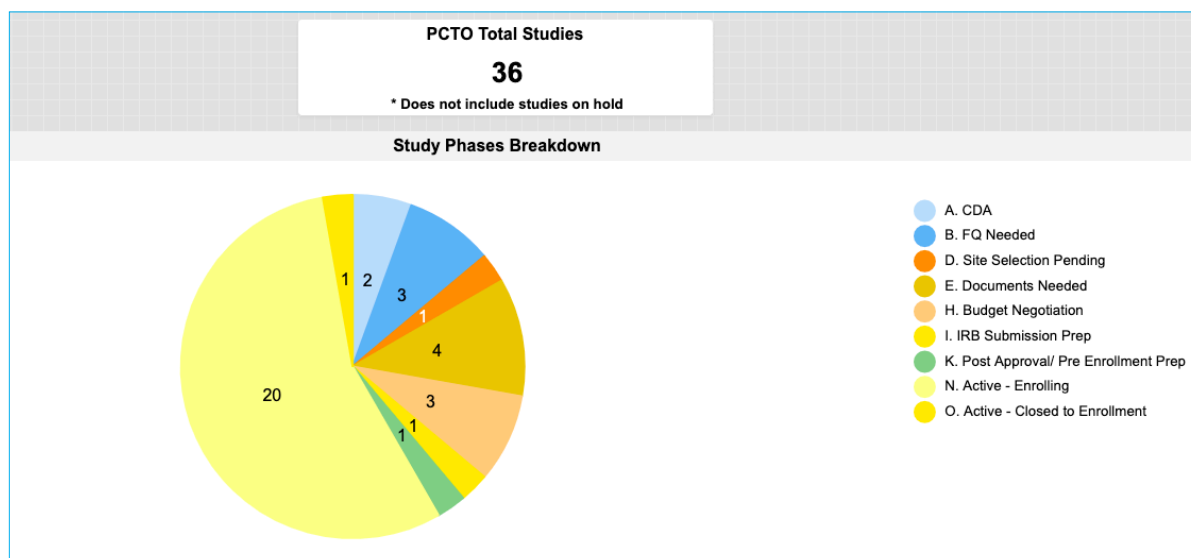
NEW in 2022

[Angela Stangarone](#) – Senior Regulatory Coordinator

[Alyssa Gontzes](#) – Clinical Research Coordinator II

[Cindy Gaytan](#) – Clinical Research Coordinator I

[Gabrielle Jonny](#) – Clinical Research Coordinator II



RESEARCH FACULTY SERVED BY PCTO

PCTO is currently serving the following investigators/divisions:

Pediatric Divisions:

Allergy

Scott Sicherer, MD

Julie Wang, MD

Amanda Cox, MD

Roxanne Oriel, MD

Mary Grace Baker, MD

Nicole Ramsey, MD

Endocrinology

Joan Han, MD

Robert Rapaport, MD

Hillary Hotchkiss, MD

Gastroenterology

Marla Dubinsky, MD

David Dunkin, MD

Keith Benkov, MD

Nancy Pittman, MD

Nephrology

Jeffrey Saland, MD

Hillary Hotchkiss, MD

Neonatal ICU

Courtney Juliano, MD

Rheumatology

Rebecca Trachtman, MD

Cardiology

Erin Paul, MD

NEW in 2022

Pediatric ICU

Sheemon Zackai, MD

Sandeep Gangadharan, MD

Shubhi Kaushik, MD

Adult Divisions With Pediatric Trials:

Allergy and Immunology

Paula Busse, MD

Dermatology

Emma Guttman, MD, PhD

CLINICAL TRIALS PORTFOLIO

Notable Projects

- PCTO is involved in the implementation and administration of an NIH funded, 7-year, multi-million-dollar consortium project.
- PCTO is working on a second trial with the NICU, for the piloting of a novel EEG headband for infants.
- PCTO ushered in the first trial in the PICU with a second multi-centered industry clinical trial in the PICU, working closely with the PICU Medical Director to create the infrastructure as well as mindset to support clinical trials in this particularly promising and important division.

Clinical Trials Open to Enrollment (21) (up from 11 in 2021)

Pediatric Divisions	Collaborations with Adult Divisions:
Cardiology – 1 (NIH sub-award)	Pediatric Allergy and Adult Dermatology - 1
Gastroenterology – 5 (up from 3)	Allergy and Immunology – 2 (NIH U01)
Endocrinology – 5 (up from 1)	
Nephrology – 1	
NICU – 1	
Rheumatology – 1	
Allergy – 5 (up from 0)	

Clinical Trials in Start-up Phase (12) (previously 13)

Pediatrics
Allergy – 3 (previously 6)
Gastroenterology – 3 (previously 6)
Endocrinology – 1 (previously 7)
Cardiology – 1
Rheumatology – 1
NICU – 1
PICU – 2

The PCTO now has a dedicated suite of offices that will be operational in early 2023 (Icahn Building, on the 3rd floor).

CENTER FOR INBORN ERRORS OF IMMUNITY

The Center for Inborn Errors of Immunity (CIEI) is a hub for collaborative research and patient care that is dedicated to improving the lives of those struggling with genetic disorders of the immune system. We strive to lay the foundation for a paradigm-shifting approach to the design of both preventative therapies (such as prophylactic drugs, vaccines, and genetic counseling) and novel treatments.

Our research is being done in the following areas:

Genetics of Immune Disorders

Primary immunodeficiencies (PIDs) are a varied group of inborn genetic errors that result in susceptibility to infections, predisposition to malignancy, or disorders of immune overactivation. Currently more than 420 genetic defects have been described. The CIEI seeks to diagnose individuals with known disease and discover new genetic etiologies of disease by using the latest sequencing and analysis technologies. Many of our member labs are working to identify and study these genetic defects. Additionally, CIEI is a part of The Mindich Child Health and Development Institute's Undiagnosed Disease Program, which is investigating the genetic underpinning of many childhood diseases, including those pertaining to the immune system. Finally, Mount Sinai's BioMe Biobank collaboration with the Regeneron Genetics Center will allow us and others to identify those individuals who might have silent or late onset conditions.

Pilot Grants

CIEI has awarded two pilot grants in 2022 to seed the discovery and research.

Dr. David Dominguez-Sola, Associate Professor of the Department of Oncological Sciences, Department of Pathology, Tisch Cancer Institute & Precision Immunology Institute, and a member of CIEI, received \$20,000 to study "Hypomorphic INO80 germline mutant variants linking immunoglobulin Class Switch Recombination defects and cancer risk."

Dr. Ki A. Goosens, Associate Professor of the Department of Psychiatry, and a member of CIEI, received \$20,000 to study "Elevated Follicle-Stimulating Hormone as a Trigger for Immune Dysregulation in Down Syndrome".

Symposia

CIEI has co-organized the 5th and 6th NYC Inborn Errors of Immunity meetings, where over 100 participants from the tri-state area listened to lectures from local and national leaders in the field, as well as presentations from MD, PhD, MD/PhD students, residents, and fellows.

Detailed Pathophysiology

Understanding the underlying mechanisms of a disease is key to developing successful treatment. To achieve this understanding, each genetic variant must be studied in isolation in order to glean key mechanistic insights. Our CIEI labs use genomics, genetics, molecular biology, cellular biology, immunology, and clinical tools to dissect these phenotypes and develop therapeutics.

Novel Therapeutics, Technologies, and Clinical Trials

The Inborn Errors of Immunity program uses the latest technologies to investigate pathophysiology, but also to unveil existing FDA-approved therapies, such as JAK inhibitor therapy. We are also facilitating the development of novel therapies, such as transient gene therapy and modRNA tools, to successfully modify immune state and cure disease.

Immune Monitoring

A key component to understanding inborn errors of immunity is the detailed mapping and functional assessment of the immune system. The Inborn Errors of Immunity program is thus working closely with the Human Immune Monitoring Center, which leverages cutting-edge technologies and deep immunological and technical expertise to provide comprehensive immune monitoring for clinical and translational studies.

PEDIATRIC PRECISION MEDICINE

Precision medicine (PM) uses individualized patient data to accurately diagnose disease, better predict the outcomes of medical issues, and treat illnesses more effectively. Currently, medical problems with strong genetic underpinnings such as congenital anomalies, neurodevelopmental disorders, and inborn errors of immunity are ones that typically manifest during infancy, childhood, and/or adolescence, and where a PM approach can be most impactful. 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.

The MCHDI is focusing on pediatric PM as one of its main strategic initiatives. The Undiagnosed Diseases Program (UDP) was established in 2017 and throughout the years has contributed to important advances in science and medicine with the discovery of novel disease genes. Additionally, the UDP has also improved patient care by identifying a unifying genetic cause for the constellation of medical problems that patients present—which gives patients and their families a much needed sense of closure and opens a new chapter where they can navigate medical care with a personalized compass.

This cutting-edge program, now led by Mafalda Barbosa, MD, PhD, is so successful because it benefits from a multidisciplinary team that includes both clinicians (including pediatricians, subspecialists, and clinical geneticists) and researchers (including PhD investigators, bioinformaticians, and laboratory geneticists). We continue to enroll infants, children, and adolescents with unsolved diseases that seem likely to have a genetic underpinning and then use new generation DNA sequencing technologies to identify the causal genetic variation. In order to improve our diagnostic yield and boost discovery, future directions of our program include moving towards third-generation sequencing and strengthen our relationship with the Functional Genomics and Disease Modeling Core.

The Functional Genomics and Disease Modeling core was established to leverage the strengths of the *Drosophila* genetic system and to develop whole animal fly models of rare gene variants in pediatric and other rare disease indications. The core uses multiple established transgenic approaches as well as newly developed assays to provide insights about how these gene variants function in vivo. The objective is that they will serve as important leads for ongoing and future studies in vertebrate models, to be ultimately translated to the clinics.

Since its inception a couple of years ago, the core has developed 20 new RASopathy fly models for various genes in the MAPK pathway encoding variants for MEK, RIT, SOS, SHOC. In addition, it has developed 5 models of rare kinase-fusion gene variants that arise in patients undergoing targeted lung cancer therapy.

Ongoing studies, using the newly developed models, have provided key new insights about the function of the RASopathy and rare cancer variants, and they were presented as a couple of posters in the Annual *Drosophila* Research Conference, San Diego, 2022.

Currently an important focus of the core is analyzing variants identified in the UDP and rarely associated in other diseases. As such, the core has developed fly models of *NDUFAF* and *MAGI2*, which represents the first time these models have been developed and studied in flies.

A new ongoing PM study focuses on validation of cord blood as a suitable specimen for genomic analysis. Doing genomic research in babies can be challenging. A common barrier to participation is the difficulty with obtaining a sample — it is not easy to draw blood from a newborn and the procedure itself causes discomfort to the baby. However, cord blood collection is noninvasive and is routinely collected for medical care. In order for us to use cord blood in future genomic studies, we have to prove that this is an appropriate specimen. There are concerns that cord blood may not be a good source of DNA for genetic studies because of possible presence of cells from the mother in the cord blood. As such, the purpose of this study is to assess if cord blood is an appropriate sample for genetic analysis. Validating cord blood as a suitable specimen for genomic studies would allow for streamlined participation of babies in large-scale genomic studies in the future.

Another important initiative of the Pediatric PM pertains to the recruitment of a pediatric cohort in the realm of the Mount Sinai Million Health Discoveries Program. We are collaborating with the Charles Bronfman Institute for Personalized Medicine in the effort of creating one of the largest repositories of sequencing data that will integrate health and research data at Mount Sinai to promote discoveries that will directly benefit our patient population. The overarching goal is to biobank genetic information from 1 million individuals that are representative of the diversity of our global population. The efforts of Dr. Gelb and Dr. Barbosa are focused on the enrollment of 100k individuals in the pediatric age range. This will constitute one of the largest and most diverse pediatric biobanks ever established and will be leveraged to allow for better understanding of the impact of genetic variations on human health and disease across the lifespan with the end goal of development of novel treatments and disease prevention.



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