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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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 development 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.
The Mindich Child Health and Development Institute

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)

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.

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
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.
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 regeneration 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.
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-omim 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
<table>
<thead>
<tr>
<th>Name</th>
<th>Position</th>
<th>Research Areas</th>
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</thead>
<tbody>
<tr>
<td>Paige M. Siper, PhD</td>
<td>(Adjunct Assistant Professor, Psychiatry)</td>
<td>Autism, intellectual disability, biomarker discovery, sensory processing</td>
</tr>
<tr>
<td>Luca Lambertini, PhD</td>
<td>(Assistant Professor, Obstetrics, Gynecology and Reproductive Science)</td>
<td>Placental biomarkers of altered fetal and child development</td>
</tr>
<tr>
<td>Behrang Mahjani, PhD</td>
<td>(Assistant Professor, Psychiatry, Genetics and Genomic Sciences, and Artificial Intelligence and Human Health)</td>
<td>Genetics of neurodevelopment disorders</td>
</tr>
<tr>
<td>Florence Marlow, PhD</td>
<td>(Associate Professor, Cell, Developmental &amp; Regenerative Biology)</td>
<td>Genetics of early patterning and germline, neurodevelopment</td>
</tr>
<tr>
<td>Marek Mlodzik, PhD</td>
<td>(Professor and Chair, Cell, Developmental &amp; Regenerative Biology; Professor, Ophthalmology, and Oncological Sciences)</td>
<td>Genetics and cell biology of planar cell polarity establishment, cell biology of Wnt-signaling and Notch-signaling</td>
</tr>
<tr>
<td>Hirofumi Morishita, MD, PhD</td>
<td>(Professor, Psychiatry, Ophthalmology, and Neuroscience)</td>
<td>Mechanisms of perceptual, cognitive, and social development relevant to neurodevelopmental disorders</td>
</tr>
<tr>
<td>Dalila Pinto, PhD</td>
<td>(Assistant Professor, Psychiatry, and Genetics and Genomic Sciences)</td>
<td>Genetics and genomics of neurodevelopmental disorders (particular focus on autism, epilepsy, schizophrenia, OCD), structural variation, transcriptomics, gene regulation, noncoding RNA</td>
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### NEURODEVELOPMENTAL DISORDERS - CONTINUED

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<tr>
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<tbody>
<tr>
<td>Avi Reichenberg, PhD</td>
<td>Professor, Psychiatry, and Environmental Medicine &amp; Public Health</td>
<td>Autism, schizophrenia, other psychiatric disorders</td>
</tr>
<tr>
<td>Andrew J. Sharp, PhD</td>
<td>Professor, Genetics and Genomic Sciences</td>
<td>Epigenomics, transcriptomics, genome function, structural variation, imprinting, congenital disorders</td>
</tr>
<tr>
<td>Annemarie Stroustrup, MD, MPH</td>
<td>Adjunct Associate Professor, Pediatrics and Environmental Medicine &amp; Public Health</td>
<td>Neurodevelopment, perinatal environmental exposures, identifying genetic etiologies of congenital disease</td>
</tr>
<tr>
<td>Shanna H. Swan, PhD</td>
<td>Professor, Environmental Medicine &amp; Public Health</td>
<td>Prenatal exposures, sexually dimorphic development, phthalates, stress, anogenital distance, neurodevelopment, analgesics, glyphosate, acetaminophen</td>
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<tr>
<td>Pilar Trelles, MD</td>
<td>Adjunct Assistant Professor, Psychiatry</td>
<td>Autism spectrum disorder, neurodevelopmental disorders, health disparities</td>
</tr>
<tr>
<td>Bryn D. Webb, MD</td>
<td>Adjunct Assistant Professor, Genetics and Genomic Sciences</td>
<td>Identifying genetic etiologies of congenital anomalies, mitochondrial disorders, undiagnosed disease</td>
</tr>
<tr>
<td>Anusha Yeshokumar, MD</td>
<td>Adjunct Assistant Clinical Professor, Pediatrics, and Neurology</td>
<td>Autoimmune encephalitis, outcomes research, inflammatory biomarkers, cognition, behavior</td>
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## OBESITY AND DIABETES

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<thead>
<tr>
<th>Name</th>
<th>Title and Affiliation</th>
<th>Research Areas</th>
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</thead>
<tbody>
<tr>
<td>Sharon Baumel-Alterzon, PhD</td>
<td>(Instructor, Medicine)</td>
<td>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</td>
</tr>
<tr>
<td>Ross L. Cagan, PhD</td>
<td>(Adjunct Professor, Cell, Developmental &amp; Regenerative Biology)</td>
<td>Drosophila as a tool to develop therapeutics for cancer, diabetes, and rare mendelian diseases</td>
</tr>
<tr>
<td>Nathalie Chami, PhD</td>
<td>(Instructor, Environmental Medicine &amp; Public Health)</td>
<td>Genetics of complex traits, monogenic disease, genetics of obesity and cardiometabolic outcomes</td>
</tr>
<tr>
<td>Lauryn Choleva, MD</td>
<td>(Instructor, Pediatrics)</td>
<td>Type 2 diabetes, type 1 diabetes, hypoglycemia</td>
</tr>
<tr>
<td>Fernando Ferrer, MD, FACS, FAAP</td>
<td>(Professor, Urology)</td>
<td>Cancer, bioactive lipids, renal injury, biomarkers, renal obstruction</td>
</tr>
<tr>
<td>Adolfo García-Ocaña, PhD</td>
<td>(Professor, Medicine)</td>
<td>Diabetes, pancreatic beta cell biology</td>
</tr>
<tr>
<td>Allan C. Just, PhD</td>
<td>(Assistant Professor, Environmental Medicine &amp; Public Health)</td>
<td>Epigenomics, environmental exposures, endocrine disruptors, air pollution, obesity, birth outcomes</td>
</tr>
<tr>
<td>Joan Han, MD</td>
<td>(Chief, Division of Pediatric Endocrinology and Diabetes; Professor, Pediatrics)</td>
<td>Pediatric obesity, neuroendocrine regulation of energy balance, genetic obesity syndromes and disorders of the leptin pathway</td>
</tr>
</tbody>
</table>
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, neurolisteriosis

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

<table>
<thead>
<tr>
<th>Name</th>
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<tbody>
<tr>
<td>Minji Byun, PhD</td>
<td>(Adjunct Assistant Professor, Medicine)</td>
<td>Genetics of immune disorders, clonal hematopoiesis, immune dysregulation</td>
</tr>
<tr>
<td>Jaime Chu, MD</td>
<td>(Assistant Professor, Pediatrics)</td>
<td>Disorders of glycosylation, cancer metabolism, liver fibrosis, environmental toxicants in liver disease</td>
</tr>
<tr>
<td>Charlotte Cunningham-Rundles, MD, PhD</td>
<td>(David S. Gottesman Professor, Medicine; Professor, Pediatrics)</td>
<td>Primary Immune Deficiency, B cells, antibody, B cell memory, hypogammaglobulinemia, immune reconstitution</td>
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<tr>
<td>Sarah Duncan-Park, PhD</td>
<td>(Assistant Professor, Pediatrics)</td>
<td>Behavioral health intervention development, psychosocial adjustment to pediatric chronic illness</td>
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<td>David Dunkin, MD</td>
<td>(Associate Professor, Pediatrics)</td>
<td>Tolerance induction and therapeutics in inflammatory bowel disease, mechanisms of inflammatory diseases of the gastrointestinal tract</td>
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<tr>
<td>Chris Gennings, PhD</td>
<td>(Professor, Environmental Medicine &amp; Public Health, and Population Health Science and Policy)</td>
<td>Biostatistical methods development for environmental health</td>
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<tr>
<td>Katherine Guttmann, MD, MBE</td>
<td>(Assistant Professor, Pediatrics)</td>
<td>Palliative care, family-centered care, parent-physician communication, research ethics</td>
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<tr>
<td>Shelley H. Liu, PhD</td>
<td>(Assistant Professor, Population Health Science and Policy)</td>
<td>Biostatistics, environmental mixtures, public health</td>
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OTHER RESEARCH FOCUSSES - 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

Prameen 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
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
- 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
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


Lee-Sarwar KA, Chen YC, Lasky-Su J, Kelly RS, Zeiger RS, O’Connor GT, ... Bunyavanich S, ... Brennan PJ. Early-life fecal metabolomics of food allergy. Allergy. 2022 Nov 30.


Trubetskoy V, Pardinas AF, Qi T, Panagiotaropoulos G, Awasthi S, Bigdeli TB, ... Reichenberg A, ... Buxbaum JD, ... O’Donovan MC. Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature. 2022 Apr;604(7906):502-8.


Wheeler MM, Stilp AM, Rao S, Halldórsson BV, Beyder D, Wen J, ... Chami N, ... Reiner AP. Whole genome sequencing identifies structural variants contributing to hematologic traits in the nhlbi topmed program. Nat.


Pham MN, Fuleihan RL, Sullivan KE, Cunningham-Rundles C. Ocular manifestations in primary immunodeficiency disorders: A report from the united states immunodeficiency network (usidnet) registry. J Allergy Clin

Aranda CJ, Gonzalez-Kozlova E, Saunders SP, Fernandes-Braga W, Ota M, Narayanan S, ... Curotto de Lafaille MA. IgG memory B cells expressing IL4R and FcER2 are associated with atopic diseases. Allergy. 2022 Nov 29.


Pavinato L, Delle Vedove A, Carli D, Ferrero M, Carestia S, Howe JL, ... De Rubeis S, Buxbaum JD, ... Brusco A. Caprin1 haploinsufficiency causes a neurodevelopmental disorder with language impairment, ADHD and ASD. Brain. 2022 Jul 27.

Campisi L, Chizari S, Ho JSY, Gromova A, Arnold FJ, Mosca L, ... De Rubeis S, ... Byun M, ... Marazzi I. Clonally expanded CD8 T cells characterize amyotrophic lateral sclerosis-4. Nature. 2022 Jun;606(7916):945-52.

Gonzalez DM, Schrode N, Ebrahim TAM, Broguiere N, Rossi G, Drakhlis L, ... Dubois NC. Dissecting mechanisms of chamber-specific cardiac differentiation and its perturbation following retinoic acid exposure. Development. 2022 Jul 1;149(13).


Willcox JAL, Geiger JT, Morton SU, McKean D, Quiat D, Gorham JM, ... Gelb B, ... Seidman CE. Neither cardiac mitochondrial DNA variation or copy number contribute to congenital heart disease risk. Am J Hum Genet. 2022 Apr 7.


Caporale N, Leemans M, Birgersson L, Germain PL, Cheroni C, Borbély G, ... Gennings C, ... Testa G. From cohorts to...


Huang J, Huffman JE, Huang Y, Do Valle I, Assimes TL, Raghavan S, Loos RJ, O'Donnell CJ. Genomics and phe~


Yengo L, Vedantam S, Marouli E, Sidorenko J, Bartell E, Sakaue S, Loos RJ, Hirschhorn JN. A saturated map of


GRANTS

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MATERIAL TRANSFER AGREEMENTS/LICENSES

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Total 10,404,770  56,302,639
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)
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)
Project Title: Whole-Genome Sequencing of Neonatal Fatalities

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

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.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:
http://icahn.mssm.edu/research/resources/shared-resource-facilities/histology
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

### PCTO Total Studies

![Pie chart showing study phases breakdown]

- **36** total studies
- *Does not include studies on hold

#### Study Phases Breakdown

- A. CDA
- B. F.O. Needed
- C. Site Selection Pending
- D. Documents Needed
- E. Budget Negotiation
- F. IRB Submission Prep
- G. Post Approval Pre Enrolment Prep
- H. Active - Enrolling
- I. Active - Closed to Enrollment
## RESEARCH FACULTY SERVED BY PCTO

PCTO is currently serving the following investigators/divisions:

### Pediatric Divisions:

<table>
<thead>
<tr>
<th>Division</th>
<th>Investigator 1</th>
<th>Investigator 2</th>
<th>Investigator 3</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Allergy</strong></td>
<td>Scott Sicherer, MD</td>
<td>Julie Wang, MD</td>
<td>Amanda Cox, MD</td>
</tr>
<tr>
<td></td>
<td>Roxanne Oriel, MD</td>
<td>Mary Grace Baker, MD</td>
<td>Nicole Ramsey, MD</td>
</tr>
<tr>
<td><strong>Endocrinology</strong></td>
<td>Joan Han, MD</td>
<td>Robert Rapaport, MD</td>
<td>Hillary Hotchkiss, MD</td>
</tr>
<tr>
<td><strong>Gastroenterology</strong></td>
<td>Marla Dubinsky, MD</td>
<td>David Dunkin, MD</td>
<td>Keith Benkov, MD</td>
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<tr>
<td></td>
<td>Nancy Pittman, MD</td>
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<tr>
<td><strong>Nephrology</strong></td>
<td>Jeffrey Saland, MD</td>
<td>Hillary Hotchkiss, MD</td>
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<tr>
<td><strong>Neonatal ICU</strong></td>
<td>Courtney Juliano, MD</td>
<td></td>
<td></td>
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<tr>
<td><strong>Rheumatology</strong></td>
<td>Rebecca Trachtman, MD</td>
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<td></td>
</tr>
<tr>
<td><strong>Cardiology</strong></td>
<td>Erin Paul, MD</td>
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</tr>
</tbody>
</table>

### NEW in 2022

<table>
<thead>
<tr>
<th>Division</th>
<th>Investigator 1</th>
<th>Investigator 2</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Pediatric ICU</strong></td>
<td>Sheemon Zackai, MD</td>
<td>Sandeep Gangadharan, MD</td>
</tr>
<tr>
<td></td>
<td>Shubhi Kaushik, MD</td>
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</tbody>
</table>

### Adult Divisions With Pediatric Trials:

<table>
<thead>
<tr>
<th>Division</th>
<th>Investigator</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Allergy and Immunology</strong></td>
<td>Paula Busse, MD</td>
</tr>
<tr>
<td><strong>Dermatology</strong></td>
<td>Emma Guttman, MD, PhD</td>
</tr>
</tbody>
</table>
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)

<table>
<thead>
<tr>
<th>Pediatric Divisions</th>
<th>Collaborations with Adult Divisions:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiology – 1 (NIH sub-award)</td>
<td>Pediatric Allergy and Adult Dermatology - 1</td>
</tr>
<tr>
<td>Gastroenterology – 5 (up from 3)</td>
<td>Allergy and Immunology – 2 (NIH U01)</td>
</tr>
<tr>
<td>Endocrinology – 5 (up from 1)</td>
<td></td>
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<tr>
<td>Nephrology – 1</td>
<td></td>
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<tr>
<td>NICU – 1</td>
<td></td>
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<tr>
<td>Rheumatology – 1</td>
<td></td>
</tr>
<tr>
<td>Allergy – 5 (up from 0)</td>
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</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Pediatrics</th>
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<tbody>
<tr>
<td>Allergy – 3 (previously 6)</td>
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<tr>
<td>Gastroenterology – 3 (previously 6)</td>
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<tr>
<td>Endocrinology – 1 (previously 7)</td>
</tr>
<tr>
<td>Cardiology – 1</td>
</tr>
<tr>
<td>Rheumatology – 1</td>
</tr>
<tr>
<td>NICU – 1</td>
</tr>
<tr>
<td>PICU – 2</td>
</tr>
</tbody>
</table>

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 genenic 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.
LEADERSHIP AND STAFF

Bruce D. Gelb, MD—Director
Elena Lum, PhD—Administrative Director
Shavez Jackson—Administrative Manager

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For more information on the MCHDI, please visit our website at www.mssm.edu/mchdi.