Icahn School of Medicine at Mount Sinai

*The Mindich Child Health and Development Institute*

# THE ANNUAL REPORT 2023

**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.**

# TABLE OF CONTENTS

# Director's Message

Bruce D. Gelb, MD, Director

In 2023, the MCHDI enjoyed an outstanding year. With the COVID-19 pandemic definitely in the rearview mirror, our in-person activities were very much back to normal. As one example, we felt comfortable inviting a family with a child whose care had been impacted through research to our annual retreat in November, something not possible the year before. That said, it sure feels like some things have changed. We are all video-conferencing savvy (okay, I admit to still forgetting to come off mute when starting to speak sometimes), which has improved our communications for the most part. On the other hand, the return to pre-pandemic levels of in-person interactiveness, particularly among trainees, remains a work in progress. Let's all commit to find more ways to support one another in 2024.

Speaking of connectivity: I noted during my remarks at the 2023 MCHDI annual retreat that 2024—and probably a couple of years beyond—was shaping up to be quite challenging with respect to NIH grant funding. I never imagined that we would still not have an approved fiscal year 2024 budget for NIH when writing these thoughts in Q1 of 2024. But here we are. The current handicapping is that we will be fortunate if NIH's budget is flat relative to last year, but a cut of 0.5 percent or 1 percent is more likely. Given that, we can anticipate that the paylines at the various NIH institutes will be dropping and award sizes will be reduced. While those prospects are unquestionably concerning, we are fortunate to have constructive steps that can mitigate them. Those steps largely depend on our connectivity: our collaborativeness within the MCHDI, and at Mount Sinai more broadly, will cushion us.

I strongly encourage all MCHDI investigators to leverage our Incubator Series and our grant specific aims reviewer program. I have used both programs for my NIH grant applications, consistently receiving useful critical feedback. I am positive that my R01 application from last June would not have achieved a fundable score without that input. Other MCHDI faculty have similar stories. I also hope that all of us will give generously of our time to one another: attending Incubator Series sessions and reviewing others' specific aims take up time, which is a precious commodity. Still, we can derive joy and pride from helping one another (and, yes, we often learn something about grant strategies).

Based on input from the MCHDI Internal Advisory Board, we will be rolling out a new program, hoping to provide additional support for grant applications. We are finishing the fine details, but, in essence, we will be funding MCHDI investigators to work with professional grant writers in order to polish their research strategies. This will require that those documents are developed sufficiently in advance of the relevant submission deadline in order to permit the writers to work their magic. Having never tried this approach, the MCHDI will be monitoring the application success rate to determine if the return on investment with respect to success rates relative to cost makes sense. We have good reason to believe it will.

I look forward to working closely with all of you this year. We should all be proud of the interactive culture that we have built together over the years. Now is the time for us to all dig a bit deeper to further enable our amazing child health-related science to garner the level of extramural support needed to advance our collective mission.

# FACULTY GROWTH

## MCHDI ANNUAL FACULTY GROWTH 2023

### In 2023

**We welcomed three new external faculty and 10 internal faculty members to our institute.**

### 97 Members

**In 2023, we welcomed three new external faculty and 10 internal faculty members to our institute. Currently, we total 97 members consisting of scientists and physician-scientists across the disciplines of Allergy & Asthma, Cardiovascular Disease, Neurodevelopmental Disorders, Obesity & Diabetes, and more.**

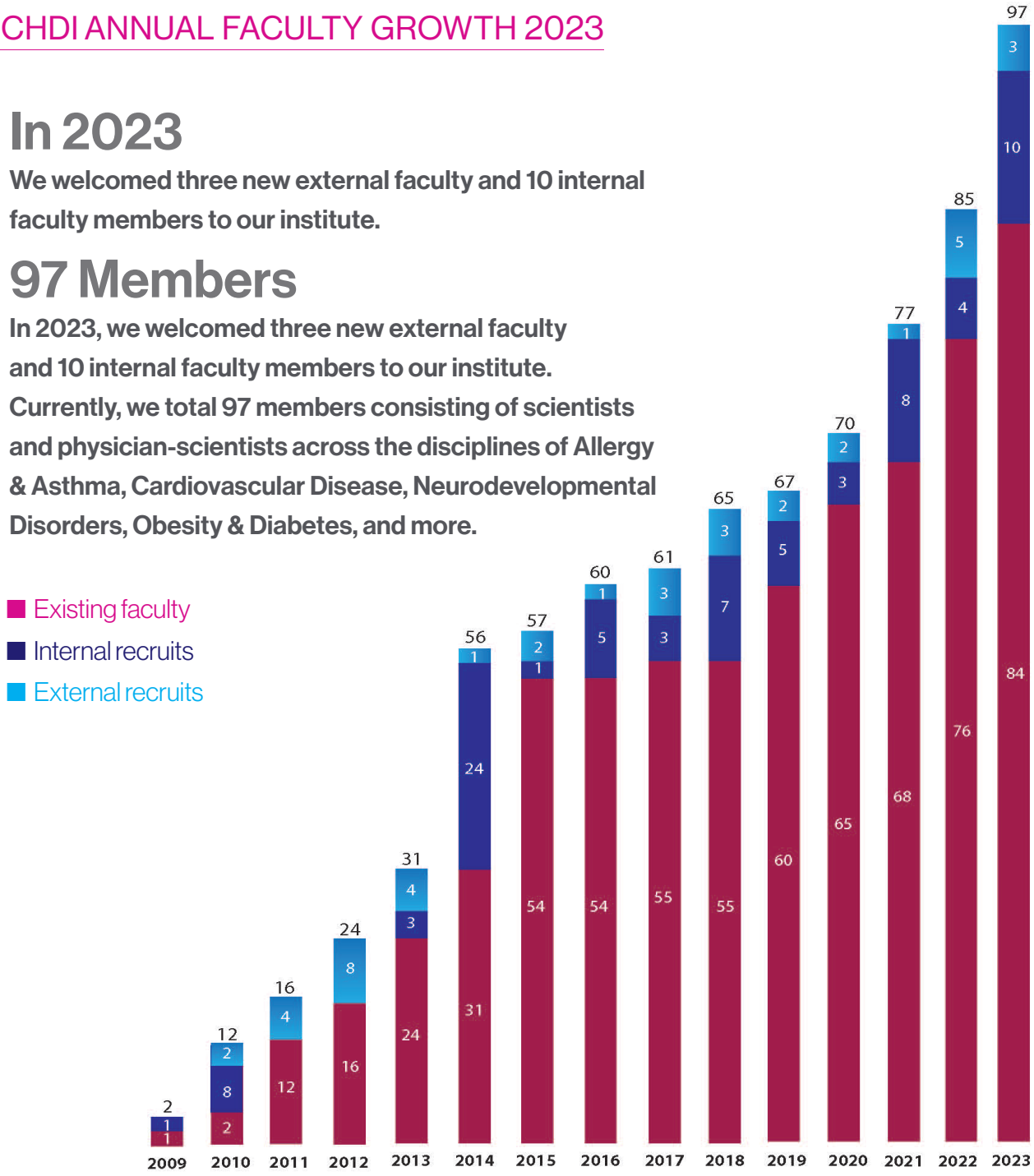



*Chart of faculty recruits since our inception in 2009. In 2023, our institute recruited three new external and 10 new internal faculty members to our institute.*

# NEW FACULTY

## NEW EXTRAMURAL FACULTY

### Brett R. Anderson, MD, MBA, MS

Brett R. Anderson, MD, MBA, MS, joins Mount Sinai as the inaugural Director for the Center for Child Health Services Research in the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai. Dr. Anderson is a pediatric cardiologist, Associate Professor in the Departments of Pediatrics, and Population Health and Policy, and an NIH-funded health services and health equity researcher.

Dr. Anderson completed her general pediatrics residency at The Children's Hospital of Philadelphia and fellowship in pediatric cardiology at NewYork-Presbyterian/Morgan Stanley Children's Hospital. She joins us after a decade at Columbia University, where she served as Director of Clinical Research, Outcomes, and Quality for the Pediatric Heart Center and Co-Director of ASPIRE! Peer Mentoring. Her significant contributions and leadership in the field are exemplified by her commitment to advancing pediatric cardiology and health services research.

Dr. Anderson brings a wealth of knowledge, employing an interdisciplinary approach that combines her medical, business, and statistical expertise. Her focus is on identifying modifiable drivers of outcomes, value, and health inequities, with a particular emphasis on children with cardiac disease. She is the founder and Director of the New York State Congenital Heart Surgery Collaborative for Longitudinal Outcomes and Utilization of Resources (CHS-COLOUR), leading an interdisciplinary collaborative that examines the etiologies of health inequities and plans for programmatic interventions. Additionally, Dr. Anderson serves as M-PI for the Pediatric Heart Network New York Consortium (PHN-NYC), a vital part of the NHLBI-funded pediatric cardiac clinical trials network.

### Romina Bevacqua, PhD

Romina Bevacqua, PhD, has been a new Assistant Professor in the Department of Medicine, Division of Endocrinology, Diabetes, and Bone Disease, and member of the Diabetes, Obesity and Metabolism Institute (DOMI) and the Regenerative Biology and Stem Cell Institute, at Mount Sinai, since January 2023 where she was selected as a Biomedical Laureate.

Previously, Dr. Bevacqua was an Instructor (2020-2021) and a postdoctoral fellow (2017-2020) in the laboratory of Professor Seung Kim, Department of Developmental Biology, at the Stanford University School of Medicine. Dr. Bevacqua's graduate studies where completed in Buenos Aires University, Argentina.

The Bevacqua Lab focuses on understanding the regulatory mechanisms governing pancreatic islet cell function and maturation, and integrates modern genetic, cell, and developmental biology, biochemical, and physiological approaches. In particular, Dr. Bevacqua's development of genetic systems, including CRISPR/Cas9, in primary human islet organoids, termed "pseudoislets," provides unprecedented methods to investigate mechanisms regulating function of mature human islets. Using these novel systems, the lab is interested in understanding how transcriptional regulators, non-coding regulatory elements, and epigenetic and external signals orchestrate crucial steps in human islet functional maturation and proliferation, and how these steps are mis-regulated in human diseases, particularly type 1 (T1D) and type 2 diabetes (T2D). The goal of the Lab is to advance and deepen the fundamental understanding of islet regulation and function, and their connection to complex traits associated with diseases, particularly T1D and T2D, knowledge that should inform islet replacement and regeneration therapies.
For more info, please refer to the Bevacqua Lab at https://labs.icahn.mssm.edu/bevacqualab/.

## NEW EXTRAMURAL FACULTY - *CONTINUED*

### Georgia Panagiotakos, PhD

Georgia Panagiotakos, PhD, is an Associate Professor of Psychiatry, and Neuroscience at the Icahn School of Medicine at Mount Sinai. She is also a member of the Seaver Autism Center for Research and Treatment, the Alper Center for Neural Development and Regeneration, the Institute for Regenerative Medicine, and the Friedman Brain Institute.

Prior to her arrival at Mount Sinai, Dr. Panagiotakos launched her independent research program as a Sandler Faculty Fellow at the University of California, San Francisco, after earning her PhD from the Stanford University School of Medicine Neurosciences Doctoral Program. The central focus of the Panagiotakos laboratory is to dissect the cellular and molecular mechanisms underlying mammalian brain development, with an eye towards uncovering the underpinnings of neuropsychiatric disorders of developmental origin. We are especially interested in understanding how electrical activity, calcium signaling, and ion channel diversity regulate developmental transitions and the generation of distinct cell types in the developing brain. To investigate this, we incorporate multiple levels of analysis and a variety of orthogonal in vivo and in vitro approaches, including genetic tools in mouse models, live imaging and sequencing technologies. In the longer term, we also aim to shed light on how activity-regulated developmental mechanisms may be reactivated in the context of adult neurological disorders and the development of brain tumors.

## NEW INTRAMURAL FACULTY

### Sandeep Gangadharan, MD

Sandeep Gangadharan, MD, is an Associate Professor of Pediatrics. His primary academic interest is in performance improvement of acute clinical care systems, whether through in situ simulation, clinical informatics, or process and implementation science.

As a member of the Improving Pediatric Acute Care Through Simulation (ImPACTS) study group, Dr. Gangadharan has been centrally involved in demonstrating both the utility of medical simulation as a diagnostic tool for evaluating complex clinical processes, such as acute resuscitation, and its ability to determine important clinical metrics that assess the quality of care. In addition to Dr. Gangadharan's interest in medical simulation, he has initiated, designed, and completed several clinical and quality research projects relevant to his field. The broader theme of many of these projects is evaluating and potentially enhancing the quality of procedural and resuscitation care in pediatric intensive care medicine utilizing multi-institutional databases. Dr. Gangadharan continues to be interested in similar work that seeks to evaluate the process of care, whether by simulation, data analytics, EMR optimization, or clinical informatics, to find areas of opportunity to make care more safe, efficient, and effective for children. Finally, Dr. Gangadharan continues to be an active member of the Pedi-Res-Q, Near-4-Kids, and Get with Guidelines collaborative efforts to study and improve resuscitative care in children. Currently, Dr. Gangadharan's focus is on clinical informatics and machine learning to develop effective clinical decision support tools and improve the process of acute care and cardiopulmonary resuscitation.

## Yolanda Garcia-Carmona, PhD

Yolanda Garcia-Carmona, PhD, is an Instructor at the Department of Medicine in the Division of Clinical Immunology at the Icahn School of Medicine at Mount Sinai. Dr. Garcia-Carmona received her PhD in 2011 in cellular and molecular biology at Autonomous University of Madrid (Spain), where she studied the crosstalk between B cells and synovial fibroblasts in the perpetuation of the inflammation in rheumatoid arthritis patients.

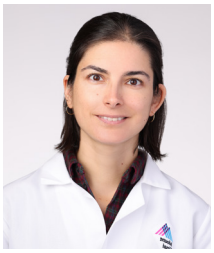
In 2011, she joined Dr. Charlotte Cunningham-Rundles' laboratory and the Immunology Institute at the Icahn School of Medicine at Mount Sinai, where she completed her postdoctoral training and was promoted to Instructor in 2017. One of her main interests is to study human B cell defects to better understand B cell biology. Working to decipher some of the genes involved in CVID, 36 percent of the United States monogenic cohort had mutations in transmembrane activator and CAML interactor (TACI). Unlike the murine TACI gene, the human TACI gene undergoes alternative splicing, to produce two isoforms (TACI-L and TACI-S), both of which are found in human B cells. When comparing functions only the TACI-S isoform, together with endogenous APRIL, sponsors the plasma cell genetic program.

Dr. Garcia-Carmona's research also involves a more bench-to-bedside approach, focusing on identifying new therapeutic targets and developing more efficient strategies to neutralize B cells in autoimmunity and B cell malignancies.

## Dirk Hubmacher, PhD

Dirk Hubmacher, PhD, is an Assistant Professor in the Department of Orthopedics, where his team investigates the role of extracellular matrix proteases, ADAMTS-like proteins, and fibrillins in the context of developmental short stature syndromes. Dr. Hubmacher received his PhD from the University of Lübeck (Germany) in 2004 where he studied iron uptake in salt-loving archaea.

He entered the field of connective tissue disorders as a postdoctoral fellow with Dr. Dieter Reinhardt (McGill University, Montreal) where he studied molecular pathomechanisms underlying Marfan syndrome and homocystinuria. In 2011, Dr. Hubmacher joined the laboratory of Dr. Suneel Apte at the Cleveland Clinic to study the function of ADAMTS proteases and ADAMTS-like proteins in mouse models of rare developmental short stature syndromes. In 2018, he moved to the Icahn School of Medicine, where his team continues to investigate pathomechanisms of these syndromes with a focus on geleophysic dysplasia, Weill-Marchesani syndrome, and Marfan syndrome. Dr. Hubmacher has received funding from the NIH/NIAMS, the Marfan Foundation, the Ines Mandl Research Foundation, and the German Academic Exchange Service. His work was recognized by the Harold and Golden Lamport Clinical Research Award (2021), the Mount Sinai Faculty Idea Prize (2019), and the Young Investigator Award from the Marfan Foundation (2005). Dr. Hubmacher served as an ad hoc reviewer on several NIH study sections and DoD review panels and served as an elected council member for the American Society for Matrix Biology (2018-2022).



## Esra Karakose, PhD

Esra Karakose, PhD, is an Assistant Professor in the Department of Medicine. The main focus of her research is to understand the genetic and epigenetic mechanisms that govern human pancreatic beta cell replication, with the aim of developing new therapies for diabetes. Since diabetes results from the lack of sufficient numbers of insulin-producing beta cells, one measure to reverse diabetes is to restore normal beta cell mass and function.

To this end, her group studies insulinoma, a benign pancreatic tumor that consists of beta cells. In their study, they found that epigenetics is important for the regulation of human beta cell replication. They also identified several novel pathways that induce proliferation in beta cells from normal human organ donors.

In a recent study, they demonstrated that TGF beta and DYRK1A signaling synergize to induce unprecedented rates of human beta cell proliferation. They uncovered that TGF beta signaling works in collaboration with Trithorax complex, which is an essential component of epigenetic regulation. Thus, their results indicated an unequivocal role for epigenetics in human beta cell replication. Further, in a more recent study, they showed that GLP1R agonists also synergize with DYRK1A signaling to induce robust levels of beta cell proliferation.

In addition to these approaches, they recently started using single cell approaches to better understand the mechanisms that control beta cell replication. In this study, the researchers analyzed the transcriptome of cadaveric human islets treated with beta cell regenerative drugs using single-cell RNA-seq. Their results revealed that the lineage dynamics in the human islets are more sophisticated than initially anticipated when islets are subject to regenerative drugs. Importantly, they showed that cycling alpha cells are the main target of regenerative drug treatment in human islets, and they have the potential to transdifferentiate into human beta cells.

## Liora S. Katz, PhD

Liora S. Katz, PhD, is an Associate Professor at the Diabetes, Obesity and Metabolism Institute, and of the Mindich Child Health and Development Institute. Dr. Katz is a beta cell biology expert. Her research spans the fields of endocrinology, thyroid dysfunction, and diabetes.

Among her notable research, she investigated the transcriptional pathways governing alpha and beta cell development, focusing on the role of Pax6. Furthermore, she has delved into cell replacement therapies for diabetes treatment, establishing a pioneering protocol for reprogramming human dermal fibroblasts into islet-like cells through a combination of epigenetic modifications and transcription factor modulation. Dr. Katz has also explored the dysregulation of proliferation in pancreatic cancer. In her recent work, Dr. Katz has placed a specific emphasis on glucose-induced adaptive expansion and the mechanisms governing beta cell failure in the context of metabolic overload. Notably, her work has shed light on the pivotal roles of ChREBP, Myc, and Nrf2 in regulating beta cell proliferation and function. Dr. Katz's research has extended to uncovering maladaptive feedback loops associated with glucotoxic beta cell failure, offering insights into genetic and pharmacological mechanisms aimed at preventing beta cell loss—an important unmet need in diabetes research. Overall, Dr. Katz's impactful research significantly advances our understanding of the molecular pathways involved in glucose-mediated adaptive responses and the various factors influencing both the health and dysfunction of beta cells.

## Corina Lesseur, MD, PhD

Corina Lesseur, MD, PhD, is an Assistant Professor in the Department of Environmental Medicine and Public Health. Dr. Lesseur received her MD at the Central University of Venezuela and a PhD in molecular and experimental medicine from Dartmouth College. She completed a postdoctoral fellowship in genetic epidemiology at the International Agency for Research in Cancer, followed by postdoctoral training in environmental health and molecular epidemiology at the Icahn School of Medicine at Mount Sinai.

Her work as a molecular epidemiologist focuses on placental epi/genomics and their link to pregnancy outcomes and early-life programming, as well as in the effects of environmental exposures (i.e., air pollution, pesticides) in the placenta and birth outcomes. She is particularly interested in maternal and infant metabolic outcomes (birth weight, obesity, and gestational diabetes). Dr. Lesseur has worked in multiple birth cohort studies evaluating placental epi/genetic features in relation to maternal and infant health, and environmental exposures. Dr. Lesseur has received funding from the NIH/NICHD, the March of Dimes and the Marie Curie COFUND.

## Alejandro Martin-Trujillo, PhD

Alejandro Martin-Trujillo, PhD, is an Assistant Professor in the Department of genetics and Genomic Sciences within the team of Dr. Andrew J. Sharp. After earning his MSc in genetics and development, he completed his PhD in biomedicine at the University of Barcelona (Spain) in 2014.

During his PhD, Dr. Martin-Trujillo characterized the extent of parent-of-origin DNA methylation in the human genome, identifying novel imprinted regions and, thus, helping to define the human imprintome. Subsequently, he investigated the deregulation of these loci in a wide range of human diseases, including the well-known imprinting disorders as well as several types of cancer. He then joined the laboratory of Dr. Sharp as a postdoctoral fellow, where his research expanded to explore epigenetic variation beyond imprinted loci as well as genetic variation at complex genomic regions that often eludes standard genetic studies. His current research primarily focuses on profiling both common and rare variation at tandem repeats (TR) from whole exome and genome sequencing data using sophisticated computational approaches on a large scale. These studies aim to identify TR variation implicated in the regulation of the genome function, phenotypic diversity, and human diseases ranging from congenital anomalies to late-onset neurodegenerative disorders.

## Nicole Ramsey, MD, PhD

Nicole Ramsey, MD, PhD, is an Assistant Professor of Pediatrics in the Division of Pediatric Allergy and Immunology 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.

Dr. Ramsey is a clinician and pediatrician-scientist who earned her BS magna cum laude from Howard University, an MD from Weill Cornell Medical College, and a PhD in biophysics and pharmacology from Weill Cornell Graduate School of Biomedical Sciences. She completed her pediatric residency in the Pediatrician-Scientist Training and Development Program at Baylor College of Medicine/ Texas Children's Hospital, and her allergy/immunology fellowship at Icahn School of Medicine at Mount Sinai. Through all of her work, Dr. Ramsey hopes to improve the availability of safe and effective medications for children.

Dr. Ramsey's research interests include food allergy treatment mechanisms (including basophil activation, allergen-specific T cell responses, proteomics, and transcriptomics), the development of new treatment modalities, exposomics, and the impact of prenatal environmental exposures on the risk of developing food allergy. She is helping to lead a clinical trial to determine the effect of an oral selective JAK inhibitor on food allergy in adult patients who have a history of eczema. Dr. Ramsey is also interested in health equity and is working on qualitative research to help improve clinical trial diversity in pediatric food allergy with focus groups and surveys.

Dr. Ramsey's areas of clinical expertise include food allergy, atopic dermatitis/eczema, allergic rhinitis, urticaria, anaphylaxis, and allergy testing.

## Anna Rommel, PhD

Anna Rommel, PhD, is an Assistant Professor in the Department of Psychiatry. Her work as a psychiatric epidemiologist focuses on environmental exposures and their link to the development of adverse health outcomes, including suboptimal pregnancy and birth outcomes, as well as adverse neurodevelopment and longer-term psychopathology.

Dr. Rommel is also interested in mental illness related to reproductive events, including pregnancy and menopause. She has been instrumental in setting up two separate birth cohorts and has conducted analyses in several existing birth cohorts to study the outcomes of early life exposure to, for example, maternal mental illness, medication, phthalates, and inflammation. Her lab applies epidemiological, genetically sensitive, and cognitive-neurophysiological (EGG) designs to study the relationship between parental and early-life factors with long-term (neuro)developmental outcomes, and the biological mechanisms underlying it. Dr. Rommel's overarching goal is the identification of modifiable risk and resilience factors, and the resulting improvement of prevention and treatment of adverse health outcomes.

## Caterina Tiozzo, MD, PhD

Caterina Tiozzo, MD, PhD, is an Associate Professor of Newborn Medicine at the Icahn School of Medicine at Mount Sinai. Dr. Tiozzo received her medical degree cum laude from the University of Padova where she also completed her first pediatric residency, neonatal fellowship, and Master of Public Health with humanitarian missions in Kenya. She then pursued her PhD through a collaboration between the University of Padova and the University of Southern California in Los Angeles.

After her humanitarian mission in Haiti during the 2010 earthquake, she decided to go back to clinical training to be able to practice in the United States so she completed her second pediatric residency and her neonatal fellowship at Columbia University. She trained and mentored many neonatal trainees over her career, both in Italy and in the United States.

She is a member of the "Society of Pediatric Research" and she is the recipient of several awards for her research discoveries from Europe and the United States.

She was selected by the Italian government for a documentary on Italians in New York during COVID and she received the title of Knight of the "Order of Croce d'Italia" by the Italian president for her work.

Dr. Tiozzo's research focuses on lung development, stem cell regeneration after lung injury, the role of the intrauterine environment in lung development, and the effect of neonatal nutrition on lung development. She is the author of more than 30 papers and is often invited to speak at national and international meetings on the topic of her research interests.

# ANNUAL RETREAT

The 11th annual retreat took place at the Harmonie Club on November 28, 2023, gathering nearly 130 participants, including faculty, trainees, staff, and volunteers. The retreat planning committee, spearheaded by Committee Chair Florence Marlow, PhD, and composed of our MCHDI Director Bruce D. Gelb, MD, and MCHDI faculty members Tirtha K. Das, PhD, Sharon Baumel-Alterzon, PhD, and Trainee Leadership Committee Chair Lauren Dierdoff, BS, worked diligently to orchestrate the event. Mustafa Khokha, MD, a distinguished Professor in the Departments of Pediatrics, Genetics, and Obstetrics, Gynecology, and Reproductive Sciences at Yale University School of Medicine, served as the keynote speaker and panelist." His keynote talk was titled "Mitochondrial Metabolism Establishes the Spemann-Mangold Organizer: Discovery From Patients With Leigh Syndrome." During the enlightening panel session titled "Science and Social Responsibility: Outreach Opportunities and Perspectives Across Academia," experienced panelists Alexander Joseph, MA, Sarah E. Millar, PhD, and Shelby Smout, PhD, shared their perspectives, and their valuable insights enriched our event.

The event also featured recognition for outstanding achievements, including the awarding of Young Investigators Competition (YIC) winners Clifford Liu, MS (PI: Bruce D. Gelb, MD), and Miranda L. Wilson (PI: Florence Marlow, PhD). Congratulations were extended to recipients of the best poster awards: Macy Akalu (PI: Dusan Bogunovic, PhD), Paloma Bravo, MS (PI: Florence Marlow, PhD), Ivianis Nieves Carril (PI: Nicole C. Dubois, PhD), Alexa von Mueffling (PI: Silvia De Rubeis, PhD), and Shrey Patel (PI: Bruce D. Gelb, MD).

# 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, MPhil**
*(Professor, Pediatrics, and Genetics and Genomic Sciences)*
**Research Areas:** Systems biology and integrative omics of asthma and allergic diseases

**Maria Curotto de Lafaille, PhD**
*(Professor, Pediatrics, and Immunology and Immunotherapy)*
**Research Areas:** Immunology of allergic diseases, B lymphocyte responses

**Nicole Ramsey, MD, PhD**
*(Instructor, Pediatrics)*
**Research Areas:** Environmental exposures and food allergy risk, food allergy treatment mechanisms, health equity in pediatric clinical trial recruitment/enrollment/retention

**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

## ASTHMA AND ALLERGY - *CONTINUED*

**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

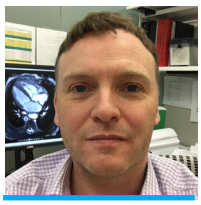
## CARDIOVASCLAR DISEASE

**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, The 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

## CARDIOVASCULAR DISEASE - *CONTINUED*

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

# NEURODEVELOPMENTAL DISORDERS - *CONTINUED*

**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

**Dorothy E. Grice, MD**
*(Professor, Psychiatry)*
**Research Areas:** Genetic and epidemiological studies of OCD, Tourette's 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

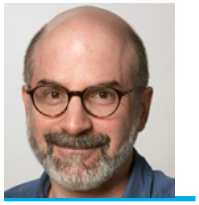
**Hala Harony-Nicolas, PhD**
*(Associate Professor, Psychiatry, and Neuroscience)*
**Research Areas:** Brain circuits of social behavior, mechanisms of action of the oxytocin hypothalamic system, animal models for autism spectrum disorder

# NEURODEVELOPMENTAL DISORDERS - *CONTINUED*

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

**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

**Alejandro Martin-Trujillo, PhD**
*(Assistant Professor, Genetics and Genomic Sciences)*
**Research Areas:** Functional genomics and epigenomics, genomic imprinting, structural variation

**Paige M. Siper, PhD**
*(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

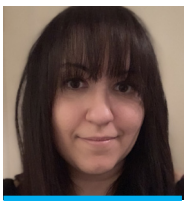
**Behrang Mahjani, PhD**
*(Assistant Professor, Psychiatry, Genetics and Genomic Sciences, and Artificial Intelligence and Human Health)*
**Research Areas:** Genetics of neurodevelopmental disorders

# NEURODEVELOPMENTAL DISORDERS - *CONTINUED*

**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



**Georgia Panagiotakos, PhD**
*(Associate Professor, Psychiatry, and Neuroscience)*
**Research Areas:** Brain development, neuropsychiatric disorders of developmental origin, autism spectrum disorders, animal and cell-based models, activity-dependent signaling

**Dalila Pinto, PhD**
*(Associate 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

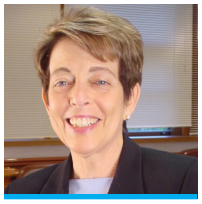
**Avi Reichenberg, PhD**
*(Professor, Psychiatry, and Environmental Medicine & Public Health)*
**Research Areas:** Autism, schizophrenia, other psychiatric disorders

**Anna Rommel, PhD**
*(Assistant Professor, Psychiatry)*
**Research Areas:** Neurodevelopmental disorders, environmental risk factors, prenatal exposures and neurophysiology

**Andrew J. Sharp, PhD**
*(Professor, Genetics and Genomic Sciences)*
**Research Areas:** Epigenomics, transcriptomics, genome function, structural variation, imprinting, congenital disorders

## NEURODEVELOPMENTAL DISORDERS - *CONTINUED*

**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 Baumel-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

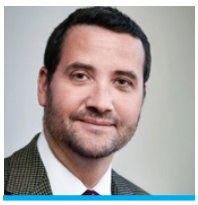
**Romina Bevacqua, PhD**
*(Assistant Professor, Medicine)*
**Research Areas:** Human islet biology, gene regulation and epigenetics of pancreatic islets, genetics of diabetes, primary organoids

# OBESITY AND DIABETES - *CONTINUED*

**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**
*(Assisant Professor, 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

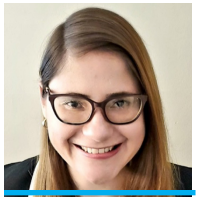
**Allan C. Just, PhD**
*(Assistant Professor, Environmental Medicine & Public Health)*
**Research Areas:** Epigenomics, environmental exposures, endocrine disruptors, air pollution, obesity, birth outcomes

**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*

**Esra Karakose, PhD**
*(Assistant Professor, Medicine)*
**Research Areas:** Diabetes, pancreatic beta cells, beta cell proliferation, alpha-to-beta cell transdifferentiation

**Liora S. Katz, PhD**
*(Associate Professor, Medicine)*
**Research Areas:** Type 1 diabetes, type 2 diabetes, β-cell proliferation, β-cell demise, glucolipotoxicity, cytokine-induced toxicity, β-cell mass preservation



**Corina Lesseur, MD, PhD**
*(Assistant Professor, Environmental Medicine & Public Health)*
**Research Areas:** Epi/genomics, environmental exposures, pregnancy outcomes, early-life metabolic programming

**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

## OBESITY AND DIABETES - *CONTINUED*

**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

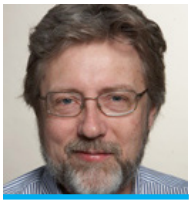
**Nita Vangeepuram, MD, MPH**
*(Associate Professor, Pediatrics, Environmental Medicine & Public Health, and Population Health Science and Policy)*
**Research Areas:** Youth diabetes prevention, community-based participatory research, health equity research

**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

**Brett R. Anderson, MD, MBA, MS**
*(Director, Child Health Services Research Center; Associate Professor, Pediatrics)*
**Research Areas:** Child health services research, health equity, econometrics



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

# OTHER RESEARCH FOCUSES - *CONTINUED*

**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

**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

# OTHER RESEARCH FOCUSES - *CONTINUED*

**David Dunkin, MD**
*(Associate Professor, Pediatrics)*
**Research Areas:** Tolerance induction and therapeutics in inflammatory bowel disease, mechanisms of inflammatory diseases of the gastrointestinal tract

**Sandeep Gangadharan, MD**
*(Associate Professor, Pediatrics)*
**Research Areas:** Clinical informatics, clinical decision support, AI, resuscitation

**Yolanda Garcia-Carmona, PhD**
*(Instructor, Medicine)*
**Research Areas:** B cell biology research, genetics of primary immunodeficiencies, therapeutic B cell neutralization

**Chris Gennings, PhD**
*(Professor, Environmental Medicine & Public Health, and Population Health Science and Policy)*
**Research Areas:** Biostatistical methods development for environmental health

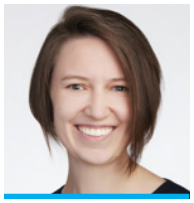
**Katherine Guttmann, MD, MBE**
*(Assistant Professor, Pediatrics)*
**Research Areas:** Palliative care, family-centered care, parent-physician communication, research ethics

**Dirk Hubmacher, PhD**
*(Assistant Professor, Orthopedics)*
**Research Areas:** Short stature syndromes, extracellular matrix, genetic connective tissue disorders

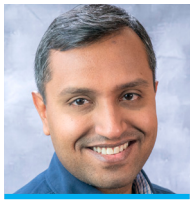
**Esra Karakose, PhD**
*(Assistant Professor, Medicine)*
**Research Areas:** Diabetes, pancreatic beta cells, beta cell proliferation, alpha-to-beta cell transdifferentiation

**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

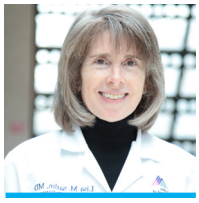


**Kaustav Mukherjee, PhD**
*(Instructor, Cell, Developmental & Regenerative Biology)*
**Research Areas:** Hematopoietic transcription regulation, genomics and epigenetics, single-cell technologies, erythroid disorders

**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

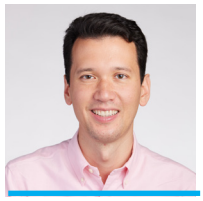
# OTHER RESEARCH FOCUSES - *CONTINUED*

**Christopher Sturgeon, PhD**
*(Associate Professor, Cell, Developmental & Regenerative Biology, and Medicine)*
**Research Areas:** Hematopoiesis, development, pluripotent stem cells, adoptive immunotherapy

**Caterina Tiozzo, MD, PhD**
*(Associate Professor, Pediatrics)*
**Research Areas:** Neonatal lung diseases, lung vascular development, lung regeneration, neonatal nutrition

**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

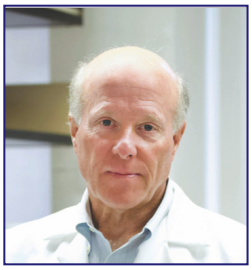


**Elvin Wagenblast, PhD**
*(Assistant Professor, Oncological Sciences, and Pediatrics)*
**Research Areas:** Blood stem cells, leukemia

# FACULTY RESEARCH INTERACTIONS

## *Faculty Highlight:* Lisa M. Satlin, MD

Bruce D. Gelb, MD

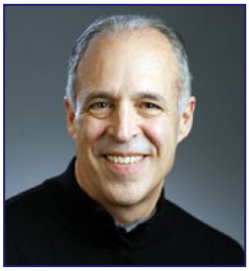


Andrew F. Stewart, MD

Scott H. Sicherer, MD

Joan Han, MD

Eyal Shemesh, MD



John Bucuvalas, MD

Jeffrey M. Saland, MD

Katherine Guttman, MD, MBE

Son Duong, MD

Lauren Choleva, MD

Nita Vangeepuram, MD, MPH

Jaime Chu, MD

David Dunkin, MD

Megan Januska, MD

Rebecca Trachtman, MD

Georgia Panagiotakos, PhD

Bryn D. Webb, MD

Marek Mlodzik, PhD

Chris Gennings, PhD

# *Faculty Highlight:* Lisa M. Satlin, MD

**Collaborators in mentorship and recruitment**
- Bruce D. Gelb, MD
- Andrew F. Stewart, MD
- Scott H. Sicherer, MD
- Joan Han, MD
- Eyal Shemesh, MD
- John Bucuvalas, MD
- Jeffrey M. Saland, MD

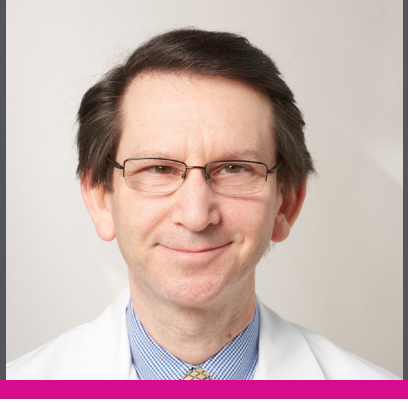
**Formal mentorship (as K award mentor)**
- Jeffrey M. Saland, MD
- Katherine Guttman, MD, MBE

**Mentorship in the Pediatric Faculty Scholar's Program (pipeline program for pediatric physician-scientists)**
- Son Duong, MD
- Lauren Choleva, MD
- Nita Vangeepuram, MD, MPH
- Jaime Chu, MD
- David Dunkin, MD
- Megan Januska, MD
- Rebecca Trachtman, MD

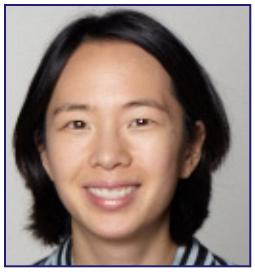
**Collaboration through the PCTO**
- Eyal Shemesh, MD

**Anticipated research study collaboration (in progress)**
- Georgia Panagiotakos, PhD

**Co-authorship of publications**
- Jeff M. Saland, MD
- John Bucuvalas, MD
- Bryn D. Webb, MD
- Bruce D. Gelb, MD
- Marek Mlodzik, PhD
- Chris Gennings, PhD



# *Faculty Highlight:* Scott H. Sicherer, MD



Supinda Bunyavanich, MD, MPH

Bruce D. Gelb, MD

Maria Curotto de Lafaille, PhD

Nicole Ramsey, MD, PhD

Jeffrey M. Saland, MD

Hugh A. Sampson, MD

Eyal Shemesh, MD

Nita Vangeepuram, MD, MPH

Julie Wang, MD

**Supinda Bunyavanich, MD, MPH**

- Collaborator on food allergy and eczema birth cohort study
- Collaborator on systems biology of early atopy analysis & bioinformatics center
- Collaborator on threshold, severity, and immunotherapy of peanut allergy
- Collaborator on gut microbiome in peanut allergy
- Collaborator on Consortium for Food Allergy Research (CoFAR)
- Collaborator on oral metabolite and immune trajectories study
- Collaborator on studies in the Food Allergy Treatment and Research Center
- Collaborator on food allergy research initiative (biosamples for pilot studies)

**Bruce D. Gelb, MD**

- Pediatric Clinical Trials Office

**Maria Curotto de Lafaille, PhD**

- Collaborator on B cell memory in human food allergy
- Collaborator on heterogeneity of T cell phenotype and function in food allergy
- Collaborator on Consortium for Food Allergy Research (CoFAR)
- Collaborator on studies in the Food Allergy Treatment and Research Center
- Collaborator on food allergy research initiative (biosamples for pilot studies)

**Nicole Ramsey, MD, PhD**

- Mentor
- Collaborator on JAK inhibition in adolescent and adult food allergy
- Collaborator on food allergy investigations for reaching equity in research

**Jeffrey M. Saland, MD**

- Collaborator as medical monitor on threshold, severity, and immunotherapy of peanut allergy

**Hugh A. Sampson, MD**

- Collaborator on Consortium for Food Allergy Research (CoFAR)
- Collaborator on threshold, severity, and immunotherapy of peanut allergy
- Collaborator on studies in the Food Allergy Treatment and Research Center
- Collaborator on food allergy research initiative (biosamples for pilot studies)

**Eyal Shemesh, MD**

- Pediatric Clinical Trials Office
- Collaborator on the EMPOWER program (food allergy studies and treatment for psychosocial and mental health)

**Nita Vangeepuram, MD, MPH**

- Collaborator on food allergy investigations for reaching equity in research

**Julie Wang, MD**

- Collaborator on food allergy patient samples for repository
- Collaborator on food allergy and eczema birth cohort study
- Collaborator on food allergy investigations for reaching equity in research
- Collaborator on food allergy clinical trials (pharmaceuticals)
- Collaborator on threshold, severity, and immunotherapy of peanut allergy
- Collaborator on Consortium for Food Allergy Research (CoFAR)
- Collaborator on studies in the Food Allergy Treatment and Research Center

# AWARDS/HONORS AND PUBLICATIONS

## FACULTY AWARDS/ HONORS

**Sharon Baumel-Alterzon, PhD**, The American Diabetes Association (ADA) 83rd Scientific Sessions, "Nrf2 Regulates Neonatal β-Cell Mass Expansion" oral talk

**Dusan Bogunovic, PhD**, International Cytokine & Interferon Society, ICIS-Luminex John R. Kettman Award for Excellence in Cytokine & Interferon Research

**Supinda Bunyavanich, MD, MPH, MPhil**, Chair, NIH Study Section: Cardiovascular and Respiratory Diseases, 2022-2024

**Bruce D. Gelb, MD**, Keynote Speaker, Pathologies of the RAS-MAPK Pathway: The Importance of a Multidisciplinary Network, Salerno, Italy, May 23, 2023

**Bruce D. Gelb, MD**, Plenary Speaker, 8th International RASopathies Symposium: Expanding Research and Care Practice, Through Global Collaboration and Advocacy, Denver, CO, August 23, 2023

**Bruce D. Gelb, MD**, Plenary Speaker, International Pediatric VAC and Heart Failure Summit 2023, St. Louis, MO, September 23, 2023

**Praveen Raju, MD, PhD**, CURE Childhood Cancer, 2023 Translation to CURE Award (T2C), 07/01/23-06/30/25, "Nanotherapeutic targeting of PPM1D inhibitors across the blood-brain barrier for pediatric brainstem tumors"

**Praveen Raju, MD, PhD**, Keynote Speaker, "Translational Hurdles in Pediatric Neuro-Oncology - The Elephants in the Room," The BrainStorm Summit - End Childhood Brain Cancer, Washington, DC, September 22, 2023

**Elvin Wagenblast, PhD**, Pew-Stewart Scholars Program for Cancer Research, 2023 Scholar

## FACULTY PUBLICATIONS

Woo JL, Nash KA, Dragan K, Crook S, Neidell M, Cook S, ... **Anderson BR**; New York State Congenital Heart Surgery Collaborative for Longitudinal Outcomes and Utilization of Resources (CHS-COLOUR). **Chronic Medication Burden After Cardiac Surgery for Pediatric Medicaid Beneficiaries.** *J Am Coll Cardiol*. 2023 Sep 26;82(13):1331-1340.

Jayaram N, Allen P, Hall M, Karamlou T, Woo J, Crook S, **Anderson BR**. **Adjusting for congenital heart surgery risk using administrative data.** *J Am Coll Cardiol.* 2023 Dec 5;82(23):2212-21.

Crook S, Dragan K, Woo JL, Neidell M, Jiang P, Cook S, ... **Anderson BR**. **Long-term health care utilization after cardiac surgery in children covered under medicaid.** *J Am Coll Cardiol.* 2023 Apr 25;81(16):1605-17.

Tran M, **Berin C**. **Reservoirs of allergic memory**. *Nat Rev Immunol*. 2023 Mar 23:1.

Rottner AK, Ye Y, Navarro-Guerrero E, Rajesh V, Pollner A, **Bevacqua RJ**, ... Gloyn AL. **A genome-wide crispr screen identifies calcoco2 as a regulator of beta cell function influencing type 2 diabetes risk.** *Nat Genet*. 2023 Jan;55(1):54-65.

Chen X, Pillay S, Lohmann F, **Bieker JJ**. **Association of ddx5/p68 protein with the upstream erythroid enhancer element (ehs1) of the gene encoding the klf1 transcription factor.** *J Biol Chem*. 2023 Dec;299(12):105489.

Malle L, Patel RS, Martin-Fernandez M, Stewart OJ, Philippot Q, Buta S, ... **Trachtman R**, ... **Bogunovic D**. **Autoimmunity in down's syndrome via cytokines, cd4 t cells and cd11c(+) b cells.** *Nature*. 2023 Mar;615(7951):305-14.

Gruber CN, Patel RS, **Trachtman R**, Lepow L, Amanat F, Krammer F, **Wilson KM**, ... **Gelb BD**, ... **Bogunovic D**. **Mapping systemic inflammation and antibody responses in multisystem inflammatory syndrome in children (mis-c).** *Cell.* 2023 Jul 20;186(15):3325.

Wigby K, Hammer M, Tokita M, Patel P, Jones MC, Larson A, ... **Bragg J**, ... Del Campo M. **Insights into the perinatal phenotype of kabuki syndrome in infants identified by genome-wide sequencing**. *Am J Med Genet A*. 2023 Apr;191(4):930-40.

Maron JL, Kingsmore S, **Gelb BD**, Vockley J, Wigby K, **Bragg J**, **Stroustrup A**, ... Davis JM. **Rapid whole-genomic sequencing and a targeted neonatal gene panel in infants with a suspected genetic disorder.** *Jama.* 2023 Jul 11;330(2):161-9.

Osman A, Mervosh NL, Strat AN, Euston TJ, Zipursky G, Pollak RM, ... **Buxbaum JD**, **Breen MS**, Kiraly DD. **Acetate supplementation rescues social deficits and alters transcriptional regulation in prefrontal cortex of shank3 deficient mice**. *Brain Behav Immun.* 2023 Aug 30;114:311-24.

Park MD, Reyes-Torres I, LeBerichel J, Hamon P, LaMarche NM, Hegde S, ... **Brown BD**, ... Merad M. **Trem2 macrophages drive nk cell paucity and dysfunction in lung cancer.** *Nat Immunol.* 2023 May;24(5):792-801.

**Brown BD**, Fauci AS, Belkaid Y, Merad M. **Rna vaccines: A transformational advance.** *Immunity*. 2023 Dec 12;56(12):2665-9.

Mayo AT, Myers CG, **Bucuvalas JC**, Feng S, Juliano CE. **Supporting robust teamwork - bridging technology and organizational science.** *N Engl J Med.* 2023 Jun 1;388(22):2019-21.

Budu-Aggrey A, Kilanowski A, Sobczyk MK, Shringarpure SS, Mitchell R, Reis K, ... **Bunyavanich S**, ...Paternoster L. **European and multi-ancestry genome-wide association meta-analysis of atopic dermatitis highlights importance of systemic immune regulation.** *Nat Commun.* 2023 Oct 4;14(1):6172.

Liu D, Meyer D, Fennessy B, Feng C, Cheng E, Johnson JS, ... **Buxbaum JD**, ... Charney AW. **Schizophrenia risk conferred by rare protein-truncating variants is conserved across diverse human populations.** *Nat Genet*. 2023 Mar;55(3):369-76.

Grandjean J, Desrosiers-Gregoire G, Anckaerts C, Angeles-Valdez D, Ayad F, Barrière DA, ... **Buxbaum JD**, ... Hess A. **A consensus protocol for functional connectivity analysis in the rat brain.** *Nat Neurosci.* 2023 Apr;26(4):673-81.

Weinstock JS, Gopakumar J, Burugula BB, Uddin MM, Jahn N, Belk JA, ... **Chami N**, ... **Loos RJF**, ... Jaiswal S. **Aberrant activation of tcl1a promotes stem cell expansion in clonal haematopoiesis.** *Nature*. 2023 Apr 12.

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# GRANTS

| AGENCY NAME | Funding from New Grants ($) | Funding from New & Existing Grants ($) |
|---|---|---|
| Department of The Army | 1,687,000 | 1,687,000 |
| National Institute of Mental Health/NIH/DHHS | 1,376,917 | 8,705,195 |
| National Institute of Environmental Health Sciences/NIH/DHHS | 813,948 | 1,625,588 |
| National Institute of Allergy And Infectious Diseases/NIH/DHHS | 808,751 | 8,536,842 |
| National Institute On Aging/NIH/DHHS | 746,162 | 915,162 |
| National Heart, Lung, And Blood Institute/NIH/DHHS | 668,965 | 3,307,547 |
| Emory University | 401,959 | 401,959 |
| Juvenile Diabetes Research Foundation | 330,883 | 330,883 |
| Alex's Lemonade Stand Foundation for Childhood Cancer | 200,000 | 200,000 |
| Damon Runyon Cancer Research Foundation | 200,000 | 200,000 |
| Eli Lilly And Company | 196,460 | 196,460 |
| Rockefeller University | 175,000 | 175,000 |
| University of Pittsburgh | 174,906 | 526,934 |
| National Institute of Child Health And Human Development/NIH/DHHS | 169,000 | 4,090,782 |
| CURE Childhood Cancer | 165,000 | 165,000 |
| Vanderbilt University Medical Center | 102,465 | 102,465 |
| Job Research Foundation | 100,000 | 100,000 |
| Children's Hospital Med. Center, Ohio | 100,000 | 150,000 |
| CureSearch for Children's Cancer | 75,000 | 75,000 |
| Pew Charitable Trusts | 75,000 | 75,000 |
| Healthfirst | 55,000 | 55,000 |
| ASE Foundation | 25,000 | 25,000 |
| Indiana University | 22,205 | 22,205 |
| Columbia University | 19,621 | 242,663 |
| University of California, San Diego | 18,104 | 18,104 |
| Food Allergy Research & Education | 399 | 100,399 |
| National Institute of Diabetes And Digestive And Kidney Diseases/NIH/DHHS | | 7,003,945 |
| National Institute of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS | | 2,689,278 |
| Benaroya Research Institute At Virginia | | 2,359,666 |
| National Institute of General Medical Sciences/NIH/DHHS | | 1,157,555 |
| Johns Hopkins University Medical School | | 1,150,931 |
| National Center for Complementary and Integrative Health/NIH/DHHS | | 704,657 |
| National Institute of Dental And Craniofacial Research/NIH/DHHS | | 605,148 |
| Albert Einstein College of Medicine | | 496,759 |
| National Eye Institute/NIH/DHHS | | 436,725 |
| University of Wisconsin-Madison | | 378,090 |
| National Center for Advancing Translational Sciences/NIH/DHHS | | 338,000 |
| Northwestern University | | 308,797 |
| Additional Ventures Foundation | | 280,459 |
| Simons Foundation | | 241,196 |
| New York University Grossman School of Medicine | | 209,122 |
| ChadTough Defeat DIPG Foundation | | 200,000 |
| President and Fellows of Harvard College | | 183,846 |
| Brigham And Women's Hospital | | 181,347 |
| Memorial Sloan-Kettering Cancer Center | | 180,579 |
| Biomarin Pharmaceutical | | 171,185 |
| Boston Children's Hospital | | 156,201 |
| Leducq Foundation (Fondation Leducq) | | 154,935 |
| University of Colorado | | 146,824 |
| Adventist Health System/Sunbelt, Inc. | | 143,871 |
| Children's Hospital Pittsburgh | | 133,246 |
| BrightFocus Foundation | | 100,000 |
| Duke (Doris) Charitable Foundation | | 92,635 |
| Virginia Commonwealth University | | 82,819 |
| American Society of Hematology | | 62,500 |
| European Commission | | 57,909 |
| University of Cambridge | | 53,820 |
| Alzheimer's Association | | 51,480 |
| FRAXA Research Foundation | | 50,000 |
| RTW Charitable Foundation | | 50,000 |
| Children's Hospital of Philadelphia | | 44,613 |
| Hirschl/Weill-Caulier Trust | | 40,000 |
| Brain and Behavior Research Foundation | | 35,000 |
| University of Southern California | | 19,117 |
| Total | 8,707,744 | 52,782,442 |

# OUTGOING MATERIAL TRANSFER AGREEMENTS/LICENSES

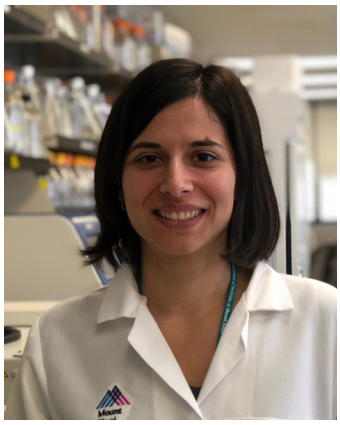
| Research Focus | Outgoing Material Transfer Agreements (#) | Licenses (#) |
|---|---|---|
| Neurodevelopmental disorders | 2 | 8 |
| Cardiovascular disease | 0 | 6 |
| Diabetes and Obesity | 1 | 5 |
| Allergy and Asthma | 1 | 8 |
| Others | 5 | 2 |
| **Total** | **9** | **29** |

| Licenses | Total Number |
|---|---|
| Antigens/Antibodies | 7 |
| Reagents/Methods/Cell Lines | 11 |
| Genes/Adapters/Vectors/Oligonucleotides | 6 |
| Gene Testing/Therapeutics | 3 |
| Mouse and Cell Models | 2 |
| **Total** | **29** |

# PILOT PROJECTS FUNDED FOR 2023

# 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.



## Marta Garcia-Forn, PhD

*Postdoctoral Fellow, Department of Psychiatry, Seaver Autism Center for Research and Treatment, Friedman Brain Institute, The Mindich Child Health and Development Institute, The Alper Center for Neural Development and Regeneration*

**Project Title:** Lineage Tracing of Glutamatergic Neurons in the Developing Cortex of a Mouse Model of DDX3X Syndrome

**Primary Mentor:**
Silvia De Rubeis, PhD
Associate Professor, Psychiatry
Seaver Autism Center for Research and Treatment, Friedman Brain Institute
The Mindich Child Health and Development Institute, The Alper Center for Neural Development and Regeneration

**Secondary Mentors:**
Mladen-Roko Rasin, MD, PhD, Neuroscience and Cell Biology
Rutgers University, RWJ Medical School
Nikolaos P. Daskalakis, MD, PhD
Harvard Medical School, McLean Hospital

## Katherine Schertz Hickey, MD

*Clinical Fellow, Pediatric Intensive Care Unit*

**Project Title:** Markers of Immune Dysregulation in Pediatric Patients With Severe Presentation of Viral Bronchiolitis

**Primary Mentor:**
Dusan Bogunovic, PhD
Director of the Center for Inborn Errors of Immunity
Associate Professor of Microbiology, Oncological Sciences, and Pediatrics
The Mindich Child Health and Development Institute, Precision Immunology Institute

**Secondary Mentors:**
Sandeep Gangadharan, MD
Medical Director of Mount Sinai Pediatric ICU
Alfin Vincencio, MD
Division Chief of Pediatric Pulmonology

# FACULTY PILOT AWARDS

Two pilot projects were selected for $75K in institutional funding for a one-year period starting March 1, 2023. 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:** Structure-Function Studies of Ara h 2 Specific Antibodies Isolated From Highly Sensitized Children With Peanut Allergy

**Principal Investigators:** Maria Curotto de Lafaille, PhD (Communicating PI), and Goran Bajic, PhD (Co-PI)

**Maria Curotto de Lafaille, PhD (Communicating PI)**
*Professor, Pediatrics, and Immunology and Immunotherapy*

**Goran Bajic, PhD (Co-PI)**
*Assistant Professor, Microbiology*

## **Project Title:** Irritable Bowel Syndrome: An Antigenic Driven Disease?

**Principal Investigators:** David Dunkin, MD (Communicating PI), and Maria Curotto de Lafaille, PhD (Co-PI)

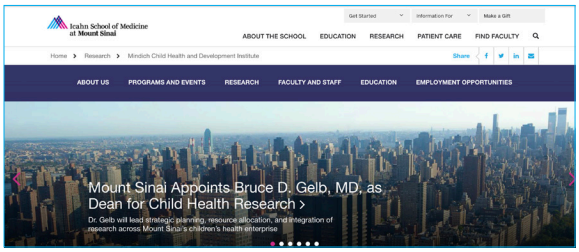
**David Dunkin, MD (Communicating PI)**
*Associate Professor, Pediatrics*

**Maria Curotto de Lafaille, PhD (Co-PI)**
*Professor, Pediatrics, and Immunology and Immunotherapy*

# COMMUNICATIONS

The 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

*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 the 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 us @MindichCHDI or visit our website at www.mountsinai.org/mchdi.


# SHARED RESOURCES

## GRANTFORWARD

GrantForward is a pre-award funding database with a comprehensive list of federal, foundation, and other funding sources. It offers a user-friendly search interface, automated email alerts, and tailored grant recommendations. GrantForward 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. The 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:
https://icahn.mssm.edu/research/ipm/programs/biome-biobank.

## 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.

# STRATEGIC PLAN IMPLEMENTATION

## PEDIATRIC CLINICAL TRIALS OFFICE

### INTRODUCTION

The Pediatric Clinical Trials Office (PCTO) within the Mindich Child Health and Development Institute (MCHDI) has emerged as a pivotal resource, addressing the research needs of investigators within the Mount Sinai Health System, particularly in the realm of pediatric clinical trials. Its inception in the aftermath of the pandemic surge in Fall 2020 underscores a strategic response to the evolving landscape of health care challenges.

A notable aspect of the PCTO's operational framework is its collaborating with the Clinical Trials Office (CTO) in Internal Medicine. This collaboration extends its purview to the adept management of a spectrum of trials, encompassing investigator-initiated, industry-funded, and federally funded endeavors. Of particular interest is the PCTO's commitment to facilitating "extension" trials, wherein the studies extend seamlessly from adult to the pediatric population. This nuanced approach speaks volumes about the adaptability and foresight in catering to the intricacies of diverse clinical research changes.

### PCTO STAFF

Michele Cohen, MS, CCRC – Co-Director/Eyal Shemesh, MD – Co-Director
Yair Bitton, MPH, MBA, CCRP – Assistant Director
Navjot Kaur – Financial Analyst
Angela Stangarone – Senior Regulatory Coordinator
Alyssa Gontzes – Clinical Research Coordinator II
Gabrielle Jonny – Clinical Research Coordinator II

In 2023, the PCTO added a Clinical Trials Manager and two additional CRCs:

Xueru Mu, CCRP – Clinical Trials Manager
Tarini Vats – Clinical Research Coordinator II
Maha Hussain – Clinical Research Coordinator II

Adding a Clinical Trials Manager and two more Clinical Research Coordinators (CRCs) demonstrates a significant expansion in capacity and capability. This expansion will undoubtedly enhance the PCTO's ability to support researchers within the Mount Sinai Health System and contribute to advancing pediatric clinical research. It's exciting to see the continued commitment to excellence and innovation in pediatric health care.

# RESEARCH FACULTY SERVED BY PCTO

PCTO is currently serving the following investigators/divisions:

## Pediatric Divisions:

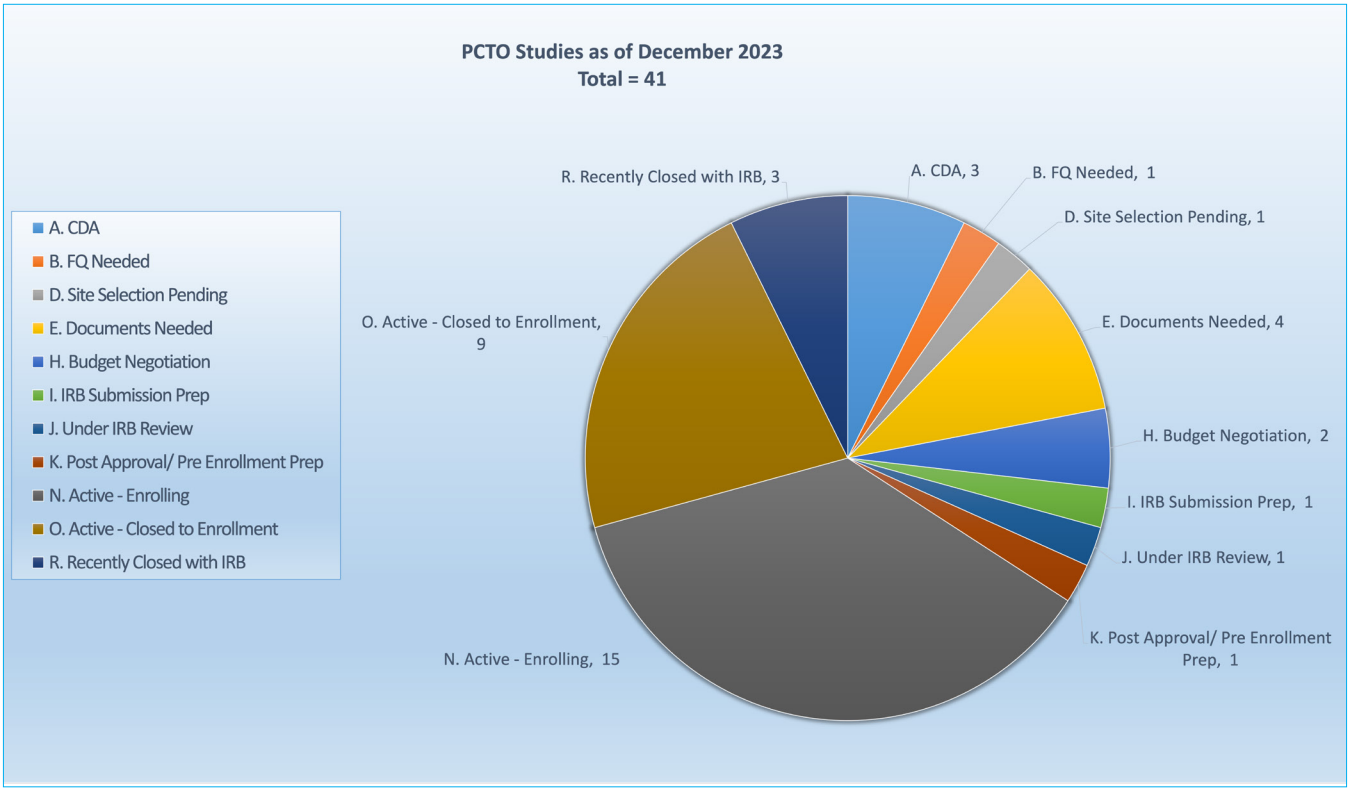
| | | |
|---|---|---|
| **Allergy** | | |
| Scott H. Sicherer, MD | Julie Wang, MD | Amanda Cox, MD |
| Roxanne Oriel, MD | Mary Grace Baker, MD | Nicole Ramsey, MD |
| **Endocrinology** | | |
| Joan Han, MD | Robert Rapaport, MD | Hillary Hotchkiss, MD |
| **Gastroenterology** | | |
| Marla Dubinsky, MD | David Dunkin, MD | Keith Benkov, MD |
| Nancy Pittman, MD | | |
| **Nephrology** | | |
| Jeffrey M. Saland, MD | Hillary Hotchkiss, MD | |
| **Neonatal ICU** | | |
| Courtney Juliano, MD | | |
| **Rheumatology** | | |
| Rebecca Trachtman, MD | | |
| **Cardiology** | | |
| Miwa Geiger, MD | Brett R. Anderson, MD, MBA, MS (new) | |
| **Pediatric ICU** | | |
| Sheemon Zackai, MD | Sandeep Gangadharan, MD | Shubhi Kaushik, MD |
| Jennifer Duchon, MD (new) | | |

## Adult Divisions With Pediatric Trials:

| | |
|---|---|
| **Dermatology** | Emma Guttman, MD, PhD |

# CLINICAL TRIALS PORTFOLIO

Tracking various phases within the clinical trials review and approval flow is crucial for maintaining competitiveness with startup timelines. By closely monitoring these phases, the PCTO can ensure efficient progress through the trial process and optimize its ability to contribute to pediatric health care advancements.




## Active Clinical Trials (24)

### Pediatric Divisions

Cardiology – 1 (NIH sub-award)

Allergy – 7 (up from 5)

Gastroenterology – 5

Rheumatology – 1

Endocrinology – 4

PICU – 2

Nephrology – 2

NICU – 1

### Collaborations with Adult Divisions:

Pediatric Allergy and Adult Dermatology – 1

## Clinical Trials in Startup Phase (15)

| Pediatrics | |
|---|---|
| Allergy – 4 | Rheumatology – 2 |
| Gastroenterology – 4 | NICU – 1 |
| Endocrinology – 2 | Nephrology – 1 |
| Cardiology – 1 | |

## NOTABLE PROJECTS

- PCTO is involved in establishing Mount Sinai as a participating site in the NIH/NHLBI-funded Pediatric Heart Network, which looks at how the heart develops and why children are born with heart problems.
- PCTO activated the first trial in the PICU treating complicated bodily infections with a new therapy versus the best available therapy. This trial has the potential to make a meaningful impact on the lives of children and families facing serious infections.
- PCTO with the Allergy Division contributed to Mount Sinai being recognized as 1 of the top 4 (out of 86 international sites) recruiting for a trial evaluating an investigational drug patch for 4- to 7-year-old children with peanut allergy where the patch is designed to simulate repeated exposures to the allergen.

# 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). They 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 their diagnostic yield and boost discovery, future directions of their program include moving towards third-generation sequencing and strengthening their relationship with the Functional Genomics and Disease Modeling Core.

The Functional Genomics and Disease Modeling Core is led by Tirtha K. Das, PhD, and was established a few years ago 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 to serve as important leads for ongoing and future studies in vertebrate models, to be ultimately translated to the clinics. The focus has currently been to develop models related to rare variants of: a) RASopathies, b) undiagnosed diseases, c) cancer.

The core has developed 20 new RASopathy fly models for various genes in the MAPK pathway. Analysis of how these variants affect lacZ reporters for the major signaling pathways have been completed, revealing some key differences that can be leads for further analysis in relevant vertebrate models.

An important focus of the core is analyzing gene variants identified in the UDP and rarely associated in other diseases. Dr. Das and his team developed fly models of NDUFAF and MAGI2, and analysis of how these variants affect lacZ reporters for the major signaling pathways have been nearly completed.

In addition, they have also developed five models of rare kinase-fusion gene variants that arise in patients undergoing targeted lung cancer therapy. Using a combination of lacZ reporter and western blot analysis key signaling differences have been identified and two manuscripts are being finalized for submission.

Finally, this year they have further used their fly expertise and initiated multiple investigator-initiated screens of drugs and novel compounds in our fly disease models. These screens serve as good first-step therapeutic index indicators, as well as to comprehend possible mechanisms of action, and helps identify lead compounds to further test in vertebrate models.

They also want to improve participation of children in genomic research. A common barrier to participation is the difficulty with obtaining a sample. However, cord blood collection is noninvasive and is routinely collected for medical care. In order to use cord blood in future genomic studies, they had 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. They hoped that 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. As such, last year a pilot study was developed that aimed at assessing if cord blood was a suitable specimen for genomic analysis. Thirty specimens of cord blood were tested and verified that there was no maternal contamination, validating their hypothesis. A manuscript is under preparation.

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. They are collaborating with the Charles Bronfman Institute for Personalized Medicine in the effort of creating a new repository of sequencing data that will integrate health and research data at Mount Sinai. The overarching goal is to biobank genetic information from 1 million individuals who 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. They are excited to announce that this groundbreaking initiative will start enrolling participants in 2024!

# CENTER FOR CHILD HEALTH SERVICES RESEARCH

The Center for Child Health Services Research in the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai serves as the primary locus for research. The Center is directed by Brett R. Anderson, MD, MBA, MS and is actively engaged in developing a robust data infrastructure to gather information securely and efficiently, all in an effort to improve child health care. It seeks to promote the sharing of interdisciplinary ideas, methodologies, data, and mentorship, to enhance productivity and creativity for investigators—both within and outside the Center. To support its initiatives, it will be partnering with existing departments, institutes, and governmental agencies. The Center's expertise focuses on improving the quality and effectiveness of the health care system for all children, with particular emphasis on providers, payers, and policy; value and effectiveness; and access and equity.

## Faculty and Staff:

### Brett R. Anderson, MD, MBA, MS

*Director*

Dr. Brett R. Anderson is the inaugural Director for the Center for Child Health Services Research in the Mindich Child Health and Development Institute. Dr. Anderson is an Associate Professor in the Departments of Pediatrics and Population Health and Policy, and an NHLBI R01-funded health services and health equity researcher. In her leadership role, she is fostering interdisciplinary collaborations to establish a primary locus for research on the quality and effectiveness of the health care system for children at the Icahn School of Medicine. Dr. Anderson's research focuses on linkage and integration of large datasets and application of econometric modeling to identify modifiable drivers of outcomes, value, and health inequities for children with cardiac disease.

### Sarah Crook, PhD

*Director of Analytics*

Dr. Crook is an Assistant Professor in the Department of Population Health. She serves as the Director of Analytics for the Center for Child Health Services Research, with expertise in biostatistics, epidemiology, and health services research. Dr. Crook earned both her BS in human physiology and MS in epidemiology and biostatistics from the University of Leeds, UK. She then completed her PhD in epidemiology and biostatistics at the University of Zurich, Switzerland, and a training fellowship in Guidelines Methodology with Cochrane and the UK's National Institute of Health and Care Excellence.

Dr. Crook will contribute to the Center's initiative through the application of her expertise and specialization in large-scale data analytics. This includes the integration of clinical registry data, administrative data, and social determinants of health.

## CENTER FOR CHILD HEALTH SERVICES RESEARCH

### Pengfei Jiang, MS

*Senior Data Analyst*

Mr. Jiang serves as the Senior Data Analyst for the Center for Child Health Services Research. He plays a pivotal role in fostering a dynamic and efficient data environment for the Center. Mr. Jiang is a DrPH student at Johns Hopkins Bloomberg School of Public Health, and holds an undergraduate degree in mathematics and economics from UCLA and a master's in biostatistics from the Mailman School of Public Health at Columbia University.

Mr. Jiang is responsible for designing and managing the data infrastructure, as well as providing essential analytic support to the faculty and collaborators.

### Yohaira Rojas Guzman

*Administrative Director*

Yohaira Rojas Guzman joins as the Administrative Director of the Center for Child Health Services Research. In her previous role, Ms. Guzman directed a mobile prostate cancer screening program, extending vital screenings and outreach to underserved communities. Her career at Mount Sinai began in 2015 as Administrative Manager of Operations at the Center for Advanced Medicine, where she supported patient care access and navigation. Ms. Guzman's background encompasses roles such as the Associate Director of Ambulatory Care Operations at Mount Sinai Hospital, where she directed construction projects and managed ambulatory practices. Additionally, she served as the Emergency Management Branch Director, overseeing planning, response, and recovery services. Before joining Mount Sinai, she worked as the Manager of Clinical Operations & Training at Rutgers University School of Dental Medicine and held several roles in revenue cycle operations at Columbia University School of Dental Medicine. As Administrative Director, she will play a pivotal role in supporting the Center's operations and cross-functional collaboration.

### Chantal Sanchez

*Research Manager*

Chantal Sanchez is the Research Manager for the Center for Child Health Services Research. Ms. Sanchez completed her undergraduate degree in biology at Columbia University and worked as a Clinical Research Coordinator before joining the team. Ms. Sanchez will focus primarily on supporting the Center's initiatives and contribute to its studies on outcomes and health disparities for children and young adults with congenital heart disease.

# LEADERSHIP AND STAFF

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

**Alisha Graham**
Administrative Coordinator
**Jennifer E. Cole**
Senior Director of Development

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M. Cecilia Berin, PhD
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Icahn School of Medicine at **Mount Sinai**

*The Mindich Child Health and Development Institute*