



Icahn School
of Medicine at
Mount
Sinai

*The Mindich
Child Health and
Development Institute*

2021 Annual Report



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

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





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Director's Message

Bruce D. Gelb, MD, Director

The year just past, 2021, will surely be remembered for its dramatic ups and downs with the COVID-19 pandemic. As we started that year, mass vaccination was rolling out, notably early for those of us at Mount Sinai, and hopes were high that this would bring an end to the pandemic, restoring life to normal. However, as we now know, SARS-CoV-2 had other plans for us. The successive variant-driven waves kept us on an emotional rollercoaster. At the MCHDI, we were fortunate enough to squeeze in our in-person retreat shortly after Thanksgiving; getting together brought an emotional high, even as we knew that the Omicron variant was likely on its way. As I write this in early 2022, our hopes are burgeoning that this wave, now waning fast, will give way to a manageable endemicity. Let us pray that this will be confirmed by this time next year.

Meanwhile, we should all take great pride in what was accomplished scientifically this past year, both for individual research teams and for the MCHDI as a whole. As recounted in this annual review, MCHDI researchers were highly successful and productive. Despite the challenges that forced computational investigators to work at a distance for much of the year and the fits and starts for wet bench research due to periods of quarantines and isolation, particularly during the Omicron wave, we all pulled together and creatively managed to keep our scientific pace high. We can look back on 2021 with pride in our fortitude and resilience. Congratulations to all!

The MCHDI has continued to expand its faculty. We highlight some of the new members in this report; welcome to our newest members! I also note the departure of two important members for new academic opportunities: Karen Wilson, MD, MPH, who co-directed the MCHDI's Pediatric Clinical Trials Office (PCTO), left for the University of Rochester, where she is the Vice Chair for Clinical and Translational Research in the Department of Pediatrics. Bryn Webb, MD, who oversaw the MCHDI's Undiagnosed Diseases Program (UDP), decamped for the University of Wisconsin at Madison. I want to thank both of them for their stalwart service to the MCHDI. Of note, Mafalda Barbosa, MD, PhD, a new MCHDI faculty member, has assumed the leadership of the UDP and will be more broadly involved in our growing efforts in pediatric precision medicine. The PCTO, which has grown spectacularly this past year, remains under the directorship of Eyal Shemesh, MD.

As we look ahead to 2022 and beyond, we should think strategically about what our post-COVID pandemic scientific world should look like. While surely some of our emphasis will be on a return to the *status quo ante*, let us not miss the opportunities that creative destruction enables. We now know that some of what we do can be done effectively from anywhere with an internet connection. We can engage scientists worldwide, for didactics and collaboration, more effectively and efficiently than previously seemed possible. I encourage everyone in the MCHDI to use this moment to contemplate how we can leverage the lessons of the past two years, painfully acquired, to enhance and accelerate our child health scientific mission.

FACULTY GROWTH

MCHDI ANNUAL FACULTY GROWTH 2021

IN 2021

We welcomed one new external faculty and eight internal faculty members to our institute.

77 Members

Currently, we total 77 members consisting of scientists and physician-scientists across the disciplines of Allergy & Asthma, Cardiovascular Disease, Neurodevelopmental Disorders, Obesity & Diabetes, and more.

■ Existing faculty
■ Internal recruits
■ External recruits

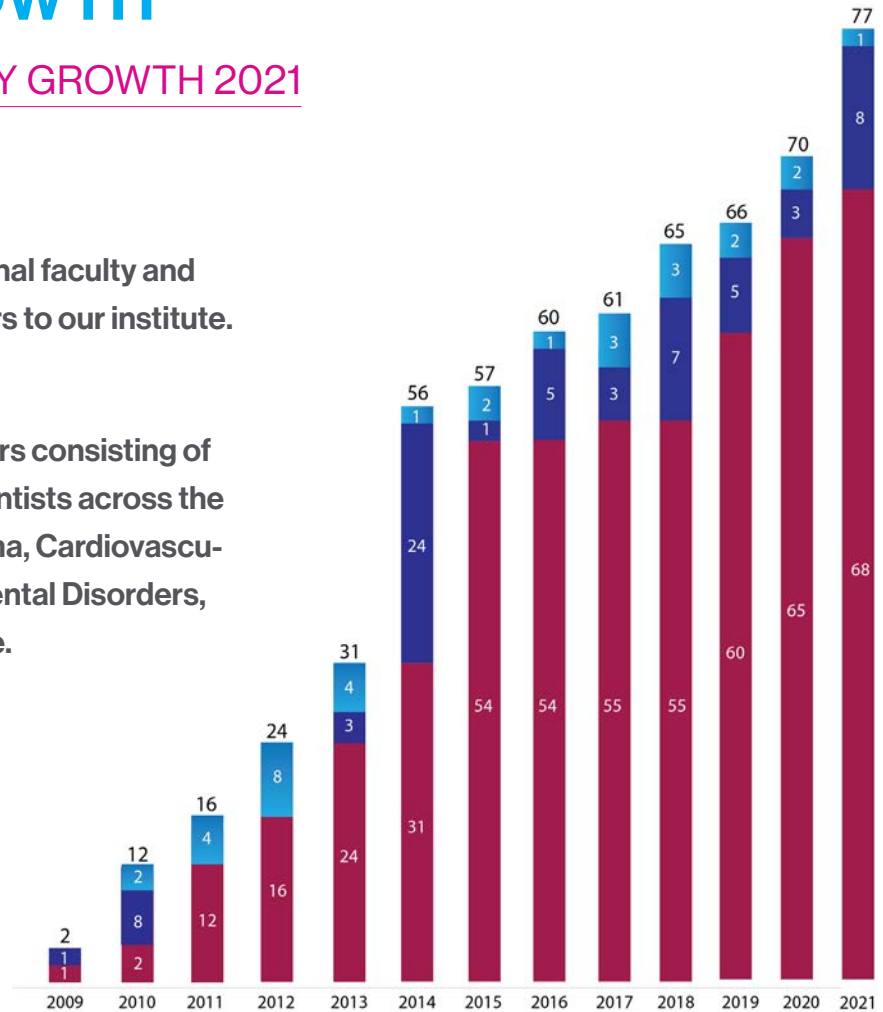


Chart of faculty recruits since our inception in 2009. In 2021, our institute recruited one new external and eight internal faculty members.

MCHDI FACULTY MEMBERS

MCHDI faculty members from left to right: Nicole C. Dubois, PhD; Adolfo García-Ocaña, PhD; Donald K. Scott, PhD; M. Cecilia Berin, PhD; Minji Byun, PhD; David Dunkin, MD; Dalila Pinto, PhD; Andrew J. Sharp, PhD; Dani Dumitriu, MD (no longer at Sinai), PhD; Bruce D. Gelb, MD; Amy R. Kontorovich, MD, PhD; Martin J. Walsh, PhD; Jia Chen, ScD; Rupangi C. Vasavada, PhD (no longer at Sinai)





NEW FACULTY

NEW EXTRAMURAL FACULTY



Lauryn Choleva, MD

Lauryn Choleva, MD, MSc, is an Instructor in the Division of Pediatric Endocrinology and Diabetes at Mount Sinai Kravis Children's Hospital. Dr. Choleva's research is conducted out of the Diabetes, Obesity and Metabolism Institute at the Icahn School of Medicine at Mount Sinai in the lab of Dr. Andrew Stewart, MD.

Her research is focused on the development of novel methods for inducing the replication and differentiation of insulin-producing beta cells for future use as regenerative pharmacotherapy for patients with Diabetes Mellitus. She is also exploring novel roles of the known cell cycle inhibitor p57KIP2 in human beta cells to understand the pathogenesis of childhood hypoglycemic syndromes.

Dr. Choleva graduated with a BSc in Anatomy and Cell Biology from McGill University and with an MSc in Clinical

Nutrition from the University of Toronto. She received her medical degree from the Sackler School of Medicine at Tel Aviv University. Dr. Choleva completed her residency in Pediatrics and her fellowship in Pediatric Endocrinology at the Icahn School of Medicine at Mount Sinai. She was a recipient of the 2020 Pediatric Scholars Award from The Icahn School of Medicine at Mount Sinai Department of Pediatrics.



NEW INTRAMURAL FACULTY



Mafalda Barbosa, MD, PhD

Mafalda Barbosa, MD, PhD, is an Assistant Professor of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai. Dr. Barbosa received her MD from the University of Porto and completed her first Medical Genetics residency in Portugal.

She then moved to the United States for her PhD studies at Mount Sinai where she specialized in novel molecular diagnostics of Neurodevelopmental Disorders. During her graduate studies she became proficient in the analysis of microarrays, exome sequencing, and methylation profiles/epimutations. Because she wanted to be able to provide patient care, after graduating from her PhD, she completed a combined Pediatrics/Genetics residency at Mount Sinai.

In the summer of 2021, Dr. Barbosa enthusiastically joined Sinai's faculty with an appointment that entails mostly

clinical work with some protected time for research, supported by The Mindich Child Health & Development Institute. Her research focus will continue to be on children with genetic conditions, including spear-heading the well-known Undiagnosed Diseases Program. Dr. Barbosa dedicated the last 15 years of her life to Medical Genetics, aiming to help children (and their families) with genetic disorders as both clinician and researcher. She is committed to continuing to serve this population as a physician-scientist while contributing to advances in Science and Medicine.

INTRAMURAL FACULTY-CONTINUED



Jennifer Bragg, MD

Jennifer Bragg, MD, is a Neonatologist and Associate Professor of Pediatrics at the Icahn School of Medicine at Mount Sinai. She is the Director of the NICU Follow-up Program and a member of the Cardiac Neurodevelopmental Outcomes Collaborative (CNOC). Dr. Bragg is the Site director for Vermont Oxford Network Critical Transitions Collaborative.

Dr. Bragg received her MD in 2007 from New York University School of Medicine. She completed her Pediatric Residency and Perinatal-Neonatal Fellowship at the Kravis Children's Hospital at Mount Sinai before an additional year as a Research Fellow with a neurodevelopmental focus.

At Mount Sinai Kravis Children's Hospital, Dr. Bragg provides inpatient intensive care and outpatient follow-up. She implemented and runs the Mount Sinai NICU Follow-up Program, a multidisciplinary program that provides neurodevelopmental care to high risk NICU graduates such as those born preterm, with congenital heart disease, seizures, hypoxic ischemic injury, and neurologic disorders.

Dr. Bragg's research focus is on identifying modifiable outcomes with a focus on sensory processing disorders, parental stress, and developmental programming. Current projects include implementation of programs including the SENSE Program (positive sensory interactions in the NICU), Reach Out and Reach and Developmental classes for families. Dr. Bragg has several Quality Improvement Initiatives such as Safe Sleep in the NICU and VON Critical Transitions. Finally, she is working with Dr. Bruce Gelb on GEMINI, a multi-center study that assesses how rapid whole genome sequencing improves time to diagnosis, delivery of care, and the economic impact of infants with rare diseases.



Jennifer Foss-Feig, PhD

Jennifer Foss-Feig, PhD, is an Assistant Professor in the Department of Psychiatry and at the Seaver Autism Center for Research and Treatment. Dr. Foss-Feig's research interests are in sensory and perceptual processing, particularly as they relate to possible underlying mechanisms of and biomarker development for autism spectrum disorder (ASD).

She studies auditory, visual, and multisensory processing, with a particular interest in temporal processing and testing for markers of excitatory/inhibitory imbalance. More recent work also examines the neurocomputational basis of proactive social behavior deficits in ASD. Dr. Foss-Feig also examines the overlap between autism and other neurodevelopmental disorders, including schizophrenia, and works on monogenic disorders that confer risk for ASD but have known etiology that maps to testable and targetable circuit-based alterations. Dr. Foss-Feig's research combines EEG, fMRI, and psychophysical approaches with clinical and behavioral assessments to explore brain-behavior relations and identify underlying alterations that may be targets for intervention. Her work

testing neural mechanisms across ASD and schizophrenia seeks to identify shared versus dissociable markers that could be useful as stratification tools for predicting psychosis risk in ASD populations. Across her research, Dr. Foss-Feig seeks to use paradigms that map to known biological alterations, are objective and reliable, and reliable, and are feasible or adaptable for individuals with neurodevelopmental disorders across the functioning spectrum. Dr. Foss-Feig has received funding from NIMH, Autism Speaks, Autism Science Foundation, the Marino Autism Research Institute, and the Brain and Behavior Research Foundation. She is a licensed clinical psychologist in the state of New York.

INTRAMURAL FACULTY-CONTINUED



Geming Lu, MD

Geming Lu, MD, MS, is an instructor in Dr. Adolfo García-Ocaña's lab of the Diabetes, Obesity, and Metabolism Institute and the Mindich Child Health and Development Institute at Icahn School of Medicine.

Dr. Lu completed his Doctor of Medicine in Harbin Medical University and finished Master of Science in the Capital University of Medical Sciences in China. After being a physician working on neuro-autoimmune disorders for 6 years, he started his scientific research career in the Immunology Institute at Mount Sinai in 2011. He focused on the molecular mechanisms of Th17 differentiation and Macrophage polarization in autoimmune animal models. He became a faculty member in his current lab after publications in Immunity, Nature communications, PNAS, and JEM et al. His recent research interests are to develop peptides and compounds treating diabetes by boosting beta cell regenerations and inhibiting auto-reactive T cells. He is

using NGS techniques to build up single-nucleus RNA seq references of human islets and target candidate genes using mass cytometry and gene editing technology. He finds HGF-cMET signaling increases MDSC populations, which can significantly inhibit auto-reactive T cells proliferation and delay T1D development; he is also exploring the feasibility of the adaptive Treg transfer and beta cell regeneration with the patent compounds in several T1D mice models.

Dr. Lu received NIH F32 fellowships from 2015 to 2017. His current research is supported by the Pilot & Feasibility grant and Immuno-Technology Core micro grant of the Einstein-Sinai Diabetes Research Center.



Kaustav Mukherjee, PhD

Kaustav Mukherjee, PhD, is an Instructor in Dr. James Bieker's lab of the Cell, Developmental and Regenerative Biology department at Icahn School of Medicine at Mount Sinai. He is also a member of the Black Family Stem Cell Institute and the Mindich Child Health and Development Institute.

A geneticist by training, he worked on yeast RNA biology during his PhD before completing his postdoctoral work at Dr. James Bieker's lab. Currently, he focuses on the biology of *in vivo* and *in vitro* erythropoiesis and associated red cell disorders such as anemia. He uses both animal models and stem cells to investigate the transcriptional regulation of gene expression during erythropoiesis by the transcription factor EKLF/Klf1 in erythroid cells, as well as macrophages that constitute the erythroblast island niche. He also uses patient-derived stem cells of congenital erythroid disorders affecting young children to study dyserythropoiesis leading to severe anemia. He extensively uses next generation sequencing, epigenetics, and single cell RNA-

seq in his research, and is skilled in the use of Bioinformatics and computational biology for analysis of NGS and sc-seq data. His research is supported by a pilot grant from the Black Family Stem Cell Institute titled "Investigating transcription regulation during erythroid differentiation of human induced pluripotent stem cells."

Dr. Mukherjee has completed his undergraduate degree from Osmania University, India, his Master's in Biotechnology from Madurai Kamaraj University, India, and his PhD in Animal Genetics from Stony Brook University, New York.

INTRAMURAL FACULTY-CONTINUED



Praveen Raju, MD, PhD

Praveen Raju, MD, PhD, is a pediatric neurologist and Associate Professor of Neurology and Pediatrics at the Icahn School of Medicine at Mount Sinai.

Dr. Raju completed his MD in 2001 at the University of Pennsylvania School of Medicine, where he also completed his PhD in Cell and Molecular Biology / Genetics as an NIH-funded MSTP Fellow. He served as a Pediatrics resident at Babies & Children's Hospital of New York / Columbia-Presbyterian Medical Center and subsequently finished his Pediatric Neurology Fellowship training at Boston Children's Hospital / Harvard Medical School in 2006 where he served as Chief Fellow during his final year. Prior to joining ISMMS, he served on the faculty at Weill Cornell Medicine for 10 years where he was the Caryl & Israel A. Englander Clinical Scholar in Children's Health and also cared for children with neurological issues at Memorial Sloan Kettering Cancer Center.

At Mount Sinai Kravis Children's Hospital, Dr. Raju's primary clinical focus is in pediatric onco-neurology and he treats patients with neurological complications of cancer including seizures, headaches, and neuropathy. His clinic also has a special interest in pediatric neurofibromatosis (NF) and neurogenetic disorders.

In addition to his clinical responsibilities, Dr. Raju directs the Laboratory for Pediatric Brain Tumor Research at ISMMS and studies the developmental origins of pediatric brain tumors with a particular translational focus on identifying improved therapies for medulloblastoma. He is also the Director of the Mount Sinai Pediatric Neurology Residency Program and Associate Director of the Mount Sinai Medical Scientist Training Program (MSTP).



Christopher M. Sturgeon, PhD

Christopher M. Sturgeon, PhD, is an Associate Professor at the Icahn School of Medicine at Mount Sinai. Chris's lab studies the development of the human hematopoietic system, using the *in vitro* differentiation of human pluripotent stem cells (hPSC) as a model system.

The ability to differentiate hPSC towards a bona fide hematopoietic stem cell (HSC) would be a major step forward for the treatment of patients in need of a suitable donor match. Similarly, hPSCs offer unprecedented access to early embryonic hematopoietic lineages, which may have untapped clinical potential. To harness these possibilities, it is essential to be able to direct the differentiation of hPSCs in a controlled fashion. To that end, Chris's research has developed defined media approaches, coupled with staged addition of recombinant morphogens such as BMP, WNT, and RA, to recapitulate these early embryonic developmental stages.

Ongoing research interests include:

- Investigating the molecular mechanisms of hematopoietic development and the immediate precursor to the HSC, hemogenic endothelium
- Characterizing the translational potential of HSC-independent immune lineages
- Identifying the developmental trajectory of nascent mesoderm as it differentiates towards blood
- Understanding the role of RNA splicing in embryonic hematopoiesis

INTRAMURAL FACULTY-CONTINUED



Nita Vangeepuram, MD, MPH

Nita Vangeepuram, MD, MPH, has had years of clinical training and experience as a general pediatrician with a special interest in management of pediatric obesity and its complications.

Her research focuses on pediatric obesity and diabetes prevention and treatment, community-engaged research, and health disparities/health equity research. She completed a joint fellowship in General Academic Pediatrics and Environmental Pediatrics and is an Assistant Professor in the Departments of Pediatrics and Population Health Science and Policy and Engagement Core Lead of Mount Sinai's Institute for Health Equity Research. Her recent Career Development Award (K23) and current R03 support a study using community-based participatory research, in addition to other novel methods (peer education and mobile health technologies), to develop models for the prevention of Type 2 diabetes among at-risk East Harlem youth. Her long-term career goal is to lead national efforts to prevent and treat conditions disproportionately impacting ethnic-minority youth and their families, by

leveraging the assets of community-academic partnerships. This goal represents a synergy of her clinical training, community outreach and advocacy work, and public health research training. She has specific experience in collaborating with community stakeholders to develop and implement health surveys, interventions, and disease prevention programs, and in conducting quantitative and qualitative research. She has led studies to examine barriers and facilitators for recruitment and retention of diverse populations for community-based and other types of research. Dr. Vangeepuram's primary interest is to ensure that patient/family perspectives guide our research through participatory approaches and that our research is representative of the diverse populations we serve.

ANNUAL RETREAT

Our 9th Annual Retreat was hosted at the Harmonie Club on November 30, 2021 with ~100 faculty, trainees, and speakers in attendance. 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 comprised of our MCHDI Director, Bruce Gelb, MD, Committee co-chairs, Hala Harony-Nicolas, PhD, and Florence Marlow, PhD, MCHDI faculty member, Robert Krauss, PhD, and our trainee leadership committee member, Adele Mossa, PhD. Keynote speaker Dr. Elizabeth Engle (Principal Investigator, F.M. Kirby Neurobiology Center and Senior Associate in Neurology, Boston Children's Hospital Professor of Neurology and Ophthalmology, Harvard Medical School Investigator, Howard Hughes Medical Institute, Associate Member, Broad Institute of MIT and Harvard), present-

ed her impressive science titled "Dissecting Coding and Noncoding Variation Underlying the Congenital Cranial Dysinnervation Disorders." Dr. Engle and other esteemed panelists including Dr. Brett Abrahams (Executive Vice-President, Research & Development, Magnolia Neurosciences) and Dr. Brateil Badal (Director of Diversity, Associated Medical Schools of New York) shared their unique perspectives and experiences in navigating careers in science. Our faculty and trainee scientists gave informative talks on a wide range of research topics and we ended the day by announcing our Young Investigators Competition winners: Nelson Rodriguez, PhD (postdoctoral division) and Winston Cuddleston (predoctoral division) as well as awardees for best posters: Joshua Morrison (judges choice), Bhavana Shewale (judges choice), Cindy Wang (judges choice) and Michael Espino (crowd favorite).



FACULTY RESEARCH AREAS

ASTHMA AND ALLERGY



M. Cecilia Berin, PhD

(Professor, Pediatrics)

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



Supinda Bunyavanich, MD, MPH

(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

(Associate 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

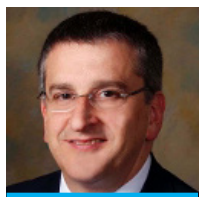


Karen M. Wilson, MD, MPH

(Adjunct Professor, Pediatrics)

Research Areas: Second hand tobacco smoke, second hand 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



Bruce D. Gelb, MD

(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



Alan Groves, MBChB, MD

(Adjunct Professor, Diagnostic, Molecular and Interventional Radiology)

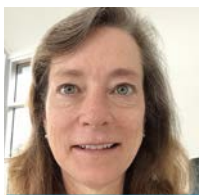
Research Areas: Hemodynamics, cardiac function, echocardiography, magnetic resonance imaging



Yuval Itan, PhD

(Assistant 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; Assistant 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)

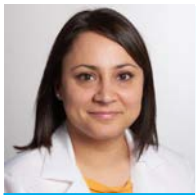
Research Areas: Environmental epigenetics, molecular epidemiology



Tirtha K. Das, PhD

(Assistant Professor, Cell, Developmental & Regenerative Biology)

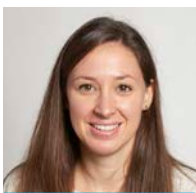
Research Areas: Integrating fly plus vertebrate disease models, cancer, rare mendelian diseases, therapeutics development



Silvia De Rubeis, PhD

(Assistant Professor, Psychiatry)

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



Jennifer Foss-Feig, PhD

(Assistant 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

(Assistant Professor, Psychiatry, and Neuroscience)

Research Areas: Brain circuits of social behavior, mechanisms of action of the oxytocin hypothalamic system, animal models for autism spectrum disorder



Megan K. Horton, PhD, MPH

(Associate Professor, Environmental Medicine & Public Health)

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



Laura Huckins, PhD

(Assistant Professor, Genetics and Genomic Sciences)

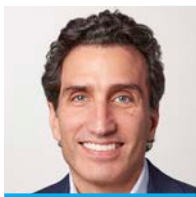
Research Areas: Psychiatric Genetics, specializing in understudied disorders and disorders affecting vulnerable populations. Particular focus on anorexia nervosa, PTSD, sexual assault, OCD. Secondary focus on machine learning algorithms; transcriptomic imputation; multi-omic eQTL-based methodologies.



Magdalena U. Janecka, PhD

(Assistant Professor, Psychiatry, Seaver Autism Center)

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



Alex Kolevzon, MD

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

Research Areas: Autism spectrum and other neurodevelopmental disorders



Robert S. Krauss, PhD

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

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

NEURODEVELOPMENTAL DISORDERS - CONTINUED



Paige M. Siper, PhD

(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



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

(Associate Professor, Psychiatry, Ophthalmology, and Neuroscience)

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



Dalila Pinto, PhD

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

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



Avi Reichenberg, PhD

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

Research Areas: Autism, schizophrenia, other psychiatric disorders



Andrew J. Sharp, PhD

(Professor, Genetics and Genomic Sciences)

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

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



Pilar Trelles, MD

(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 Professor, Pediatrics, and Neurology)

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

OBESITY AND DIABETES



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

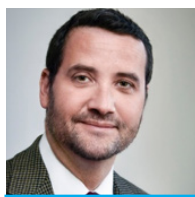


Lauryn Choleva, MD

(Instructor, Pediatrics)

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

OBESITY AND DIABETES - CONTINUED



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



Ruth J.F. Loos, PhD

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

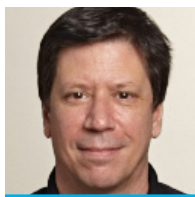
Research Areas: Genetics of obesity and related cardiometabolic traits, genetic epidemiology, epidemiology



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)

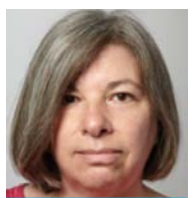
Research Areas: Metabolic regulation of transcription, beta cell regeneration and preservation, diabetes



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, and drug discovery



Susan Teitelbaum, PhD

(Professor, Environmental Medicine & Public Health)

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

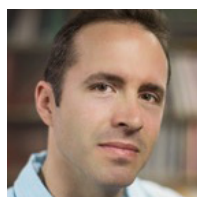
OBESITY AND DIABETES - CONTINUED



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



Ryan W. Walker, PhD

(Assistant Professor, Environmental Medicine & Public Health)

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



Martin J. Walsh, PhD

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

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

OTHER RESEARCH FOCUSES



James J. Bieker, PhD

(Professor, Cell, Developmental & Regenerative Biology)

Research Areas: Transcriptional regulation of gene expression in erythroid cells



Dusan Bogunovic, PhD

(Professor, Microbiology, Oncological Sciences, and Pediatrics)

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



Brian D. Brown, PhD

(Professor, Genetics and Genomic Sciences)

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



John Bucuvalas, MD

(Professor, Pediatrics)

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

OTHER RESEARCH FOCUSES- CONTINUED



Minji Byun, PhD

(Assistant Professor, Medicine)

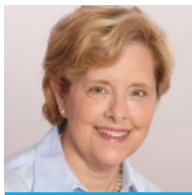
Research Areas: Genetics of immune disorders, clonal hematopoiesis, immune dysregulation



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



David Dunkin, MD

(Associate Professor, Pediatrics)

Research Areas: Tolerance induction and therapeutics in inflammatory bowel disease, mechanisms of inflammatory diseases of the gastrointestinal tract



Chris Gennings, PhD

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

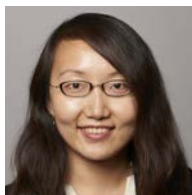
Research Areas: Biostatistical methods development for environmental health



Katherine Guttman, MD, MBE

(Assistant Professor, Pediatrics)

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



Shelley H. Liu, PhD

(Assistant Professor, Population Health Science and Policy)

Research Areas: Biostatistics, environmental mixtures, public health



Kaustav Mukherjee, PhD

(Instructor, Cell, Developmental & Regenerative Biology)

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

OTHER RESEARCH FOCUSES - CONTINUED



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)

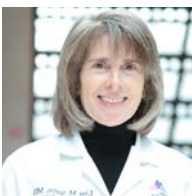
Research Areas: Stem cells, hair regeneration, morphogenesis



Jeffrey M. Saland, MD, MSCR

(Chief, Pediatric Nephrology, and Hypertension)

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 System 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, multi-site and multi-disciplinary 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



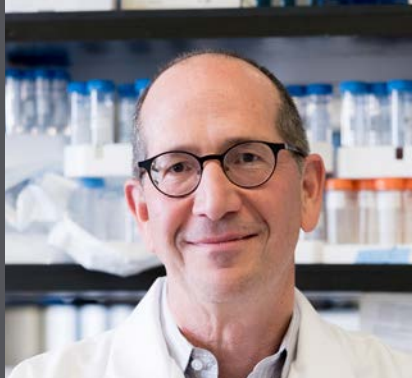
Ernest Turro, PhD

(Associate Professor, Genetics and Genomic Sciences)

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



FACULTY RESEARCH INTERACTIONS



Faculty Highlight: Joseph D. Buxbaum, PhD



Mafalda Barbosa, MD, PhD



Michael S. Breen, PhD



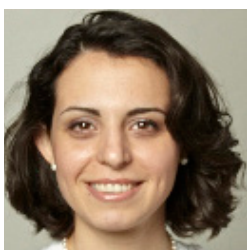
Jennifer Foss-Feig, PhD



Silvia De Rubeis, PhD



Dorothy E. Grice, MD



Hala Harony-Nicolas, PhD



Laura Huckins, PhD



Magdalena U. Janecka, PhD



Alex Klevzon, MD



Hirofumi Morishita, MD, PhD



Avi Reichenberg, PhD



Paige M. Siper, PhD



Shanna H. Swan, PhD

Mafalda Barbosa, MD, PhD

- Collaborator on genetics of neurodevelopmental disorders

Michael S. Breen, PhD

- Mentor
- Collaborator on psychiatric and neurodevelopmental disorders
- Co-investigator in the CommonMind Consortium

Jennifer Foss-Feig, PhD

- Mentor
- Collaborator on translational and clinical autism research

Silvia De Rubeis, PhD

- Mentor
- Collaborator on model systems for autism and autism genetics
- Collaborator on the "Autism Sequencing Consortium"

Dorothy E. Grice, MD

- Collaborator on autism and OCD
- Collaborator on "Rare Genetic Variation and Risk for Obsessive Compulsive Disorder"

Hala Harony-Nicolas, PhD

- Mentor
- Collaborator on model systems for autism

Laura Huckins, PhD

- Collaborator on psychiatric genomics

Magdalena U. Janecka, PhD

- Mentor
- Collaborator on autism epidemiology and epigenetics
- Collaborator on "Maternal health in pregnancy and autism risk – genetic and non-genetic mechanisms"

Alex Klevzon, MD

- Collaborator on translational and clinical autism research
- Collaborator on Rare Disease Clinical Research Network

Hirofumi Morishita, MD, PhD

- Collaborator on rodent models of autism

Avi Reichenberg, PhD

- Collaborator on autism epidemiology
- Collaborator on "Autism Risk and Maternal Cardiometabolic Health (ARCH) study"

Paige M. Siper, PhD

- Mentor
- Collaborator on translational and clinical autism research
- Collaborator on "Treatment and Outreach Programs for Needy Jewish Children with Autism and Related Disorders"

Shanna H. Swan, PhD

- Collaborator on autism epidemiology



Faculty Highlight: Supinda Bunyavanich, MD, MPH



M. Cecilia Berin, PhD



Joseph D. Buxbaum, PhD



Nicole C. Dubois, PhD



Bruce D. Gelb, MD



Megan K. Horton, PhD, MPH



Hugh A. Sampson, MD



Scott H. Sicherer, MD



Andrew J. Sharp, PhD



Julie Wang, MD

M. Cecilia Berin, PhD

- Collaborator, co-investigator on U19AI136053 "Immune Basis & Clinical implications of Threshold-Based Phenotypes of Peanut Allergy"
- Co-author on several papers including "Food allergy and the microbiome: Current understandings and future directions" by Bunyavanich et al. JACI 2019

Joseph D. Buxbaum, PhD

- Co-member of MCHDI Internal Advisory Board

Nicole C. Dubois, PhD

- Co-member of MCHDI Internal Advisory Board

Bruce D. Gelb, MD

- Mentor

Megan K. Horton, PhD, MPH

- Co-chair for Child Health Research Series (past)

Hugh A. Sampson, MD

- Mentor
- Collaborator, co-investigator on U19AI136053 "Immune Basis & Clinical implications of Threshold-Based Phenotypes of Peanut Allergy"
- Co-author on several papers including "Early-life gut microbiome and egg allergy" by Fazlollahi et al. Allergy 2018

Scott H. Sicherer, MD

- Mentor
- Collaborator, co-investigator on U19AI136053 "Immune Basis & Clinical implications of Threshold-Based Phenotypes of Peanut Allergy"
- Collaborator, co-investigator on R01 AI147028 "Gut Microbiome Dynamics in Peanut Allergy"
- Co-author on several papers including "Multidimensional study of the oral microbiome, metabolite, and immunologic environment in peanut allergy" by Ho et al. JACI 2021

Andrew J. Sharp, PhD

- Co-member of MCHDI Internal Advisory Board
- Co-author on several papers including "Integrative transcriptomic analysis reveals key drivers of acute peanut allergic reactions" by Watson et al. Nat Comm 2017

Julie Wang, MD

- Collaborator, co-investigator on U19AI136053 "Immune Basis & Clinical implications of Threshold-Based Phenotypes of Peanut Allergy"
- Co-author on several papers including "Epinephrine autoinjector prescribing patterns in an urban pediatric population" by Sann et al. JACI Pract. 2016



AWARDS/HONORS AND PUBLICATIONS

FACULTY HONORS/AWARDS

Supinda Bunyavanich, MD, MPH, Mount Sinai Endowed Chair, Professor in Allergy and Systems Biology

Jamie Chu, MD, Mount Sinai Endowed Chair, Professor in Pediatric Liver Research

Tirtha K. Das, PhD, RASopathies Network 2021, Junior Investigator Oral Presentation Award

Yuval Itan, PhD, Keynote presentation at the International Symposium on Bioinformatics, December 2021

Amy R. Kontorovich, MD, PhD, 2021 Cullman Family Award for Excellence in Physician Communication

Marek Mlodzik, PhD, Recipient of the Gregor Johann Mendel medal and lecture for "Outstanding Achievement in Science," November 2021

Lisa M. Satlin, MD, Member of the Board of Directors of the American Physician Scientist Association

Lisa M. Satlin, MD, Chair, Research Committee of the Association of Medical School Pediatric Department Chairs

Lisa M. Satlin, MD, Appointed as the US/PSDP representative to the Selection Committee of the Canadian Child Health Clinical Scientist Program

Scott H. Sicherer, MD, Jerome Glaser Distinguished Service Award, Section on Allergy and Immunology, American Academy of Pediatrics

Ernest Turro, PhD, Invited Keynote Speaker, European Mathematical Genetics Meeting (EMGM), Paris, 22-23 April 2021

Ernest Turro, PhD, Invited Speaker in the Educational Session "Bayesian methods applied in clinical settings", European Society of Human Genetics (ESHG) Meeting, Glasgow, August 28-31, 2021

FACULTY PUBLICATIONS

Lozano-Ojalvo D, Tyler SR, **Berin MC**. **Is the plasticity of the th17 subset a key source of allergenic th2 responses?** *Allergy*. 2021 Oct;76(10):3238-40.

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Ogishi M, Yang R, Aytekin C, Langlais D, Bourgey M, Khan T, ... **Bogunovic D**, ... Casanova JL. **Inherited pd-1 deficiency underlies tuberculosis and autoimmunity in a child.** *Nat Med*. 2021 Sep;27(9):1646-54.

Taft J, Markson M, Legarda D, Patel R, Chan M, Malle L, ... **Bogunovic D**. **Human tbk1 deficiency leads to autoinflammation driven by tnf-induced cell death.** *Cell*. 2021 Aug 19;184(17):4447-63.e20.

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Casanova-Acebes M, Dalla E, Leader AM, LeBerichel J, Nikolic J, Morales BM, ... **Brown BD**, ... Merad M. **Tissue-resident macrophages provide a pro-tumorigenic niche to early nscL cells.** *Nature*. 2021 Jul;595(7868):578-84.

Rose SA, Wroblewska A, Dhainaut M, Yoshida H, Shaffer JM, Bektesevic A, ... **Brown BD**. **A microRNA expression and regulatory element activity atlas of the mouse immune system.** *Nat Immunol*. 2021 Jul;22(7):914-927.

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Sikich L, **Kolevzon A**, King BH, McDougale CJ, Sanders KB, Kim SJ, ... **Trelles MDP**, ..., **Siper P**, ... Veenstra-Vander-Weele J. **Intranasal oxytocin in children and adolescents with autism spectrum disorder.** *N Engl J Med.* 2021 Oct 14;385(16):1462-73.

Kontorovich AR, Tang Y, Patel N, Georgievskaya Z, Shadrina M, Williams N, ... **Itan Y**, ... **Gelb BD**. **Burden of cardiomyopathic genetic variation in lethal pediatric myocarditis.** *Circ Genom Precis Med.* 2021 Aug;14(4):e003426.

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Belbin GM, Cullina S, Wenric S, Soper ER, Glicksberg BS, Torre D, ... **Loos RJF**, ... Kenny EE. **Toward a fine-scale population health monitoring system.** *Cell.* 2021 Apr 15;184(8):2068-83.e11.

Huang LO, Rauch A, Mazzaferro E, Preuss M, Carobbio S, Bayrak CS, ... **Itan Y**, ... **Loos RJF**. **Genome-wide discovery of genetic loci that uncouple excess adiposity from its comorbidities.** *Nat Metab.* 2021 Feb;3(2):228-43.

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Ray EC, Carrisoza-Gaytan R, Al-Bataineh M, Marciszyn AL, Nkashama LJ, **Chen J**, ... **Satlin LM**. **L-wnk1 is required for bk channel activation in intercalated cells.** *Am J Physiol Renal Physiol*. 2021 Aug 1;321(2):F245-f54.

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GRANTS

AGENCY_NAME	Funding from New Grants (\$)	Funding from New & Existing Grants (\$)
Pfizer Inc., U.S. Pharmaceuticals Group	2,495,710	2,495,710
National Heart, Lung, And Blood Institute/NIH/DHHS	1,248,245	4,430,819
National Institute Of Allergy And Infectious Diseases/NIH/DHHS	119,053	9,043,693
National Institute Of Diabetes And Digestive And Kidney Diseases/NIH/DHHS	118,165	12,670,744
National Institute Of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS	1,032,756	2,658,234
National Institute Of Mental Health/NIH/DHHS	802,540	7,285,557
National Institute Of Child Health And Human Development/NIH/DHHS	764,170	1,627,742
National Institute Of Neurological Disorders And Stroke/NIH/DHHS	465,058	1,767,137
University of Wisconsin-Madison	276,255	284,730
STEMCELL Technologies	270,951	270,951
Simons Foundation	250,000	250,000
AllerGenis	224,588	224,588
Anonymous	207,654	207,654
Brigham And Women's Hospital	183,178	183,178
National Institute On Aging/NIH/DHHS	167,763	167,763
ImmunityBio, Inc.	125,842	125,842
Massachusetts General Hospital	108,050	108,050
Children's Hospital Pittsburgh	107,400	107,400
Cigna Foundation	100,000	100,000
The BRAIN Foundation	98,646	98,646
National Institute Of Environmental Health Sciences/NIH/DHHS	93,934	4,509,076
Van Andel Institute	82,928	82,928
Oryzon	72,602	72,602
Broad Institute	69,977	69,977
Ann & Robert H. Lurie Children's Hospital of Chicago	56,643	56,643
Akoea Therapeutics, Inc.	55,559	55,559
Washington University School Of Medicine In St. Louis	50,850	50,850
University of Cambridge	50,439	50,439
RTW Charitable Foundation	50,000	50,000
Ball State University	48,356	48,356
Virginia Commonwealth University	42,348	42,348
Washington University	38,138	156,263
Vanderbilt University Medical Center	34,372	11,912
Lab11 Therapeutics LLC	29,257	29,257
Albireo AB	28,812	28,812
Jounce Therapeutics	27,857	27,857
Baylor College Of Medicine	22,035	22,035
University Of California - Davis	20,611	20,611
Benaroya Research Institute At Virginia	12,814	2,334,631
Children's Hospital Med. Center, Ohio	10,000	10,000
National Human Genome Research Institute/NIH/DHHS		2,016,562
National Institute Of General Medical Sciences/NIH/DHHS		1,098,774
Johns Hopkins University Medical School		1,052,063
National Eye Institute/NIH/DHHS		940,158
Aging/NIH/DHHS		852,907
Columbia University		842,999
Albert Einstein College Of Medicine		794,882
National Center for Complementary and Integrative Health/NIH/DHHS		723,980
The Leona M. and Harry B. Helmsley Charitable Trust		535,629
Emory University		380,025
New York State Stem Cell Board		363,550
Rutgers		336,417
Duke (Doris) Charitable Foundation		330,000
University of Pittsburgh		233,164
Klarman Family Foundation		232,042
University of Copenhagen		229,499
Johns Hopkins University		200,001
President and Fellows of Harvard College		180,422
Adventist Health System/Sunbelt, Inc.		144,710
Seaver Foundation		115,000
Food Allergy Research & Education		100,000
University of Washington		66,767
University of North Carolina		66,471
Stanford University		61,708
Cincinnati Children's Hospital		52,110
Tufts University		43,738
New York University, Washington Square		43,578
US-Israel Binational Science Foundation		40,000
Karlstad University		36,023
Brain and Behavior Research Foundation		35,000
Hirschl/Weill-Caulier Trust		35,000
European Commission		31,744
Children's Hospital of Philadelphia		5,725
Total	12,135,554	64,159,242

MATERIAL TRANSFER AGREEMENTS/LICENSES

Research Focus	Outgoing Material Transfer Agreements (#)	Technology Licenses (#)
Neurodevelopmental disorders	3	8
Cardiovascular disease	2	4
Diabetes and Obesity	2	4
Allergy and Asthma	0	0
Others	6	2
Total	13	18

Licenses	Total Number
Antigens/Antibodies	6
Reagents/Methods/Cell Lines	3
Genes/Adapters/Vectors/Oligonucleotides	6
Gene Testing/Therapeutics	3
Total	18



PILOT PROJECTS FUNDED FOR 2020-2021

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.



Keerthi Thirtamara Rajamani, PhD
Postdoctoral Fellow, Psychiatry

Investigator: Keerthi Thirtamara Rajamani, PhD, Postdoctoral Fellow, Psychiatry, Seaver Center for Autism Research and Treatment

Project Title: Oxytocin Modulation of the Supramammillary Nucleus, a Social Novelty Associated Brain Region and Its Role in Autism Spectrum Disorder

Primary Mentor: Hala Harony-Nicolas, PhD, Assistant Professor, Psychiatry, Neuroscience, Seaver Center for Autism Research and Treatment, Mindich Child Health and Development Institute

Secondary Mentor: Hirofumi Morishita, MD, PhD, Associate Professor, Psychiatry, Neuroscience, Ophthalmology, Friedman Brain Institute, Mindich Child Health and Development Institute



Carol L. Shen, MD
Clinical Fellow, Division of Pediatric Nephrology and Hypertension

Investigator: Carol L. Shen, MD, Clinical Fellow, Pediatric Nephrology and Hypertension

Project Title: Aberrant JAK/STAT Signaling in Nephrotic Syndrome

Primary Mentor: Dusan Bogunovic, PhD, Professor of Microbiology, Oncological Sciences, Pediatrics, Mindich Child Health and Development Institute

Secondary Mentors: Jeffrey Saland, MD, MSCR, Professor of Pediatrics, Division of Pediatric Nephrology and Hypertension, Mindich Child Health and Development Institute

FACULTY PILOT AWARDS

Three pilot projects were selected for \$70K in institutional funding for a one-year period starting March 1, 2022. The purpose of the pilot program is to provide MCHDI faculty with funding for initial stages of research projects, with the goal of generating sufficient data to apply for larger, external grants. Projects are encouraged that are likely to a) improve children's health, b) promote collaboration within the MCHDI, and c) leverage additional extramural funding for the Principal Investigators (PIs).

Project Title:

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

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



**Nicole C. Dubois, PhD
(Communicating PI)**

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



Sander Houten, PhD (Co-PI)

*Associate Professor, Genetics
and Genomic Sciences*



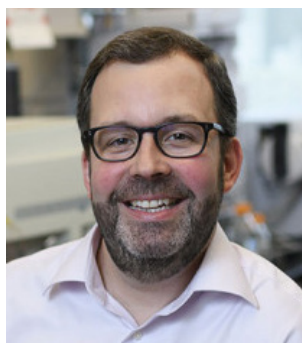
**George Diaz, MD, PhD
(Co-I)**

*Professor, Genetics and
Genomic Sciences and
Pediatrics*



Nenad Bursac, PhD (Co-I)

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



Justin Cross, PhD (Co-I)

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

FACULTY PILOT AWARDS - CONTINUED

Project Title:

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

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



**Rebecca Trachtman, MD
(Communicating PI)**

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



Dusan Bogunovic, PhD (Co-PI)

*Professor, Pediatrics, Microbiology,
and Oncological Sciences
Mindich Child Health and
Development Institute
Precision Immunology Institute*

Project Title:

Whole-Genome Sequencing of Neonatal Fatalities

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



**Ernest Turro, PhD
(Communicating PI)**

*Associate Professor,
Genetics and Genomic
Sciences
Mindich Child Health and
Development Institute
Charles Bronfman Institute
for Personalized Medicine*



Felix Richter, MD, PhD (Co-I)

*Pediatrics Resident Physician
Mindich Child Health and
Development Institute
Pediatrics Residency at The
Mount Sinai Hospital*



**Katherine Guttman,
MD, MBE (Co-PI)**

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



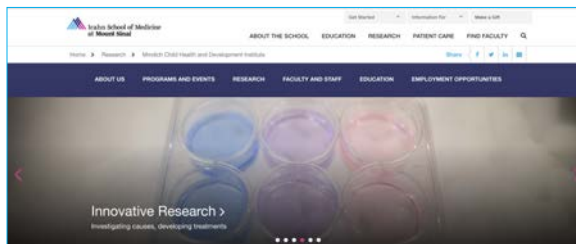
**Mafalda Barbosa, MD, PhD
(Co-PI)**

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

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 bi-annual newsletter distributed internally to faculty, trainees, and other Institute affiliates to highlight important research breakthroughs, publications, awards, and events within MCHDI. View our latest newsletters featured on <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 on 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 >1,000 enrollees. The pediatric cohort is comprised of samples from diverse racial and ethnic groups.

For more information please visit:

www.icahn.mssm.edu/research/institutes/institute-for-personalized-medicine/innovation-and-technology/biome-platform

BIOREPOSITORY CORE SHARED RESOURCE FACILITY

The biorepository CORE facility provides basic histology services such as processing and embedding section fixed and frozen tissues from animal or human sources. In addition, services include DNA/RNA/miRNA extractions, preparing and analyzing tissue microarrays, and supporting functions for tissue procurement, both from consented and anonymized collections.

For a full list of their services visit their website at:

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

STRATEGIC PLAN IMPLEMENTATION

PEDIATRIC CLINICAL TRIALS OFFICE

INTRODUCTION

Mindich Child Health and Development Institute (MCHDI) leadership created a clinical trials program to serve the needs of researchers within the Mount Sinai health system who wish to conduct pediatric clinical trials. The program got off the ground in February 2020 and was immediately put on hold because of the pandemic surge. It was not until Fall 2020 that the program was able to begin recruiting investigators and their trials to offer expertise in regulatory, budgeting, and operations of trials that involve children and their families. We are partnered with the Clinical Trials Office (CTO) in Internal Medicine to provide services to existing and “in development” trials, whether they involve investigator-initiated, industry-funded, or federally funded efforts. Of note, in addition to the traditional services offered by the CTO, we support “extension” trials (adult studies that wish to extend into pediatric populations) and NIH-funded clinical research.

This report covers the period from June 2020 – December 2021 and includes projected growth for 2022.

PCTO STAFF

In March 2020, Yair Bitton was recruited Program Manager for the PCTO, working closely with co-Directors Michele Cohen (head of PCTO) and Dr. Eyal Shemesh (a clinical trials investigator and division chief in Pediatrics) to set the foundation for the program and begin hiring staff. In Spring 2021, we added two part-time employees: Jeanna Johnson-Zalsos was hired to assist with regulatory support, and Michelle Mendiola was hired as research coordinator to support five studies for pediatric GI that were transferred to the PCTO as well as a NICU new trial. In Fall 2021, a part-time finance analyst, Navjot Kaur, was hired to maintain the financial dashboard for all PCTO investigators. On November 1 we hired Catherine Swarts (mainly to support a new NIH funded clinical trial). At the end of 2021, one more clinical research coordinator was recruited in time for a 2022 start date for a large industry funded clinical trial run in a collaboration between the departments of dermatology and pediatrics. We are looking to hire two more staff in the beginning of 2022. Current and “in-recruitment” staff are listed below:

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 (100% NIH Grant)

Pam Pak – Clinical Research Coordinator II (100% NIH Grant)

Michelle Mendiola – Clinical Research II Coordinator (part-time PCTO Funding)

Navjot Kaur – Financial Analyst (part-time); to move to Full Time

Jeanna Zalsos-Johnson – Regulatory Coordinator (part-time); to move off projects in July

RESEARCH FACULTY SERVED BY PCTO

PCTO is currently serving the following investigators / divisions:

Pediatric Divisions:

Allergy

Scott Sicherer, MD

Julie Wang, MD

Amanda Cox, MD

Roxanne Oriel, MD

Mary Grace Baker, MD

Nicole Ramsey, MD

Endocrinology

Joan Han, MD

Robert Rapaport, MD

Gastroenterology

Marla Dubinsky, MD

David Dunkin, MD

Keith Benkov, MD

Nancy Pittman, MD

Nephrology

Jeffrey Saland, MD

Hillary Hotchkiss, MD

Neonatal ICU

Courtney Juliano, MD

Rheumatology

Rebecca Trachtman, MD

Cardiology

Erin Paul, MD

Adult Divisions with pediatric trials

Dermatology

Emma Guttman, MD, PhD

Allergy and Immunology

Paula Busse, MD

Cardiology (Heart Center)

Amy R. Kontorovich, MD, PhD

CLINICAL TRIALS PORTFOLIO

Notable Projects

- **NIH:** PCTO is involved in the implementation and administration of an NIH funded, 7 year, multi-million-dollar consortium project.
- **Adult trials with pediatric extensions:** one of the PCTO missions is to enhance adult-pediatric collaborations, which is a potential but largely untapped strength of the Mount Sinai Health System (as compared with stand-alone pediatric hospitals). This is done in collaboration with the Medicine CTO (MCTO). The PCTO has already been successful in supporting those “across the lifecycle” specific types of clinical trials:
 - Through an investigator-initiated application of an extension trial of a drug that is currently only approved in adults, the PCTO has helped secure over \$1 million in funding during 2021 for an approved allergy medication used in a novel setting.
 - PCTO/MCTO collaborated with Pediatric Nephrology on another adult/pediatric trial evaluating the efficacy and safety of Lumasiran in children and adults with primary hyperoxaluria type 1 (PH1).
 - PCTO has negotiated and finalized contracting on a heart center trial (PI – Dr. Kontorovich).
- **Development and support of clinical trials in divisions who previously were not engaged in such activity:** For example, PCTO ushered in the first (out of many, we hope) clinical trial in the NICU, working closely with the NICU chief to create the infrastructure as well as mindset to support clinical trials in this particularly promising and important division.

Clinical Trials Open to Enrollment (11)

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

Clinical Trials in Start-up phase (23)

Pediatrics

Allergy – 6
Gastroenterology – 6
Endocrinology – 7
Nephrology – 1
Cardiology – 1
Rheumatology – 1

Allergy and Immunology (Adult/Peds) – 1 sub-contract for existing U01 awarded but still in start-up

CENTER FOR INBORN ERRORS OF IMMUNITY

The Center for Inborn Errors of Immunity (CIEI) seeks to illuminate the elusive pathogenesis behind 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 to seed the discovery and research enterprise surrounding our mission.

Dr. Horowitz, Assistant Professor of Oncological Sciences and a member of CIEI, received \$20,000 to study "The role of HLA class I polymorphisms on NK cell development and susceptibility to bone marrow failure and acute myeloid leukemia."

Dr. Miorin, Assistant Professor in Microbiology, and a member of CIEI, received \$20,000 to study "Role of ANKEF1 deficiency in life-threatening respiratory infection."

Symposia

CIEI has co-organized the 3rd and 4th NYC Inborn Errors of Immunity meetings, where over 100 participants listened to lectures from the 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 such as 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 Model Core.

The Functional Genomics and Disease Modeling Core was established to leverage the strengths of the *Drosophila* genetic system and develop whole animal fly models of rare gene variants found in pediatric and other rare disease indications. The core uses multiple established transgenic approaches as well as newly developed assays to provide insights about the mechanism of action of these variants in human disease.

Building on our past multi-disease fly model expertise, over the last year the core has developed 20 new RASopathy fly models for various genes in the MAPK pathway including variants for *MEK*, *RIT1*, *SOS*, and *SHOC2*. The core has also started to develop models of variants implicated in other rare non-RASopathy diseases. Currently the focus is on a couple of gene variants identified as part of the UDP and rarely implicated in other diseases, like ones in *NDUFAF* and *MAGI*.

In low-occurrence cancers, rare gene variants also arise in tumors of patients receiving anti-cancer drug therapy, which leads to reduced drug efficacy. The core has developed five such models of rare kinase-fusion gene variants that arise in patients undergoing targeted cancer therapy.

These ongoing studies in conjunction with the core have provided key new insights about the mechanism of action of these RASopathy and rare cancer variants, which will serve as leads for ongoing and future studies in vertebrate models.

In addition, two MCHDI faculty, Jennifer Bragg and Bruce Gelb, are serving as site Principal Investigators for a clinical trial called Genomic Medicine for Ill Neonates and Infants (GEMINI). This multi-site study, which recently completed recruitment, is comparing the utility of a newborn-specific targeted gene panel to rapid genome sequencing for critically ill patients under 1 year of age who are suspected to have an underlying genetic disorder. While the official results of GEMINI are still pending, the experience for families and the Newborn Medicine staff was extremely positive. This led to Mount Sinai Hospital approving the use of rapid genome sequencing clinically as a 4-month trial, which is underway.



Icahn
School of
Medicine at
Mount
Sinai

*The Mindich
Child Health and
Development Institute*

LEADERSHIP AND STAFF

Bruce D. Gelb, MD-Director
Elena Lum, PhD-Administrative Director
Risa Slaughter-Administrative Manager

Shavez Jackson-Administrative Coordinator
Jennifer E. Cole-Senior Director of Development

FACULTY

Mafalda Barbosa, MD, PhD
M. Cecilia Berin, PhD
Harold S. Bernstein, MD, PhD
James J. Bieker, PhD
Dusan Bogunovic, PhD
Jennifer Bragg, MD
Michael S. Breen, PhD
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