Mount Sinai

The Mindich Child Health and Development Institute

Annual Report 2019

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.

Contents

Message From the Director

In 2019, the Mindich Child Health and Development Institute (MCHDI) took substantial steps to expand its mission and execute two major strategic goals to establish a Pediatric Clinical Trials Office and Inborn Errors of Immunity Center as well as continue efforts to recruit key faculty to support ongoing initiatives. The MCHDI family has grown to 66 faculty members who pursue research relevant to a broad range of health issues for infants, children and adolescents. Our new members include two faculty recruited extramurally (Katherine Guttmann, MD, MBE and Maria Curotto de Lafaille, PhD) and five Mount Sinai faculty (Michael S. Breen, PhD, Magdalena U. Janecka, PhD, Florence Marlow, PhD, Pilar Trelles, MD, and Ryan W. Walker, PhD). Our members have been actively engaged in research, contributing to over 330 publications this past year.

In 2019, the MCHDI continued to make significant progress in the implementation of our strategic plan. One of our initiatives centers on developing the infrastructure to support clinical trials with pediatric subjects, both interventional and observational ones. We identified two experienced MCHDI investigators, Drs. Eyal Shemesh and Karen M. Wilson, to lead this new endeavor. They organized a working group that prioritized our needs and goals. Emerging from this was an exciting partnership with the Department of Medicine's Clinical Trials Office (CTO). Working with Michele Cohen, the Administrative Director of the CTO, we identified new space for additional administrative staff, which will provide the capacity for overseeing pediatric clinical trials. A search is currently underway to identify a manager for the pediatric clinical trials resource. Once that person has begun, we anticipate phasing in this resource for all investigators wishing to utilize its services over the next 12 months. The goals are to facilitate start-up and maintenance of pediatric clinical trials, improve economic efficiency, and provide enhanced assistance for the burdensome regulatory aspects of these activities. In addition, we anticipate collaborations with Annetine Gelijns and her colleagues at the International Center for Health Outcomes and Innovation Research, who provide expertise in novel trial design, a potentially fruitful approach for the rarer traits that we tend to study in children.

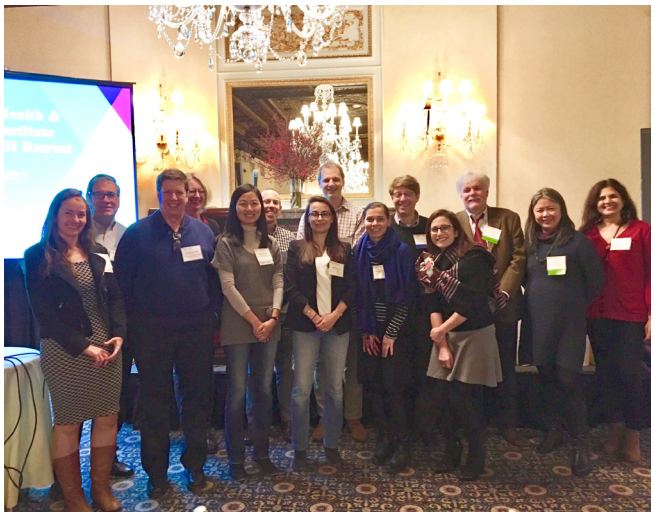
For the pediatric precision medicine initiative, we also made significant advances in the past year. Our Undiagnosed Diseases Program continues to enroll patients with perplexing disorders that appear to be genetic. Now under the leadership of Bryn D. Webb, MD, the program has solved several cases this year, including ones leading to novel gene discoveries. The NHGRI-funded project NYCKidSeq (Mount Sinai's Principal Investigators are Carol Horowitz, Eimear Kenny and myself), which is studying a novel method for communicating genetic testing results, has been recruiting vigorously from diverse New York City populations who tend to be underserved in genomic medicine. Dr. Annemarie Stroustrup is the site Principal Investigator on a project called GEMINI, a subaward from Tufts for an NCATS-funded project, that is comparing a new gene resequencing panel to rapid genome sequencing for newborns and young infants with apparently genetic disorders. Recruitment for GEMINI has also been robust this year. Working with Dr. George Diaz, Chief of Clinical Genetics as well as the leadership of Pediatric Medical Education, we are initiating a novel educational program in pediatric genomic medicine for Mount Sinai's pediatric trainees, which will roll out in the spring of 2020. Finally, we are in the midst of some faculty recruitment that will enhance our research efforts in pediatric precision medicine.

Last year, the MCHDI announced the creation of the Center for Inborn Errors of Immunity, which is a joint venture with the Precision Immunology Institute. When Isabel Meyts, whom we had recruited from Belgium to lead this, needed to withdraw her acceptance for personal reasons, we were delighted to name Dusan Bogunovic, PhD, an outstanding MCHDI faculty member who is a rising star in the field of inborn errors of immunity, as the new Director. His vision is to focus this new Center on the molecular and immunological understanding of these rare immunogenetic traits that present as severe infections or autoinflammatory disorders. The Center will aim at improved diagnosis of these rare diseases as well as at offering therapeutic options for reducing their morbidities. Joining him at the Center from its outset will be three other MCHDI members—Minji Byun, PhD, Charlotte Cunningham-Rundles, MD, PhD, and David Dunkin, MD.

Bruce D. Gelb, MD, Director

Faculty Growth

Chart of faculty recruits since our inception in 2009. In 2019, our Institute recruited two new external and five internal 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 Mount Sinai), PhD; Bruce D. Gelb, MD; Amy R. Kontorovich, MD, PhD; Martin J. Walsh, PhD; Jia Chen, ScD; Rupangi C. Vasavada, PhD (no longer at Mount Sinai)

Chart of faculty recruits since our inception in 2009. In 2019, our Institute recruited two new external and five internal faculty members to our Institute.

New Faculty

New Extramural Faculty

Katherine Guttmann, MD, MBE
Assistant Professor, Pediatrics

Katherine Guttmann, MD, MBE

Katherine Guttmann, MD, MBE, is an Assistant Professor at the Icahn School of Medicine and an attending neonatologist in the Mount Sinai Health System. She completed medical school at the University of Pennsylvania, followed by residency and fellowship at the Children's Hospital of Philadelphia where she also served as Chief Fellow. Dr. Guttmann conducts research focusing on ethics and communication with families. Past work has investigated parental perspectives on conversations related to diagnosis and prognosis of cerebral palsy, success of goals of care discussions in the NICU, parents' experiences of research participation, and telemedicine as a novel means of improving communication with families in the NICU. Additional academic interests include Neonatal Palliative Care and Research Ethics. She holds a Master's degree in Bioethics from the University of Pennsylvania, which informs her work. She is committed to understanding and improving the family experience of NICU hospitalization.

Maria Curotto de Lafaille, PhD
Associate Professor, Pediatrics

Maria Curotto de Lafaille, PhD

Maria Curotto de Lafaille, PhD, is an Associate Professor of Pediatrics and member of the Jaffe Food Allergy Institute and the Precision Immunology Institute at the Icahn School of Medicine. Dr. Lafaille obtained her PhD degree in Immunology from the University of São Paulo in Brazil, and trained as a postdoctoral fellow in Infectious Diseases at Harvard University. Before joining Mount Sinai, Dr. Lafaille held faculty positions at the Agency for Science, Research and Technology in Singapore and at New York University Medical School. Dr. Lafaille has a long-standing interest in allergic diseases, having made important contributions to the understanding of basic mechanisms of mucosal tolerance and allergic sensitization. Among them was the pioneer work on the essential role of outside-thymus induced regulatory T cells in the prevention of allergic inflammation. Studies on the mechanisms of IgE regulation in mice revealed unique aspects of the differentiation of IgE-producing cells that generated new paradigms for the allergy field. Current studies in Dr. Lafaille's laboratory aim to elucidate the mechanisms that maintain the B cell memory of allergic responses in mice and human. In collaboration with colleagues at the Jaffe Food Allergy Institute and the Precision Immunology Institute, her group works to understand how immunological memory shapes the evolution of food allergy toward resolution or persistence of the disease.

New Intramural Faculty

Michael S. Breen, PhD
Assistant Professor, Psychiatry
Assistant Professor, Genetics and Genomic Sciences

Michael S. Breen, PhD

Michael S. Breen, PhD, is an Assistant Professor in the Departments of Psychiatry and Genetics and Genomic Sciences. Dr. Breen was a Computational Biologist at the Center of Genomic Regulation (Barcelona, ES) where he developed statistical methods to measure epistatic interactions and their influence on complex traits. He later received a PhD in Genomics and Bioinformatics from the University of Southampton, UK, with a focus on neuropsychiatric traits. As a postdoctoral fellow at the Icahn School of Medicine, his research has been sponsored by the NARSAD Brain and Behavior Research Foundation, the Autism Science Foundation, the National Institute of Mental Health, and the Beatrice and Samuel A. Seaver Foundation. Dr. Breen's research is at the intersection of genomics and neuroscience, utilizing a number of transcriptomic (e.g. RNA-Seq, single cell sequencing, long-read sequencing) and functional genomic approaches to investigate gene expression, RNA editing, and function in the human brain and in neurodevelopmental disorders. His laboratory generates genomic data sets from patient-derived tissues, including iPSC-derived neurons, postmortem brain tissue, peripheral blood and cord blood, and subsequently analyzes these data under a prism of computational and biostatistical methods. In doing so, his work strives to construct biologically plausible mechanistic models of disease, which can be validated both functionally and clinically.

Magdalena U. Janecka, PhD
Assistant Professor, Psychiatry

Magdalena U. Janecka, PhD

Magdalena U. Janecka, PhD, is an Assistant Professor in the Department of Psychiatry, and a member of the Seaver Autism Center. Dr. Janecka received her undergraduate degree from the University of St. Andrews, Scotland (double major in Psychology and Biology), and subsequently completed her PhD in Social, Genetic, and Developmental Psychiatry at King's College London. After her first postdoctoral position at the University of Oxford, she was awarded the Seaver Foundation Postdoctoral Fellowship, and moved to the Icahn School of Medicine in 2016. Dr. Janecka's research focuses on understanding why certain parental and early-life factors are associated with a higher risk of neurodevelopmental disorders in children. In order to better understand the causal mechanisms underlying this risk, she integrates insights from epidemiology, epigenetics, and genetics. The goal of her research is to elucidate how the environments impact long-term developmental outcomes, contributing to identification of modifiable risk factors, prevention, and patient stratification.

New Intramural Faculty continued

Florence Marlow, PhD (Associate Professor, Cell, Developmental & Regenerative Biology)

Florence Marlow, PhD

Florence Marlow, PhD, is an Associate Professor in the Cell, Development, & Regenerative Biology Department at the Icahn School of Medicine at Mount Sinai. She serves as Co-Director of the Development, Regeneration, and Stem Cells Graduate Training Area and Associate Director of the MSTP program. The Marlow lab uses genetic, molecular, cell biological, and embryological approaches to investigate the molecular pathways and cell biological events that regulate specification and maintenance of the first embryonic axes and germline, and that maintain polarity and function in oocytes and in neurons. Her lab identified RNAbps that interact with a key regulator of oocyte polarity and identified novel factors required for sex-specific differentiation of germline cells. In addition, her group has used genetic approaches in zebrafish to investigate the development of the nervous system and generate new animal models of disease. Dr. Marlow graduated from Rensselaer Polytechnic Institute with a Bachelor of Science. She earned her PhD in Molecular Biology from Vanderbilt University. She was a recipient of a Damon Runyun Postdoctoral Fellowship and completed her postdoctoral training at the University of Pennsylvania. She joined the Mount Sinai faculty in 2016.

Pilar Trelles, MD
Assistant Professor, Psychiatry

Pilar Trelles, MD

Pilar Trelles, MD, is a child and adolescent psychiatrist, and Assistant Professor of Psychiatry. She splits her time between conducting patient-oriented research at the Seaver Autism Center at Mount Sinai Hospital, and providing clinical care for individuals with developmental disabilities at the Developmental Disability Center at Mount Sinai West, where she leads the psychiatric clinic. Dr. Trelles' research and clinical work aims to address care and research disparities in NDD. As such, she works closely with community agencies, both locally and internationally, treating children with ASD and other NDDs, to address inequalities in patient care and ethnic disparities in genomic research. Dr. Trelles enjoys teaching, and has been an invited speaker in regional, national, and international conferences.

Ryan W. Walker, PhD
Assistant Professor, Environmental Medicine & Public Health

Ryan W. Walker, PhD

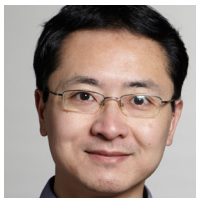
Ryan W. Walker, PhD, is an Assistant Professor in the Department of Environmental Medicine & Public Health at the Icahn School of Medicine at Mount Sinai. He is a clinical obesity and type 2 diabetes scientist with expertise in clinical nutrition, exercise physiology, and the genetics of metabolic disorders. He has extensive experience in clinical interventions. Dr. Walker is currently studying the early establishment of the infant gut microbiome and the role of intestinal bacteria in the etiology of obesity and related diseases. He has particular interests in the relationships between diet, microbiome and host health, and the identification of modifiers of the gut microbiome to establish targets for interventions that improve health outcomes and prevent disease.

Faculty Research Areas

Asthma and Allergy

M. Cecilia Berin, PhD (Professor, Pediatrics)
Research Areas: Immune mechanisms of food allergy and regulation of immune tolerance

Supinda Bunyavanich, MD, MPH (Associate Professor, Pediatrics, and Genetics and Genomic Sciences)
Research Areas: Integrative genomics of asthma and allergic diseases



Ke Hao, ScD (Associate Professor, Genetics and Genomic Sciences)
Research Areas: Genetic pleiotropy, mendelian randomization, inflammatory bowel disease, placenta biology, ambient air particulate matter exposure

Hugh A. Sampson, MD (Kurt Hirschhorn Professor, Pediatrics)
Research Areas: Immunopathogenesis of food allergy and anaphylaxis

Scott H. Sicherer, MD (Director, Jaffe Food Allergy Institute; Division Chief, Pediatric Allergy; Elliot Roslyn Jaffe Professor, Pediatrics)
Research Areas: Food allergy epidemiology, treatments, natural course, quality of life

Julie Wang, MD (Professor, Pediatrics)
Research Areas: Novel therapeutics for food allergy, epidemiology and management of food allergy in minority, urban populations

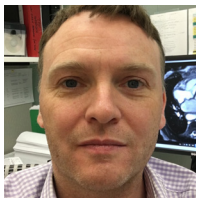
Karen M. Wilson, MD, MPH (Debra and Leon Black Division Chief, General Pediatrics; Vice-Chair for Clinical and Translational Research, Pediatrics; Professor, Pediatrics)
Research Areas: Secondhand tobacco smoke, secondhand marijuana smoke, inpatient respiratory illness

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

Anne Moon, MD, PhD (Adjunct Professor, Pediatrics)
Research Areas: Developmental biology of congenital heart disease and limb defects, functions of Tbx and fibroblast growth factors

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

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

Neurodevelopmental Disorders

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

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

Lisa Eiland, MD (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-omit eQTL-based methodologies.

Neurodevelopmental Disorders continued

Magdalena U. Janecka, PhD (Assistant Professor, Department of 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

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

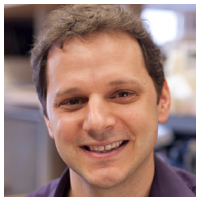
Marek Mlodzik, PhD (Professor and Chair, Cell, Developmental & Regenerative Biology; Professor, Ophthalmology and Oncological Sciences)
Research Areas: Genetics and cell biology of planar cell polarity establishment, cell biology of Wnt-signaling and Notch-signaling

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

Neurodevelopmental Disorders continued

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

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, non-coding RNA



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

Annemarie Stroustrup, MD, MPH (Associate Professor, Pediatrics, Obstetrics, Gynecology and Reproductive Science, 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

Pilar Trelles, MD (Assistant Professor, Psychiatry)
Research Areas: Autism spectrum disorder, neurodevelopmental disorders, health disparities

Bryn D. Webb, MD (Assistant Professor, Genetics and Genomic Sciences and Pediatrics)
Research Areas: Identifying genetic etiologies of congenital anomalies, mitochondrial disorders, undiagnosed disease

Anusha Yeshokumar, MD (Assistant Professor, Neurology and Pediatrics)
Research Areas: Autoimmune encephalitis, outcomes research, inflammatory biomarkers, cognition, behavior

Obesity and Diabetes

Ross L. Cagan, PhD (Director, Center for Personalized Cancer Therapeutics; Professor, Cell, Developmental & Regenerative Biology, Oncological Sciences, and Ophthalmology)
Research Areas: Drosophila as a tool to develop therapeutics for cancer, diabetes, and rare mendelian diseases

Adolfo García-Ocaña, PhD (Professor, Medicine)
Research Areas: Diabetes, pancreatic beta cell biology

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

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

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

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

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

Psychiatric Disorders

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

Dorothy E. Grice, MD (Professor, Psychiatry)
Research Areas: Genetic and epidemiological studies of OCD, Tourette disorder, autism, and related childhood-onset neuropsychiatric disorders, prenatal exposures, including smoking, functional analysis of identified risk genes

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

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

Other Research Focuses

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

Dusan Bogunovic, PhD (Associate Professor, Microbiology, 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, and biotechnology

Other Research Focuses continued

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

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

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

Charlotte Cunningham-Rundles, MD, PhD (David S. Gottesman Professor, Medicine; Professor, Pediatrics)
Research Areas: Primary Immune Deficiency, B cells, antibody, B cell memory, hypogammaglobulinemia, immune reconstitution

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

Chris Gennings, PhD (Professor, Environmental Medicine & Public Health, 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

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

Other Research Focuses continued

Shelley H. Liu, PhD (Assistant Professor, Population Health Science and Policy)
Research Areas: Biostatistics, environmental mixtures, public health

Michael Rendl, MD (Professor, Cell, Developmental & Regenerative Biology, and Dermatology)
Research Areas: Stem cells, hair regeneration, morphogenesis

Jeffrey M. Saland, MD (Associate Professor, Pediatrics)
Research Areas: Kidney disease in children, lipoprotein metabolism in children with CKD, hemolytic uremic syndrome

Lisa M. Satlin, MD (Herbert H. Lehman Professor and System Chair, Pediatrics)
Research Areas: Ontogeny and mechanoregulation of epithelial ion channels in secretory epithelia, 3D bioprinting of kidney tubules

Rebecca Trachtman, MD (Assistant Professor, Pediatrics)
Research Areas: Biomarkers, patient-reported outcomes in juvenile idiopathic arthritis

Faculty Research Interactions

Vignette Highlight: Eyal Shemesh, MD

Bruce D. Gelb, MD

Gogel Family Professor and Director, MCHDI

Professor, Pediatrics

Professor, Genetics and Genomic Sciences

Collaborating on launch of pediatric clinical trials office

Karen M. Wilson, MD, MPH

Debra and Leon Black Chief, Division of General Pediatrics

Vice-Chair, Clinical and Translational Research

Professor, Pediatrics

Eyal Shemesh, MD

Chief, Division of Behavioral and Developmental Health

Director, Child Health Clinical Trials Service

Professor, Pediatrics

Professor, Psychiatry

Partnering to implement patient-reported outcomes into the work of the pediatric liver transplant clinic

Collaborating on the EMPOWER program to study and support patients living with food allergy (PMID:28117270)

Jaime Chu, MD

Assistant Professor, Pediatrics

Collaborating on clinical trial to improve medical adherence in pediatric liver transplant recipients (NIDDK UO1)

John Bucuvalas, MD

Chief, Division of Hepatology

Director, Solid Organ Transplant Outreach for the Recananti-Miller Transplant Institute

Vice Chair of Faculty Affairs, Pediatrics

Professor, Pediatrics

Scott H. Sicherer, MD

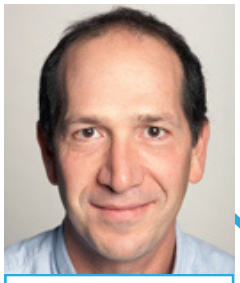
Director, Jaffe Food Allergy Institute

Division Chief, Pediatric Allergy

Elliot Roslyn Jaffe Professor, Pediatrics

Faculty Research Interactions

Vignette Highlight: Silvia De Rubeis, PhD

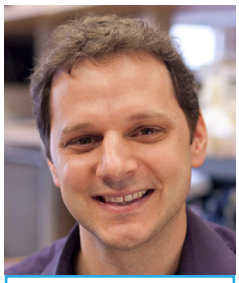


Joseph D. Buxbaum, PhD

Director, Seaver Autism Center

Professor, Psychiatry, Neuroscience, and Genetic and Genomic Sciences

Collaborating on defining the genetic bases of autism spectrum disorder



Andrew J. Sharp, PhD

Associate Professor, Genetics and Genomic Sciences

Collaborating on identifying blood-based epigenetic signatures in rare genetic neurodevelopmental disorders

Michael S. Breen, PhD

Assistant Professor, Psychiatry and Genetics and Genomic Sciences

Collaborating on integrating pre-clinical and clinical findings in DDX3X syndrome

Dorothy E. Grice, MD

Professor, Psychiatry

Silvia De Rubeis, PhD

Assistant Professor, Psychiatry

Collaborating on integrating pre-clinical and clinical findings in ADNP syndrome

Paige M. Siper, PhD

Assistant Professor, Psychiatry

Awards and Publications

Faculty Awards/Honors

Supinda Bunyavanich, MD, MPH, Castle Connolly Exceptional Woman in Medicine 2019

Joseph D. Buxbaum, PhD, Fellow of the International Society for Autism Research

Jaime Chu, MD, AASLD Foundation, AASLD Foundation Bridge Award

Bruce D. Gelb, MD, President of the American Pediatric Society

Bruce D. Gelb, MD, Treasurer of the American Society of Human Genetics

Dorothy E. Grice, MD, Distinguished Fellow of the American Academy of Child and Adolescent Psychiatry (AACAP)

Hala Harony-Nicolas, PhD, Friedman Brain Institute Scholar Award, "Implication of the Hypothalamic Oxytocin System in Autism-Associated Social Deficits"

Amy R. Kontorovich, MD, PhD, Department of Medicine at Mount Sinai, Junior Faculty Translational Collaborative Research Initiative Award

Robert S. Krauss, PhD, Mount Sinai Student Body, 2019 Outstanding Teaching by a Faculty Member Award

Andrew J. Sharp, PhD, ASHG conference, Plenary session, "PgmNr95: A survey of epigenetic variation in >23,000 individuals identifies many disease-relevant epimutations and novel CGG expansions"

Trainee Honors/Awards

Carolina Cappi, PhD, PI: Dalila Pinto, Mount Sinai, MCHDI Pilot Grant, "Assessing the role of microRNAs in Obsessive-Compulsive Disorder (OCD)"

Conor Gruber, PI: Dusan Bogunovic, Mount Sinai, MCHDI Pilot Grant, "Assessing the Role of Monoallelic Expression in Primary Immunodeficiency"

Corina Lesseur, MD, PhD, PI: Jia Chen, NICHD, K99, "Integrative Analysis of Human Placental Epi/genome in Relation to Fetal Growth"

Anna S. Rommel, PhD, PI: Shanna Swan, Mount Sinai, Promising Young Investigator (Travel) Award

Publications

Gowthaman U, Chen JS, Zhang B, Flynn WF, Lu Y, Song W, ... **Berin MC**, ... Eisenbarth SC. **Identification of a t follicular helper cell subset that drives anaphylactic ige.** *Science*. 2019 Aug 30;365(6456).

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Grants

Agency	Funding from New Grants ($)	Funding from Existing & New Grants ($)
National Institute Of Mental Health/NIH/DHHS	$2,529,585	$7,196,683
National Institute Of Child Health And Human Development/NIH/DHHS	$1,005,002	$1,845,729
National Institute Of Allergy And Infectious Diseases/NIH/DHHS	$821,533	$7,022,649
National Institute Of Arthritis & Musculoskeletal & Skin Diseases/NIH/DHHS	$471,771	$1,276,985
Helmsley Foundation	$414,682	$414,682
Johns Hopkins University Medical School	$414,603	$414,603
Duke (Doris) Charitable Foundation	$330,000	$330,000
Alnylam Pharmaceuticals, Inc.	$280,490	$280,490
University of Copenhagen	$225,000	$225,000
Johns Hopkins University	$200,000	$200,000
Astellas Pharma US, Inc.	$197,392	$197,392
Sean Parker Foundation	$179,622	$179,622
Simons Foundation	$150,000	$150,000
Benaroya Research Institute At Virginia	$138,577	$2,215,772
Rainwater Charitable Foundation	$133,334	$133,334
University Of Michigan	$116,531	$116,531
Seaver Foundation	$115,000	$115,000
American Association For The Study Of Liver Diseases	$100,000	$100,000
Harvard University	$82,800	$82,800
Children's Medical Center of Dallas	$81,700	$81,700
University Of Pittsburgh	$76,683	$212,283
University Of Pennsylvania	$74,830	$74,830
American Epilepsy Society	$50,000	$50,000
Childhood Arthritis and Rheumatology Research Alliance	$50,000	$50,000
Brain and Behavior Research Foundation	$35,000	$70,000
Hirschl/Weill-Caulier Trust	$35,000	$70,000
CSL Behring	$33,161	$33,161
European Commission	$31,744	$31,744
Regeneron Pharmaceuticals, Inc.	$9,011	$9,011
National Center for Advancing Translational Sciences/NIH/DHHS		$6,288,994
National Institute Of Diabetes And Digestive And Kidney Diseases/NIH/DHHS		$5,714,042
National Institute Of Environmental Health Sciences/NIH/DHHS		$5,019,859
National Human Genome Research Institute/NIH/DHHS		$3,784,706
National Heart, Lung, And Blood Institute/NIH/DHHS		$2,920,907
Office of the Director, National Institutes of Health/NIH/DHHS		$1,987,420
National Institute Of Neurological Disorders And Stroke/NIH/DHHS		$1,433,468
National Eye Institute/NIH/DHHS		$1,180,882
National Institute Of General Medical Sciences/NIH/DHHS		$1,014,329
Albert Einstein College Of Medicine		$924,479
National Cancer Institute/NIH/DHHS		$677,685
Columbia University		$539,777
National Institute For Occupational Safety & Health/CDC/DHHS		$499,991
National Institute Of Dental And Craniofacial Research/NIH/DHHS		$497,855
New York State Stem Cell Board		$363,550
Emory University		$343,668
National Institute On Drug Abuse/NIH/DHHS		$211,875
American Academy Of Pediatrics		$165,197
University Of Southern California		$149,999
American Diabetes Association, Inc.		$115,000
Boston Children's Hospital		$114,016
Cancer Research Institute, Inc.		$100,000
University Of North Carolina		$83,088
Immune Deficiency Foundation		$82,272
University Of Washington		$80,192
Moebius Syndrome Foundation		$80,000
Stanford University		$61,726
University Of California, San Francisco		$48,378
Tufts University		$36,288
Duke University		$34,190
SIDRA MEDICAL AND RESEARCH CENTER		$15,686
Total	**$8,383,051**	**$57,749,520**

Material Transfer Agreements

Research Focus	Outgoing Material Transfer Agreements (#)	Technology Licenses (#)
Neurodevelopmental disorders	0	6
Cardiovascular disease	0	5
Diabetes and Obesity	8	4
Allergy and Asthma	0	5
Others	1	1
Total	**9**	**21**

Licenses

Licenses	Total Number
Antigens/Antibodies	7
Reagents/Methods/Cell Lines	6
Genes/Adapters/Vectors/Oligonucleotides	6
Gene Testing/Therapeutics	2
Total	**21**

Pilot Projects Funded for 2019-2020

Trainee Pilot Awards

Two new trainee awardees were selected for the second annual trainee pilot program in the amount of $10K over a one-year period. The purpose of the program is to support postdoctoral/clinical fellows or PhD/MD-PhD students in pursuing an independently funded research project that is separate from their ongoing projects under their current Principal Investigator/mentor. Successful 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.

Investigator:	Carolina Cappi, PhD, Postdoctoral fellow, Department of Psychiatry
Project Title:	Assessing the role of microRNAs in Obsessive-Compulsive Disorder (OCD)
Primary Mentor:	Dalila Pinto, PhD, Assistant Professor of Psychiatry, and Genetics and Genomic Sciences
Secondary Mentor:	Thomas V. Fernandez, MD, Assistant Professor in the Yale Child Study Center and of Psychiatry, Yale University School of Medicine, New Haven

Investigator:	Conor Gruber, MD, PhD Candidate, Department of Microbiology and Precision Immunology Institute
Project Title:	Assessing the Role of Monoallelic Expression in Primary Immunodeficiency
Primary Mentor:	Dusan Bogunovic, PhD, Associate Professor in the Department of Microbiology, Department of Pediatrics
Secondary Mentor:	Brad Rosenberg, MD, PhD, Assistant Professor in the Department of Microbiology

Carolina Cappi, PhD
Postdoctoral fellow, Department of Psychiatry

Conor Gruber, MD, PhD
MD, PhD Candidate, Department of Microbiology and Precision Immunology Institute

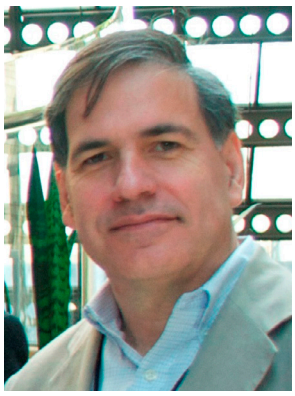
Pilot Projects Funded for 2019-2020 continued

Faculty Pilot Awards

Three pilot projects were selected for $70K in institutional funding for a one-year period starting January 31, 2020. The purpose of the pilot program is to provide MCHDI faculty with funding for initial stages of research projects, with the goal of generating sufficient data to apply for larger, external grants. Projects are encouraged that are likely to: a) improve children's health, b) promote collaboration within the MCHDI, and c) leverage additional extramural funding for the Principal Investigators (PIs).

Principal Investigators: Adolfo García-Ocaña, PhD, and Sarah A Stanley, MBBCh, PhD
Project Title: Neural control of pancreatic endocrine function in the development of type 1 diabetes

Principal Investigators: Jaime Chu, MD, Lauren M. Petrick, PhD, and Sanjiv Harpavat, MD, PhD
Project Title: Identifying the role of early environmental toxicants in newborns with biliary atresia



Adolfo García-Ocaña, PhD
Professor, Medicine

Sarah A. Stanley, MBBCh, PhD
Assistant Professor, Medicine

Jaime Chu, MD
Assistant Professor, Pediatrics
Associate Chief, Division of Pediatric Hepatology
Director, Pediatric Physician-Scientist Residency Program

Lauren M. Petrick, PhD
Assistant Professor and Head of Metabolomics, Environmental Medicine & Public Health

Sanjiv Harpavat, MD, PhD
Assistant Professor, Department of Pediatrics
Texas Children's Hospital
Baylor College of Medicine

Annual Retreat

The 7th Annual MCHDI Retreat was held at the Harmonie Club on November 12, 2019, with over 100 faculty members, trainees, and guests in attendance. The retreat planning committee this year was comprised of faculty members: Adolfo García-Ocaña, Hala Harony-Nicolas, Jeff Saland and Trainee Leadership Committee member Oscar Rodriguez. We introduced a new live polling application to introduce our speakers, and incorporated a lunch survey competition. Dr. Bruce Gelb moderated the panel on big data resources with panelists Joseph Finkelstein, MD, PhD, Shelley Liu, PhD, and Andrew J. Sharp, PhD. We awarded the winners of our Young Investigators Competition: Sharon Alterzon-Bamuel, PhD (Postdoctoral Division, PI: Donald Scott, PhD) and Justin Taft (Predoctoral Division, PI: Dusan Bogunovic, PhD) and Poster Competition: Nicole B. Ramsey, MD, PhD (PI: M. Cecilia Berin, PhD) and David Rodriguez (PI: Nicole Dubois, PhD). Dr. Gelb also moderated the parents' perspective segment, where we heard from a family with a child affected by Crohn's disease. We were able to get a unique perspective from the child's maternal grandmother who suffers from colitis.

Communications

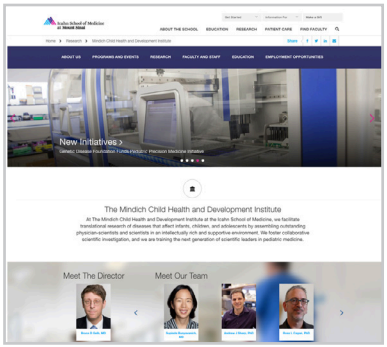
MCHDI delivers the latest updates on research advancements, events, and news, both internally and externally via various communication channels. Below is information about the MCHDI website, newsletter, and social media platforms:

Website ▲ Our website includes detailed information about our signature programs, shared resources, trainee education, and employment opportunities. You can also find our complete list of faculty and links to their research websites as well as the latest press releases featuring our faculty. Our annual reports and MCHDI newsletters are also accessible via our website. Please visit our website at **www.mountsinai.org/mchdi.**

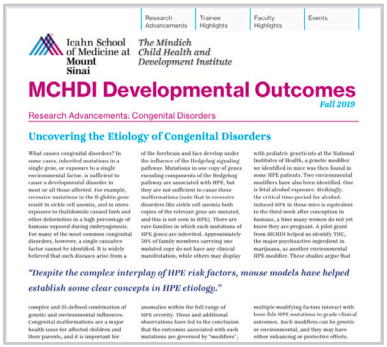
Newsletter ▲ The MCHDI Developmental Outcomes is a biannual newsletter distributed internally to faculty, trainees, and other Institute affiliates to highlight important research breakthroughs, publications, awards, and events within MCHDI. View our latest newsletters featured on **icahn.mssm.edu/research/mindich/about/newsletters.**

Facebook ▲ Our official MCHDI Facebook page was launched in 2014 and currently has 500+ likes and followers. Our team posts almost daily to share updates on faculty research, seminars, events, and other information relevant to children's health. Please like and follow our page at **www.facebook.com/mindichchdi.**

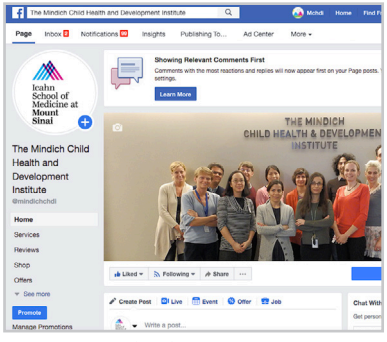
Twitter ▲ Our tweets are streaming on our website in real time. Follow or tweet to us @MindichCHDI or on our website at **www.mountsinai.org/mchdi.**



MCHDI website



MCHDI newsletter



MCHDI Facebook

MCHDI Twitter

Shared Resources

Grant Forward

Grant Forward is a pre-award funding database with a comprehensive list of federal, foundation, and other funding sources. It offers a user-friendly search interface, automated email alerts, and tailored grant recommendations. Grant Forward subscriptions for MCHDI faculty and trainees are covered by our Institute. To sign up, please visit: **www.grantforward.com.**

BioMe Biobank

The BioMe Biobank contains the largest collection of DNA and plasma samples at Mount Sinai, enabling high-throughput disease genotyping and phenotyping, while maintaining patient confidentiality through the Epic electronic medical record (EMR). The goal is to integrate patient clinical care information and research data. Observational epidemiologic studies of children have expanded in the past decade in response to the rising prevalence of childhood diseases, including obesity, autism, and asthma, and of environmental risk factors, such as lead and pesticides; and the ability to genotype DNA has enabled further inquiry into the genetic basis of childhood diseases. MCHDI, in collaboration with the Charles R. Bronfman Institute for Personalized Medicine, is funding the collection of DNA samples from pediatric patients with allergies, and since February 2012, the Jaffe Food Allergy Institute has recruited more than 1000 enrollees. The pediatric cohort is comprised of samples from diverse racial and ethnic groups.

For more information, please visit: **www.icahn.mssm.edu/research/institutes/institute-for-personalized-medicine/innovation-and-technology/biome-platform.**

Biorepository CORE Shared Resource Facility

The biorepository CORE facility provides basic histology services, such as processing and embedding section fixed and frozen tissues from animal or human sources. In addition, services include DNA/RNA/miRNA extractions, preparing and analyzing tissue microarrays, and supporting functions for tissue procurement, both from consented and anonymized collections. For a full list of their services, visit their website at: **icahn.mssm.edu/research/resources/shared-resource-facilities/histology.**

Strategic Plan Implementation

Pediatric Clinical Trials Office

Clinical Trials are the final step that is required to prove that a therapeutic method can work in humans. Most such trials are done in adults, but the results of trials in adults are not necessarily entirely applicable to children. Pediatric clinical trials are therefore required to evaluate the efficacy of therapeutic or diagnostic strategies in pediatric populations, and the conduct of such trials has long been recognized as a priority by regulatory and funding agencies. However, conducting clinical trials in children necessitate specific approaches to study design and conduct that require special expertise. For example: the approach to consent is different (consent is required from the parents, and an assent, which is not a full consent, is required of children – depending on the age and cognitive ability), study design and processes may be quite different in different developmental stages, medication doses are calculated per patient / patient characteristics (e.g., body mass index) rather than kept as a constant, and so on. It is furthermore recognized that patient recruitment to pediatric trials could be more complicated and that because pediatric diseases are generally less common, trials are likely to involve multisite designs.

For those reasons, MCHDI leadership decided to create a clinical trials program that will serve the needs of researchers within the Mount Sinai health system who wish to conduct pediatric clinical trials. The program will offer expertise in regulatory, budgeting, and operations of trials that involve children and families. We have partnered with the Clinical Trials Office (CTO) in Internal Medicine to provide this unique service which would offer its services to existing and "in development" trials, whether they involve investigator-initiated, industry-funded, or federally-funded efforts. Under the support and supervision of the CTO Director, MCHDI leadership, and a dedicated group of pediatric investigators who are already successfully engaged in clinical trials research, a program manager will be charged with creating an operational team that will provide knowledgeable, efficient and convenient support to our diverse initiatives (which have been run in separate silos up until now).

Our vision is to enhance and expand current efforts, allow the development of future ideas and initiatives, reduce the startup costs and time-to-implementation of novel therapeutics, while promoting a collaborative multidisciplinary research enterprise in order to improve the health and quality of life of children.

Inborn Errors of Immunity Program

Inborn errors of immunity comprise over 400 different monogenic disorders. These disorders present in various clinical features: infection (life-threatening or recurrent), malignancy, autoinflammation, autoimmunity or severe allergy. Most of these conditions manifest in childhood – although some can first manifest in adulthood.

Inborn errors of immunity carry important morbidity and mortality, and are a significant burden to health economics systems. Moreover, uncovering the genetic diagnosis in an early stage of the disease is crucial for designing the optimal treatment, be it a drastic measure (hematopoietic stem cell transplantation or gene therapy), a therapy targeted to the cellular pathway that is defective, or a generic treatment with antibiotics/immunoglobulins to prevent irreversible end-organ damage.

As such, this Center has found an excellent niche within the Mindich Child Health and Development Institute: The Center is truly a "bed to bench and back" program. Hence, the Center for Inborn Errors of Immunity program aims to embrace all physicians, physician-

Strategic Plan Implementation continued

scientists, and scientists working on the immune system and the organ systems affected by a defect in the immune system. This broad research effort encompassing genomic strategies as well as classic molecular

immunology aims to unravel the pathophysiology of known but poorly described inborn errors of immunity and to decipher new inborn errors of immunity. Most importantly, we aim to identify and develop new tools for precision therapy in children and adults affected by inborn errors of immunity.

Pediatric Precision Medicine Program

Precision medicine (PM) uses individualized patient data to accurately and rapidly diagnose disease, better predict the outcomes of medical issues, and treat illnesses more precisely and effectively. Currently, medical problems with strong genetic underpinnings such as birth defects, neurodevelopmental delays, and inborn errors of immunity are ones that typically manifest during infancy, childhood, and/or adolescence, and where a PM approach can be transformative. Moreover, these types of conditions can lead to diagnostic odysseys, during which young patients are subjected to extensive medical testing for months or years, families wait anxiously for definitive answers, and effective therapies, when available, are delayed. Through the MCHDI's exciting new Pediatric Precision Medicine Program, we will improve outcomes by applying state-of-the-art genomic technologies as early as possible in the course of a child's disorder.

GDF
GENETIC DISEASE FOUNDATION
KNOW YOUR GENES

Recent advances in genomic medicine have enabled the PM approach that we will undertake. Using just a few drops of blood from the child, we are able to perform high-capacity DNA sequencing to examine the genes that provide instructions for all of the body's proteins. Especially when compared to similar sequencing of the patient's parents in order to identify the rare differences, our ability to pinpoint disease-causing DNA mutations is unparalleled in medical history. To date, our experiences have proven that this approach can solve medical mysteries, identifying known disease genes presenting in unexpected ways as well as allowing us to pinpoint novel ones.

Through the generous support of the **Genetic Disease Foundation**, we have offered this PM approach to infants, children, and teenagers with some of the most complex and difficult-to-diagnose medical issues. In addition to accelerating and improving their care, this program will advance medical education by preparing the young physicians in pediatric training at Mount Sinai to use genomic medicine effectively, allowing them to better serve the community in their future practices.

Mount Sinai

The Mindich Child Health and Development Institute

Leadership and Staff

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Program Manager Elena Lum, PhD
Administrative Manager Risa Slaughter
Administrative Coordinator Shavez Jackson
Senior Executive Director of Development Monica Sohn

Faculty

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James J. Bieker, PhD
Dusan Bogunovic, PhD
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For more information on the MCHDI, please visit our website at **www.mssm.edu/mchdi.**