



Icahn School  
of Medicine at  
Mount  
Sinai

*The Mindich  
Child Health and  
Development Institute*

# MCHDI Developmental Outcomes

Spring 2018

## Research Advancements: Precision Medicine

### Mindich Institute Launches Pediatric Precision Medicine Initiative

In every pediatric population, there are children with conditions that remain undiagnosed despite extensive evaluation and testing by multiple medical specialties. There are also children who present for the first time in the hospital, prior to any evaluations, with rare syndromes that do not fit any particular disease category or known diagnosis. These “medical mysteries” often have either already undergone or will lead to lengthy diagnostic odysseys for patients and their families, spanning numerous hospitalizations, medical specialties, and inconclusive tests. With support from the Genetic Disease Foundation, the Mindich Child Health and Development Institute led by Dr. Bruce Gelb launched this year the Mount Sinai Pediatric Precision Medicine Initiative to find diagnoses for these patients. Along with a handful of other Undiagnosed Disease Programs across the country, this exciting program brings new hope for families caring for children with undiagnosed diseases.

A specific and accurate diagnosis is the foundation of medical care without which it is challenging to provide a prognosis for families or target the root cause of the disease. Next-generation genome sequencing (NGS) has emerged as a critical tool for identifying the genetic causes of rare diseases in infants and children with complex disorders that do not readily fall into known disease categories. NGS has been used in other programs to pinpoint the causal genetic mutations in about 25% of previously undiagnosed cases that had already undergone extensive yet

inconclusive medical evaluations. Aside from ending the frustrating diagnostic odysseys that these families experience, the genetic discoveries can be critical for therapeutic decision-making— sometimes suggesting promising therapies, providing prognostic information to families, and other times just making it clear that the right thing to do is palliative care. In addition to bringing closure to families, this knowledge can also be critical for future reproductive choices for the parents and can link families to resources nationally or internationally that are focused on advancing care for their specific disease. Without a diagnosis, families are often unable to join forces with other families grappling with the same diagnosis for support and to advocate for research. In some instances, undiagnosed disease programs even discover completely new diseases previously unknown in the medical literature.

So far, the Mount Sinai Pediatric Precision Medicine Initiative has enrolled 75 individuals from 25 families (about 1/3 from minority populations) from across all pediatric specialties (hematology, oncology, endocrinology, gastrointestinal, rheumatology, immunology, etc). More than half of the cases have been analyzed, and the program has found a definite or likely genetic diagnosis for 25% of cases. Among the solved cases is an entirely new disease caused by a previously unknown disease gene that happens to have an already FDA-approved targeted treatment. Other solved cases have identified new mutations in other ultra-rare disease

genes (most of these diseases have fewer than 10 cases worldwide). For several cases, research collaborations have been initiated in order to perform specialized tests specific to the syndrome or gene in order to confirm the diagnosis. This close interaction between the clinic and personalized research is a window into the future of pediatrics and precision medicine.

Future goals for the program include expanding the number of referrals from across the Mount Sinai Health System, genomics education for pediatrics residents to prepare them for the near future when every child may have their genome sequenced, and integration with other precision medicine programs at Mount Sinai such as NYCKidSeq (an NIH genomics program focused on minority populations) and the NIH Undiagnosed Diseases Network. The Pediatric Precision Medicine Initiative is a glimpse into the future of pediatrics and the profound changes that genetics will have on day-to-day clinical work, in particular for complex diseases.

For more information about the program, please contact [bruce.gelb@mssm.edu](mailto:bruce.gelb@mssm.edu) or [pedsprecisionmed@mssm.edu](mailto:pedsprecisionmed@mssm.edu).



**Gilad Evrony, MD, PhD**  
Resident, Pediatrics

*The Pediatric Precision Medicine Initiative is a glimpse into the future of pediatrics and the profound changes that genetics will have on day-to-day clinical work, in particular for complex diseases.*

# MCHDI Researchers Identify Six Genes Driving Peanut Allergy Reactions

## Genes Could Be Targeted for New Therapies To Treat Peanut Allergy

A research team led by MCHDI faculty member Supinda Bunyavanich, MD, MPH has identified six genes that activate hundreds of other genes in children experiencing severe allergic reactions to peanuts. This is the first study to identify genes driving acute peanut allergic reactions using a double blind placebo-controlled study conducted in humans with comprehensive sequencing of genes expressed before, during, and after they ingested peanut. Because children were studied over the course of their allergic reactions, each subject could serve as their own reference, allowing the researchers to accurately detect gene expression changes resulting from peanut allergic reactions.

The results of the study were published in *Nature Communications* on December 5th, 2017. The study was partially funded by an MCHDI Pilot Grant awarded to Dr. Bunyavanich. Several coauthors are also members of MCHDI and Mount Sinai.

The standard treatment for peanut allergic individuals includes avoidance and prompt care of allergic reaction. Immunotherapy has demonstrated progress, but it is not effective for all individuals, carries adverse side effects, and has not been approved by the FDA.

“This study highlights genes and molecular processes that could be targets for

new therapies to treat peanut allergic reactions and could be very important to understanding how peanut allergy works overall,” said the study’s senior author Supinda Bunyavanich, MD, MPH, Associate Professor and member of the MCHDI and Departments of Pediatrics and Genetics and Genomic Sciences at Mount Sinai. “We still don’t completely understand everything that happens in the body during peanut allergic reactions. We can use these genes to direct our studies of peanut allergy and one day hopefully predict how strongly someone with peanut allergy will react.”

The research team collected blood samples from 40 peanut allergic children before, during, and after randomized, double-blinded, placebo-controlled oral food challenges. Subjects ingested incremental amounts of peanut at 20 minute intervals until an allergic reaction occurred or a cumulative dose of 1.044 grams of protein was ingested. In a similar fashion on a different day, the same subjects ingested incremental doses of placebo oat powder and blood samples were drawn before, during, and after the challenge. The team then performed comprehensive RNA sequencing on the blood samples followed by network-based analyses to determine which genes and cells were being activated and driving these allergic reactions.

“Other studies have looked at genes expressed in people with food allergies and compared them to people who don’t have food allergies,” said Bunyavanich. “One of the strengths of our study is that we looked at genes expressed over time in children actively reacting to peanut, following that person throughout their reaction, which provided a detailed and comprehensive picture of what’s happening on the genetic and molecular level during a peanut allergic reaction.” Further, the team was also able to leverage data-driven approaches to identify six genes that were activated at the most upstream level, thus pointing the way to high-yield targets for therapeutic intervention.

To address if their findings may be relevant to food allergies other than peanut allergy, Bunyavanich and the research team plan to conduct future studies targeting other common food allergens such as milk and egg.



**Supinda Bunyavanich, MD, MPH**  
Associate Professor, Pediatrics  
Associate Professor, Genetics and Genomic Sciences

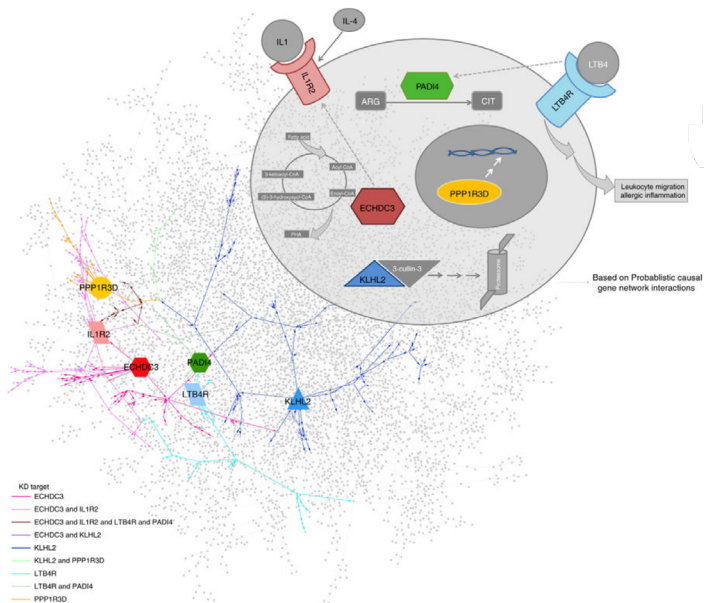


Figure 1: Key drivers interact within the probabilistic causal gene network and cellular environment. A cartoon cell schematic of the key drivers identified as primary causal regulators of the acute-phase response module and peanut response genes is shown in the upper right, demonstrating their locations of activity in the cellular context based on prior knowledge. Activation of LTB4R by LTB4 binding leads to macrophage, T cell, and neutrophil chemotaxis. PADI4 converts arginine (ARG) to citrulline (CIT) residues and plays a role in granulocyte and macrophage development. Induced and released by IL4, IL1R2 is a decoy receptor that inhibits IL1 activity. ECHDC3 is an enzyme involved in fatty acid biosynthesis. PPIR3D regulates protein serine/threonine phosphatase activity, and KLHL2 is involved in proteasomal degradation and reorganization of actin cytoskeleton for cell projection by oligodendrocyte precursors. At the bottom left, the constructed probabilistic causal gene network is displayed with key drivers indicated by enlarged, labeled nodes, with their shape and color corresponding to the cell schematic. Edges are colored based on the interaction of each key driver with downstream genes at a path length of seven, displaying the singular and combinatorial downstream effects that each key driver can have on this network. Interactions between the key drivers inferred from the probabilistic causal gene network are indicated in the cell schematic by the dashed-line arrows.

## New Extramural Faculty

### John Bucuvalas, MD

**John Bucuvalas, MD** is the Chief of the Hepatology and Vice Chair of Faculty Affairs in the Jack and Lucy Clark Department of Pediatrics at the Icahn School of Medicine at Mount Sinai and the Kravis Children's Hospital at Mount Sinai. He also serves as the Director of Solid Organ Transplant Outreach for the Recananti-Miller Transplant Institute. He graduated from Harvard College, Magna Cum Laude in biology and then obtained his medical degree at Harvard Medical School. He completed his pediatric residency including a year as Chief Resident before his gastroenterology fellowship at Cincinnati Children's Hospital. He is board certified in pediatric gastroenterology with a certificate of added qualification in transplant hepatology. He advanced to Professor of Pediatrics at the University of Cincinnati and served as Director of the Integrated Solid Organ Transplant Program before coming to Mount Sinai. The overall goal of Dr. Bucuvalas's research is to give children and adolescents liver transplant candidates and recipients the promise of full and meaningful life by delivering

#### Recent Publications:

- Shemesh E, **Bucuvalas JC**, Anand R, Mazariegos GV, Alonso EM, Venick RS, Reyes-Mugica M, Annunziato RA, Shneider BL. The Medication Level Variability Index (MLVI) Predicts Poor Liver Transplant Outcomes: A Prospective Multi-Site Study. *Am J Transplant*. 2017 Oct;17(10):2668-2678.
- Hsu EK, Shaffer ML, Gao L, Sonnenday C, Volk ML, **Bucuvalas J**, Lai JC. Analysis of Liver Offers to Pediatric Candidates on the Transplant Wait List. *Gastroenterology*. 2017 Oct;153(4):988-995.
- Feng S, **Bucuvalas J**. Tolerance after liver transplantation: Where are we? *Liver Transpl*. 2017 Dec;23(12):1601-1614.
- Grippa F, Palazzolo M, **Bucuvalas JC**, Gloor, PA. Supporting Development Efforts Of Clinical Care Teams. *International Journal of Organizational Design and Engineering*. 2012;2(2):149.
- Ng VL, Alonso EM, **Bucuvalas JC**, Cohen G, Limbers CA, Varni JW, Mazariegos G, Magee J, McDiarmid SV, Anand R; Studies of Pediatric Liver Transplantation (SPLIT) Research Group. Health Status of Children Alive 10 Years after Pediatric Liver Transplantation Performed in the US and Canada: Report of the Studies of Pediatric Liver Transplantation Experience. *J Pediatr*. 2012;160:820-6.

reliable state of the art care integrated across disciplines ensuring that we acquire and apply new knowledge and improve processes in the constantly evolving delivery system. His primary research efforts, funded by NIH, focus on clinical and translational to define predictors of operational tolerance, to predict risk and determine the mechanism of long term structural liver allograft injury

and to define strategies to mitigate non-adherence in transplant recipients.



**John Bucuvalas, MD**  
Professor, Pediatrics  
Chief, Division of Hepatology  
Vice Chair, Faculty Affairs,  
Pediatrics

## New Intramural Faculty

### Silvia De Rubeis, PhD

**Silvia De Rubeis, PhD** is an Assistant Professor at the Seaver Autism Center for Research and Treatment, Department of Psychiatry, and Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai. Dr. De Rubeis is a molecular neuroscientist and geneticist interested in understanding the genetic, molecular and cellular mechanisms underlying autism spectrum disorder (ASD) and intellectual disability (ID). Dr. De Rubeis completed her PhD in Cellular and Molecular Biology at the University of Rome "Tor Vergata" in Italy. During her first postdoctoral training with Dr. Claudia Bagni at the Katholieke Universiteit Leuven and Vlaams Instituut voor Biotechnologie (VIB) in Leuven, Belgium, she studied how the regulation of mRNA translation shapes the synaptic development in the context of Fragile X syndrome, a leading monogenic cause of ASD and the most common inherited form of ID. She then joined ISMMS for a second postdoctoral training in Genetics

#### Recent Publications:

- De Rubeis S**, He X, Goldberg AP, ... Devlin B, Daly MJ, Buxbaum JD. Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*. 2014 Nov 13;515(7526):209-15.
- De Rubeis S**, Pasciuto E, Li KW, ... Posthuma D, Smit AB, Bagni C. CYFIP1 coordinates mRNA translation and cytoskeleton remodeling to ensure proper dendritic spine formation. *Neuron*. 2013 Sep 18;79(6):1169-82.
- Siper PM\*, **De Rubeis S\***, Trelles MDP, ... Bernier RA, Kolevzon A, Buxbaum JD. Prospective investigation of FOXP1 syndrome. *Mol Autism*. 2017 Oct 24;8:57. \*Equal contribution
- Lim ET, Uddin M, **De Rubeis S**, ... Scherer SW, Buxbaum JD, Walsh CA. Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. *Nat Neurosci*. 2017 Sep;20(9):1217-1224.
- Zalfa F, Eleuteri B, Dickson KS, Mercaldo V, **De Rubeis S**, ... Bagni C. A new function for the fragile X mental retardation protein in regulation of PSD-95 mRNA stability. *Nat Neurosci*. 2007 May;10(5):578-87.

and Genomics in the lab of Dr. Joseph Buxbaum. While there, she studied the role of rare genetic variation in ASD through large-scale exome sequencing analyses and discovered novel genes and loci conferring risk. Her lab studies developmental defects resulting from disruptive mutations in novel high-risk genes identified from genomic studies in ASD and ID. Her research takes a genetics-first approach for functional analyses in cellular and mouse

models and strives to take into account clinically relevant aspects that emerge from patient-based research.



**Silvia De Rubeis, PhD**  
Assistant Professor,  
Psychiatry

### Alison P. Sanders, PhD

**Alison P. Sanders, PhD** is an environmental health engineer & molecular epidemiologist in the Department of Pediatrics and the Department of Environmental Medicine and Public Health at the Icahn School of Medicine at Mount Sinai. Alison earned a BS in Biomedical Engineering from the University of Virginia, an MS in Environmental Engineering from the University of Wisconsin-Madison, and her PhD in Environmental Sciences and Engineering with a minor in Epidemiology from the University of North Carolina-Chapel Hill. Dr. Sanders conducts research that combines exposure science, population health, and toxicology to characterize the developmental effects of prenatal and early-life exposure to metals, and is PI of a K99/R00 award from the NIEHS. She has received two pilot awards from the Mount Sinai P30 Core Center and two awards from the Burroughs Wellcome Fund. Her studies utilize large birth cohorts in the US and Mexico to investigate relationships between exposure to toxic metals and birth

#### Recent Publications:

- Sanders AP, Saland JM, Wright RO, Satlin L.** Perinatal and childhood exposure to environmental chemicals and blood pressure in children: A review of literature 2007-2017. *Pediatric Research*. [In Press]
- Rodosthenous R, Burris HH, **Sanders AP**, Just AC, Dereix A, Svensson K, Solano M, Tellez-Rojo MM, Wright RO, Baccarelli AA. Second trimester extracellular microRNAs in maternal blood and fetal growth: an exploratory study. *Epigenetics*. 2017 Sep;12(9):804-810.
- Sanders AP**, Burris HH, Just AC, Motta V, Svensson K, Mercado-Garcia A, Pantic I, Schwartz J, Tellez-Rojo MM, Wright RO, Baccarelli AA. MicroRNA expression in the cervix during pregnancy is associated with length of gestation. *Epigenetics*. 2015;10(3):221-8.
- Sanders AP**, Smeester L, Rojas D, DeBussycher T, Wu MC, Wright FA, Zhou Y, Laine JE, Rager JE, Swamy G, Ashley-Koch A, Miranda ML, Fry RC. Cadmium exposure and the epigenome: Exposure-associated patterns of DNA methylation in leukocytes from mother-baby pairs. *Epigenetics* 2014 Feb;9(2):212-21.
- Sanders AP**, Messier KP, Shehee M, Rudo K, Serre ML, Fry RC. Arsenic in North Carolina Domestic Wells: Public Health Implications. *Environment International*. 2012 Jan;38(1):10-6.

outcomes with poorer renal outcomes later in childhood. She also directs a trainee development program that enhances early career scientists' skillsets in teaching, translating, and communicating research findings to audiences ranging from 10-year-olds to graduate-level scientists.



**Alison P. Sanders, PhD**  
Instructor, Pediatrics & Environmental Medicine and Public Health

### Paige M. Siper, PhD

**Paige M. Siper, PhD** is a licensed clinical psychologist, Chief Psychologist of the Seaver Autism Center for Research and Treatment, and an Assistant Professor in the Department of Psychiatry. She has expertise in the diagnosis, neuropsychological assessment, and treatment of individuals with a variety of neurodevelopmental disorders, including autism spectrum disorder (ASD) and intellectual disability. Dr. Siper's research focuses on biomarker discovery and sensory processing using electrophysiological and behavioral approaches. Dr. Siper is the co-developer of the Sensory Assessment for Neurodevelopmental Disorders (SAND), which is the first clinician-administered observation and corresponding caregiver interview to quantify sensory reactivity according to DSM-5 criteria for ASD. Dr. Siper is currently using a type of EEG known as a visual evoked potential, along with the SAND and comprehensive clinical characterization of patients, to identify biological and bio-behavioral markers of idiopathic and single-gene forms of ASD. Her long-term research goals involve the integration of neural and neuropsychological findings to identify subtypes, monitor disease trajectory, and objectively measure treatment response. Dr. Siper has a strong interest in ensuring

#### Recent Publications:

- Siper PM**, Kolevzon A, Wang AT, Buxbaum JD, Tavassoli T. A clinician-administered observation and corresponding caregiver interview capturing DSM-5 sensory reactivity symptoms in children with ASD. *Autism Research*. 2017 Jun;10(6):1133-1140.
- Siper PM**, Zemon V, Gordon J, George-Jones J, Lurie S, Zweifach J, Tavassoli T, Wang AT, Jamison J, Buxbaum JD, Kolevzon A. Rapid and Objective Assessment of Neural Function in Autism Spectrum Disorder Using Transient Visual Evoked Potentials. *PLoS One*. 2016 Oct 7;11(10):e0164422.
- Siper PM\***, De Rubeis S\*, Trelles MDP, Durkin A, Di Marino D, Muratet F, Frank Y, Lozano R, Eichler EE, Kelly M, Beighley J, Gerdtts J, Wallace AS, Mefford HC, Bernier RA, Kolevzon A, Buxbaum JD. Prospective investigation of FOXP1 syndrome. *Mol Autism*. 2017 Oct 24;8:57. \*Equal Contribution
- De Rubeis S\*, **Siper PM\***, Durkin A, Weissman J, Muratet F, Halpern D, Trelles MDP, Frank Y, Lozano R, Wang AT, Holder JL Jr, Betancur C, Buxbaum JD, Kolevzon A. Delineation of the genetic and clinical spectrum of Phelan-McDermid syndrome caused by SHANK3 point mutations. *Mol Autism*. 2018 Apr 27;9:31. \*Equal Contribution
- Trelles MP, **Siper PM**, Grice DE. Current Treatment for Pediatric Psychiatric Disorders. *Neurobiology of Mental Illness*, Fifth Edition. Ed. Dennis S. Charney, Ed. Eric J. Nestler, Ed. Pamela Sklar, Ed. Joseph D. Buxbaum. New York: Oxford University Press, 2017. 915-932. Print.

inclusion of severely affected individuals in research studies.

Dr. Siper graduated with honors and distinction from Cornell University with a Bachelor of Science. She earned her PhD in Clinical Psychology from Yeshiva University and completed her clinical psychology internship at the University of Miami Miller School of Medicine. Dr. Siper was the recipient of an Autism Speaks Meixner Translational Research

Postdoctoral Fellowship, which she completed at the Seaver Autism Center at Mount Sinai. She joined Mount Sinai faculty in 2014.



**Paige M. Siper, PhD**  
Assistant Professor, Psychiatry

### Trainee Pilot Projects: 2018 Awardees

#### **Project Title: The Oral Microbiome and Metabolic Alterations in Food Allergy**

**Investigator:** *Hsi-en Ho, MD, Clinical Fellow, Department of Pediatrics*

**Primary Mentor:** *Supinda Bunyavanich, MD, MPH, MPhil, Department of Pediatrics and Genetics and Genomic Sciences*

**Secondary Mentor:** *Scott H Sicherer, MD, Chief, Department of Pediatrics and Director, Jaffe Food Allergy Institute*

**Secondary Mentor:** *Alexander V. Grishin, PhD, Department of Pediatrics*

**Abstract:** Food allergy has become a major health problem in the US with an estimated 8% of children affected. In addition to the risk of causing life-threatening anaphylaxis, food allergy

has significant impacts on the nutritional and psychosocial health of affected children. Data from murine models of food allergy strongly support a role of commensal bacteria and their metabolites in regulating oral tolerance. However, the link between symbiotic bacteria and the immune system in children with or without food allergy remains obscure. In this pioneering study, we are applying next generation sequencing to comprehensively profile the unique characteristics of oral microbial community (i.e., the oral microbiota) in children who develop food allergy. In addition, through metagenomics analysis, we aim to identify the key bacterial metabolic pathways altered in food-allergic children. Our long-term goal is to dissect the biological mechanisms linking key environmental

factors, such as the microbiota, to the immune regulation of oral tolerance. By identifying the distinct microbiota and metabolites affecting the development of food allergy, our data have the potential to identify novel biomarkers, discover new therapeutic targets, and provide guidance for primary prevention of food allergy.



**Hsi-en Ho, MD**

Clinical Fellow, Department of Pediatrics

*“Our long-term goal is to dissect the biological mechanisms linking key environmental factors, such as the microbiota, to the immune regulation of oral tolerance.”*

*—Hsi-en Ho, MD*

#### **Project Title: A cell-type specific in vitro model to rapidly screen modulators of neurodevelopmental plasticity**

**Investigator:** *Milo R. Smith, PhD, Postdoctoral fellow, Department of Neuroscience, Genetics and Genomic Sciences, Psychiatry, and Ophthalmology*

**Primary Mentors:** *Hirofumi Morishita, MD, PhD, Department of Neuroscience, Genetics and Genomic Sciences, Psychiatry, and Ophthalmology and Joel T. Dudley, PhD, Department of Genetics and Genomic Sciences*

**Secondary Mentor:** *Nan Yang, PhD, Department of Neuroscience*

**Abstract:** Critical periods are childhood windows of neuroplasticity that respond to sensory and social experience to enable development of optimal cognition and behavior. Disruption of critical periods can lead to neurodevelopmental disorders — for example, normal visual processing

in the brain can be disrupted by early eye problems such as a childhood cataract. If caught early, the resulting amblyopia can be corrected and good vision can be restored. However, if the cataract is not removed until after the critical period has closed, the condition becomes permanent impacting 3% of adults. Discovering drugs that can reactivate critical period plasticity after a critical period has closed would be a boon for treating plasticity-related neurodevelopmental disorders, such as amblyopia. My colleague Masato Sadahiro and others have identified a key inhibitory interneuron subtype marked by somatostatin that when transiently activated in adult rodents reactivates critical period plasticity. Inspired by this finding, we are setting out in collaboration with stem cell expert Nan Yang (Neuro) to develop a human induced stem cell based screen to identify drugs that specifically activate the somatostatin subtype. Together with Dr. Yang’s previously established 1-step method to induce a

pure inhibitory neuronal population from stem cells, we will use CRISPR to mark those that express somatostatin with red fluorescence and those that are activated by a drug with green as a proof of principle screen to discover drugs that may reactivate critical period plasticity. We hope this work will be a stepping stone towards discovering neurodevelopmental therapeutics for improved child health.



**Milo R. Smith, PhD**

Postdoctoral fellow, Department of Neuroscience, Genetics and Genomic Sciences, Psychiatry, and Ophthalmology

*“We hope this work will be a stepping stone towards discovering neurodevelopmental therapeutics for improved child health.”*

*—Milo R. Smith, PhD*

## Publications, Grants, Awards/Honors

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### Trainee Grants

**Evan S. Bardot**, PI: Nicole Dubois, NHLBI, F31, “Investigating Specification of Ventricular Cardiovascular Cells in the Gastrulating Mouse Embryo”

**Alejandro Martin-Trujillo**, PhD, PI: Andrew Sharp, American Heart Foundation, Postdoctoral Fellowship, “Epigenetic defect in congenital heart defects”

### Trainee Awards

**Giovanna Collu**, PhD, PI: Marek Mlodzik & Kathryn Bambino, PhD, PI: **Jaime Chu**, Genetics Society of America, Career Development Symposium Award

**Maya Deyssenroth**, PhD, PI: **Jia Chen**, Teratology Society Meeting, Travel Award 2018

### Faculty Grants

**Brian D. Brown**, PhD, NIH/NICHD, R21, “T cell Mediated Gene Replacement Therapy”

**Minji Byun**, PhD, Castleman Disease Collaboration Network, “The Role of DNMT3A in Idiopathic Multicentric Castleman Disease”

**Jaime Chu**, MD, Art in Giving, The Rachel Molly Markoff Foundation Award

**David Dunkin**, MD, NIH NIDDK, RO3, “Epicutaneous Tolerance Induction with Flagellin Epitopes for the Treatment of Colitis”

**Laura M. Huckins**, PhD, BBRF-NARSAD Young Investigator Award, “Analyzing gene expression in 60,000 bipolar disorder and schizophrenia cases to characterize shared and distinct genomic and molecular pathologies”

**Alexander Kolevzon**, PhD & **Paige M. Siper**, PhD, NINDS, R01, “Electrophysiological Markers for Interventions in Phelan-McDermid Syndrome and Idiopathic ASD”

**Dalila Pinto**, PhD, NIH/NIMH, R01, “Mapping the role of long noncoding RNAs in gene regulatory networks in schizophrenia”

### Faculty Awards/Honors

**Bruce D. Gelb**, MD, Icahn School of Medicine at Mount Sinai, Jacobi Medallion Award 2018

**Lisa M. Satlin**, MD, University of Alabama at Birmingham, The 2nd Annual James A. Schafer Lectureship, “Cell-specific Function and Regulation of Mechanosensitive Ion Channels in the Distal Nephron”

**Lisa M. Satlin**, MD, Experimental Biology Annual Meeting in San Diego, 2018 Carl W. Gottschalk Award of the American Society of Physiology (Renal Section)

## Faculty Highlights

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### Publications

Claus Henn B, **Austin C**, Coull BA, Schnaas L, **Gennings C**, Horton MK, ... **Wright RO**, **Arora M**. **Uncovering neurodevelopmental windows of susceptibility to manganese exposure using dentine microspatial analyses.** *Environ Res.* 2018 Feb;161:588-98.

Chiang D, Chen X, Jones SM, Wood RA, **Sicherer SH**, Burks AW, ..., **Sampson HA**, ... **Berin MC**. **Single-cell profiling of peanut-responsive t cells in patients with peanut allergy reveals heterogeneous effector th2 subsets.** *J Allergy Clin Immunol.* 2018 Jan 31.

Agudo J, Park ES, Rose SA, Alibo E, Sweeney R, Dhainaut M, ... **Brown BD**. **Quiescent tissue stem cells evade immune surveillance.** *Immunity.* 2018 Feb 20;48(2):271-85.e5.

Lurz E, Quammie C, Englesbe M, Alonso EM, Lin HC, Hsu EK, ... **Bucvalas J**, ... Kamath BM. **Frailty in children with liver disease: A prospective multicenter study.** *J Pediatr.* 2018 Mar;194:109-15.e4.

Fazlollahi M, Chun Y, Grishin A, Wood RA, Burks AW, Dawson P, ... **Sampson HA**, **Sicherer SH**, **Bunyavanich S**. **Early-life gut microbiome and egg allergy.** *Allergy.* 2018 Jan 10.

Kerur N, Fukuda S, Banerjee D, Kim Y, Fu D, Apicella I, ... **Buxbaum JD**, ... Ambati J. **Cgas drives noncanonical-inflammasome activation in age-related macular degeneration.** *Nat Med.* 2018 Jan;24(1):50-61.

**Sonoshita M**, Scopton AP, Ung PMU, Murray MA, Silber L, Maldonado AY, ... **Cagan RL**, Dar AC. **A whole-animal platform to advance a clinical kinase inhibitor into new disease space.** *Nat Chem Biol.* 2018 Mar;14(3):291-8.

Everson TM, Punshon T, Jackson BP, **Hao K**, **Lambertini L**, **Chen J**, ... Marsit CJ. **Cadmium-associated differential methylation throughout the placental genome: Epigenome-wide association study of two u.S. Birth cohorts.** *Environ Health Perspect.* 2018 Jan 22;126(1):017010.

**Bambino K**, Zhang C, **Austin C**, Amarasiriwardena C, **Arora M**, **Chu J**, Sadler KC. **Inorganic arsenic causes fatty liver and interacts with ethanol to cause alcoholic liver disease in zebrafish.** *Dis Model Mech.* 2018 Feb 26;11(2).

DeWitt SJ, Bradley KA, Lin N, Yu C, **Gabbay V**. **A pilot resting-state functional connectivity study of the kynurenine pathway in adolescents with depression and healthy controls.** *J Affect Disord.* 2018 Feb;227:752-8.

Meriney SD, Tarr TB, Ojala KS, Wu M, Li Y, Lacomis D, **Garcia-Ocaña A**, ... Wipf P. **Lambert-eaton myasthenic syndrome: Mouse passive-transfer model illuminates disease pathology and facilitates testing therapeutic leads.** *Ann N Y Acad Sci.* 2018 Jan;1412(1):7381.

Manheimer KB, **Richter F**, Edelmann LJ, D'Souza SL, Shi L, Shen Y, ... **Gelb BD**. **Robust identification of mosaic variants in congenital heart disease.** *Hum Genet.* 2018 Feb;137(2):183-95.

Gilsbach R, Schwaderer M, Preissl S, Gruning BA, Kranzhofer D, Schneider P, ... **Gelb BD**, Hein L. **Distinct epigenetic programs regulate cardiac myocyte development and disease in the human heart in vivo.** *Nat Commun.* 2018 Jan 26;9(1):391.

Grant LP, **Gennings C**, Wickham EP, Chapman D, Sun S, Wheeler DC. **Modeling pediatric body mass index and neighborhood environment at different spatial scales.** *Int J Environ Res Public Health.* 2018 Mar 8;15(3).

Yip BHK, Bai D, Mahjani B, Klei L, Pawitan Y, Hultman CM, **Grice DE**, ... **Buxbaum JD**, Devlin B, **Reichenberg A**, Sandin S. **Heritable variation, with little or no maternal effect, accounts for recurrence risk to autism spectrum disorder in sweden.** *Biol Psychiatry.* 2018 Apr 1;83(7):589-97.

Dyke JP, Garfinkel AC, **Groves AM**, Kovanlikaya A. **High-resolution rapid neonatal whole-body composition using 3.0 tesla chemical shift magnetic resonance imaging.** *Pediatr Res.* 2018 Mar;83(3):638-44.

### Publications, continued

- Zhang J, Peng S, Cheng H, Nomura Y, Di Narzo AF, **Hao K**. Genetic pleiotropy between nicotine dependence and respiratory outcomes. *Sci Rep*. 2017 Dec 4;7(1):16907.
- Uban KA, **Horton MK**, Jacobus J, Heyser C, Thompson WK, Tapert SF, ... Sowell ER. Biospecimens and the abcd study: Rationale, methods of collection, measurement and early data. *Dev Cogn Neurosci*. 2018 Mar 16.
- Pardinas AF, Holmans P, Pocklington AJ, Escott-Price V, Ripke S, Carrera N, ... **Huckins LM**, ... Walters JTR. Common schizophrenia alleles are enriched in mutation-intolerant genes and in regions under strong background selection. *Nat Genet*. 2018 Mar;50(3):381-9.
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## 6<sup>th</sup> Annual MCHDI Retreat

# Save the Date

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**Date:** November 27, 2018

**Time:** TBA

**Location:** Harmonie Club  
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