Icahn School of Medicine at Mount Sinai
The Mindich Child Health and Development Institute

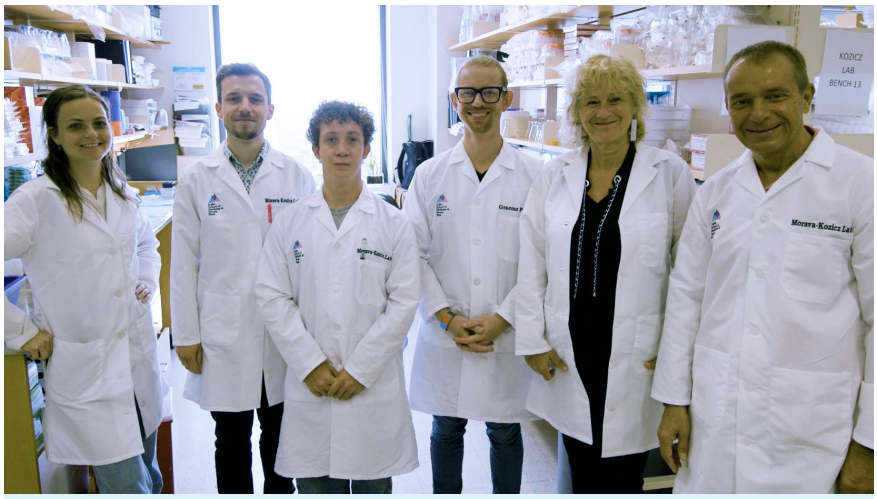
MCHDI Developmental Outcomes

FALL 2026

Research Advancements: From Drug Screening to Cortical Brain Organoids and Clinical Trials

Developing Patient Cell Derived Models for Treatment Development in Metabolic Disease

Our research team has been studying DHDDS-related juvenile parkinsonism, a progressive neurodegenerative disease that presents in childhood. Recently, we uncovered new clues about how this rare neurological disorder affects the brain and identified a potential treatment approach that warrants further investigation.



The Morava-Kozicz Lab team

Using patient-derived fibroblast cell lines, we created cortical brain organoids to study this rare monogenic disease. In these miniature human brain models, we found disruptions in several important processes, including mitochondrial energy production, lipid metabolism involving dolichol and phospholipids, and protein processing. Surprisingly, abnormal cholesterol trafficking, specifically localized to astrocytes, also appeared to play a major role in the disease.

In collaboration with a drug-screening laboratory using yeast avatars carrying the most common DHDDS variant, we found that NAD^+-related molecules had a positive effect on yeast growth and survival. Among the potential candidates for drug repurposing, niacin and nicotinamide mononucleotide (NMN) demonstrated the strongest NAD^+-boosting effects.

We then evaluated the effects of NMN treatment in our brain organoid models. NMN improved energy balance, basal and maximal respiration, and cellular function, as measured using a multi-electrode array assay.

Our team also followed six patients who took NMN under medical supervision. Using AI-based home video follow-up, we quantified changes in gait, posture, and coordination while the patients were taking the supplement. All six patients showed significant clinical improvement during follow-up.

These findings are notable because this is the first study to use patient-derived brain organoids to investigate DHDDS-related juvenile parkinsonism and to connect laboratory findings with early clinical observations in patients.

The next step is to conduct larger, controlled clinical trials, including a randomized, placebo-controlled trial, to determine whether NMN is safe and effective and to better understand its mechanism of action. We are currently enrolling patients in an open-label NMN trial to further evaluate its safety. This trial is

funded by the CDG-UK patient association. (Clinicaltrials.gov: NCT07572825)

We also hope that our findings will help guide the development of targeted treatments for DHDDS-related juvenile parkinsonism and shed light on other neurological disorders involving mitochondrial dysfunction and abnormal lipid metabolism.

Our related publications are currently under review at *Brain* and *Genetics in Medicine.*



Eva Morava, MD, PhD
Professor, Genetics and Genomic Sciences and Pediatrics



Tamas Kozicz, MD, PhD
Professor, Genetics and Genomic Sciences and Neuroscience

Key Publications:

1. Muffels IJJ, Kantautas KA, MacDonald G, Garapati K, Pasupuleti RR, Tinker RJ, Shah R, Thevandavakkam MA, Donnelly J, Hrstka R, Smith D, Van Klinken J, Vaz F, Pandey A, Perlstein EO, Kozicz T, **Morava E.** DHDDS-related juvenile parkinsonism is caused by impaired lipid metabolism, glycosylation, and mitochondrial dysfunction, which can be rescued by NAD⁺ treatment. *medRxiv* [Preprint]. 2026 Jun 5:2026.

2. Da Silva A, Tene Tadoum SB, Muffels IJJ, Budhraja R, Sturiale L, Messina A, Giladi M, Taherzadeh M, Fazeli M, Bonneil É, Khan S, Te Vruchte D, Yamanaka Y, Di Cristo G, Hamdan FF, Platt FM, Tomatsu S, Haitin Y, Kozicz T, Thibault P, Garozzo D, Pandey A, **Morava E,** Rossignol E, Pshezhetsky AV. Mono-allelic p.R37H Dehydrodolichyl Diphosphate Synthase variants lead to protein glycosylation defects, aberrant lipid profiles and interneuron scarcity in a novel mouse model of progressive epileptic encephalopathy. *bioRxiv* [Preprint]. 2025 Aug 21:2025.

3. Muffels IJJ, Sadek M, Kozicz T, **Morava E.** Assessing age of onset and clinical symptoms over time in patients with heterozygous pathogenic DHDDS variants. J Inherit Metab Dis. 2024 Sep;47(5):935-944. doi: 10.1002/jimd.12769. Epub 2024 Jun 21. PMID: 39540616.

Resetting Cellular Age for the Next Generation

Every new generation begins young, even though the egg that gives rise to it develops within an adult body. How germ cells erase the effects of cellular age and prepare to support a new embryo remains one of the most compelling questions in reproductive and developmental biology.

Our laboratory studies this process using the developing egg of the fruit fly, Drosophila melanogaster. Although flies and humans differ in many respects, the fundamental cellular machinery governing egg development is remarkably conserved. This system allows us to observe with exceptional spatial and temporal precision how an undifferentiated germ cell acquires the capacity to produce an egg that is "functionally youthful" and can give rise to the next generation.

We call this developmental period the germ cell-to-maternal transition. During this transition, germ cells leave their earlier identity, activate a differentiation program, and begin producing the RNAs and proteins needed by the future embryo [1-4]. Our work has shown that this transformation is coordinated at several levels: genes are turned on and off, RNAs are selectively translated, and the genome itself is reorganized within the nucleus. We have found that transcription factors work with chromatin insulators and nuclear pore proteins to establish boundaries between genomic neighborhoods. These findings connected changes in gene expression to changes in cellular architecture and led us to ask a broader question: Does the germ cell also replace the molecular machines that execute these programs?

Our studies of the nuclear pore complex provided the first evidence that it may. Nuclear pore complexes are large structures that regulate communication between the nucleus and cytoplasm while also helping organize the genome. We discovered that nuclear pore complexes are reduced during a precise stage of germ-cell development and that new nuclear pore complexes are subsequently produced. Disrupting either their removal or their rebuilding prevents germ cells from acquiring the correct developmental identity[5]. Nuclear pore complex re-

modeling may therefore be crucial for the germ cell to establish a new program while preventing older cellular states from being carried into the egg.

We are now investigating whether the same principle applies to ribosomes, the cellular machines that translate genetic information into proteins. Ribosomes can accumulate damage or lose functional precision over time. Our emerging work suggests that developing germ cells remove older ribosomes and synthesize a new population as the egg is specified. We are examining how nutrient-sensing pathways, mitochondrial metabolism, and protein-quality-control systems coordinate this process.

Together, these studies suggest that rejuvenation may require more than resetting gene expression. Germ cells may physically replace essential cellular machines inherited from earlier developmental stages, creating a renewed cellular foundation for embryogenesis. Understanding this natural process could ultimately reveal new ways to improve egg quality, enhance regenerative capacity, or restore function in aged and damaged tissues.



Prashanth Rangan, PhD
Professor, Stem Cell Biology and Regenerative Medicine and Pediatrics

Key Publications:

1. Sarkar K, Kotb NM, Lemus A, Martin ET, McCarthy A, Camacho J, Iqbal A, Valm AM, Sammons MA, **Rangan P.** A feedback loop between heterochromatin and the nucleopore complex controls germ-cell-to-oocyte transition during *Drosophila* oogenesis. *Dev Cell*. 2023 Nov 20;58(22):2580-2596.e6.
2. Blatt P, Wong-Deyrup SW, McCarthy A, Breznak S, Hurton MD, Upadhyay M, Bennink B, Camacho J, Lee MT, **Rangan P.** RNA degradation is required for the germ-cell to maternal transition in *Drosophila*. *Curr Biol.* 2021 Jul 26;31(14):2984-2994.e7.
3. Kotb NM, Ulukaya G, Chavan A, Nguyen SC, Proskauer L, Joyce EF, Hasson D, Jagannathan M, **Rangan P.** Genome organization regulates nuclear pore complex formation and promotes differentiation during Drosophila oogenesis. *Genes Dev.* 2024 Jun 25;38(9-10):436-454.
4. Kotb NM, Ulukaya G, Ramamoorthy A, Park LS, Tang J, Hasson D, **Rangan P.** TORC1-dependent translation drives chromatin remodeling during the germ-cell-to-maternal transition in *Drosophila*. EMBO J. 2026 Mar;45(5):1648-1671.
5. Venkat S, Nguyen T, Blangini C, Pollak M, Schindler K, Capelson M, **Rangan P.** Developmentally programmed nuclear pore complex replacement enables oocyte specification. *bioRxiv* [Preprint]. 2026 Feb 16:2026.02.13.705775.

Trainee Highlights

Karen Levinson Mindich Trainee Pilot Program: 2026 Awardees

Project Title: Mapping Fetal Brain-Behavior Coupling in Late Gestation: A Framework for Early Neurodevelopmental Trajectories

Investigator: Ezra Aydin, PhD, T32 Postdoctoral Fellow, Psychiatry

Primary Mentor: Vanessa Babineau, PhD, Director, Resilient Beginnings Lab, Women's Mental Health Center, and Assistant Professor of Psychiatry, OBGYN, and Reproductive Sciences

Secondary Mentors: Veerle Bergink, MD, PhD, Director, Women's Mental Health Centre, and Professor of Psychiatry, OBGYN, and Reproductive Sciences

Lazar Fleysher, PhD, Instructor of Diagnostic, Molecular, and Interventional Radiology

Abstract: Fetal movement is one of the earliest observable signs of nervous system development, yet little is known about how fetal behavior relates to the developing brain. Advances in fetal MRI now allow us to measure brain function before birth, while 4D ultrasound provides a direct window into spontaneous fetal behavior. However, these aspects of development have largely been studied separately, limiting our understanding of how neural and behavioral development unfold together in utero.

To address this gap, I will combine fetal resting-state functional magnetic resonance imaging (rsfMRI) with 4D ultrasound to examine fetal brain–behavior relationships during late gestation. Pregnant participants enrolled in an ongoing study will

Karen Levinson Mindich Trainee Pilot Program: 2026 Awardees

complete both assessments between 30 and 34 weeks of pregnancy. Fetal rsfMRI will be used to measure functional connectivity within the developing sensorimotor system, with a primary focus on communication between corresponding regions across the two cerebral hemispheres. During 4D ultrasound, spontaneous fetal movements and facial behaviors will be recorded and coded to quantify movement frequency, behavioral complexity, and organized or goal-directed actions.



Ezra Aydin, PhD
T32 Postdoctoral Fellow, Psychiatry

First, I will establish the feasibility of collecting and integrating high-quality fetal MRI and ultrasound data within the same developmental window. I will then test whether individual differences in fetal sensorimotor connectivity are associated with differences in observable fetal behavior. This work will establish a framework for directly studying coordinated brain and behavioral development before birth and provide foundational data for future longitudinal studies examining how fetal neurobehavioral development relates to newborn and later developmental trajectories.

Project Title: Immunometabolism in Lysinuric Protein intolerance

Investigator: Patrick O'Connell, DO, PhD, Chief Resident in Pediatrics, Genetics, and Genomic Sciences

Primary Mentor: Jaya Ganesh, MD, Professor, Genetics and Genomic Sciences

Secondary Mentors: Daniel Puleston, PhD, Director, Metabolomics Laboratory, and Assistant Professor of Immunology and Immunotherapy
Miriam Merad, MD, PhD, Dean of Translational Research and Therapeutic Innovation, Chair of Immunology and Immunotherapy, Director, Precision Immunology Institute, and Professor of Oncological Sciences and Medicine

Abstract:
Lysinuric Protein Intolerance (LPI) is a rare Inborn Error of Metabolism (IEM) resulting from bi-allelic pathogenic variants in SLC7A7. This results in loss of expression of the dibasic amino acid transporter, γ+LAT1, and subsequent dysregulation of arginine, lysine, and ornithine transport throughout multiple organ systems. The clinical features resulting from this uncommon urea cycle disorder include failure to thrive, osteoporosis, pulmonary alveolar proteinosis (PAP), anemia, hepatosplenomegaly, progressive renal disease, susceptibility to infections, and smoldering hemophagocytic lymphohistiocytosis (HLH). The significant number of immune symptoms these patients present with, particularly the smoldering HLH, are a significant cause of morbidity and mortality, yet there has been little research to understand the mechanisms tying dysregulated animo acid transportation to immune dysfunction. As such, there are no agreed upon immunomodulatory therapies for these patients.

This project aims to recruit a small cohort of LPI patients for a comprehensive, systems biology-based study into the extent, and underlying mechanisms linking metabolic dysfunction to immune dysfunction. We will employ unbiased plasma proteomics and metabolomics along with single-cell RNA-seq to obtain a holistic picture of the global metabolome and immune landscape. In vitro functional studies trialing various FDA approved immune modulators will also be performed. Through this work we hope to gain an understanding of the key drivers of immune dysfunction in LPI patients which will inform clinical decisions regarding immunomodulatory strategies and broaden our general understanding of the interplay between metabolism and the immune system in humans.



Patrick O'Connell, DO, PhD
Chief Resident, Pediatrics and Genetics and Genomic Sciences

MCHDI TLC: Building Community and Supporting Trainee Development

The Mindich Child Health and Development Institute (MCHDI) Trainee Leadership Committee (TLC) is dedicated to supporting the professional growth of our trainees. Our goal is to create opportunities for individuals at all stages and from all backgrounds to connect, exchange ideas, and build a strong sense of community within MCHDI.

In 2021–2022, the TLC launched the MCHDI Trainee Incubator Series, giving postdocs, PhD students, and master's students the chance to lead seminars and receive feedback on project proposals, grant applications, job talks, and conference presentations. This program was the first in a growing set of TLC initiatives. Guided by input from trainees, we continue to develop new programming focused on:

1. Career development, grant writing, and data analysis
2. Encouraging collaboration across disciplines and training levels
3. Creating more opportunities for faculty–trainee networking

We value your feedback and aim to tailor our events and resources to what will be most useful, so please let us know what sessions you'd like to see!

Since 2017, the TLC has also run a pilot grant program designed to help postdoctoral/clinical fellows and PhD/MD-PhD students launch new independent projects, an important step toward academic independence. Over the past ten years, eighteen trainees have been awarded.

This year's first awardee is Dr. Ezra Aydin, a postdoctoral fellow in the Department of Psychiatry mentored by Dr. Vanessa Babineau. The second awardee is Dr. Patrick O'Connell, a Chief Resident within Pediatrics and Genetics and Genomic Sciences mentored by Dr. Jaya Ganesh. Our awardees will share their research at the 14th annual MCHDI retreat this November.

Looking ahead, applications for the 2027–2028 academic year will open next spring, and we strongly encourage all trainees with exciting proposals to apply.

We are also deeply grateful to past and present TLC members for their dedication and hard work. Last year's committee was led by Shrey Patel, Aditi Prasad, Lauren Dierdorff, and Silvia De Rubeis. This year's committee includes Shrey Patel who serves as Chair, Aditi Prasad, Alexandra Trouilloud, and Silvia De Rubeis.

In addition to leading programs and events, one TLC member each year joins the planning committee for the MCHDI Annual Retreat. This year, Shrey will represent trainees on the retreat committee, ensuring their voices and perspectives are reflected. We're excited for another meaningful and impactful year ahead with the MCHDI trainee community!



Shrey Patel, BS
PhD Candidate, Biomedical Sciences



Aditi Prasad, MS
PhD Candidate, Developmental Regenerative and Stem Cell Biology



Alexandra Trouilloud
PhD Candidate, Stem Cell Biology and Regenerative Medicine



Silvia De Rubeis, PhD
Associate Professor, Psychiatry

Faculty Grants/Honors/Awards

Supinda Bunyavanich, PhD and **Scott Sicherer, PhD,** NIAID, U19, Immunologic Trajectories of Peanut Desensitization, May 2026-April 3031

Supinda Bunyavanich, PhD, Natasha's Foundation, Selected for Natasha's Prize Thinkery, a gathering of 35 experts selected from over 250 global applicants to work on food allergy prevention with a 10M British Pound prize

Jaime Chu, MD, Mirum Pharmaceuticals, "Defining fibrogenic niches in PFIC2"

Silvia De Rubeis, PhD, Co-Director of the PhD Program in Neuroscience

Silvia De Rubeis, PhD, Member of the Developmental Brain Disorders Study Section (DBD), Brain Disorders and Clinical Neuroscience Integrated Review Group (BDCN), NIH

Bruce D. Gelb, MD, NHLBI, UG3, "1/2 Clinical Trial of a MEK Inhibitor for RASopathy-Associated Severe Infantile Hypertrophic Cardiomyopathy"

Bruce D. Gelb, MD, NHLBI, P01, "Elucidating the contributions of genetic variation to congenital heart disease causality and outcomes"

Yuval Itan, PhD and **Shannon Kelleher, PhD**, NICHD, R01, "Investigating the genetic and genomic mechanisms of human lactation disorders"

Liora S. Katz, PhD, NIH/NIDDK, R01, "Molecular Glues for 14-3-3 and ChREBPalpha: Mechanisms and Therapeutic Potential for Protecting Pancreatic Beta-Cells"

Liora S. Katz, PhD, HIRN/NIDDK, R03, "Targeting ChREBPbeta to Protect beta-Cells from Metabolic and Inflammatory Stress in T1D"

Amy R. Kontorovich, MD, PhD, NHLBI, R01, "Am I ATTRisk? Health empowerment through population genomic screening for transthyretin amyloidosis"

Maria A. Curotto de Lafaille, PhD, NIAID, R01, "The biology of type 2 memory B cells and their role in allergy persistence"

Kaustav Mukherjee, PhD, NCATS, R03 Pilot Studies on Understudied proteins associated with rare diseases, "Contribution of the Integrator subunit INTS12 in transcription elongation control during human erythropoiesis and in a rare congenital erythroid disorder"

Kaustav Mukherjee, PhD, Cooley's Anemia Foundation Research Fellowship Award, "Role of Integrator complex subunits INTS11 and INTS12 in erythroid transcription control and hemoglobin switching in normal and ineffective erythropoiesis"

Dalila Pinto, PhD, NIMH, R01, "Characterization of splicing and isoform dysregulation in ASD at cellular resolution"

Trainee Grants/Honors/Awards

John Hong, MD, PI: Jaime Chu, MD, NIDDK, K08 Mentored Clinical Scientist Development Award "MMP7+ Hepatic Stellate Cells as Drivers of Fibrosis in Biliary Atresia"

Arushi Samal, PhD candidate, PI: Nicole Dubois, PhD, American Heart Association Basic Cardiovascular Sciences Meeting, "Spatiotemoral Mechanisms of Human Ventricular Chamber Development", presentation

Daniel R. Schecter, MD, MS, PI: Jaya Ganesh, MD, Icahn School of Medicine at Mount Sinai, Resident/Fellow Publication of the Month Award, "Resident/Fellow Publication of the Month – June 2026"

Daniel R. Schecter, MD, MS, PI: Jaya Ganesh, MD, United Mitochondrial Disease Foundation (UMDF), Mitochondrial Medicine Travel Scholarship, "2026 Mitochondrial Medicine Conference Travel Scholarship"

Daniel R. Schecter, MD, MS, PI: Jaya Ganesh, MD, United Mitochondrial Disease Foundation (UMDF), Mitochondrial Medicine Pathway for Advanced Clinical Training (M-PACT) Grant, "2027 Mitochondrial Medicine Pathway for Advanced Clinical Training (M-PACT) Fellowship"

Publications

Sanchez CM, **Crook S**, Jiang P, Dragan K, Woo JL, Newburger JW, ... **Anderson BR**. Congenital heart diseases involving congenitally diseased pulmonary valve phenotypes: Novel insights into the health care burden across the life course. *JTCVS Open*. 2026 Aug;32:101846.

Babineau V, Courtney K, Monk C, Widom CS. Childhood maltreatment and pregnancy-related reproductive health in middle adulthood: A prospective investigation. *Am J Obstet Gynecol MFM*. 2026 Jul;8(7):101984.

Ibrahim AY, Levine E, Iverson A, Sharma S, Forman L, Cuccia C, **Barbosa M**. Expansion of the clinical and molecular spectrum of lins1-associated disease. *Am J Med Genet A*. 2026 Sep 13.

Thinakaran A, **Foss-Feig J**, Cai S, Savino J, Suh M, Lavi A, ... **Buxbaum JD**, **Kolevzon A**, **Beker S**. Age-related increases in 40hz neural synchrony are specific to typical development: A cross-sectional study of autism spectrum disorder. *bioRxiv*. 2026 Aug 12.

Publications

Ravindranath Y, Craig DB, Parajuli P, Bagla S, Saad A, Cukovic D, ... **Bieker JJ**. Interplay between fibroblast growth factor 9 (fgf9) and cd44 underlies 46,xy disorders of sex development in kruppel-like factor 1 (klf-1)-e325k-associated congenital dyserythropoietic anaemia (cda-iv). *Br J Haematol.* 2026 Sep 17.

Breen MS, Tao R, Yang A, Wang X, Amini P, Rodriguez de Los Santos M, ... **Buxbaum JD**, **Grice DE**. Convergent molecular signatures across eating disorders and obsessive-compulsive disorder in the human brain. *Cell Rep.* 2026 Jun 23;45(6):117514.

Charney AW, Liharska LE, Vornholt E, Valentine A, Lund A, Hashemi A, ... **Breen MS**, .. Kopell BH. A transcriptional program associated with neurotransmission in the living human brain. *Mol Psychiatry.* 2026 May;31(5):2727-38.

Falcomatà C, Schaefer MM, Singh B, Chhamalwan D, Tepper A, Nielsen SR, ... **Brown BD**. A serpin-myeloid axis in pancreatic cancer heterogeneity and immune evasion. *Nature.* 2026 Sep 9.

Mattiuz R, Boumelha J, Aerakis E, Le Berichel J, Hamon P, Halasz L, ... **Brown BD**, ... Merad M. Dendritic cells control tertiary lymphoid structure development and maintenance in cancer. *Science.* 2026 Jul 16;393(6808):eady1678.

Zhang L, Chun Y, Valeiron S, Grishina G, Lo T, **Wang J**, **Sicherer S**, **Bunyavanich S**. Coordinated changes in oral propionate, oral microbiota, and peripheral blood inflammatory processes during peanut oral immunotherapy. *J Allergy Clin Immunol.* 2026 Sep 2.

Rossi F, Sportelli L, Kikidis GC, Grassi G, Di Camillo F, Bertolino A, ... **Buxbaum JD**, ... Pergola G. Co-expression-based models improve eqtl predictions for transcriptome-wide association studies and highlight new schizophrenia-associated genes. *Nat Genet.* 2026 Jul;58(7):1559-72.

Palmer DS, Hill B, Hodgson S, Jõeloo M, Kalantzis G, Kousathanas A, ... **Chami N**, ... **Itan Y**, ... Neale BM. The biobank rare variant consortium powers the discovery of rare genetic associations through global collaboration. *medRxiv.* 2026 May 24.

Amreen B, **Lesseur C**, Jagani R, Patel H, Katz D, Hamburger J, ... **Rommel AS**, ... **Gennings C**, ... **Chen J**. Prevalent detection of multiclass environmental chemicals in paired csf and plasma samples and their potential associations with postpartum depressive symptoms in pregnant women. *Environ Res.* 2026 Aug 1;302:124663.

Chorniy A, Owen AL, Kang RH, Cherupally M, Liss DT, French DD, ... Ackermann RT. Progression to diabetes by adults with prediabetes who use the national diabetes prevention program: A natural experiment. *Diabetes Care.* 2026 Sep 1;49(9):1576-84.

Swartz T, Cavazos JE, Horwitz M, Hu PJ, Sampson B, Scharschmidt T, ... **Chu J**, ... Williams CS. Navigating the path: Advice to physician-scientists on choosing a clinical specialty. *Elife.* 2026 Jul 3;15.

Slack HR, Byrom N, Bate C, Hościłowicz A, Homer S, Fuhrmann D, ... **Crook S**, Long E. From familiar faces to new spaces: Perceived social network change and its association with loneliness during the transition to university. *J Adolesc.* 2026 Aug 6.

Olguín Calderón D, Kilpatrick LE, Conil C, Philippot Q, Ogishi M, ... **Cunningham-Rundles C**, ... Boisson-Dupuis S. Humans homozygous for rare or common hypomorphic il23r variants are prone to tuberculosis. *J Exp Med.* 2026 Aug 3;223(8).

Kethidi N, Pothukuchi S, Aleman A, Charytonowicz D, Kuhn C, Wong T, ... **Cunningham-Rundles C**, ... Mehandru S. Persistence of mucosal car-t cells and inflammatory remodeling in enterocolitis associated with bcma car-t cell therapy. *Nat Med.* 2026 Sep 23.

Streit F, Awasthi S, Hall ASM, Braun A, Niarchou M, ... **Davis LK**, ... Witt SH. Genome-wide association analyses of borderline personality disorder identify 11 loci and highlight shared risk with mental and somatic disorders. *Nat Genet.* 2026 Aug;58(8):1831-44.

Dierdorff L, El Khaldi I, Garcia-Forn M, Wang W, Lukin J, Smith CM, ... **De Rubeis S**. *Ddx3x*-deficiency in mice impairs corticofugal motor circuits and causes motor deficits that are attenuated by early-life exercise. *PNAS*. In Press.

Banker SM, Schafer M, Barkley S, Trayvick J, Chen A, Peters AW, ... **Foss-Feig JH**, Schiller D. Neural tracking of social navigation in autism spectrum disorder. *Biol Psychiatry.* 2026 Aug 15;100(4):389-98.

Schecter DR, **Tinker RJ**, O'Connell P, Pang J, SzeKing Lee S, Kars ME, ... **Itan Y**, **Morava E**, **Kozicz T**, ... **Ganesh J**. Enrichment of rare mitochondrial DNA variants among individuals with kidney disease reveals undiagnosed mitochondrial disease. *Kidney Int Rep.* 2026 Jul;11(7):106578.

Schecter DR, Donnelly C, White A, Raynes H, Heller G, Rajan D, ... **Ganesh J**. Homozygosity for a clinically significant galc haplotype associated with late-infantile krabbe disease detected on newborn screening: Implications for clinical management and genetic counseling. *Int J Neonatal Screen.* 2026 Jul 29;12(3).

Gelb BD. Pediatric genomic medicine-rapid progress, but not too fast: American pediatric society's 2026 john howland award lecture. *Pediatr Res.* 2026 Jun 21.

Lieberman-Cribbin W, Spear E, **Gennings C**, Martinez M, Arora M, Galanti S, ... **Teitelbaum SL**, ... Stroustrup A. Assessing preterm infant metal exposures in shed teeth: Findings from the developmental impact of nicu exposures (dine) study. *J Expo Sci Environ Epidemiol.* 2026 Aug 20.

Acevedo-Sánchez J, Zajac L, Khurana S, Cloonan P, May P, Patlolla L, ... **Gennings C**, ... Mogilner L. Impact of an innovative community-academic partnership connecting families facing food insecurity with a food pantry. *Acad Pediatr.* 2026 Jul 2;26(7):103379.

Omar M, Agbareia R, Salah RA, Bragazzi NL, Apakama DU, **Glicksberg BS**, **Gelb BD**, **Holmes E**, ... Klang E. Sociodemographic variability in pediatric emergency decisions by ai. *Pediatrics.* 2026 Jun 1;157(6).

Klang E, Sorin V, Korfiatis P, Sawant AS, Freeman R, Charney AW, ... **Glicksberg BS**. Large language models are poor clinical administrators: An evaluation of structured queries in real-world electronic health records. *PLOS Digit Health.* 2026 May;5(5):e0001326.

Weiss EM, Brickler A, Beretta A, Ahmad KA, Dickert NW, Dorner RA, ... **Guttmann K**, ... Kraft SA. Ethical considerations for using alternative methods of consent in neonatal clinical trials: Recommendations from a modified delphi consensus process. *Arch Dis Child Fetal Neonatal Ed.* 2026 Apr 30.

Bankier S, Talukdar H, Khan M, Mocci G, Sukhavasi K, **Hao K**, ... Björkegren JLM. Plasma proteins are integral to cross-tissue gene regulatory networks implicated in cardiometabolic disorders and coronary artery disease. *Cell Syst.* 2026 Sep 16:101735.

Schmitz EG, Paul AJ, Ghosh R, Saucier N, Kolicheski A, Risma SI, ... **Itan Y**, ... Cooper MA. Targeted deep sequencing identifies mosaicism in patients with immune dysregulation. *J Allergy Clin Immunol.* 2026 May;157(5):1179-94.

Kayaalp B, Çil K, Conil C, Cobat A, Kars ME, **Itan Y**, ... Özçelik T. Prediction of human missense variant effects from functional evidence. *Nat Genet.* 2026 Sep;58(9):2350-60.

Levy T, Lapidus D, Friedman K, Siper P, Glass L, Squires L, ... **Buxbaum JD**, **Kolevzon A**. Prevalence of phelan mcdermid syndrome estimated to be ~1:7300 using a multisource model. *Autism Res.* 2026 Aug;19(8):e70297.

Publications

Li E, Liao SL, Trivieri MG, Gorevic P, Fettig V, Lala A, ... **Kontorovich AR**. Early-onset vitreous and cardiac amyloidosis with ttr trp61leu. *JACC Case Rep.* 2026 Jun 17;31(24):108337.

Shammas I, Iaali H, Watzlawik JO, Vidal Folch N, Dasari S, Preston G, ... **Kozicz T**, ... Naddaf E. Multilevel impairment of mitochondrial respiration in inclusion body myositis. *Brain.* 2026 Aug 2.

Da Silva A, Fallah MS, Tene Tadoum SB, Fazeli M, Muffels IJJ, Wang S, ... **Kozicz T**, ... **Morava E**, ... Pshezhetsky AV. Mice with the mono-allelic p.R37h dhdds variant show aberrant glycosylation and interneuron deficits. *Brain.* 2026 Sep 11.

Carroll KN, **Lesseur C**, Brunst KJ, Colicino E, Chiu YM, Hartman TJ, ... Wright RJ. Maternal lifetime stress and placental mitochondrial DNA mutational load: Effect modification by maternal characteristics. *Placenta.* 2026 Aug;182:158-66.

Halvorsen MW, Wang S, Miller-Fleming TW, Yu D, Topaloudi A, de Schipper E, ... **Mahjani B**, **Buxbaum JD**, **Grice DE**, ... **Davis LK**, ... Crowley JJ. Persistent tic disorders are associated with 17q12 duplications. *Mol Psychiatry.* 2026 Sep 16.

Caballero M, Satterstrom FK, **De Rubeis S**, **Buxbaum JD**, **Mahjani B**. Identification of moderate effect size genes in autism spectrum disorder through a novel gene pairing approach. *Commun Biol.* 2026 May 30;9(1).

Hathazi D, Lyons C, Lagos D, Podmanicky O, Zarate-Mendez M, Nie Y, ... **Morava E**, **Kozicz T**, ... Horvath R. Mitochondrial DNA heteroplasmy drives cortical neuronal disturbances in human organoids harbouring the common m.3243a>g mutation. *Nat Commun.* 2026 Jun 21;17(1).

Elsharkawi I, Preston G, **Kozicz T**, **Morava E**. Phosphodiesterase-5 inhibition as an off-label therapy in mitochondrial disease. *Mol Genet Metab.* 2026 Sep 16;149(3):110261.

Termorshuizen JD, Davies HL, Lee SH, Dennis JK, Hübel C, Johnson JS, ... **Pinto D**, ... **Davis LK**, ... Coleman JRI. Genomic meta-analyses of binge-eating behavior and anorexia nervosa yield insights into the unique and shared biology of eating disorder phenotypes. *Nat Ment Health.* 2026;4(9):1361-77.

Butta GM, Alburquerque B, Kearns C, Hadas Y, VanDyck MW, Scaglioni S, ... **Pinto D**, ... Simon V. Three immunoregulatory signatures define non-productive hiv infection in stem cell memory cd4(+) t cells. *Nat Commun.* 2026 Jun 25;17(1).

Ashley SA, Merritt K, Solmi F, Luque Laguna P, Logeswaran Y, Jones DK, ... **Reichenberg A**. Course of head growth from birth to adolescence across the psychosis spectrum. *Psychol Med.* 2026 Sep 9;56:e267.

Ghuwalewala S, Cao J, Rezza A, Rangl M, Kwiatkowski A, Brown A, ... **Rendl M**. Molecular signatures and signaling interactions of the hair follicle stem cell niche. *J Invest Dermatol.* 2026 Jul;146(7):1787-806.e18.

Cohen LE, Aydin E, Kaplan C, Patel D, Rooney T, Tubassum R, ... **Rommel AS**. Examining the association between prenatal inflammation and postpartum depression in a large, prospective pregnancy cohort in new york city. *Brain Behav Immun.* 2026 Aug;136:106593.

Amreen B, Milazzo F, Gigase F, D'Souza D, Samper N, Martin JT, ... **Chen J**, **Lesseur C**, **Rommel AS**. Impact of gestational maternal sars-cov-2 infection on neonatal inflammatory biomarkers. *Am J Reprod Immunol.* 2026 Aug;96(2):e70313.

Tsuang AJ, Chung SA, Lin AA, Baloh CH, Ruiz CA, Svedenhag P, ... **Sampson HA**. Elevated neonatal electrical impedance spectroscopy measurements are associated with atopic dermatitis in high-risk infants. *Allergy.* 2026 Jul;81(7):2557-9.

Kuraoka S, Higashi Y, Saito S, Pourgonabadi S, Honjoh H, Ishigaki S, ... **Satlin LM**, ... Morizane R. Deciphering the impact of rac1-sptan1 in arpkd cystogenesis using multifaceted models. *Adv Sci (Weinh).* 2026 May;13(25):e24001.

LaFlamme CW, Clarkson C, Ibañez K, Wang JY, Sengupta S, Lord J, ... **Sharp AJ**, ... Mefford HC. Multiomic approaches identify a rare ccg repeat expansion in bclaf3 in neurodevelopmental disorders. *Genome Med.* 2026 Jul 22.

Garg P, Jadhav B, Shadrina M, Martin-Trujillo A, **Sharp AJ**. A phenome-wide association study of cnvs genotyped from genome sequencing read depth in the uk biobank. *Am J Hum Genet.* 2026 Jun 4;113(6):1297-318.

Shemesh E, Anand R, Annunziato RA, Fredericks EM, Garcia J, Gupta NA, ... Shneider BL. A remote intervention to improve medication nonadherence guided by a marker of risk derived from the electronic health records of adolescent transplant recipients. *Am J Transplant.* 2026 Sep;26(9):2099-110.

Ponda P, Bird JA, Bufford JL, Cianferoni A, Faig W, Kleiner GI, ... **Sicherer SH**, Assa'ad A. Food oral immunotherapy patient selection, dosing, and monitoring practices among a network of us food allergy centers. *J Allergy Clin Immunol Pract.* 2026 Jul 8.

Pers TH, Alhadeff AL, Myers MG, Jr., Adriaenssens AE, Fenselau H, **Stanley SA**, ... Mutlu B. Decoding neural signals to build metabolic therapies. *Cell Metab.* 2026 Aug 4;38(8):1513-7.

Wang P, Kumar K, **Lambertini L**, Liu H, Wood O, Chen S, ... **Scott DK**, ... **Garcia-Ocana A**, **Karakose E**, ... **Stewart AF**. Harmine selectively enhances human beta cell identity markers and function via protein kinase a pathways. *Diabetologia.* 2026 Sep 21.

Thai BQ, Sargent EJ, Luff SA, Fields CA, Churko JM, Young JN, ... **Sturgeon CM**, Bhattacharya D. Wnt signaling in human pluripotent stem cells promotes hdac2-dependent epigenetic programs and development of retinoic acid-responsive mesoderm. *Stem Cell Reports.* 2026 Jul 14;21(7):102964.

Krasnov H, Hung W, Knobel P, Kloog I, Ko F, Thompson H, ... **Teitelbaum SL**, ... Yitshak-Sade M. Urban exposures, frailty, and mental illness in world trade center health program responders. *Nat Commun.* 2026 Sep 11;17(1).

Shyr C, **Tinker RJ**, Brown RF, Wright A, Peterson JF, Phillips JA, 3rd, ... Bastarache L. An ehr-based framework for modeling growth curves and constructing growth centile charts for genetic disorders. *NPJ Genom Med.* 2026 Jul 3;11(1).

Carney M, Campbell K, Maddali R, Kim M, Gao Q, Thompson J, **Tiozzo C**. Association between essential fatty acid source and holman index in preterm infants: A prospective cohort study. *JPEN J Parenter Enteral Nutr.* 2026 Sep 27.

Ramaekers K, Tran M, Lunghi M, Hermans A, Thys C, De Wispelaere K, **Turro E**, ... Freson K. Reduced platelet formation associated with serine metabolic dysregulation in integrin αiibβ3-deficient megakaryocytes. *Blood.* 2026 Sep 10;148(11):1466-80.

Freed G, Quijada Álamo M, Lee L, Wang K, Poddar N, Autar S, ... **Wagenblast E**. A crispr-based humanized model reveals cooperative role of stag2 loss in familial gata2-deficient mds progression. *Blood Adv.* 2026 Sep 24.

Lama C, Isakov D, Rosenberg S, Quijada-Álamo M, Saurty-Seerunghen MS, Moein S, ... **Wagenblast E**, ... Nam AS. Type 1 interferon perturbates clonal competition by reshaping human blood development. *Nat Genet.* 2026 Sep 15.

Bikou O, Halouani A, Sun Y, Bisserier M, Eisenacher C, Antar SA, ... **Walsh MJ**, ... Sassi Y. Microrna-224 orchestrates bmp and tgfβ signaling and is a therapeutic target in preclinical pulmonary arterial hypertension. *Sci Transl Med.* 2026 Sep 2;18(865):eaef6676.

Publications

Wang J, Khoshbakht S, Lee KW, Jones SM, Huffaker MF, Plaut M, ... **Sampson HA**. Epitope-specific ige profiles are predictive of desensitization and remission in the impact trial. *J Allergy Clin Immunol*. 2026 Jul 24.

Janevic T, Kim H, Ng A, Howell FM, Liu SH, Fox A, **Weber E**. Racial inequities in postpartum coverage during medicaid continuous coverage: Evidence from affordable care act expansion versus nonexpansion states. *Am J Prev Med*. 2026 Jul;71(1):108320.

Luke MJ, **Wood SM**, Beck AF. Use of the child opportunity index in adolescent and young adult health research. *J Adolesc Health*. 2026 Aug;79(2):183-5.

Zhou EG, Day S, Argenio K, Schwartz AE, Elbel B. Factors associated with covid-19 primary and booster vaccine uptake among new york city public school students. *Am J Public Health*. 2026 Aug 6:e1-e11.

SAVE THE DATE

14th Annual MCHDI Retreat

Date: November 10, 2026

Time: 8:00AM – 6:00PM

Location: Harmonie Club

Ballroom, 1st Floor

4 E 60th St, New York, NY 10022

Icahn School of Medicine at Mount Sinai

The Mindich Child Health and Development Institute

Icahn School of Medicine at Mount Sinai

The Mindich Child Health and Development Institute

Website: www.mountsinai.org/mchdi
Email: mchdi@mssm.edu
Facebook: www.facebook.com/mindichchdi
Twitter: @MindichCHDI
Contact: Tel: (212) 824-8938 Fax: (212) 241-3310
Address: 1470 Madison Avenue, 8th Floor
Hess Center for Science and Medicine at Mount Sinai
New York, NY 10029-6542