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

MCHDI Developmental Outcomes

SPRING 2026

Research Advancements: Diabetes and Drug Development

Mount Sinai's Diabetes Obesity Metabolism Institute and Drug Discovery Institute Make Major Advances In Human Beta Cell Regenerative Drug Development for Types 1 and 2 Diabetes

The Discovery of DYRK1A Inhibitors for Diabetes. All diabetes results from inadequate production of insulin by the "beta cells" in the pancreas. Dr. Andrew Stewart was recruited to Mount Sinai in 2012 with the *vision* that human beta cell regenerative therapy was possible, and with the *goal* of identifying a simple oral pill that could be taken by the 800 million people in the world with diabetes. This vision was realized in 2015 when Drs. Stewart and Peng Wang, using a robotic high-throughput screen of 102,000 candidate drugs, found one, called harmine, that for the first time is able to induce beta cells from human organ donors to proliferate or regenerate. Harmine targets and inhibits an enzyme in beta cells named "DYRK1A". In 2019, a graduate student in the group, Courtney Ackeifi, showed that harmine not only makes human beta cells proliferate, it also enhances their ability to make and secrete insulin: it makes "more and better beta cells". The group also reported that combining any one of a number of DYRK1A inhibitor drugs with currently existing GLP1 receptor agonists (GLP1RA's) such as semaglutide, exenatide, liraglutide and others, produced a synergistic further increase in human beta cell proliferation to high levels.

Animal Studies Demonstrate Efficacy. All of the work described thus far was performed in tissue culture (think of petrie dishes). In 2024, in an effort to show that these events could occur in living animals, Drs. Adolfo Garcia-Ocana and Sarah Stanley developed a system in which human pancreatic beta cells could be transplanted into mice that lack an immune system. Thus, the human beta cells would not be rejected as "foreign", and the mice containing human beta cells could be treated with harmine. These experiments made the critical observation that harmine and other DYRK1A inhibitors not only made human beta cells proliferate in a petrie dish ("in vitro"), they also did this in live mice with human beta cell grafts ("in vivo"). Specifically, harmine alone was able to increase human beta cell mass by 300% over three months, and if a GLP1RA was added to harmine, this increased to 700%. This answered the question: "could DYRK1A inhibitors prove to be be effective in people living with T1D or T2D?" with a resounding "yes". Equally importantly, these *in vivo* experiments showed that harmine not only increased human beta cell mass, it also rapidly (within a week) reversed diabetes.

Human Studies Reveal Safety. But there were two problems. First, harmine is a natural compound derived from plants like *Peganum harmala* and *Banisteriopsis caapi*. Since these plants are used as "recreational" psychoactive brews, this created worries that harmine might cause hallucinations if used in humans with diabetes. To directly address this question, Drs. Jessica Ables and James Murrough in Mount Sinai's Department of Psychiatry performed a Phase 1 trial in normal volunteers and found that, in doses predicted to be effective in human diabetes, harmine is not psychoactive, and was well tolerated.

Development of Novel Next-Gen Compounds. The second problem was that since harmine is a naturally existing molecule, it cannot be patented, and therefore is not attractive for commercial development. To address this challenge,

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Drs. Robert DeVita and Kunal Kumar in Mount Sinai's Drug Discovery Institute began a campaign to chemically synthesize more than 1000 novel potential DYRK1A inhibitors. This program yielded a list of next-generation, entirely novel compounds, several of which are considerably more potent and effective than harmine in inducing human beta cells to regenerate, and in reversing diabetes.

Intellectual Property and Commercialization. The studies described above have led to a series of patents filed through Mount Sinai Innovative Partners (MSIP), Mount Sinai's technology transfer office. These patents were recently licensed to a biotechnology company for full commercialization of Mount Sinai's next-generation DYRK1A inhibitors. This is now moving rapidly.

What's Next? As with most new drugs, the FDA requires that they are safe and effective, but that does not necessarily mean that their mechanism of action is fully understood. In the case of DYRK1A inhibitors, we know that they make *more better beta cells*. But there is much to learn: although we have a deep understanding as to how they make human beta cells *proliferate*, we have little understanding as to how they further enhance human beta cell *function*. Thus, the main current projects in the Stewart/Wang lab focus on understanding this important, beneficial, but unresolved mechanism of action. A new paper is under review at present. The main current projects in the DeVita/Kumar lab are the development of even more effective, more potent, more selective DYRK1A inhibitors, and their potential applications to other diseases. The current efforts in the Garcia-Ocana labs, now at City of Hope in California, are showing that, and how, DYRK1A inhibitors are effective in the autoimmune setting of Type 1 diabetes. Great and rapid progress is continues in all three labs.

Support. This work has been supported since 2012 with multiple Mindich Institute Pilot Awards to Drs. Lauryn Choleva, Sarah Stanley, Esra Karakose and others. (The work has also received continuous support from the National Institutes of Health and BreakthroughT1D (formerly JDRF).

Andrew F. Stewart, MD
Director, Diabetes Obesity and Metabolism Institute
Professor, Medicine, Endocrinology,
Diabetes and Bone Disease

Key Publications:

1. Wang P, Felsenfeld DP, Liu H, Sivendran S, Bender A, Kumar A, Alvarez-Perez JC, Garcia-Ocana A, Sanchez R, Scott DK, Stewart AF. A high-throughput chemical screen reveals that harmine-mediated inhibition of DYRK1A increases human pancreatic beta cell replication. *Nature Medicine* 21:383-388, 2015.
2. Kumar K, Wang P, Sanchez R, Swartz EA, Stewart AF, DeVita RJ. Development of kinase-selective, harmine-based DYRK1A inhibitors that induce human pancreatic beta cell proliferation. *J Med. Chem.* 61:7687-7699, 2018.
3. Kumar K, Ung P M-U, Wang P, Wang H, Li H, Andrews MK, Stewart AF, Schlessinger A, DeVita RJ. Novel Selective Thiadiazine DYRK1A Inhibitor Lead Scaffold with Human Pancreatic β-cell Proliferation Activity. *Eur J Med Chem.* 157:1005-1016, 2018.
4. Wang P, Karakose E, Liu H, Swartz E, Ackeifi C, Zlatanic V, Wilson J, Argmann C, Scott DK, Garcia-Ocana A, Stewart AF. Combined Inhibition of DYRK1A, SMAD and Trithorax Pathways Synergizes to Induce Robust Replication in Adult Human Beta Cells. *Cell Metabolism* 29:638-52, 2019.
5. Ackeifi C, Wang P, Karakose E, Manning Fox JE, González BJ, Liu H, Wilson J, Swartz E, Berrouet C, Li Y, Kumar K, MacDonald PE, Sanchez R, Thorens B, DeVita R, Homann D, Egli D, Scott DK, Garcia-Ocaña A, Stewart AF. GLP1 Receptor Agonists Synergize with DYRK1A Inhibitors To Potentiate Functional Human Beta Cell Regeneration. *Science Translational Medicine* 12, eaaw9996, 2020.
6. Wang P, Karakose E, Argmann C, Wang H, Balev M, Brody R, Rivas H, Liu X, Wood O, Liu H, Choleva C, Hasson D, Paulo JA, Scott DK, Lambertini L, DeCaprio JA, Stewart AF. Disrupting the DREAM Complex Enables Proliferation of Adult Human Pancreatic Beta Cells. *J Clin Invest.* 132:e157086.
7. Karakose E, Wang X, Wang P, Caracamo S, Demircioglu D, Lambertini L, Wood O, Kang R, Lu G, Scott DK, Garcia-Ocana A, Argmann C, Sebra R, Hasson D, Stewart AF. Cycling Alpha Cells In Regenerative Drug-Treated Human Pancreatic Islets May Serve As Key Beta Cell Progenitors. *Cell Reports Medicine*.
8. Rosselot C, Li Y, Wang P, Alvarsson A, Beliard K, Lu G, Kang R, Li R, Liu H, Gillespie V, Tzavaras N, Kumar K, DeVita RJ, Stewart AF, Stanley SA, Garcia-Ocaña A. Harmine and Exendin-4 Combination Therapy Safely Expands Human Beta Cell Mass in Vivo In A Mouse Xenograft System. *Science Translational Medicine.* 10;16(755):eadg3456.
9. Ables JL, Israel L, Wood O, Govindarajalu U, Freemont R, Banerjee R, Liu H, Cohen J, Wang P, Kumar K, Lu G, DeVita RJ, Garcia-Ocaña A, Murrough J, Stewart AF. Clinical Outcomes In Healthy Subjects Receiving A Single Oral Dose Of Harmine. *Psychopharmacology* 38:910-919, 2024.

Interprofessional perspectives on communication quality in the Neonatal Intensive Care Unit

With advances in care for the most vulnerable infants in the Neonatal Intensive Care Unit (NICU), parents of NICU babies find themselves grappling with complex information and making difficult decisions. As a result, communication between NICU parents and clinicians has never been more important. Communication quality directly affects patient and parent outcomes in the NICU. Despite its importance, NICU clinicians often have limited communication skills training and may struggle to communicate optimally in the face of increasing clinical demands and staffing shortages.

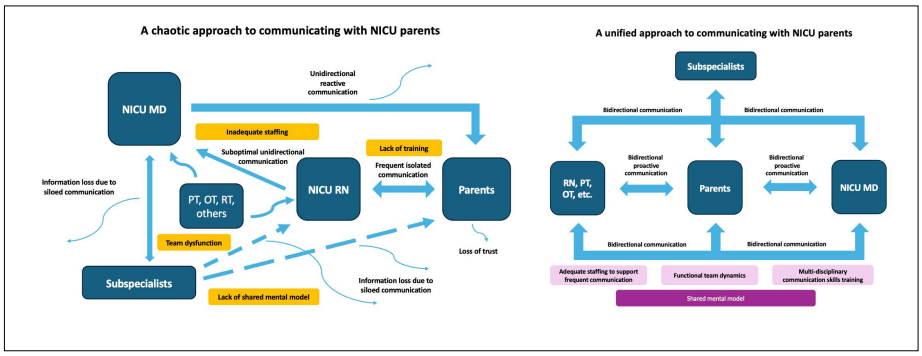


Figure: A chaotic vs. unified approach to communicating with parents in the NICU

A range of provider types comprise the interprofessional NICU team, each bringing a unique perspective. Yet there is limited literature reflecting these diverse points of view. Our team used a novel 1-question qualitative interview approach to capture the perspectives of a broad range of NICU staff on communication quality.

The team interviewed 62 clinicians (nurses, physicians, physician assistants, nurse practitioners, occupational therapists, social workers, child life specialists, a music therapist, chaplain, discharge coordinator, and a patient care associate). Using thematic analysis, researchers identified two categories of themes: messaging and dynamics. 'Messaging' themes centered around clarity (setting goals; using clear and direct language/avoiding jargon; ensuring parental understanding) or timing and approach (unhurried, frequent, timely, proactive). Within 'dynamics', themes were related to team interactions (inclusivity, respect, consistency) and parent-centeredness (viewing parents as key stakeholders, identifying and adapting to communication needs, creating a safe space, responding with empathy, offering flexible modes of communication).

Staff members prioritize use of clear and direct language to ensure parents understand clinical updates. Participants emphasized a need for timely and proactive communication, which has been previously recognized as inconsistently provided in the NICU. Nurses identified inclusive and respectful team communication as essential to parent-clinician communication, while physicians were less likely to include team dynamics in a definition of high-quality communication. Non-physician team members were more focused on ensuring parent-centeredness than were physicians.

Study findings indicate that low-quality approaches to communication may lead to loss of trust, loss of information, and parent distress, and are exacerbated by systems level issues including lack of training, inadequate staffing, and team dysfunction. In contrast, high-quality communication acknowledges the key role of parents, is bidirectional, and is founded on a shared mental model. This collective understanding underlies functional team dynamics and a cohesive approach to training (see Figure). The conceptual framework shown here can serve as a first step towards such a shared mental model. Investing in improving team dynamics, equipping teams with a shared model of and approach to high quality communication and ensuring adequate staffing may be beneficial for improving communication with parents.

Our ongoing research will contribute to the model developed following completion of this study by incorporating data from interviews with parents and ethnographic observations in the NICU. Future work will aim to develop communication tools and interventions to begin to improve communication in the NICU and thereby improve outcomes for NICU babies and their parents.

Katherine Guttmann, MD, MBE
Associate Professor, Pediatrics

Mindich Scholars Faculty Pilot Awards Program: 2026 Awardees

Project Title: Defining Exercise-Induced Pathways of Cardioinflammation in Genetically Susceptible Myocardium

Principal Investigators: Amy Kontorovich, MD, PhD (Communicating PI) and Cameron McAlpine, PhD (Co-PI)

Abstract: The American Heart Association recommends that school-aged children engage in at least 60 minutes of moderate-to-vigorous intensity exercise per day. However, for children harboring certain genetic variants, this level of exercise may be harmful or even fatal. Specifically, in individuals with putatively damaging variants in desmosomal (i.e., PKP2, DSP, DSC2, DSG) genes or the gene encoding dystrophin (DMD), exercise exceeding the low intensity level can trigger the onset or progression of cardiomyopathy, ventricular tachyarrhythmias or sudden death. Variant carriers are understandably advised to avoid exercise, but this can lead to sedentary lifestyle, obesity and unintended accrual of atherosclerotic risks. Prior rodent students in these genotypes suggest that the root cause of this myocardial injury is cardioinflammation, highlighting immune and metabolic derangements. However, surprisingly little is known about why or how alterations in genes encoding structural elements of the cardiomyocyte can drive inflammation. The complex molecular and cell-cell networks in the heart that associate host cardiomyocyte genetics with cardiac macrophage activation and environmental triggers like exercise in outcomes of myocardial inflammation remain surprisingly understudied. We hypothesize that genetically-mediated changes in cardiomyocyte structural integrity induce basal alterations of mitochondrial function and impairments in innate immune signaling. We also hypothesize that upon exposure to exercise, genetically susceptible myocardium succumbs to inflammation through different immunometabolic-mediated pathologic mechanisms: basal IFN activation in DMD deficiency leads to innate immune exhaustion, while in PKP2 deficiency, NF-kB-mediated changes in mitochondrial function promote fulminant disease and unresolved cardiac inflammation. In this highly translational study, we will uncover novel pathways of myocardial inflammation via host genetic, immune and environmental (exercise) interactions using hiPSC-derived cardiomyocyte and mouse models bearing relevant PKP2, DSP and DMD mutations. Defining genotype-specific pathways and exercise thresholds in these models will enable future studies toward more nuanced, safe and effective physical activity recommendations for children and adults at elevated genetic risk.

Amy Kontorovich, MD, PhD (Communicating PI)
Associate Professor, Medicine, Cardiology and Genomic Medicine
Director, Center for Inherited Cardiovascular Diseases

Cameron McAlpine, PhD (co-PI)
Associate Professor, Medicine, Cardiology
Fuster Heart Hospital

Project Title: Impact of Blood-Borne Factors on Social Deficits Induced by Early Life Social Isolation

Principal Investigators: Hirofumi Morishita, MD, PhD (Communicating PI) and Joseph Castellano, PhD (Co-PI)

Abstract: Early life social isolation can have long-lasting consequences on risk for developing psychiatric disorders including substance-use disorder (SUD). Determining the mechanistic impact of early life social isolation will guide new treatments that can be applied to reduce risk of psychiatric conditions. Animal studies have shown that juvenile social isolation dysregulates social and cognitive behaviors. However, the specific molecular and circuit mechanism driving the deficits remain poorly understood. Notably, social isolation not only impacts the brain but also affects the body, a process that may be bidirectional. Studies in the aging field revealed a critical contribution of blood-borne factors on brain function, but far fewer studies on blood-to-brain axis are conducted in the field of child health, and none in the context of early social isolation. The goal of this pilot study is to assess the impact of blood-borne factors on social and cognitive behavioral changes in a mouse model induced by juvenile social isolation at behavioral (Aim1), cellular/circuit (Aim2), and molecular levels (Aim3). Based on our intriguing preliminary data supporting a protective role of plasma from juvenile group-housed mice on isolation-induced social behavior, we will test the hypothesis that juvenile social isolation induces specific molecular changes in the systemic environment that modulate sub-cortical circuits to trigger social rebound behavior, which is associated with later SUD-related behaviors. To test this hypothesis, we will combine innovative approaches, including, but not limited to, machine learning-based behavioral characterization, circuit analysis in behaving animals, and aptamer-based multiplexed proteomic technology. We are well qualified to conduct the proposed study. Dr. Morishita (co-PI) has a demonstrated expertise on examining the impact of juvenile social isolation on social circuit and behavior, and Dr. Castellano (co-PI) has extensive expertise in studying the impact of blood-borne factors on brain function, making both PIs highly suited to conduct the proposed collaboration.

Hirofumi Morishita, MD, PhD (Communicating PI)
Professor, Psychiatry, Neuroscience, and Ophthalmology
Mindich Child Health and Development Institute

Joseph Castellano, PhD (Co-PI)
Associate Professor, Neuroscience, Neurology, and Stem Cell Biology & Regenerative Medicine Black Family Stem Cell Institute

Mindich Scholars Faculty Pilot Awards Program: 2026 Awardees

Project Title: Natural History and Mechanistic Studies of ReNU and ReNU2 Syndromes

Principal Investigators: Ernest Turro, PhD (Communicating PI) and Mafalda Barbosa, MD, PhD (Co-PI)

Abstract: We have recently shown that variants in RNU4-2 and RNU2-2 — two paralogs of genes that are transcribed into small nuclear RNA (snRNA) components of the major spliceosome — are prevalent causes of neurodevelopmental disorders (NDDs). Mutations in RNU4-2 cause the most prevalent known autosomal monogenic NDD; mutations in RNU2-2 cause an NDD with a prevalence ≈20% that of RNU4-2 syndrome; and biallelic variants in RNU2-2 cause the most prevalent known recessive NDD. In aggregate, >100,000 people are likely affected, but few have received a genetic diagnosis because the gold standard diagnostic assay, whole-exome sequencing, does not capture variation in these genes. Although the functions of canonical snRNAs U4 and U2 in the major spliceosome are well understood, there is limited evidence that pathogenic variants in RNU4-2 and RNU2-2 impair splicing, suggesting other mechanisms may be at play.

In Aim 1, we will recruit families with a potential RNU4-2/RNU2-2-related NDD to join our IRB-approved INDEED study. We will capture phenotype information to describe the natural histories of the disorders and identify phenotype-genotype correlations. In Aim 2, we will differentiate wildtype and edited iPSCs with pathogenic RNU4-2 mutations (already available) into neuronal stem cells and neurons, and perform transcriptomic and molecular localization studies. In Aim 3, we will reprogram primary fibroblasts from RNU2-2 patients in the INDEED study into iPSCs for future investigation (patient samples are required because, in contrast to RNU4-2, RNU2-2 cannot be edited by CRISPR/Cas9 without significant off-target effects).

This pilot project is intended to generate preliminary data to support NIH grant applications seeking to establish the INDEED study as a center of excellence for RNU4-2 and RNU2-2 related NDDs and to pursue mechanistic studies using cell lines. Our interdisciplinary team, with expertise in genetics/statistics/computing (Turro), clinical genetics (Barbosa) and neuroscience/natural history studies (Morava), has the ideal composition to pursue this work.

Ernest Turro, PhD (Communicating PI)
Associate Professor, Genetics and Genomic Sciences

Mafalda Barbosa, MD, PhD (Co-PI)
Assistant Professor, Genetics and Genomic Sciences and Pediatrics

New Extramural Faculty

Vanessa Babineau, PhD

Vanessa Babineau, PhD, is an Assistant Professor in the Departments of Psychiatry and Obstetrics, Gynecology and Reproductive Science (Ob/Gyn) at the Icahn School of Medicine at Mount Sinai (ISMMS), and Director of the Resilient Beginnings Laboratory (REBEL). She is the recipient of NIH K99/R00 and BBRF grants. Dr. Babineau studies intergenerational trauma and infant brain and behavior development. She identifies biological and behavioral pathways of intergenerational trauma and risk for mental health disorders to future generations in the context of mental health during pregnancy (e.g., depression, anxiety, PTSD, bipolar disorder) among persons who have a history of trauma including experiences of childhood maltreatment. Mother-child dyads are followed longitudinally to identify associations with newborn amygdala connectivity, infant emotion regulation, mother-infant interactions, parenting sensitivity, and risk for psychopathology. Dr. Babineau's research promotes resilient beginnings by supporting the development of trauma-responsive preventive perinatal interventions integrated in Ob/Gyn care, with a two-generation impact.

Dr. Babineau serves as a licensed Clinical Psychologist in the Women's Mental Health Center, a mental health service in collaboration with Mount Sinai's Ob/Gyn practices. She specializes in the trauma-responsive care and the treatment of mood and anxiety disorders in the perinatal period.

Key Publications:

1. **Babineau V,** Courtney K, Monk C, Widom, CS. Childhood maltreatment and pregnancy-related reproductive health in middle adulthood: A prospective investigation. *American Journal of Obstetrics & Gynecology MFM*. [In Press]
2. **Babineau V**, *Lavallée A, D'Antonio K, Werner E, Drysdale AT, Osbourne M, Grubb M, Moise N, Reuveni I, Zhang Z, Lee S, Dumitriu D, D'Alton M, Monk C. Mental health service utilization in a novel insurance-based Ob/Gyn integrated model for women across the lifespan. *Arch Womens Ment Health*. 2026 Jan 5;29(1):5. *Co-first author
3. **Babineau V**, McCormack CA, Feng T, Lee S, Berry O, Knight BT, Newport JD, Stowe ZN, Monk C. Pregnant women with bipolar disorder who have a history of childhood maltreatment: Intergenerational effects of trauma on fetal neurodevelopment and birth outcomes. *Bipolar Disord*. 2022 Sep;24(6):671-682.
4. **Babineau V**, Fonge YN, Miller ES, Grobman WA, Ferguson PL, Hunt KJ, Vena JE, Newman RB, Guille C, Tita ATN, Chandler-Laney PC, Lee S, Feng T, Scorza P, Takács L, Wapner RJ, Palomares KT, Skupski DW, Nageotte MP, Sciscione AC, Gilman S, Monk C. Associations of Maternal Prenatal Stress and Depressive Symptoms With Childhood Neurobehavioral Outcomes in the ECHO Cohort of the NICHD Fetal Growth Studies: Fetal Growth Velocity as a Potential Mediator. *J Am Acad Child Adolesc Psychiatry*. 2022 Sep;61(9):1155-1167.

Dr. Babineau completed a PhD at McGill University, followed by a Postdoctoral Research Fellowship at Columbia University and subsequent appointment as Assistant Professor. She joined ISMMS in 2026 where she is a member of the Women's Mental Health Center, the Mindich Child Health and Development Institute, the Friedman Brain Institute, and the BioMedical Engineering and Imaging Institute (BMEII).

Vanessa Babineau, PhD
Assistant Professor, Psychiatry and Obstetrics, Gynecology and Reproductive Science
Director, Resilient Beginnings Lab
Women's Mental Health Center

Key Publications:

5. Berry OO, **Babineau V**, Lee S, Feng T, Scorza P, Werner EA, Monk C. Perinatal depression prevention through the mother-infant dyad: The role of maternal childhood maltreatment. *J Affect Disord*. 2021 Jul 1;290:188-196.

Rory J. Tinker, MBChB

Rory J. Tinker, MBChB, is an Assistant Professor in the Department of Genetics and Genomic Sciences, with a secondary appointment in Pediatrics, at the Icahn School of Medicine at Mount Sinai. A board certified Clinical Geneticist and Pediatrician, Dr. Tinker's research integrates genomics, population biobanks, and electronic health records (EHRs) to accelerate diagnosis and discovery in rare genetic diseases.

His work focuses on quantifying diagnostic delay, developing EHR-based phenotypes, and implementing AI-enabled tools to identify genetic diseases earlier and more equitably. Dr. Tinker trained in Neuroscience and Medicine at the University of Manchester before completing fellowships in mitochondrial genetics at the University of Newcastle and the University of Pennsylvania, followed by a combined Pediatrics and Medical Genetics residency at Vanderbilt University Medical Center. His research has yielded more than 40 peer-reviewed publications, including first-author papers in Genetics in Medicine and American Journal of Medical Genetics, and was recognized with the ACMG Richard King Trainee Award. Dr. Tinker has developed novel pipelines for variant interpretation and characterized diagnostic trajectories across Mendelian disorders. His current K08 award proposal aims to combine artificial intelligence and implementation science to reduce diagnostic delays in rare diseases, under the mentorship of Drs. Ron Do, Eva Morava, and Bruce Gelb. His long-term goal is to lead a translational research program that bridges computational discovery and clinical application to improve outcomes for patients with rare genetic disorders.

Rory J. Tinker, MBChB
Assistant Professor, Genetics and Genomic Sciences

Key Publications:

1. Schecter DR, **Tinker RJ,** O'Connell P, Pang J, SzeKing Lee S, Kars ME, Guvenek A, Preuss M, Itan Y, Morava E, Kozicz T, Hirano M, Naini A, Ganesh J. Enrichment of Rare mtDNA Variants Among Individuals with Kidney Disease Reveals Undiagnosed Mitochondrial Disease. *Kidney International Reports* (2026).
2. Al-Shahrani H, Szabó E, Staccone C, MacDonald G, Furuta Y, Schecter D, Edmondson AC, McRae A, Baker J, Morava E, **Tinker RJ.** Expanded Clinical Spectrum of Autosomal-Dominant STT3A-CDG. *Biomolecules*. 2026 Mar 12;16(3):418.
3. Furuta Y, Agrawal NS, Owen NN, Bastarache LA, Shyr C, Geiger HU, Jackson S, Lauderdale CJ, Carter CR, Morgan KA, Hamid R, Phillips Iii JA, **Tinker RJ**. Sex-Specific Diagnostic Inequality in Fabry Disease: Lessons Learned from Analysis of Newborn Screening and Cascade Testing in Tennessee from 2017 to 2024. *Public Health Genomics*. 2026;29(1):73-82.
4. **Tinker RJ**, Richter LD, Furuta Y, Shyr C, Shirazi SM, Sucre J, Gatta LA, Phillips JA 3rd, Peterson JF, Bastarache L. Evaluating pregnancy and neonatal outcomes in mothers with genetic disease using electronic health care records. *Genet Med.* 2026 Mar;28(3):101681.
5. **Tinker RJ**, Jacob N, Syed MG, Kelkar J, Donnelly C, Elsharkawi I, Ganesh J, Gelb B, Pejaver V, Kozicz T, Morava E. Drivers of Diagnostic Delay in Mitochondrial Disease: Missed Recognition of Canonical Features. *medRxiv* [Preprint]. 2025 Oct 14:2025.10.09.25337582.

Cecile Yama, MD

Cecile Yama, MD, is an Assistant Professor in the Center for Child Health Services Research at the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai and a general pediatrician in the Department of Pediatrics. She also holds a co-appointment in the Department of Population Health Science and Policy. Dr. Yama's clinical, research, and advocacy work bridges policy and clinical care to foster healthy environments for children, beginning in early childhood. Clinically, she is a board-certified pediatrician with specialized expertise in trauma-informed care for young children with behavioral, developmental, and medical complexity. She has advanced training in infant complex care, NICU follow-up, and primary care psychiatry. Her clinical practice centers on building long-term, family-centered partnerships, supporting families as they navigate complex medical and social systems, streamlining care across providers, and integrating behavioral and social supports into primary care.

Her research examines how economic and social policies influence health outcomes for low- and middle-income families. Using mixed methods, she rigorously evaluates how policy changes affect child health and family well-being. Through interdisciplinary collaborations, she also designs, implements, and evaluates clinic-based strategies that connect families to policies that address social needs. She works closely with nonprofits and think tanks to contribute a child health perspective to evidence-based reports on social policy. Dr. Yama completed the highly prestigious National Clinical Scholars Program at the University of California, Los Angeles, where she also earned a Master's in Health Policy and Management. Her scholarship has been published in leading journals, including JAMA Pediatrics, JAMA Network Open, JAMA Health Forum, and Perinatology. Her research has advanced understanding of the role of anti-poverty policy interventions in improving outcomes for U.S. children and families. She was selected for the Academic Pediatric Association's Young Investigator Award—an honor given annually to four promising early-career pediatric investigators—and has secured funding from the Robert Wood Johnson Foundation and other organizations to support her work.

Key Publications:

1. **Yama C**, Rook JM, Wisk LE, Dudovitz R, Hernández D, Eisenman DP, Leifheit KM. Expiration of the Expanded Child Tax Credit and Energy Insecurity in US Households With Children, 2021-2022. *Am J Public Health*. 2025 Aug;115(8):1312-1321.
2. Rook JM, **Yama CL**, Schickedanz AB, Feuerbach AM, Lee SL, Wisk LE. Changes in Self-Reported Adult Health and Household Food Security With the 2021 Expanded Child Tax Credit Monthly Payments. *JAMA Health Forum*. 2023 Jun 2;4(6):e231672.
3. **Yama CL**, Rook JM. The Child Tax Credit-Tax Policy as Health Policy. *JAMA Pediatr*. 2024 Nov 1;178(11):1097-1098.
4. Choi K, **Yama C**, Leon Y, Kersey L. Pilot of health care workers promoting tax filing and receipt of tax credits for low income families. *The Journal for Nurse Practitioners*. 2026;22(1).
5. **Yama CL**, Greenberg RG, Johnson E, Mago-Shah DD. Social needs and healthcare utilization in NICU graduates. *J Perinatol*. 2024 Dec;44(12):1732-1737.

Cecile Yama, MD
Assistant Professor, Pediatrics
and Population Health Science and Policy

Eva Morava, MD, PhD

Eva Morava, MD, PhD is a board-certified clinical geneticist, pediatrician and metabolic specialist. She is a full professor and director for the Program for Inherited Metabolic Diseases at the Department of Genetic and Genomic Sciences. She did her specialty trainings in Europe, Hungary and in the Netherlands and in the US at Tulane Medical Center in LA. Her major clinical expertise is inborn errors of metabolism (IEM). She has decades of experience in diagnostics, care and treatment in IEM, especially in congenital disorders of glycosylation (CDG) and mitochondrial disorders.

Dr Morava has a research laboratory, focusing on translational research in mitochondrial disorders and congenital disorders of glycosylation. She is actively involved in translational research, developing novel therapeutic approaches in genetic disorders. She is also the PD/PI of the U54 FCDGC consortium https://www.rarediseasesnetwork.org/fcdgc studying congenital disorders of glycosylation, and the P01 consortium focusing on establishing and characterizing model systems in CDG. She has more than 450 publications.

Dr Morava is the editor in chief of Molecular genetics and Metabolism. She was editor in Chief of JIMD and council member of the Society for the Study on Inborn Errors of Metabolism (SSIEM). She has been on the newborn screening committee in different states. She is working in close collaboration with the UMDF and CDG-CARE, where she is an advisory board member. She is a co-chair for the MSSM site for NORD.

Dr Morava is passionate about education, especially patient education. She is the director for the categorical and combined genetics residency and fellowship programs and the Genetics T32 program at MSSM. She is active in course development and education at the North American Metabolic Academy.

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

Key Publications:

1. Preston G, Shammas I, Pinto E Vairo F, Ligezka A, Aschoff CAM, Poswar F, Schwartz IVD, Kozicz T, **Morava E**. Reversible Metabolic and Liver Disease in Complex III Deficiency: Novel Variants Expand the Reported UQCRC2-Associated Phenotype. *Cells*. 2026 Mar 27;15(7):596.
2. Edmondson AC, Honzík T, Lam C, Õunap K, McWilliams P, **Morava E**. Incidence and prevalence of phosphomannomutase 2-congenital disorder of glycosylation: Past, present, and future. *Mol Genet Metab*. 2025 Sep-Oct;146(1-2):109188.
3. Muffels IJJ, Kozicz T, Perlstein EO, **Morava E**. The Therapeutic Future for Congenital Disorders of Glycosylation. *J Inherit Metab Dis*. 2025 Mar;48(2):e70011.
4. Lam C, Scaglia F, Berry GT, Larson A, Sarafoglou K, Andersson HC, Sklirou E, Tan QKG, Starosta RT, Sadek M, Wolfe L, Horikoshi S, Ali M, Barone R, Campbell T, Chang IJ, Coles K, Cook E, Eklund EA, Engelhardt NM, Freeman M, Friedman J, Fu DYT, Botzo G, Rawls B, Hernandez C, Johnsen C, Keller K, Kramer S, Kuschel B, Leshinski A, Martinez-Duncker I, Mazza GL, Mercimek-Andrews S, Miller BS, Muthusamy K, Neira J, Patterson MC, Pogorelc N, Powers LN, Ramey E, Reinhart M, Squire A, Thies J, Vockley J, Vreugdenhil H, Witters P, Youbi M, Zeighami A, Zemet R, Edmondson AC, **Morava E**. Frontiers in congenital disorders of glycosylation consortium, a cross-sectional study report at year 5 of 280 individuals in the natural history cohort. *Mol Genet Metab*. 2024 Aug;142(4):108509.
5. Muthusamy K, Perez-Ortiz JM, Ligezka AN, Altassan R, Johnsen C, Schultz MJ, Patterson MC, **Morava E**. Neurological manifestations in PMM2-congenital disorders of glycosylation (PMM2-CDG): Insights into clinico-radiological characteristics, recommendations for follow-up, and future directions. *Genet Med*. 2024 Feb;26(2):101027.

Tamas Kozicz, MD, PhD

Tamas Kozicz, MD, PhD, is a physician–scientist and Professor of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai. He leads a translational research program focused on understanding the molecular and cellular mechanisms underlying rare neurodevelopmental and metabolic disorders, with particular emphasis on congenital disorders of glycosylation (CDG).

Dr. Kozicz's research integrates patient-derived induced pluripotent stem cells, human cortical organoids, multi-omics profiling, and functional neuronal network analyses to define how defects in protein glycosylation disrupt brain development and function. His laboratory is especially interested in neuron–glia interactions, metabolic reprogramming, lipid homeostasis, and neuroinflammatory signaling as drivers of disease progression in pediatric-onset neurological disorders.

He currently serves as a Project Principal Investigator on an NIH-funded Program Project Grant investigating neural pathogenic mechanisms in CDG and is an active member of the NIH-supported

Key Publications:

1. Radenkovic S, Preston G, Budhraja R, Muffels I, Ligezka A, Staff NP, Hrstka R, Balakrishnan B, Shah R, Verberkmoes S, Shammas I, Bosnyak I, Stiers KM, Lai K, Beamer LJ, Pandey A, Morava E, **Kozicz T.** PGM1 deficiency is linked to sarcomeric and mitochondrial dysfunction in patient-derived iPSC-cardiomyocytes. *J Transl Med*. 2026 Feb 21;24(1):430. doi: 10.1186/s12967-026-07808-9. PMID: 41723528; PMCID: PMC13032676.
2. Shah R, Budhraja R, Radenkovic S, Preston G, King AT, Sabry S, Bleukx C, Shammas I, Young L, Chandran J, Byeon SK, Hrstka R, Smith DY 4th, Staff NP, Drake R, Sloan SA, Pandey A, Morava E, **Kozicz T.** Network Hypoactivity in ALG13-CDG: Disrupted Developmental Pathways and E/I Imbalance as Early Drivers of Neurological Features in CDG. *Cells*. 2026 Jan 14;15(2):147.
3. Muffels IJJ, Budhraja R, Radenkovic S, Shah R, Pandey A, Morava E, **Kozicz T.** Are viral vector-mediated therapies compatible with aberrant glycosylation? *Mol Ther Methods Clin Dev*. 2025 Jul 22;33(3):101540.

Frontiers of Congenital Disorders of Glycosylation Consortium. Through close collaboration with clinicians, patient advocacy organizations, and basic scientists, his work aims to translate mechanistic insight into biomarkers and therapeutic strategies that can improve outcomes for affected children and families.

Dr. Kozicz has co-led multiple clinical–translational studies defining disease progression and variability in rare genetic disorders and has contributed to the development of early-stage therapeutic approaches, including small-molecule interventions and gene-targeted strategies. His research has supported the initiation of clinical trials in several CDG subtypes.

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

Key Publications:

4. Muffels IJJ, Budhraja R, Shah R, Radenkovic S, Morava E, **Kozicz T.** Predicting disease-overarching therapeutic approaches for congenital disorders of glycosylation using multi-OMICS. *Mol Genet Metab*. 2025 Sep-Oct;146(1-2):109195.
5. Morava E, Elsharkawi I, **Kozicz T.** Reframing primary mitochondrial disease as a sterile interferonopathy. *Mol Genet Metab*. 2025 Sep-Oct;146(1-2):109217.

At Mount Sinai, Dr. Kozicz is committed to interdisciplinary collaboration, trainee mentorship, and advancing child health research through integration of genetics, neuroscience, and precision medicine.

Chelsea Lowther, PhD

Chelsea Lowther, PhD, is an Assistant Professor in the Department of Genetics and Genomic Sciences and a member of the Institute for Genomic Health at the Icahn School of Medicine at Mount Sinai (ISMMS). She joined ISMMS in 2024 after completing her postdoctoral research fellowship at Massachusetts General Hospital, Harvard Medical School, and the Broad Institute, and her PhD at the University of Toronto.

Her research applies state-of-the-art genomics technologies and computational approaches to understand the genetic architecture of human disease, with a particular emphasis on the contribution of structural variants (SVs; genomic rearrangements greater than 50 base pairs in size). SVs contribute to a broad spectrum of conditions across the developmental continuum, ranging from fetal structural anomalies to pediatric neurodevelopmental disorders and later-onset neuropsychiatric illnesses. As the Principal Investigator on a NICHD K99/R00 award, she is currently developing new statistical methods to improve the functional annotation of SVs by investigating how disruptions to 3D genome organization influence gene regulation. She also serves on the American College of Medical Genetics and Genomics (ACMG) CNV Technical Standards Working Group, where she contributes to the development of evidence-based guidelines that support consistent and accurate clinical interpretation of SVs across diagnostic laboratories.

Chelsea Lowther, PhD
Assistant Professor, Genetics and Genomic Sciences

Key Publications:

1. **Lowther C**, Valkanas E, Giordano JL, Wang HZ, Currall BB, et al. Systematic evaluation of genome sequencing for the diagnostic assessment of autism spectrum disorder and fetal structural anomalies. *American Journal of Human Genetics*. 2023 Sep 7;110(9):1454-1469.
2. **Lowther C,** Mehrjouy MM, Collins RL, Bak M, Dudchenko O, et al. Balanced chromosomal rearrangements offer insights into coding and noncoding genomic features associated with developmental disorders. *medRxiv* (2022).
3. Raca G, Astbury C, Behlmann A, De Castro MJ, Hickey SE, MD, Karaca E, **Lowther C**, et al. Points to consider in the detection of germline structural variants using next-generation sequencing: a statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Medicine*. 2022 Dec 9:S1098-3600(22)00955-8.
4. Collins RL, Brand H, Karczewski KJ, Zhao X, Alföldi J, Khera AV, **Lowther C**, et al. A structural variation reference for medical and population genetics. *Nature*. 2020 May;581(7809):444-451.
5. **Lowther C**, Merico D, Costain G, Waserman J, Boyd K, Noor A, Speevak M, Stavropoulos DJ, Wei J, Lionel AC, Marshall CR, Scherer SW, Bassett AS. Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. *Genome Medicine*. 2017 Nov 30;9(1):105.

Faculty Grants/Honors/Awards

Romina Bevacqua, PhD, Career Development Award, BreakthroughT1D, "Protective Interventions Against Loss of Function and Destruction of β cells in Type 1 Diabetes"

Son Q. Duong, MD, NY Community Trust Foundation Grant, "Artificial Intelligence Screening for Congenital Heart Disease in Fetuses and Neonates"

Marek Mlodzik, PhD, Icahn School of Medicine at Mount Sinai, 2026 Jacobi Medallion recipient.

Lisa Satlin, MD, Election to the American Society of Clinical Investigation as an Honorary Member, April 2026.

Lisa Satlin, MD, Founder's award, American Society of Pediatric Nephrology, April 2026.

Ernest Turro, PhD, Discovery and characterization of the dominant and recessive forms of ReNU2 syndrome, Closing Keynote, Fifth Interdisciplinary Congress on Human Genetics, Granada, Spain, April 17 2026.

Julie Wang, MD, American Academy of Allergy, Asthma and Immunology, 2026 Distinguished Clinician Award.

Anna-Sophie Rommel, PhD, Wellcome Trust, "Mental illness in perimenopause: Longitudinal predictors and preventive treatment opportunities"

Trainee Grants/Honors/Awards

Yuna Lee, PI: Tirtha K. Das, PhD, TERRA-NYC-Stem Fair - **2nd Place** (USA Metric Association Award), "Novel Drug Cocktails for Lung Cancer: Enhancing Standard of Care Treatments using the Drosophila Melanogaster Model System"

Patrick O'Connell, DO, PhD, Immune Deficiency Foundation Research Grant,"Immunometabolism in Lysinuric Protein Intolerence"

Kaya Parikh, PI: Tirtha K. Das, PhD, Regeneron ISEF Fair 2026- **1st Place** Biochemistry ($6000), "Semaglutide and Counterfeit Semaglutide in Drosophila Model of Type 2 Diabetes and Obesity"

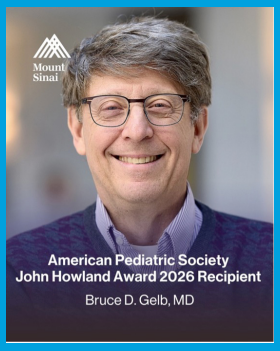
Kaya Parikh, PI: Tirtha K. Das, PhD, Regeneron ISEF Fair 2026 - **Special Award** (The Scientific and Technological Council of Turkiye; $1000), "Semaglutide and Counterfeit Semaglutide in Drosophila Model of Type 2 Diabetes and Obesity"

Kaya Parikh, PI: Tirtha K. Das, PhD, TERRA-NYC-Stem Fair - **1st Place** (Qualified for ISEF, Arizona, May 2026), "Semaglutide and Counterfeit Semaglutide in Drosophila Model of Type 2 Diabetes and Obesity"

Danny Shing, PI: Tirtha K. Das, PhD, TERRA-NYC-Stem Fair - **1st Place** (Qualified for ISEF, Arizona, May 2026), "Identifying Novel Cell Signaling Components Required for KIF5B-RET-Driven Lung Cancer: A Genetic Approach"

Award Highlight

American Pediatric Society Announces Bruce D. Gelb, MD, as Recipient of Its Prestigious 2026 APS John Howland Award



Congratulations to Bruce D. Gelb, MD, Gogel Family Chair and Director of the Mindich Child Health and Development Institute and Dean for Child Health Research on being selected by the American Pediatric Society as the recipient of the 2026 John Howland Award—widely regarded as the highest honor in academic pediatrics. The award was presented at the APS Presidential Plenary during the 2026 Pediatric Academic Societies Meeting in Boston in April.

> Read the full press release here: https://www.mountsinai.org/about/newsroom/2025/american-pediatric-society-announces-bruce-d-gelb-md-as-recipient-of-its-prestigious-2026-aps-john-howland-award

Publications

Jayaram N, Hall M, Spertus JA, Chauhan D, Karamlou T, St Louis JD, ... **Anderson BR**. Public reporting and case selection in congenital heart surgery: Signals from a multicenter observational study. *J Am Coll Cardiol.* 2026 Jan 28.

Breen MS, Yang A, Wang X, Rodriguez de Los Santos M, Tao R, Weinberger DR, ... **Buxbaum JD**. Trisomy 21 drives adarb1 overexpression and premature rna recoding in the developing fetal brain. *Nat Commun.* 2026 Mar 31;17(1).

Mateus-Tique J, Lakshmi A, Singh B, Iyer R, Sánchez-Paulete AR, Falcomatà C, ... **Brown BD**. Armored macrophage-targeted car-t cells reset and reprogram the tumor microenvironment and control metastatic cancer growth. *Cancer Cell.* 2026 Mar 9;44(3):534-50.e11.

Marks A, Siu S, Bianchini F, Wang C, Lakshmi A, Phelan M, ... **Brown BD**. Mrna vaccine immunity is enhanced by hepatocyte detargeting and not dependent on dendritic cell expression. *Nat Biotechnol.* 2026 Apr 29.

Kelly M, Ali S, Huerta A, Hsu EK, Squires JE, Campbell KM, ... **Bucuvalas JC**, ... Wadhwani SI. Characterizing barriers to living donor liver transplantation for pediatric recipients: Qualitative results from the social-tx study. *J Pediatr.* 2026 Apr 21:115112.

Zhang L, Chun Y, Reed K, Grishina G, Lo T, **Wang J**, **Sicherer S**, **Bunyavanich S**. Oral immunotherapy suppresses peripheral blood transcriptomic response to peanut in peanut allergy. *J Allergy Clin Immunol.* 2026 Feb;157(2):398-408.

Yoon Y, Chun Y, Zhang L, Grishina G, Grishin A, **Bunyavanich S**. Unraveling mechanisms of allergen sensitization and allergic rhinitis via the nasal transcriptome. *J Allergy Clin Immunol.* 2026 Jan;157(1):110-7.

Natividad Avila M, Jung S, Satterstrom FK, Fu JM, Levy T, Sloofman LG, Klei L, ... **Kolevzon A**, ... **Siper PM**, ... **Mahjani B**, **De Rubeis S**, ... **Buxbaum JD**. Deleterious coding variation associated with autism is shared across ancestries. *Nat Med.* 2026 Apr;32(4):1519-29.

Spetko N, Song Y, Orui H, Angell-James C, **Cassidy M**, Liu K, ... Strom JB. Distance and likelihood of cardiovascular imaging receipt among medicare beneficiaries: Cardiovascular imaging deserts among medicare beneficiaries. *JACC Cardiovasc Imaging.* 2026 Apr;19(4):447-59.

Amreen B, **Lesseur C**, Sadikki H, Saul SR, Kazi S, Leung AM, ... **Chen J**, van Gerwen M. Preliminary insights into methylation patterns in agent orange exposed thyroid cancers: A pilot study. *Environ Pollut.* 2026 Feb 1;390:127475.

Chorniy A. Lasting consequences of early environments on health. *Bmj.* 2026 Apr 22;393:s702.

Zhang X, Sivakumar V, Paredes-Marin A, Leviton A, Villanueva A, Bansal MB, ... **Bucuvalas J**, **Chu J**. Real-world comparison of steatosis-associated fibrosis estimator and fibrosis-4 scores for assessing metabolic dysfunction-associated steatotic liver disease-related fibrosis in a highly diverse urban metabolic dysfunction-associated steatotic liver disease population. *Clin Gastroenterol Hepatol.* 2026 Jan;24(1):261-3.e3.

Meares R, Hamiduzzaman M, McLennan V, Miles S, **Crook S**, Grove L, ... Flood V. Resident and family carer perspectives on the impact of allied health student placements on service delivery to residents in northern nsw aged care homes: A qualitative study. *Aust J Rural Health.* 2026 Apr;34(2):e70160.

Ho HE, Radigan L, Meffre E, **Cunningham-Rundles C**. Iga defects in cvid lead to bacterial translocation, increased serum γ-interferon, and baff. *J Hum Immun.* 2026 Mar 2;2(2):e20250080.

Thorpe HHA, Cupertino RB, Pakala SR, Fontanillas P, Jennings MV, Yang J, ... **Davis LK**, ... Sanchez-Roige S. Genome-wide association study of delay discounting identifies 11 loci and reveals transdiagnostic associations across mental and physical health. *Mol Psychiatry.* 2026 Apr;31(4):2081-93.

Strom NI, Verhulst B, Bacanu SA, Cheesman R, Purves KL, Gedik H, ... **Davis LK**, ... Hettema JM. Genome-wide association study of major anxiety disorders in 122,341 european-ancestry cases identifies 58 loci and highlights gabaergic signaling. *Nat Genet.* 2026 Feb;58(2):275-88.

Moolhuijsen LME, Zhu J, Mullin BH, Pujol-Gualdo N, Actkins KV, Mack JA, ... **Davis LK**, ... Day FR. Genomic analyses implicate hormonal and metabolic dysregulation in polycystic ovary syndrome. *Nat Genet.* 2026 Apr 23.

Li X, Raisinghani N, Gallinat A, Santos-Gallego CG, Zhang S, La Salvia S, ... **Dubois NC**, ... Sahoo S. Circulating extracellular vesicles in the pathogenesis of heart failure in patients with chronic kidney disease. *Circulation.* 2026 Jan 13;153(2):94-114.

Duong SQ, Vaid A, Jiang P, Bitterman Y, Krishnamurthy Y, Chiu IM, ... **Glicksberg BS,** ... **Anderson BR**, ... Mayourian J. Development and multicentre validation of an artificial intelligence electrocardiogram model for ventricular remodeling in repaired tetralogy of fallot. *Eur Heart J Digit Health.* 2026 Mar;7(2):ztag015.

Lang SH, Cottingham N, Donnelly C, Risen S, Muncher RM, Brewer ED, **Saland JM**, ... **Ganesh J**. Outcomes of kidney transplantation in three patients with single large-scale mitochondrial DNA deletion syndromes. *Mol Genet Metab.* 2026 Mar;147(3):109731.

Lavelle TA, Maron JL, Kingsmore SF, Lin CH, Zhu Y, Sweigart B, Reed D, **Gelb BD**, Vockley J, Davis JM. Rapid genome sequencing compared with a gene panel in critically ill infants with a suspected genetic disorder: An economic evaluation. *J Pediatr.* 2026 Feb;289:114889.

Kim SW, Parfenov M, Rodriguez-Murillo L, Conner DA, Sharma A, Peter I, Xiao F, Layton O, Tai A, Ward T, Wasson LK, Gorham JM, Mazaika E, Lagomarsino VN, Young-Pearse TL, Goldmuntz E, Wakimoto H, Agopian AJ, McKean DM, DePalma SR, Pu WT, Seidman CE, **Gelb BD**, Seidman JG. Robo2 variants associated with atrial septal defect define a novel regulatory element. *Circ Genom Precis Med.* 2026 Apr;19(2):e004918.

Edlund S, Haglund N, Bornehag CG, **Gennings C**, Kiviranta H, **Kolevzon A**, ... **Reichenberg A**, **Swan S**, Källén K. Perinatal and maternal factors associated with autism spectrum disorder. *PLoS One.* 2026;21(3):e0316968.

Omar M, Sorin V, Wieler LH, Charney AW, Kovatch P, Horowitz CR, ... **Glicksberg BS,** ... Klang E. Mapping the susceptibility of large language models to medical misinformation across clinical notes and social media: A cross-sectional benchmarking analysis. *Lancet Digit Health.* 2026 Jan;8(1):100949.

Veenstra-VanderWeele J, Brown LK, Martin A, Muhle RA, Jacob S, Drury SS, ... **Grice DE,** ... Szatmari P. Developing physician-scientists through integrated child and adolescent psychiatry clinical and research training programs. *J Am Acad Child Adolesc Psychiatry.* 2026 Mar 23.

Dalldorf K, Milburn S, Francisco B, Ahle G, Arguello-Angarita S, Aris K, ... **Guttmann KF**, Weintraub AS. Outcomes of universal newborn g6pd deficiency screening in a large urban cohort. *Pediatrics.* 2026 Jan 1;157(1).

von Scheidt M, Adkar SS, Krefting J, Hoermann G, Meggendorfer M, Bauer S, ... **Hao K**, ... Schunkert H. Clonal haematopoiesis of indeterminate potential and mortality in coronary artery disease. *Eur Heart J.* 2026 Jan 24;47(4):453-69.

Rechtman E, **Reichenberg A**, Invernizzi A, Fleysher L, Rebello V, ... **Gennings C**, ... **Horton MK**. Fetal and postnatal metal metabolism-related changes in brain function are associated with childhood behavioral deficits. *Sci Adv.* 2026 Apr 24;12(17):eadz1340.

Publications, continued

Schmitz EG, Paul AJ, Ghosh R, Saucier N, Kolicheski A, Risma SI, ... **Itan Y**, ... **Bogunovic D**, ... Cooper MA. Targeted deep sequencing identifies mosaicism in patients with immune dysregulation. *J Allergy Clin Immunol.* 2026 Feb 5.

Koenen M, Becher T, Pagano G, Del Gaudio I, Barrero JA, Montezano AC, ... **Itan Y**, ... Cohen P. Ablation of prdm16 and beige fat identity causes vascular remodeling and elevated blood pressure. *Science.* 2026 Jan 15;391(6782):306-13.

Fyfe C, Winell H, Dougherty J, Gutmann DH, **Kolevzon A**, Marrus N, ...Sandin S. Time trends in the male to female ratio for autism incidence: Population based, prospectively collected, birth cohort study. *Bmj.* 2026 Feb 4;392:e084164.

Jabagi H, Shaw RE, **Kontorovich AR**, Alemany VS, Ciallella C, Burns P, Grau JB. Utilization of current acc/aha genetic testing recommendations for thoracic aortic disease at a large adult aortic center. *Genet Med.* 2026 Apr;28(4):102069.

Pecorelli S, Lucchini R, **Lambertini L**, Novelli G. Fathers matter too: Sperm-borne epigenetic messages link paternal exposures to next-generation health. *Nat Rev Urol.* 2026 Feb 4.

Prada D, Morava-Kozicz E, Rajendrakumar AL, Kupsco A, **Lesseur C**, Irizar H, ... Parks R. Blood mitochondrial heteroplasmic variants and cognitive performance in late midlife: Regards study. *BMC Neurol.* 2026 Apr 21;26(1).

Liu SH, Chen Y, Feuerstahler L, Koelmel JP, Godri Pollitt KJ, Xu Y, ... Manz KE. Quantifying pfas-omics burden scores for nontargeted analysis using multidimensional item response theory: An exploratory analysis of novel and legacy pfas in cord blood. *Environ Sci Technol.* 2026 Mar 3;60(8):6322-40.

Liu X, Smout S, **Mahjani B**, Munk-Olsen T, Blæhr EE, Robakis TK, Bergink V. Risk of relapse in psychotic and bipolar disorders after prenatal antipsychotic discontinuation. *JAMA Netw Open.* 2026 Mar 2;9(3):e260682.

Vuong LT, **Mlodzik M**. Nuclear translocation of β-catenin in wg/wnt signaling via the ift-a microtubule-associated complex requires pasovec/gid8 proteins. *Sci Adv.* 2026 Apr 3;12(14):eaea3382.

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):1648-71.

Rapp AM, Ponting C, Ramos G, Escovar E, Hazlett C, Tan PZ, ... Chavira DA. A data-driven approach to identifying determinants of depression in rural latine adolescents. *Cultur Divers Ethnic Minor Psychol.* 2026 Jan 8.

Merritt K, Palmer ER, Laguna PL, Sethi A, Rogers JC, Evans CJ, **Reichenberg A**, ... David AS. Inflammatory markers (il-6 and crp) in childhood and their association with brain structure and psychotic experiences in adulthood. *Brain Behav Immun.* 2026 Mar;133:106247.

Kwiatkowski A, **Rendl M**. Two's company, three's a hair cycle? Hfdscs join the hair germdermal papilla regulatory interface. *J Invest Dermatol.* 2026 Mar;146(3):577-80.

Grubner A, Wang A, Fisher P, **Renny MH**. Lower back pain in a healthy 17-year-old adolescent. *Pediatr Rev.* 2026 Mar 1;47(3):163-6.

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 Apr 7:106593.

Saland JM, Lieske JC, Groothoff JW, Frishberg Y, Shasha-Lavsky H, Magen D, ... Hulton SA. Efficacy and safety of lumasiran in patients with primary hyperoxaluria type 1: Results from a phase iii clinical trial. *Kidney Int Rep.* 2024 Jul;9(7):2037-46.

Dribin TE, **Sampson HA**, Schnadower D, Anagnostou A, Fox AT, Golden DBK, ... **Wang J**, ... **Sicherer SH**, ... Camargo CA, Jr. Non-injectable versus injectable epinephrine treatment thresholds for acute allergic reactions in the community. *J Allergy Clin Immunol Pract.* 2026 Apr;14(4):976-80.

Dribin TE, **Sampson HA**, Anagnostou A, Camargo CA, Jr., Fox AT, Golden DBK, ... **Wang J**, ... **Sicherer SH**, ... Schnadower D. Epinephrine and emergency medical services activation recommendations during acute allergic reactions in community settings: International consensus report. *J Allergy Clin Immunol.* 2026 Feb;157(2):429-41.

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 Feb 26:e24001.

Shemesh E, Ng VL, Dunphy C, Logan S, Chessell J, Swami N, ... Mazariegos GV. Patient-caregiver discrepancy score: Multisite evaluation of a novel pediatric outcome measure. *J Pediatr.* 2026 Mar;290:114900.

Jeong S, **Sicherer SH**. Improved food allergy and anaphylaxis knowledge and comfort among internal medicine residents following a brief educational intervention. *J Allergy Clin Immunol Glob.* 2026 Mar;5(2):100643.

Levy T, Silver H, Benrey N, Siegel A, Layton C, Zemon V, ... **Buxbaum JD**, **Kolevzon A**, **Siper PM**. Transient visual evoked potential abnormalities in adnp syndrome. *J Neurodev Disord.* 2026 Apr 1.

Fernandez NA, Durland LJ, Garcia A, Atkins MH, Kennedy M, **Sturgeon CM**, Keller G. Retinoic acid regulates the development of human definitive hematopoiesis in a non-cell-autonomous manner. *Development.* 2026 Apr 15;153(8).

Hua J, Rochester JR, Foley JM, Hahn LB, Min MY, Kenfield SA, ... **Swan SH**. Targeting plastic exposure in infertile couples: A pilot intervention study. *Toxics.* 2026 Mar 16;14(3).

Stingone JA, Bengoa S, Valle C, Masters J, Cadenhead T, Rabin A, ... **Gennings C**, ... **Teitelbaum SL**. The development of the human health exposure analysis resource (hhear) data repository for environmental epidemiology research. *Environ Int.* 2026 Jan;207:109967.

Rea-Moreno M, Tian L, Tavakol TN, Yang MC, Pek NM, Gulati S, ... **Januska MN**, ... **Tiozzo C**, Chen YW. Unveiling alternate pathways for sars-cov-2 infection via extracellular vesicle-mediated transfer of ace2 and tmprss2. *Nat Commun.* 2026 Apr 10.

Yarman Y, Zhao X, Ahn H, Thomson H, Sarkar A, Yuan T, ... **Turro E**, ... Ma P. Understanding how a highly prevalent grk5 polymorphism affects platelets and enhances thrombotic risk. *Blood.* 2026 Apr 16;147(16):1873-84.

Greene D, Mendez R, Lees J, **Barbosa M**, Bruselles A, Chiriatti L, ... **Turro E**. Biallelic variants in rnu2-2 cause the most prevalent known recessive neurodevelopmental disorder. *Nat Genet.* 2026 Apr;58(4):774-81.

Ansari SA, Brake MA, Pathak N, Flaumenhaft JT, Ludington JG, Panwar N, **Turro E**, Schulman S. Human missense variants in f3 impair the initiation of blood coagulation. *Blood.* 2026 Feb 5;147(6):689-701.

Wang K, Saniei S, Poddar N, Martinez IG, Chao C, Autar S, ... **Wagenblast E**. Ontogeny dictates oncogenic potential, lineage hierarchy, and therapy response in pediatric leukemia. *Cancer Discov.* 2026 Mar 2;16(3):541-70.

Smith GR, Zhao B, Lindholm ME, Raja A, Viggars M, Pincas H, ... **Walsh MJ**, ... Sealfon SC. Multi-omic identification of key transcriptional regulatory programs during endurance exercise training in rats. *Nat Commun.* 2026 Mar 21.

Publications, continued

Sindher SB, Chehade M, Dellon ES, Jones SM, Spergel JM, Lin A, ... **Wang J**, **Sicherer SH**, ... Chinthrajah RS. Management of gastrointestinal symptoms during oral immunotherapy: Guidance from outmatch investigators. *J Allergy Clin Immunol Pract.* 2026 Apr;14(4):772-9.

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 aca expansion vs non-expansion states. *Am J Prev Med.* 2026 Feb 14:108320.

Zhou EG, Brownstein JS & Rader B. Assessing MMR vaccination coverage gaps in US children with digital participatory surveillance. *Nat. Health* 1, 138–144 (2026).

SAVE THE DATE

14th Annual MCHDI Retreat

Date: November 10, 2026

Time: TBA

Location: Harmonie Club

Ballroom, 1st Floor

4 E 60th St, New York, NY 10022

Icahn School of Medicine at Mount Sinai

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