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

MCHDI Developmental Outcomes

FALL 2024

Research Advancements: Neurodevelopmental Disorders

Mutations in *RNU4-2* cause one of the most prevalent monogenic neurodevelopmental disorders

Neurodevelopmental disorders (NDDs) are a group of conditions that result from an abnormal development of the brain. Intellectual disability is a common feature of these disorders and refers to limitations in intellectual functioning (e.g., learning, reasoning, problem-solving) and adaptive behavior (e.g., social and practical skills). Intellectual disability often has a genetic basis: approximately 1,500 genes have been implicated in intellectual disability. Despite this knowledge, most patients today do not receive a genetic diagnosis following genetic testing.

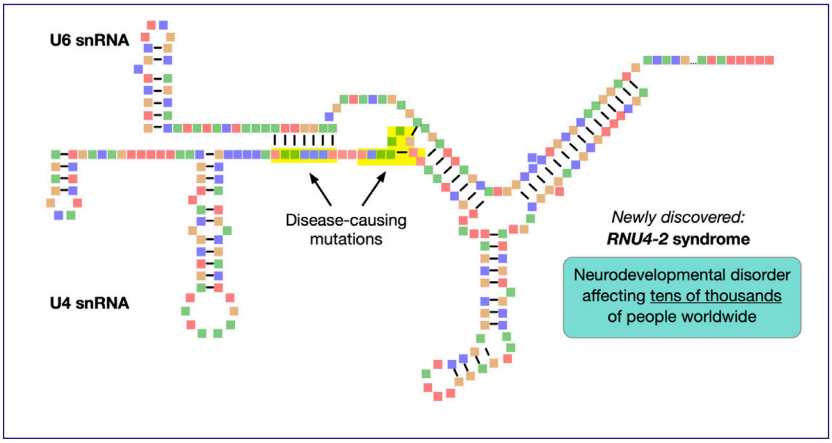

Figure: Representation of the U4/U6 snRNA duplex. The critical regions harboring the pathogenic mutations responsible for *RNU4-2* syndrome are highlighted.

There are two types of gene: protein coding and non-coding genes. Non-coding genes are transcribed into RNA, but the RNA is not subsequently translated into a protein. Previous large genetic studies of NDDs used a technology called whole-exome sequencing, which by and large only analyzes coding genes. This is one of the reasons that only about 10 non-coding genes have been implicated in NDDs. An approach called whole-genome sequencing, on the other hand, covers the entire genome, including non-coding genes.

To discover whether genetic variants in non-coding genes might cause intellectual disability, we analyzed whole-genome sequencing data in the UK's National Genomic Research Library. This library contains data on patients consented to participate in research through the UK's National Health Service. We compared the genetic data for all 41,132 known non-coding genes in about 5,529 study participants with intellectual disability and 46,401 controls.

Our statistical analysis found that genetic variants in a very small non-coding gene, only 141 bases long, are responsible for an NDD that features intellectual disability and other traits, such as short stature, small heads, seizures and low muscle mass. Despite these features, the patients were not previously recognized as clinically distinct from other patients with NDDs. This contrasts with well-known syndromes such as Down's syndrome, which was described as a distinct disorder in 1862 and explained genetically a century later.

The gene we implicated is called RNU4-2. It encodes a small RNA component of a large molecular complex called a spliceosome. Spliceosomes play a crucial role in a basic biological function of cells, called gene splicing, which is present in all animals, plants and fungi.

We were struck by how common this disorder is. We estimate it is one of the most common NDDs caused by genetic variants in a single gene. It is likely to affect between 1 in 20,000 and 1 in 25,000 young people. Furthermore, remarkably, a single genetic variant accounts for about two thirds of patients with *RNU4-2* syndrome.

This discovery is feeding into updated clinical genetic tests for NDDs. This should allow tens of thousands of parents to receive a genetic diagnosis for their affected children's condition, bringing closure to what in many cases will have been a diagnostic odyssey lasting many years.

Almost all affected individuals have what is called a spontaneous or de novo mutation. This means that the parents did not pass on the genetic variants, but rather the variants appeared spontaneously due to an error during DNA replication. This is important for parents because it means that, with few exceptions, the same disorder is unlikely to reoccur in future pregnancies.

Our findings are reported in: Mutations in the U4 snRNA gene *RNU4-2* cause one of the most prevalent monogenic neurodevelopmental disorders. Greene D, Thys C, Berry IR, Jarvis J, Ortibus E, Mumford AD, Freson K, Turro E. *Nat Med.* 2024 May; 8(30):2165–2169.

Ernest Turro, PhD
Associate Professor, Genetics and Genomic Sciences

The neurodevelopmental origins of adult psychiatric disorders

Schizophrenia effects 1% of the population and is defined as a disorder of aberrant thought and perception and persistent social withdrawal and flattened affect. The first diagnosis of schizophrenia typically occurs in early adulthood and the disorder is persistent and severely debilitating. Despite the focus on the abnormalities in thought and perception ("hallucinations" and "delusions") for diagnosis, the early description of schizophrenia by Kraepelin more than 130 years ago highlighted slow cognitive decline and other developmental signs occurring before symptoms manifest. While studies addressing early developmental manifestation of schizophrenia began in the 1960s, it was not until 1987 that an etiological model of schizophrenia was outlined, suggesting schizophrenia as a disorder of neurodevelopmental origins [1]. This so-called neurodevelopmental hypothesis of schizophrenia has become the dominant etiological model of the disorder.

Research in my group is focused on understanding the developmental origins of schizophrenia using large population-based resources combining epidemiological approaches, behavioral and molecular genetic methods. Early in my career my work demonstrated that in apparently healthy adolescents who will go on to develop schizophrenia, there is a cognitive impairment that equals approximately half a standard deviation, or 8 IQ points deficit [2]. In an upcoming critical review of the nascent literature on childhood and adolescence cognitive functioning in schizophrenia [3], we illustrate that (i) the cognitive dysfunction that predates schizophrenia is not static, it worsens from early childhood onwards through to the onset of psychosis, reaching an equivalent of 15 IQ points deficit after schizophrenia is first diagnosed. At the same time, (ii) early cognitive dysfunction is not universal in schizophrenia; and (iii) a substantial role for genes affecting both schizophrenia and early life cognitive functioning, a common hypothesis for the relation between cognitive deficits and schizophrenia, has not been unequivocally proven.

We have been equally interested in the origins of the social deficit in schizophrenia, and showed that, like IQ, early social isolation and social withdrawal are progressively present in future schizophrenia patients. In earlier work, Sven Sandin and colleagues demonstrated that schizophrenia, autism spectrum disorder, intellectual disability and ADHD clusters in families, suggesting shared neurodevelopmental origins for these disorders [4,5]. In latest work Adam Socrates and Eva Velthorst in the group designed a study to characterize the genetics of social isolation [6]. First, a genome-wide associations study (GWAS) was carried in 400,000 individuals from the UK Biobank, to identify genetic contribution to social isolation. A derived polygenic score for social isolation was then tested in the Avon Longitudinal Study Cohort in the UK, an independent, developmental cohort. The polygenic score was predictive of scores measuring friendship quality and quantity in late adolescence. Finaly, the work demonstrated genetic correlations with schizophrenia, as well as autism spectrum disorder, major depressive disorder, educational attainment, extraversion, and loneliness.

Taken together our work supports a rethink of schizophrenia as a neurodevelopmental disorder with psychosis as a potentially preventable outcome. Future research should focus on understanding the (possibly unique) etiology, clinical characteristics and outcome in the group of neurodevelopmental schizophrenia patients, leading to improved diagnostic and treatment.

References

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3. **Reichenberg A,** Kahn RS. Cognitive dysfunction preceding the first psychosis episode in schizophrenia. *Mol Psychiatry.* 2024. [In Press]
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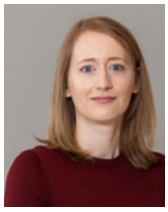
6. Socrates AJ, Mullins N, Gur RC, Gur RE, Stahl E, O'Reilly PF, **Reichenberg A,** Jones H, Zammit S, Velthorst E. Polygenic risk of social isolation behavior and its influence on psychopathology and personality. *Mol Psychiatry.* 2024 May 30.

Avi Reichenberg, PhD
Professor, Psychiatry and
Environmental Medicine & Public Health

Sarah Crook, PhD

Sarah Crook, PhD is an Assistant Professor in the Department of Population Health. She serves as the Director of Analytics the Center for Child Health Services Research in the Mindich Child Health and Development Institute at the Icahn School of Medicine. With expertise in biostatistics, epidemiology, and health services research. Dr. Crook contributes to the center's initiatives through the application of her expertise and specialization in large-scale data analytics. This includes the integration of clinical registry data, administrative data, and social determinants of health.

Dr. Crook earned both her BS in Human Physiology and MS in Epidemiology and Biostatistics from the University of Leeds, UK. She then completed her PhD in Epidemiology and Biostatistics at the University of Zurich, Switzerland, and a training fellowship in Guidelines Methodology with Cochrane and the UK's National Institute of Health and Care Excellence.



Sarah Crook, PhD
Assistant Professor, Population Health

Key Publications

1. Pavlyha M, Li Y, **Crook S**, Anderson BR, Reyes-Soffer G. Race/ethnicity and socioeconomic status affect the assessment of lipoprotein(a) levels in clinical practice. *J Clin Lipidol.* 2024 Jul 22:S1933-2874(24)00211-3.
2. **Crook S**, Dragan K, Woo JL, Neidell M, Nash KA, Jiang P, ... Anderson BR; New York State Congenital Heart Surgery Collaborative for Longitudinal Outcomes & Utilization of Resources (CHS-COLOUR). Impact of Social Determinants of Health on Predictive Models for Outcomes After Congenital Heart Surgery. *J Am Coll Cardiol.* 2024 Jun 18;83(24):2440-2454.
3. Jayaram N, Allen P, Hall M, Karamlou T, Woo J, **Crook S**, Anderson BR. Adjusting for congenital heart surgery risk using administrative data. *J Am Coll Cardiol* 2023; 82(23): 2212-2221. Jayaram N, Allen P, Hall M, Karamlou T, Woo J, **Crook S**, Anderson BR. Adjusting for Congenital Heart Surgery Risk Using Administrative Data. *J Am Coll Cardiol.* 2023 Dec 5;82(23):2212-2221.
4. **Crook S**, Dragan K, Woo JL, Neidell M, Jiang P, Cook S, ... Anderson BR; New York State CHS-COLOUR. Long-Term Health Care Utilization After Cardiac Surgery in Children Covered Under Medicaid. *J Am Coll Cardiol.* 2023 Apr 25;81(16):1605-1617.
5. Woo JL, Nash KA, Dragan K, **Crook S**, Neidell M, Cook S, ... Anderson BR; New York State Congenital Heart Surgery Collaborative for Longitudinal Outcomes and Utilization of Resources (CHS-COLOUR). Chronic Medication Burden After Cardiac Surgery for Pediatric Medicaid Beneficiaries. *J Am Coll Cardiol.* 2023 Sep 26;82(13):1331-1340.

Emma Holmes, MD

Emma Holmes is an Assistant Professor in the Division of Newborn Medicine. Having received her MD from the University of Connecticut and completed residency and fellowship at Mount Sinai in Pediatrics and Neonatal-Perinatal Medicine respectively, Dr. Holmes has developed a robust clinical and research profile. She works with the A.I.M.S lab within the Charles Bronfman Institute of Personalized medicine and specializes in the application of artificial intelligence (AI) to neonatal medicine, employing cutting-edge techniques, including deep learning and graph technology, to enhance the understanding and treatment of critical conditions such as apnea of prematurity and culture-negative sepsis. Her area of interest is in the how physiologic waveforms, such as ECG and pulse oximetry, can be used to provide insight on the pathophysiologic changes occurring within neonates. This research has been presented at national conferences, highlighting the potential of AI to revolutionize neonatal care. In addition to a strong research agenda, Dr. Holmes is committed to fostering a collaborative and inclusive environment. She believes in delivering compassionate, evidence-based care and collaborates with multidisciplinary teams to optimize patient outcomes. Through their clinical and research endeavors, Dr. Holmes aims to drive transformative change in neonatal medicine, ultimately improving health outcomes for vulnerable populations worldwide.

Emma Holmes, MD
Assistant Professor, Pediatrics

Key Publications

Holmes E, Kauffman J, Juliano C, Duchon J, Nadkarni GN. Distinguishing neonatal culture-negative sepsis from rule-out sepsis with artificial intelligence-derived graphs. *Pediatr Res.* 2024 Aug 15.

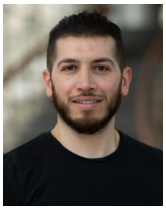
Polidoro E, Weintraub AS, Guttmann KF. Federal regulations and neonatologists' views on care of seriously ill infants: changes over time. *Pediatr Res.* 2022 Oct;92(4):1059-1063.

St John Sutton EM, **Polidoro E,** Ratner V, McKinsey S, Groves A. Rapid simulator-based training for heart rate assessment in the newborn. Resuscitation. 2023 Jan;182:109670.

Emilio Merheb, PhD

Emilio Merheb, PhD, is an instructor at the Diabetes, Obesity, and Metabolism Institute (DOMI) at the Icahn School of Medicine at Mount Sinai. Under the mentorship of Dr. Romina Bevacqua, he is utilizing novel genetic tools to target different genes in human islets to investigate mechanisms contributing to islet maturation and function. These efforts have led to the identification of transcription factors and pathways that drive islet maturation and function, as well as the deciphering of non-coding regulatory mechanisms involved in islet cell maturation and dysregulation in disease.

His research has been presented at both national and international conferences, including the Society for Neuroscience (SfN) and the OddPols 2021 International Conference on Transcriptional Mechanism and Regulation. In addition to oral presentations and posters at various conferences, such as the NYC*T1D symposium, the XV European Meeting on Glial Cells in Health and Disease, and Science Strong, he has authored or co-authored multiple peer-reviewed publications on topics such as biochemistry, neuroscience, molecular biology, genetics, and metabolism.



Emilio Merheb, PhD
Instructor, Medicine

Key Publications

1. Moir RD, **Merheb E,** Chitu V, Stanley ER, Willis IM. Molecular basis of neurodegeneration in a mouse model of Polr3-related disease. *bioRxiv [Preprint].* 2024 Jul 2:2023.12.12.571310.
2. Bevacqua RJ, Zhao W, **Merheb E,** Kim SH, Marson A, Gloyn AL, Kim SK. Multiplexed CRISPR gene editing in primary human islet cells with Cas9 ribonucleoprotein. *iScience.* 2023 Dec 8;27(1):108693.
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4. **Merheb E,** Cui MH, DuBois JC, Branch CA, Gulinello M, Shafit-Zagardo B, Moir RD, Willis IM. Defective oligodendrocyte development and function in an RNA polymerase III mutant leukodystrophic mouse. *Proceeding of the National Academy of Sciences.* 2021.
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He has also mentored and collaborated with research associates, rotation students, postdoctoral fellows, and PhD students on multiple projects, resulting in poster presentations at major conferences and various publications.

Felix Richter, MD, PhD

Felix Richter, MD, PhD is a Neonatal Hospitalist in the Division of Newborn Medicine in the Jack and Lucy Clark Department of Pediatrics at the Icahn School of Medicine at Mount Sinai and Mount Sinai West. He graduated with a BA in Integrated Science, Biological Sciences, and Chemistry from Northwestern University. He completed his MD, PhD, and pediatrics residency training at the Icahn School of Medicine at Mount Sinai. During his graduate training, he developed software and statistical learning approaches to integrate whole-genome sequencing, epigenomic, and RNAseq datasets. He leveraged these tools to discover a role for noncoding de novo variants in congenital heart disease (CHD). He also described the contribution of inherited, mosaic, and large structural variants to CHD. During the height of the pandemic, he developed scalable phenotyping methods to describe COVID-19

Felix Richter, MD, PhD
Instructor, Pediatrics

Key Publications

1. **Richter F,** Morton SU, Kim SW, Kitaygorodsky A, Wasson LK, Chen KM, Zhou J, Qi H, Patel N, DePalma SR, Parfenov M, Homsy J, Gorham JM, Manheimer KB, Velinder M, Farrell A, Marth G, Schadt EE, Kaltman JR, Newburger JW, Giardini A, Goldmuntz E, Brueckner M, Kim R, Porter GA Jr, Bernstein D, Chung WK, Srivastava D, Tristani-Firouzi M, Troyanskaya OG, Dickel DE, Shen Y, Seidman JG, Seidman CE, Gelb BD. Genomic analyses implicate noncoding de novo variants in congenital heart disease. *Nat Genet.* 2020 Aug;52(8):769-777.
2. **Richter F,** Rutherford KD, Cooke AJ, Meshkati M, Eddy-Abrams V, Greene D, Kosowsky J, Park Y, Aggarwal S, Burke RJ, Chang W, Connors J, Giannone PJ, Hays T, Khattar D, Polak M, Senaldi L, Smith-Raska M, Sridhar S, Steiner L, Swanson JR, Tauber KA, Barbosa M, Guttmann KF, Turro E. A Deep Intronic PKHD1 Variant Identified by SpliceAI in a Deceased Neonate With Autosomal Recessive Polycystic Kidney Disease. *Am J Kidney Dis.* 2024 Jun;83(6):829-833.
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clinical trajectories. This was featured in the New York Times. His clinical and research expertise includes pediatrics, genetics, bioinformatics, and statistical learning, and he is a long-standing editor on Wikipedia.

Together with other MCHDI faculty, Felix started and currently leads a multi-site study to perform genomic autopsies across 14 neonatal intensive care units (NICU) for unexplained neonatal deaths (https://www.nicunet.com/). Through this work, his team discovered new causes of cardiac, genitourinary, and cerebrovascular disease. These results were provided back to families and were featured on the TODAY Show. He also initiated and continues to lead research on using computer vision from video-EEG data to predict sedation, cerebral dysfunction, and seizures in neonates from the Mount Sinai NICU.

Key Publications

4. Somani SS, **Richter F,** Fuster V, De Freitas JK, Naik N, Sigel K; Mount Sinai COVID Informatics Center; Bottinger EP, Levin MA, Fayad Z, Just AC, Charney AW, Zhao S, Glicksberg BS, Lala A, Nadkarni GN. Characterization of Patients Who Return to Hospital Following Discharge from Hospitalization for COVID-19. *J Gen Intern Med.* 2020 Oct;35(10):2838-2844.
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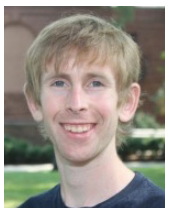
New Extramural Faculty

Michael Cassidy, PhD

Michael Cassidy, PhD is an Assistant Professor of Pediatrics and Population Health Sciences and Policy in the Center for Child Health Services Research in the Mindich Child Health and Development Institute at the Icahn School of Medicine at Mount Sinai. He is an applied microeconomist who uses the science of causal inference to study the social, economic, and environmental determinants of wellbeing across the lifespan. His current work focuses on housing, homelessness, education, and children's health.

Prior to Mount Sinai, Mike was a postdoc and associate research scholar in the Center for Health and Wellbeing at Princeton University's School of International and Public Affairs. He has also held positions at The Century Foundation and the New York City Office of Management and Budget.



Michael Cassidy, PhD
Assistant Professor, Pediatrics and Population Health Sciences and Policy

He holds a Ph.D. in Economics from Rutgers University, a Masters in Public Affairs from Princeton University, and a B.A. in Communication and Political Science from the University of Pennsylvania.

Mike is an invited researcher at the Abdul Latif Jameel Poverty Action Lab (J-PAL), North America, and a faculty affiliate at the Wilson Sheehan Lab for Economic Opportunities (LEO) at the University of Notre Dame.

Key Publications

1. **Cassidy M,** Currie J, Glied S, Howland R. Child Mental Health, Homelessness, and the Shelter System: Evidence from Medicaid in New York City. *Am J Epidemiol.* 2024 Sep 9:kwae345.
2. **Cassidy M,** Currie J. The Effects of Legal Representation on Tenant Outcomes in Housing Court: Evidence from New York City's Universal Access Program. *Journal of Public Economics.* 2023; 222 (104844).
3. Treglia D, **Cassidy M,** Bainbridge J. Improving School Attendance among Homeless Children: Evaluating the Attendance Matters Program. *Children and Youth Services Review.* 2023; 149 (106880).

Anna Chorniy, PhD

Anna Chorniy, PhD is a health economist interested in studying and improving public health insurance programs, including Medicaid, especially for children with common chronic conditions, such as asthma and ADHD. She completed her undergraduate and M.S. degrees at the Lomonosov Moscow State University, where she studied Economics with the focus on Risk Management and Insurance. She earned her PhD in Economics from Clemson University and completed additional postdoctoral training at Princeton University. Prior to joining Icahn School of Medicine, Dr. Chorniy served as an Assistant Professor at Northwestern University Feinberg School of Medicine.

In her current work, she studies the impact of Medicaid on children's health and well-being, physician agency and treatment choices, the relationship between health insurance and labor market outcomes, and industrial organization of health care markets. This research is centered on evaluating health policy impacts and often relies on quasi-experimental econometric methods and large administrative data (e.g. claims). For example, Dr. Chorniy analyzed the transition from the fee-for-service (FFS) reimbursement model to the Medicaid managed care (MMC) payment system — a major shift that affected 80% of states by 2016. This research highlighted that this transition significantly influenced the dramatic rise in ADHD diagnoses and treatment over recent decades. She further showed that these changes are not necessarily beneficial for the diagnosed children. While ADHD medication improves behavioral outcomes, its long-term benefits on academic performance are limited.

Anna Chorniy, PhD
Assistant Professor, Pediatrics and
Population Health Science and Policy

Key Publications

1. **Chorniy A,** Currie J, Sonchak L. Exploding Asthma and ADHD Caseloads: The Role of Medicaid Managed Care. *Journal of Health Economics.* 2018; 60, 1-15.
2. Arbogast A, **Chorniy A,** Currie J. Administrative Burdens and Child Medicaid and CHIP Enrollments. *American Journal of Health Economics.* 2024; 10(2): 237-271.
3. **Chorniy A,** Kitashima L. Sex, Drugs, and ADHD: The Effects of ADHD Pharmacological Treatment on Teens' Risky Behaviors. *Labour Economics.* 2016; 43, 87–105.
4. **Chorniy A,** Moffa MA, Seltzer, RR. Expanding Access to Home-Based Behavioral Health Services for Children in Foster Care. *Administration and Policy in Mental Health and Mental Health Services Research.* 2024; 51:525–52.
5. Foster CC, **Chorniy A,** Kwon S, Kan K, Heard-Garris N, Davis, MM. Children with special health care needs and forgone family employment. *Pediatrics.* 2021; 148(3).

Lea K. Davis, PhD

Lea K. Davis, PhD is a Professor of Medicine, Psychiatry, and Genetics and Genomic Sciences. She is the Scientific Director of the Sinai Million Health Discoveries Program (SMHDP), one of the largest such sequencing projects of its kind. The SMHDP aims to integrate health and research data at Mount Sinai to promote discoveries that will directly benefit our patient population. Dr. Davis's research spans many domains of medicine and psychiatry from neurodevelopmental disorders to women's health and employs a population level approach to investigate the genetic basis of a wide range of complex health conditions, diseases, and common traits. Her lab's research integrates genetics, clinical informatics, and social determinants of health to study health outcomes captured in real world clinical data found in the electronic health record. In addition to her work on complex trait genomics, Dr. Davis has a long-standing commitment to furthering social justice through science, research ethics, genomic privacy, and responsible data sharing.

Lea K. Davis, PhD
Professor, Medicine, Psychiatry,
and Genetics and Genomic Sciences
Population Health Science and Policy

Key Publications

1. Singh K, Lee H, Sealock JM, Miller-Fleming T, Straub P, Cox NJ, ...**Davis LK**.. Genes associated with depression and coronary artery disease are enriched for cardiomyopathy and inflammatory phenotypes. *Nat. Mental Health* 2, 574–582 (2024).
2. Sealock JM, Lee YH, Moscati A, Venkatesh S, Voloudakis G, Straub P, Singh K, Feng YA, Ge T, Roussos P, Smoller JW, Chen G, **Davis LK**. Use of the PsycheMERGE Network to Investigate the Association Between Depression Polygenic Scores and White Blood Cell Count. *JAMA Psychiatry.* 2021 Dec 1;78(12):1365-1374.
3. Actkins KV, Jean-Pierre G, Aldrich MC, Velez Edwards DR, **Davis LK**. Sex modifies the effect of genetic risk scores for polycystic ovary syndrome on metabolic phenotypes. *PLoS Genet.* 2023 May 31;19(5):e1010764.
4. Miller-Fleming TW, Allos A, Gantz E, Yu D, Isaacs DA, Mathews CA, Scharf JM, **Davis LK**. Developing a phenotype risk score for tic disorders in a large, clinical biobank. *Transl Psychiatry.* 2024 Jul 28;14(1):311.
5. Lencz T, Sabatello M, Docherty A, Peterson RE, Soda T, Austin J, ... **Davis LK.** Concerns about the use of polygenic embryo screening for psychiatric and cognitive traits. *Lancet Psychiatry.* 2022 Oct;9(10):838-844.

Sarah Wood, MD

Sarah Wood, PhD is the Division Chief of Adolescent Medicine and an Associate Professor in the Icahn School of Medicine at Mount Sinai. She is a physician scientist with a career mission to improve equitable delivery of sexual, reproductive, and mental health care for adolescents and young adults. A former community health worker and HIV tester, Dr. Wood received her undergraduate degree at DePaul University and her M.D. at Drexel University of College of Medicine. After completing residency at the Children's Hospital of Philadelphia (CHOP), Dr. Wood continued her training as an Adolescent Medicine fellow and received a Masters of Science in Health Policy Research at the University of Pennsylvania Perelman School of Medicine. In 2024, she joined the faculty at Mount Sinai to carry on the mission and the vision of the Division of Adolescent Medicine and the AHC to provide high quality comprehensive care to vulnerable adolescents. Her research, funded by the National Institute of Mental Health and Gilead Sciences, focuses on developing and implementing HIV prevention interventions in adolescent primary care. She is also a faculty member in the Center for Child Health Services Research at Mount Sinai. She has published over 50 publications in peer-reviewed journals, and is a frequent speaker on podcasts, webinars, and at national scientific. Clinically, she is an Adolescent Medicine and HIV specialist committed to improving the care for youth locally and globally.

Sarah Wood, MD
Associate Professor, Pediatrics
Division Chief, Adolescent Medicine
Director, Mount Sinai Adolescent Health Center

Key Publications

1. **Wood SM,** Bauermeister J, Fiks AG, Phillips AW, Richardson HM, Garcia SM, Maleki P, Beidas RS, Young JF, Gross R, Dowshen N. Adolescent Preferences for a Pediatric Primary Care-based Sexually Transmitted Infection and HIV Prevention Intervention. *J Adolesc Health.* 2024 Jun;74(6):1231-1238.
2. Teixeira da Silva D, Makeneni S, Wall H, Bauermeister JA, **Wood S.** Measuring quality STI care among adolescent female primary care patients in Philadelphia. *Sex Transm Infect.* 2023 Jun;99(4):272-275.
3. Krass P, Tam V, Min J, Joslin I, Khabie L, Wilkinson TA, **Wood SM.** Adolescent Access to Federally Funded Clinics Providing Confidential Family Planning Following Changes to Title X Funding Regulations. *JAMA Netw Open.* 2022 Jun 1;5(6):e2217488.
4. **Wood SM,** Meanley S, Bonett S, Torres ME, Watson DL, Williams JL, Brady KA, Bauermeister JA. Strengthening HIV Prevention Services Through an Implementation Science Approach: Perspectives From the HIV Testers in Philadelphia. *J Acquir Immune Defic Syndr.* 2022 Jul 1;90(S1):S90-S97.
5. Wood S, Min J, Tam V, Pickel J, Petsis D, Campbell K. Inequities in Chlamydia trachomatis Screening Between Black and White Adolescents in a Large Pediatric Primary Care Network, 2015-2019. *Am J Public Health.* 2022 Jan;112(1):135-143.

Eric G. Zhou, PhD

Eric Geng Zhou, PhD is an Instructor of Pediatrics at the Center for Child Health Services Research, with a joint appointment in the Department of Population Health Science and Policy at the Mindich Child Health and Development Institute, Icahn School of Medicine at Mount Sinai. He is a health economist specializing in the health and development of underserved children. His research focuses on how healthcare resources, public policies, and environmental factors influence child well-being and contribute to health disparities.

Dr. Zhou's recent work, supported by the National Institute of Nursing Research, examines the real-world effects of COVID-19 vaccinations on children's health and academic outcomes in New York City public schools. This research, conducted in collaboration with various city government agencies, aims to inform public health strategies that promote health and mitigate disparities. His research has been published in journals such as JAMA Network Open and Obesity (Silver Spring). Through his work, Dr. Zhou contributes to evidence-based strategies aimed at promoting equitable healthcare access and delivery, while improving the well-being of vulnerable populations.

Eric G. Zhou, PhD
Instructor, Pediatrics and
Population Health Science and Policy

Key Publications

1. **Zhou EG,** Schwartz AE, Elbel B. Gentrification and childhood obesity: Evidence from New York City public school students in public housing. *Obesity (Silver Spring).* 2024 Feb;32(2):390-397.
2. Elbel B, **Zhou GE,** Lee DC, Chen W, Day SE, Konty KJ, Schwartz AE. Analysis of School-Level Vaccination Rates by Race, Ethnicity, and Geography in New York City. *JAMA Netw Open.* 2022 Sep 1;5(9):e2231849.

New Extramural Faculty- *Continued*

He obtained his Ph.D. in Public Policy (Health Economics track) from the Wagner School of Public Service at New York University, an M.A. in Public Policy Analysis from the Harris School of Public Policy at the University of Chicago, and an LL.B. in Sociology from Xiamen University.

TLC: Building the MCHDI Trainee Community

The Mindich Child Health and Development Institute (MCHDI) Trainee Leadership Committee (TLC) is dedicated to fostering the professional development of MCHDI trainees. Our mission is to create opportunities for individuals from diverse backgrounds and career stages to connect, share ideas, and build a strong community within MCHDI.

We seek to gather input from MCHDI trainees to develop new series focused on:

1. Career development, grant writing, and data analysis.
2. Enhancing collaboration among trainees from various research areas and educational backgrounds.
3. Facilitating networking opportunities between faculty and trainees.

Past initiatives that exemplify this include the MCHDI Trainee Incubator Series and involvement in the Child Health Research Seminar Series (CHRS). From 2021 to 2022, the TLC launched the MCHDI Trainee Incubator Series, which provided postdocs, PhD students, and master's students a platform to lead seminars and receive feedback on their project ideas, grant applications, job interviews, and major conference presentations. The TLC also hosted one to three workshops for the CHRS, chaired by Dr. Rebecca Trachtman. The series featured speakers at all career levels studying pediatric health from various institutes and organizations. We look forward to hearing from trainees on what new initiatives could be helpful in the future.

In 2017, the TLC established a pilot grant program to support postdoctoral/clinical fellows and PhD/MD-PhD students in pursuing new independent research projects—a crucial step towards academic independence. Over the past seven years, more than ten trainees have completed this program. This year's recipient is Dr. Jeronimo Lukin, a postdoctoral fellow from the De Rubeis lab, who will present his work at the MCHDI annual retreat in November. Applications for the 2025-2026 academic year will open next spring, and we encourage all MCHDI trainees with compelling proposals to apply!

We also extend our gratitude to all past and present TLC members for their incredible contributions over the years. The TLC was led by Lauren Dierdorff, Carolina Cappi, Shrey Patel, Yvette Carbajal, and Dr. Silvia De Rubeis last academic year. The 2024 TLC committee includes Lauren Dierdorff (Committee Chair), Shrey Patel, Dr. Silvia De Rubeis, and new member Aditi Prasad. In addition to organizing TLC programs and events, one committee member helps coordinate our MCHDI fall annual retreat each year. This year, Aditi joined the retreat committee as a trainee representative to ensure that trainees' perspectives and interests are reflected.

We're excited for another impactful year ahead with MCHDI trainees!

Lauren Dierdorff, BS
PhD Candidate, Psychiatry

Shrey Patel, BS
PhD Candidate, Biomedical Sciences

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

Silvia De Rubeis, PhD
Associate Professor, Psychiatry

Karen Levinson Mindich Trainee Pilot Program: 2024 Awardee

Project Title: Experience-driven changes in cortical circuits and gene expression in a mouse model of autism

Investigator: Jeronimo Lukin, PhD, Postdoctoral Fellow, Department of Psychiatry

Seaver Autism Center for Research and Treatment, Friedman Brain Institute, The Mindich Child Health and Development Institute

Primary Mentor:
Silvia De Rubeis, PhD

Associate Professor, Psychiatry

Seaver Autism Center for Research and Treatment

Friedman Brain Institute

The Mindich Child Health and Development Institute

Secondary Mentor:
Zhuhao Wu, PhD

Assistant Professor, Neuroscience

Weill Cornell Medicine Feil Family Brain & Mind Research Institute

Abstract:
Altered cortical circuits in the developing brain have been strongly linked to autism spectrum disorders (ASD) as well as ASD-relevant mouse models. Mutations in DDX3X cause a neurodevelopmental condition called DDX3X syndrome often presenting with ASD. Our lab generated the first mouse model for DDX3X sndrome and showed that Ddx3x+/- female mice have abnormal neocortical development and abnormal behavior in open field exploration (OF).

In collaboration with Dr. Zhuhao Wu (Weill Cornell Medicine), we mapped neural activity of the Ddx3x+/-mice brain after Open Field (OF) exploration using 3D c-Fos immunostaining on brains cleared with iDISCO technique and found an overactivation of several sensory cortical regions, including the anterolateral visual cortex. Based on these preliminary data and the phenotypes of Ddx3x+/- mice and patients, I plan to explore the gap between behavioral phenotypes and neuronal circuits alterations. My hypothesis is that circuits involving the visual cortex in Ddx3x+/- mice may exhibit altered neural activity and molecular identity contributing to the anxiety-like behaviors observed. To test this hypothesis, I will use chemogenetics to manipulate neuronal circuits and determine the role of visual cortical circuits on Ddx3x+/- mice anxiety-like behavior. Additionally, I will perform transcriptomics experiments to identify the experience-driven molecular signatures of Ddx3x mutations in the visual cortex.

Understanding how Ddx3x regulates brain circuits might offer a new key to decipher the complexity of circuit alterations in ASD. This project aims at revealing a salient circuit amenable for developing more broadly therapeutic intervention in children and individuals with DDX3X mutations and ASD.

Jeronimo Lukin, PhD
Postdoctoral Fellow, Psychiatry
Seaver Autism Center for Research and Treatment
Friedman Brain Institute
The Mindich Child Health and Development Institute

Faculty Grants/Awards/Honors

Bruce D. Gelb, MD and **Manisha Balwani, MD,** NINDS, U01, "Mount Sinai Center for Undiagnosed Diseases"

Bruce D. Gelb, MD and **Ross Cagan, PhD**, NHLBI, R01, "Therapy development for genetic disorders of the RAS/MAPK pathway"

Dorothy E. Grice MD (PI) and **Sidney Hankerson, MD, MBA** (M-PI), NIMH, R01, "Phenotypic and genetic architecture of OCD in African Americans"

Elvin Wagenblast, PhD, NCI, R01, "Overcoming Therapy Resistance in Fusion Oncoprotein Driven Pediatric Leukemia"

Brian Brown, PhD, NIDDK, R01, "MicroRNA-controlled mRNA therapeutics"

Brian Brown, PhD, **Julio Aguirre-Ghiso, PhD** (M-PI) and **Maria Sosa, PhD** (M-PI), NCI, U01, "Investigating disseminated cancer cell clonal cooperation and immune control in dormancy and metastasis"

Lauryn Choleva, MD, Integrated Islet Distribution Program Islet Initiative Award, "Exploring the Function of p57KIP2 in Diseases of The Human Pancreatic Beta Cell"

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SAVE THE DATE

12th Annual MCHDI Retreat

Date: November 20, 2024

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

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