



Icahn School
of Medicine at
Mount
Sinai

*The Mindich
Child Health and
Development Institute*

MCHDI Developmental Outcomes

FALL 2023

MCHDI Launches the Child Health Services Research Center Directed by Brett R. Anderson, MD, MBA, MS

We are pleased to announce that Dr. Brett R. Anderson, MD, MBA, MS has joined Mount Sinai as the inaugural Director for the Center for Child Health Services Research in the Mindich Child Health and Development Institute at the Icahn School of Medicine.

Dr. Anderson is a pediatric cardiologist and NIH-funded health services researcher, who blends her medical, business, and statistical backgrounds to bring together interdisciplinary investigators and data for the purposes of identifying modifiable drivers of outcomes, value, and health inequities, with a particular focus on children with cardiac disease. She has examined the effects of provider characteristics, surgical timing, social determinants of health, and various measures of healthcare access. She is the founder and Director of the New York State Congenital Heart Surgery Collaborative for Longitudinal Outcomes and Utilization of Resources (CHS-COLOUR), an interdisciplinary collaborative that brings together leadership and data from all congenital heart surgical centers in New York State, health services researchers, and the Department of Health, to examine etiologies of health inequities and to plan for programmatic interventions. Dr. Anderson completed her general pediatrics residency at The Children's Hospital of Philadelphia and fellowship in pediatric cardiology at NewYork-Presbyterian/Morgan Stanley Children's Hospital, prior to joining Columbia University Irving Medical Center's faculty in 2013. Dr. Anderson joins us after a decade at Columbia, where she served as Director of Clinical Research, Outcomes, and Quality for the Pediatric Heart Center and Co-Director of ASPIRE! Peer Mentoring.

The new Center for Child Health Services Research will serve as the primary locus for research on the healthcare system for children at The Icahn School of Medicine. The Center seeks to promote the sharing of interdisciplinary ideas, methodologies,

data, and mentorship, to enhance productivity and creativity for investigators—both within and outside the Center. Partnering with existing departments, institutes, and governmental agencies, the Center's expertise will focus on improving the quality and effectiveness of the healthcare system for all children, with particular emphasis on: **Providers, Payers, & Policy; Value & Effectiveness;** and **Access & Equity.**

Key Publications

1. Woo JL, Nash KA, Dragan K, Crook S, Neidell M, Cook S, Hannan EL, Jacobs M, Goldstone AB, Petit CJ, Vincent R, Walsh-Spoonhower K, Mosca R, Kumar TKS, Devejian N, Kamenir SA, Alfieris GM, Swartz MF, Meyer D, Paul EA, Newburger JW, Billings J, Davis MM, **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.
2. Hannan EL, Wu Y, Cozzens K, **Anderson BR**. The Neighborhood Atlas Area Deprivation Index for Measuring Socioeconomic Status: An Overemphasis On Home Value. *Health Aff (Millwood)*. 2023 May;42(5):702-709.
3. Crook S, Dragan K, Woo JL, Neidell M, Jiang P, Cook S, Hannan EL, Newburger JW, Jacobs ML, Bacha EA, Petit CJ, Vincent R, Walsh-Spoonhower K, Mosca R, Kumar TKS, Devejian N, Kamenir SA, Alfieris GM, Swartz MF, Meyer D, Paul EA, Billings J, **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.
4. Allen P, Zafar F, Mi J, Crook S, Woo J, Jayaram N, Bryant R 3rd, Karamlou T, Tweddell J, Dragan K, Cook S, Hannan EL, Newburger JW, Bacha EA, Vincent R, Nguyen K, Walsh-Spoonhower K, Mosca R, Devejian N, Kamenir SA, Alfieris GM, Swartz MF, Meyer D, Paul EA, Billings J, **Anderson BR**; New York State CHS-COLOUR. Risk Stratification for Congenital Heart Surgery for ICD-10 Administrative Data (RACHS-2). *J Am Coll Cardiol*. 2022 Feb 8;79(5):465-478.
5. **Anderson BR**, Fieldston ES, Newburger JW, Bacha EA, Glied SA. Disparities in Outcomes and Resource Use After Hospitalization for Cardiac Surgery by Neighborhood Income. *Pediatrics*. 2018 Mar;141(3):e20172432.



Brett R. Anderson, MD, MBA, MS

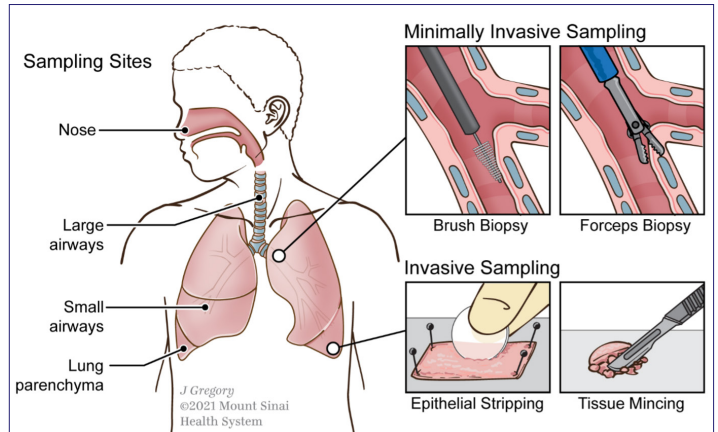
Director, Child Health Services Research Center
Associate Professor, Pediatrics
Mindich Child Health and Development Institute

Pediatric Cystic Fibrosis Research at Mount Sinai Goes Single Cell

Cystic fibrosis (CF) is a multisystemic, autosomal recessive disorder caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. The CFTR gene encodes a membrane protein and anion channel of the same name that is found in various epithelia throughout the human body, including the respiratory tract as well as the gastrointestinal tract, pancreas, hepatobiliary system, urogenital tract, and sweat gland. Defective or deficient CFTR protein leads to the common manifestations of CF, including obstructive lung disease, pancreatic insufficiency, and male infertility. Despite the tremendous advances afforded by CFTR modulator therapies – small molecules designed to target the underlying defects in the CFTR protein – a significant proportion of people with CF (pwCF) – many from minoritized racial and ethnic groups – are presently unable to benefit due, in large part, to genotypic heterogeneity (1). As such, CF lung disease remains a critical area of study.

Leveraging single-cell RNA sequencing (scRNA-seq) – a powerful tool that allows for the comparison of individual cellular transcriptomes through the identification of RNA molecules on a genomic scale and with high resolution – Drs. Megan Januska, Yifei Sun, and Martin Walsh are working to define disease-related changes in immune and respiratory epithelial cell populations in the pediatric CF lung from minimally invasive respiratory specimens obtained during flexible bronchoscopy (2). The use of scRNA-seq has allowed the research team to not only identify discrete immune and respiratory epithelial cell populations and subpopulations – some unique to the pediatric CF lung – with regard to their distinctive gene expression profiles but also characterize the cellular and molecular alterations that accompany CF lung disease. While scRNA-seq has been used to profile immune cell populations from expectorated sputa and bronchoalveolar lavage fluid from adults with CF and epithelial cell populations from adults with CF undergoing lung transplantation, this is the first study performed exclusively in the pediatric population and leveraging multiple specimen types (3-5).

The research team hopes the ongoing study will help inform novel approaches to the treatment of CF lung disease through the molecular characterization of disease-related immune and respiratory epithelial cell populations and subpopulations and a description of immune-epithelial cell interactions. Indeed, the heterogeneity of cell populations and subpopulations among children with CF suggests that specific signatures may predict CF lung disease progression and response to intervention as well as offers opportunities for the development of genotype agnostic therapies that could benefit all pwCF.



The present study leverages minimally invasive respiratory specimens, including tracheal brush biopsies, bronchial brush biopsies, and bronchoalveolar lavage (not depicted), to study the pediatric CF lung.

References

1. **Januska MN**, Marx L, Walker PA, Berdella MN, Langfelder-Schwind E. The CFTR variant profile of Hispanic patients with cystic fibrosis: Impact on access to effective screening, diagnosis, and personalized medicine. *J Genet Couns*. 2020 Aug;29(4):607-615.
2. **Januska MN, Walsh MJ**. Single-cell RNA sequencing reveals new basic and translational insights in the cystic fibrosis lung. *Am J Respir Cell Mol Biol*. 2023 Feb;68(2):131-139.
3. Schupp JC, Khanal S, Gomez JL, Sauler M, Adams TS, et al. Single-cell transcriptional archetypes of airway inflammation in cystic fibrosis. *Am J Respir Crit Care Med*. 2020 Nov 15;202(10):1419-1429.
4. Li X, Kolling FW, Aridgides D, Mellinger D, Ashare A, Jakubzick CV. ScRNA-seq expression of IFI27 and APOC2 identifies four alveolar macrophage superclusters in healthy BALF. *Life Sci Alliance*. 2022 Jul 12;5(11):e202201458.
5. Carraro G, Langerman J, Sabri S, Lorenzana Z, Purkayastha A, et al. Transcriptional analysis of cystic fibrosis airways at single-cell resolution reveals altered epithelial cell states and composition. *Nat Med*. 2021 May;27(5):806-814.



Martin J. Walsh, PhD

Professor, Pharmacological Sciences, Pediatrics, and Genetics and Genomic Sciences



Megan N. Januska, MD

Assistant Professor, Pediatrics and Genetics and Genomic Sciences

Exploring the Genetic Risk of OCD

Obsessive-compulsive disorder (OCD) has an estimated prevalence of about 2% and can lead to serious and life-long challenges. Despite its severity, and despite a historical understanding that OCD is highly heritable, research on the causes of OCD – including its genetic architecture – has lagged behind other psychiatric conditions. Chronic tic disorders, including Tourette syndrome, are related to OCD, as are anxiety and eating disorders. Genetic research in these disorders is also lagging.

Dr. Dorothy Grice and her team have been working to address this knowledge gap in genetic risk of OCD and related psychiatric disorders that, until recently, have not been at the forefront of genetic risk research. Dr. Grice leads an interdisciplinary research program focused on understanding the genetics and biology underlying OCD and tic disorders, applying the research methods used to identify genetic factors in other psychiatric illness to these underexplored areas.

Much of the team's work has utilized large-scale family cohorts to better understand the role of genetic risk for OCD. Their earlier work exploring familial risk patterns in a Danish birth cohort suggested a role for both direct genetics and maternal effect in OCD risk. Maternal effect, or genetic nurture, is an indirect genetic relationship of maternal factors impacting offspring behavior and other traits. More recently, the team developed a large multi-generational epidemiological study cohort in Sweden using national health records and DNA samples from individuals with OCD and confirmed the important role for both direct and indirect genetic effects (including maternal effect) in risk for OCD. This research also demonstrated a previously unilluminated aspect of OCD genetic risk: that changes across the allelic frequency spectrum – ranging from common to rare genetic changes – play a role in the risk for OCD.

Rare genetic changes, including rare spontaneous (*de novo*) mutations, are of particular interest to Dr. Grice and her team, who are now using sequencing techniques to study the ~20,000 protein-coding genes in the human genome to look for rare and *de novo* mutations associated with OCD. To

date, for this gene discovery research their team has aggregated over 22,000 samples from ten independent sources, including the Swedish study cohort, the Mount Sinai BioMe Biobank, the US national All of Us study, the UK Biobank, and smaller published sequence studies from 4 other research groups. So far, they have identified several genes that, when carrying spontaneous mutations, lead to increased risk for OCD. One of the most prominent genes identified, *CHD8*, is known to contribute to other psychiatric conditions, like autism.

These new insights into OCD genetic risk have been driven by the unprecedented number of participants – both cases and controls – in the study cohorts. As they continue to add to their sample collection, an urgent priority for Dr. Grice and her team is to expand and diversify the pool of OCD genetic samples.

Per Dr. Grice, “Although OCD affects every community and racial and ethnic group that has been studied, the genetic collections to date do not reflect this diversity – the vast majority of existing OCD samples have been collected from individuals of European ancestry.”

Indeed, from the 22,000 samples that have been analyzed thus far – which leverages two databases that are considered diverse – less than 100 Black American individuals with OCD were included. The team has therefore joined with collaborators, including Dr. Sidney Hankerson, and colleagues who work with Black communities and developed teams in New York and in North Carolina to engage Black adults in this important research, “to ensure that we learn about OCD risk across racial and ethnic groups.”

Dorothy E. Grice, MD

Professor, Psychiatry
Director, Tics, OCD and Related
Disorders Program



TLC Past and Future Goals

The Mindich Child Health and Development (MCHDI) Trainee Leadership Committee (TLC) continues to support MCHDI trainees in their professional development and growth. The TLC's mission is to create events for trainees from different backgrounds and at various career levels to come together to share ideas and build community within the MCHDI. The TLC activity highlights are listed below.

From 2021-2022, the TLC hosted the MCHDI Trainee Incubator Series, offering postdocs, PhD students, and masters students the opportunity to lead seminars and get feedback on new project ideas, grant and fellowship applications, job interviews and presentations at major conferences. The Incubator Series was just the beginning of initiatives employed by the TLC. We continue to gather insights from MCHDI trainees and plan new series and meetings to: 1) address career development, grant writing, and data analysis interests; 2) enhance interactions and collaborations amongst trainees from different research areas and educational levels; 3) facilitate networking between faculties and trainees. The TLC welcomes and implements suggestions from trainees to facilitate the most useful resources and events.

Every year, the TLC hosts at least three workshops for the Child Health Research Seminar Series (CHRS) chaired by Dr. Rebecca Trachtman. The TLC recruits speakers that study pediatric health at any career level and from different institutes or companies.

In 2017, the TLC launched a trainee pilot grant program in order to support postdoctoral/clinical fellows or PhD/MD-PhD students in pursuing a new independently funded line of research as a critical step towards academic independence. Over the past seven years of this unique program, more than ten trainees have been awarded and successfully completed the program. This year's recipients are postdoctoral fellow Dr. Marta Garcia-Forn from the De Rubeis lab and clinical fellow Dr. Katherine Schertz Hickey from the Bogunovic lab. The recipients will host oral presentations during our MCHDI annual retreat in November. Applications for the 2024-2025 academic year will open next Spring, and we encourage all MCHDI trainees with striking proposals to apply!

Finally, we would like to thank all the past and present TLC members for their amazing contributions and work managed and ensured during these past years. During the last academic year, the TLC was led by Carolina Cappi, Adele Mossa, Vahe Khachadourian, Nefatiti Anderson, and Silvia De Rubeis. The 2023 TLC committee includes Lauren Dierdorff, who has been selected as Committee Chair, Shrey Patel, Yvette Carbajal, Carolina Cappi, and Silvia De Rubeis. We are looking forward to expanding the TLC and recruiting new members. Besides the commitment in organizing TLC programs and events, every year one of the TLC members is involved in the organization of our MCHDI fall annual retreat. This year, Lauren has joined the retreat committee as a trainee representative, to provide feedback and converge the trainees' perspectives and interests. We look forward to another exciting year ahead with MCHDI trainees!



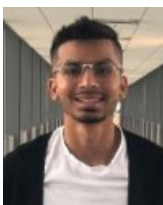
Carolina Cappi, PhD
Postdoctoral Fellow,
Psychiatry



Yvette Carbajal, BA
Associate Researcher,
Pediatrics



Lauren Dierdorff, BS
PhD Candidate, Psychiatry



Shrey Patel, BS
Student, Biomedical Sciences



Silvia De Rubeis, PhD
Associate Professor,
Psychiatry

Trainee Pilot Project: 2023 Awardees

Project Title: Lineage Tracing of Glutamatergic Neurons in the Developing Cortex of a Mouse Model of DDX3X Syndrome

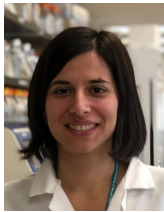
Investigator: Marta Garcia-Forn, PhD, Postdoctoral Fellow, Psychiatry, Seaver Autism Center for Research and Treatment, Friedman Brain Institute, The Mindich Child Health and Development Institute, The Alper Center for Neural Development and Regeneration

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, The Alper Center for Neural Development and Regeneration

Secondary Mentors:

Mladen-Roko Rasin, MD, PhD, Neuroscience and Cell Biology
Rutgers University, RWJ Medical School
Nikolaos P. Daskalakis, MD, PhD, Harvard Medical School, McLean Hospital



Marta Garcia-Forn, PhD

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

Abstract: Pediatric congenital brain malformations are the result of disrupted prenatal neocortical development, which is a critical nexus of risk for neurodevelopmental disorders including Autism Spectrum Disorder (ASD). Mutations in the ASD-risk gene DDX3X are the cause of DDX3X syndrome, a leading cause of intellectual disability and neurodevelopmental delays in females, often co-morbid with ASD and brain malformations.

This proposal aims to unravel the mechanisms underlying congenital brain malformations in neurodevelopmental disorders by focusing on the mechanisms of cortical development. Human and murine cortical development has been intensively studied and species-specific differences have emerged. Yet, the need for distinct cortical progenitors in either species is still elusive. Here, I will use cutting-edge in utero techniques and our mouse model for DDX3X syndrome, reported to show cortical malformations, to study the generation of glutamatergic neurons during cortical development in health and disease, setting the focus on the two existing routes of neurogenesis: direct neurogenesis, through which glutamatergic neurons are born directly from radial glia cells (RGC); and indirect neurogenesis, through which glutamatergic neurons are born from RGC-derived intermediate progenitors (IP).

This project is expected to bring a new understanding of how Ddx3x shapes cortical development and ultimately begin to understand the co-morbidity between neurodevelopmental disorders and congenital brain malformations, which will lay the basis for developing novel therapeutics with great impact on children's health.

Project Title: Markers of Immune Dysregulation in Pediatric Patients With Severe Presentation of Viral Bronchiolitis

Investigator: Katherine Schertz Hickey, MD, Clinical Fellow, Pediatric Intensive Care Unit

Primary Mentor:

Dusan Bogunovic, PhD
Director of the Center for Inborn Errors of Immunity
Associate Professor, Microbiology, Oncological Sciences, and Pediatrics
Mindich Child Health and Development Institute
Precision Immunology Institute

Secondary Mentors:

Sandeep Gangadharan, MD
Medical Director, Mount Sinai Pediatric ICU
Alfin Vincencio, MD
Division Chief of Pediatric Pulmonology

Abstract: Pediatric bronchiolitis is a condition caused by an acute viral infection of the lower airways. It results in a significant number of hospitalizations, morbidity, and mortality. Bronchiolitis can differ significantly in its clinical presentation. It can result in ICU hospitalization or can be managed while patient is at home, even

when caused by the same virus. This leads us to a hypothesis that host immune response significantly differs between those hospitalized in the ICU as compared to those who are not. Currently there is a paucity of studies in pediatric bronchiolitis patients that document markers of immune dysfunction and clinical presentation. Therefore, more in-depth analysis of immunologic profiling may offer insight to the immunologic mechanisms that drive disease severity. We hypothesize that in pediatric patients with severe presentation of acute lower airway disease, there will be significant differences in immunologic profiling compared to those with less severe disease. The objective of this pilot study is to determine the immunologic response map difference, including immune cells, immunoglobulins, and cytokines, in patients with moderate to severe clinical disease as compared to those with mild clinical disease and to perform whole exome genomic to determine if there are innate genetic differences among those who develop severe clinical presentation of disease compared to those who do not.



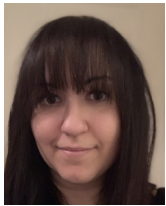
Katherine Schertz Hickey, MD

Clinical Fellow, Pediatric Intensive Care Unit

New Extramural Faculty

Georgia Panagiotakos, PhD

Georgia Panagiotakos, PhD is an Associate Professor of Psychiatry and Neuroscience at the Icahn School of Medicine at Mount Sinai. She is also a member of the Seaver Autism Center for Research and Treatment, the Alper Center for Neural Development and Regeneration, the Institute for Regenerative Medicine, and the Friedman Brain Institute. Prior to her arrival at Mount Sinai, Dr. Panagiotakos launched her independent research program as a Sandler Faculty Fellow at the University of California, San Francisco, after earning her Ph.D. from the Stanford University School of Medicine Neurosciences Doctoral Program. The central focus of the Panagiotakos laboratory is to dissect the cellular and molecular mechanisms underlying mammalian brain development, with an eye towards uncovering the underpinnings of neuropsychiatric disorders of developmental origin. We are especially interested in understanding how electrical activity, calcium signaling, and ion channel diversity regulate developmental transitions and the generation of distinct cell types in the developing brain. To investigate this, we incorporate multiple levels of analysis and a variety of orthogonal in vivo and in vitro approaches,



rate multiple levels of analysis and a variety of orthogonal in vivo and in vitro approaches,

Georgia Panagiotakos, PhD
Associate Professor, Psychiatry
and Neuroscience

including genetic tools in mouse models, live imaging and sequencing technologies. In the longer term, we also aim to shed light on how activity-regulated developmental mechanisms may be reactivated in the context of adult neurological disorders and the development of brain tumors.

Key Publications

1. Meng X, Yao D, Imaizumi K, Chen X, Kelley KW, Reis N, Thete MV, McKinney AA, Kulkarni S, **Panagiotakos G**, Bassik MC, Pasca SP. **Assembloid CRISPR Screens Reveal Impact of Disease Genes in Human Neurodevelopment.** *Nature.* 2023 Sep 27.
2. McKinney AA, Petrova R, **Panagiotakos G.** **Calcium and Activity-dependent Signaling in the Developing Cerebral Cortex.** *Development.* 2022 Sep 1;149(17):dev198853.
3. **Panagiotakos G**, Pasca SP. **A matter of space and time: Emerging roles of disease-associated proteins in neural development.** *Neuron.* 2022 Jan 19;110(2):195-208.
4. Braun SMG, Petrova R, Tang J, Krokhotin A, Miller EL, Tang Y, **Panagiotakos G***, Crabtree GR*. **BAF subunit switching regulates chromatin accessibility to control cell cycle exit in the developing mammalian cortex.** *Genes Dev.* 2021 Mar 1;35(5-6):335-353. (*co-senior authors)
5. **Panagiotakos G**, Haveles C, Arjun A, Petrova R, Rana A, Portmann T, Pasca SP, Palmer TD, Dolmetsch RE. **Aberrant calcium channel splicing drives defects in cortical differentiation in Timothy syndrome.** *Elife.* 2019 Dec 23;8:e51037.

New Intramural Faculty

Sandeep Gangadharan, MD

Sandeep Gangadharan, MD's, primary academic interest is in performance improvement of acute clinical care systems, whether through in situ simulation, clinical informatics, or process and implementation science. As a member of the Improving Pediatric Acute Care through Simulation (IMPACTS) study group, Dr. Gangadharan has been centrally involved in demonstrating both the utility of medical simulation as a diagnostic tool for evaluating complex clinical processes, such as acute resuscitation, and its ability to determine important clinical metrics that assess the quality of care. In addition



Sandeep Gangadharan, MD
Associate Professor, Pediatrics

Key Publications

1. Auerbach M, Whitfill T, Gawel M, Kessler D, Walsh B, **Gangadharan S**, Hamilton MF, Schultz B, Nishisaki A, Tay KY, Lavoie M, Katznelson J, Dudas R, Baird J, Nadkarni V, Brown L. **Differences in the Quality of Pediatric Resuscitative Care Across a Spectrum of Emergency Departments.** *JAMA Pediatr.* 2016 Oct 1;170(10):987-994.
2. Chime NO, Katznelson J, **Gangadharan S**, Walsh B, Lobner K, Brown L, Gawel M, Auerbach M; International Network for Simulation-based Pediatric Innovation, Research, and Education (INSPIRE) IMPACTS Investigators. **Comparing Practice Patterns Between Pediatric and General Emergency Medicine Physicians: A Scoping Review.** *Pediatr Emerg Care.* 2017 Apr;33(4):278-286.
3. Silver LJ, Pan S, Bucuvalas JC, Reid-Adam JA, Oishi K, Ofori-Amanfo G, **Gangadharan S.** **Acute Kidney Injury Following Pediatric Liver Transplant.** *J Intensive Care Med.* 2022 Jan;37(1):107-113.

New Intramural Faculty- Continued

to Dr. Gangadharan's interest in medical simulation, he has initiated, designed, and completed several clinical and quality research projects relevant to his field. The broader theme of many of these projects is evaluating and potentially enhancing the quality of procedural and resuscitation care in pediatric intensive care medicine utilizing multi-institutional databases. Dr. Gangadharan continues to be interested in similar work that seeks to evaluate the process of care, whether by simulation, data analytics, EMR optimization, or clinical informatics, to find areas of opportunity to make care more safe, efficient, and effective for children. Finally, Dr. Gangadharan continues to be an active member of the Pedi-Res-Q, Near-4-Kids, and Get with Guidelines collaborative efforts to study and improve resuscitative care in children. Currently, Dr. Gangadharan's focus is on clinical informatics and machine learning to develop effective clinical decision support tools and improve the process of acute care and Cardiopulmonary Resuscitation.

Corina Lesseur, MD, PhD

Corina Lesseur, MD, PhD is an Assistant Professor in the Department of Environmental Medicine and Public Health. Dr. Lesseur received her MD at the Central University of Venezuela and a PhD in Molecular and Experimental Medicine from Dartmouth College. She completed a postdoctoral fellowship in Genetic Epidemiology at the International Agency for Research in Cancer, followed by postdoctoral training in environmental health and molecular epidemiology at the Icahn School of Medicine at Mount Sinai.

Her work as a molecular epidemiologist focuses on placental epi/genomics and their link to pregnancy outcomes and early-life programming, as well as in the effects of environmental exposures (i.e., air pollution, pesticides) in the placenta and birth outcomes. She is particularly interested in maternal and infant metabolic outcomes (birth weight, obesity, and gestational diabetes). Dr. Lesseur has worked in multiple birth cohort studies evaluating placental epi/genetic features in relation to maternal and infant health, and environmental exposures. Dr. Lesseur has received funding from the NIH/

NICHD, the March of Dimes and the Marie Curie cofund.



Corina Lesseur, MD, PhD

Assistant Professor, Environmental Medicine and Public Health

Key Publications—Continued

4. Ambati SR, Tamuz M, DeVoe B, Rotjan A, Lesser M, **Gangadharan S**. Improving Resuscitation Timing: Random Assignment of Interprofessional Team Leaders in Simulated Resuscitation. *Pediatr Emerg Care*. 2022 Feb 1;38(2):e978-e982.
5. Langhan ML, Emerson BL, Nett S, Pinto M, Harwayne-Gidansky I, Rehder KJ, Krawiec C, Meyer K, Giuliano JS Jr, Owen EB, Tarquinio KM, Sanders RC Jr, Shepherd M, Bysani GK, Shenoi AN, Napolitano N, **Gangadharan S**, Parsons SJ, Simon DW, Nadkarni VM, Nishisaki A; for Pediatric Acute Lung Injury and Sepsis Investigators (PALISI) and National Emergency Airway Registry for Children (NEAR4KIDS) Investigators. End-Tidal Carbon Dioxide Use for Tracheal Intubation: Analysis From the National Emergency Airway Registry for Children (NEAR-4KIDS) Registry. *Pediatr Crit Care Med*. 2018 Feb;19(2):98-105.

Key Publications

1. **Lesseur C**, Kaur K, Kelly SD, Hermetz K, Williams R, Hao K, Marsit CJ, Caudle WM, Chen J. Effects of prenatal pesticide exposure on the fetal brain and placenta transcriptomes in a rodent model. *Toxicology*. 2023 May 15;490:153498.
2. Li Q, **Lesseur C**, Srirangam P, Kaur K, Hermetz K, Caudle WM, Fiedler N, Panuwet P, Prapamontol T, Naksen W, Suttiwan P, Baumert BO, Hao K, Barr DB, Marsit CJ, Chen J. Associations between prenatal organophosphate pesticide exposure and placental gene networks. *Environ Res*. 2023 May 1;224:115490.
3. **Lesseur C**, Jessel RH, Ohrn S, Ma Y, Li Q, Dekio F, Brody RI, Wetmur JG, Gigase FAJ, Lieber M, Lieb W, Lynch J, Afzal O, Ibroci E, Rommel AS, Janevic T, Stone J, Howell EA, Galang RR, Dolan SM, Bergink V, De Witte LD, Chen J. Gestational SARS-CoV-2 infection is associated with placental expression of immune and trophoblast genes. *Placenta*. 2022 Aug;126:125-132.
4. Kaur K, **Lesseur C**, Deyssenroth MA, Kloog I, Schwartz JD, Marsit CJ, Chen J. PM2.5 exposure during pregnancy is associated with altered placental expression of lipid metabolic genes in a US birth cohort. *Environ Res*. 2022 Aug;211:113066.
5. **Lesseur C**, Pathak KV, Pirrotte P, Martinez MN, Ferguson KK, Barrett ES, Nguyen RHN, Sathyanarayana S, Mandrioli D, Swan SH, Chen J. Urinary glyphosate concentration in pregnant women in relation to length of gestation. *Environ Res*. 2022 Jan;203:111811.

New Intramural Faculty- Continued

Alejandro Martin-Trujillo, PhD

Alejandro Martin Trujillo is an Assistant Professor in the Department of Genetics and Genomic Sciences within the team of Dr. Andrew Sharp. After earning his MSc in Genetics and Development, he completed his PhD in Biomedicine at the University of Barcelona (Spain) in 2014. During his PhD, Dr. Martin Trujillo characterized the extent of parent-of-origin DNA methylation in the human genome, identifying novel imprinted regions and thus, helping to define the human imprintome. Subsequently, he investigated the deregulation of these loci in a wide range of human diseases including the well-known imprinting disorders as well as several types of cancer. He then joined the laboratory of Dr. Sharp as a postdoctoral fellow, where his research expanded to explore epigenetic variation beyond imprinted loci as well as genetic variation at complex genomic regions that often eludes standard genetic studies. His current research primarily focuses on profiling both common and rare variation at tandem repeats (TR) from whole exome and genome

sequencing data using sophisticated computational approaches on a large scale. These studies aim to identify TR variation implicated in the regulation of the genome



Alejandro Martin-Trujillo, PhD

Assistant Professor, Genetics and Genomic Sciences

Caterina Tiozzo, MD, PhD

Caterina Tiozzo, MD, PhD is an Associate Professor of Newborn Medicine at the Icahn School of medicine. Dr. Tiozzo received her medical degree cum laude from Padova's University where she also completed her first pediatric residency, neonatal fellowship and Master of Public Health with humanitarian missions in Kenya. She then pursued her PhD through a collaboration between University of Padova and University of Southern California in Los Angeles.

After her humanitarian mission in Haiti during the 2010 earthquake, she decided to go back to clinical training to be able to practice in the United States so she completed her second pediatric residency and her neonatal fellowship at Columbia University. She trained and mentored many neonatal trainees over her career, both in Italy and in the U.S.



Caterina Tiozzo, MD, PhD

Associate Professor, Pediatrics (Newborn Medicine)

function, phenotypic diversity, and human diseases ranging from congenital anomalies to late-onset neurodegenerative disorders.

Key Publications

Martin-Trujillo A, Garg P, Patel N, Jadhav B, Sharp AJ. [Genome-wide evaluation of the effect of short tandem repeat variation on local DNA methylation.](#) *Genome Res.* 2023 Feb;33(2):184-196.

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She is a member of the "Society of Pediatric Research" and she is the recipient of several awards for her research discoveries from Europe and United States.

Key Publications

1. **Tiozzo C**, De Langhe S, Yu M, Londhe VA, Carraro G, Li M, Li C, Xing Y, Anderson S, Borok Z, Bellusci S, Minoo P. [Deletion of Pten expands lung epithelial progenitor pools and confers resistance to airway injury.](#) *Am J Respir Crit Care Med.* 2009 Oct 15;180(8):701-12.
2. **Tiozzo C**, Carraro G, Li A, Baptista S, Danopoulos S, Lavarreda-Pearce M, Li C, Chan B, De Langhe S, Borok Z., Bellusci S and Minoo P. [Mesodermal Pten inactivation leads to alveolar capillary dysplasia-like phenotype.](#) *J Clin Invest.* 2012 Nov;122(11):3862-72.
3. Chao CM, Yahya F, Moiseenko A, **Tiozzo C**, Shrestha A, Ahmadvand N, El Agha E, Quantius J, Dilai S, Kheirollahi V, Jones M, Wilhem J, Carraro G, Ehrhardt H, Zimmer KP, Barreto G, Ahlbrecht K, Morty RE, Herold S, Abellar RG, Seeger W,

New Intramural Faculty- Continued

She was selected by the Italian government for a documentary on Italians in New York during COVID and she received the title of Knight of the “Order of Croce d’Italia” by the Italian President for her work.

Dr. Tiozzo’s research focuses on lung development, stem cells regeneration after lung injury, the role of the intrauterine environment in lung development and the effect of neonatal nutrition on lung development. She is the author of over 30 papers and often invited to speak at national and international meetings on the topic of her research interests.

Schermuly R, Zhang JS, Minoo P, Bellusci S. Fgf10 deficiency is causative for lethality in a mouse model of bronchopulmonary dysplasia. *J Pathol.* 2017 Jan;241(1):91-103

4. Clauss, C, Thomas, S, Khodak I, Tack, V, Akerman M, Hanna N, **Tiozzo C.** Hydrocortisone and bronchopulmonary dysplasia: variables associated with response in premature infants. *J Perinatol.* 2020 Sep;40(9):1349-1357.

5. Clauss C, Tack V, Macchiarulo M, Akerman M, El-Chaar G, Hanna N, **Tiozzo C.** Light protection of parenteral nutrition, cholestasis, and other prematurity-related morbidities in premature infants. *Front Pediatr.* 2022 Aug 3;10:900068.

Grants, Awards, and Honors

Faculty Grants/Awards/Honors

Sharon Baumel-Alterzon, PhD, The Einstein/Sinai Diabetes Research Center Pilot and Feasibility Micro-grant Award, “The role of Nrf2 in beta-cell development from human pluripotent stem cells”

Supinda Bunyavanich, MD, MPH, MPhil (PI) and Scott Sicherer, MD (M-PI), NIAID, U19, “Threshold, Severity, and Immunotherapy of Peanut Allergy”

Supinda Bunyavanich, MD, MPH, MPhil, Chair, NIH Study Section: Cardiovascular and Respiratory Diseases, 2022-2024

Bruce D. Gelb, MD, Keynote Speaker, Pathologies of the RAS-MAPK Pathway: The Importance of a Multidisciplinary Network, Salerno, Italy, May 23, 2023

Bruce D. Gelb, MD, Plenary Speaker, 8th International RASopathies Symposium: Expanding Research and Care Practice, Through Global Collaboration and Advocacy, Denver, CO, August 23, 2023

Bruce D. Gelb, MD, Plenary Speaker, International Pediatric VAC and Heart Failure Summit 2023, St. Louis, MO, September 23, 2023

Praveen Raju, MD, PhD, CURE Childhood Cancer, 2023 Translation to CURE Award (T2C), 07/01/23-06/30/25, “Nanotherapeutic targeting of PPM1D inhibitors across the blood-brain barrier for pediatric brainstem tumors”

Praveen Raju, MD, PhD, Keynote Speaker, “Translational Hurdles in Pediatric Neuro-Oncology - The Elephants in the Room”, The BrainStorm Summit - End Childhood Brain Cancer, Washington, DC, September 22, 2023

Eyal Shemesh, MD, Research Grant, Department of Global Health and Health System Design, and the Arnhold Institute for Global Health, in partnership with New York City Health + Hospitals (NYC H+H)/Elmhurst and NYC H+H/Queens, “CP&R – Cardiovascular Precision Medicine & Remote Intervention”

Ernest Turro, PhD, NHLBI, R01, “Integrative analysis of whole genomes and transcriptomes from multiple cell types in rare disease patients.”

Elvin Wagenblast, PhD, Pew-Stewart Scholars Program for Cancer Research, 2023 Scholar

Trainee Grants/Awards/Honors

Lauren Dierdorff, PhD candidate, Federation of European Neuroscience Societies (FENS) Chen Institute NeuroLéman Summer School: “Motor Control: From Thought to Action” stipend awardee

Lauren Dierdorff, PhD candidate, Federation of European Neuroscience Societies (FENS) Chen Institute NeuroLéman Summer School: “Motor Control: From Thought to Action”, “Dissecting Cortical Circuits Driving Motor Deficits in a Mouse Model of DDX3X syndrome” poster presentation

Lauren Dierdorff, PhD candidate, Seaver Autism Center for Research and Treatment, Beatrice and Samuel A. Foundation Predoctoral Fellowship

Tasneem Ebrahim, BA, Weinstein Conference for Cardiovascular Development and Regeneration, “Dissecting mechanisms of cardiac specification and differentiation using large-scale CRISPR-based screens” presentation award

Miranda Wilson, BS, NICHD, F31 Parent Diversity Grant, “Molecular Mechanisms of Sex-Specific Differentiation”

Miranda Wilson, BS, 9th Vertebrate Sex Determination Meeting, “The RNA-binding protein, Rbpms2, regulates mTOR signaling via the GATOR2 complex protein, Mios, to promote oogenesis and female fate in zebrafish” oral talk presentation

Miranda Wilson, BS, 9th Vertebrate Sex Determination Meeting, “The RNA-binding protein, Rbpms2, regulates mTOR signaling via the GATOR2 complex protein, Mios, to promote oogenesis and female fate in zebrafish”, *FEBS Letters* “Best Talk” award

Publications

- 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;82(13):1331-1340.
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Publications, continued

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Webb

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