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

MCHDI Developmental Outcomes

Spring 2021

Research Advancements: Environmental Health

Decreasing Sperm Counts and Prenatal Phthalate Exposure

In late July 2017, it seemed as if every media outlet around the globe had become obsessed with the state of human sperm counts, with alarming headlines worldwide. By the end of the year, the paper Shanna H. Swan, PhD and her coauthors had published in Human Reproduction Update (Vol.23, No.6 pp. 646–659, 2017) "Temporal Trends in Sperm Count: A Systematic Review and Meta-Regression Analysis," (Levine et al 2017) which sparked these stories was ranked 26th by Altmetrics, among all referenced scientific papers published worldwide in 2017.

The scope of the meta-analysis was broad. With the help of Rachel Pinotti (MSSM Reference Librarian) they used bibliographic data bases to identify all English language studies of human sperm count published in 1981–2013. Following a predefined protocol 7518 abstracts were screened and 2510 full articles reporting primary data on SC were reviewed. The resulting meta-regression analysis of trends in sperm count and concentration included data from 185 studies based on data from 42,935 men who provided semen samples in 1973-2011. It was the largest such meta-analysis ever done.

They reported that between 1973 and 2011, the first and lasts years of sample collection, the median sperm count in Western countries fell from 99 mill per ml to 47 mill per ml, a decline of 52% in 39 years. (There were too few studies to obtain a reliable estimate of slope for those countries). When they compared this slope to those for recent periods the slopes did not change significantly, suggesting that the decline was not becoming less marked and concluded there had been a robust significant and continuing decline in sperm counts in Western countries.

This paper did not address causes of this decline. However, a series of studies that Dr. Swan and her colleagues have been conducting since 2000 have identified a class of chemicals that have the ability to significantly impair male reproductive function; the phthalates. Multiple animal studies published around 2000 demonstrated that prenatal exposure to certain phthalates (DEHP, DBP, BBzP) altered the development of male genitals, producing a cluster of outcomes termed the *phthalate syndrome* (Foster 2006). In particular, one measure of reproductive development, the anogenital distance (AGD), was reduced in male rodents following early prenatal exposure to these phthalates as well as smaller penile and scrotal size and undescended testes. In their multicenter pregnancy cohort study (Study for Future Families, SFF) they examined concentrations of phthalate metabolites in stored urine samples in relation to genital measures in male infants and identified the Phthalate Syndrome in humans (Swan et al 2005, Swan et al 2008). They subsequently replicated this finding in a second multi-center pregnant study (The Infant Development and the Environment Study (TIDES) (Swan et al 2015). To examine the reproductive consequences of a short male AGD, they conducted a study of college students at the University of Rochester and showed that AGD was directly related to sperm concentration (Mendiola et al 2011). Studies in Stanford confirmed this finding and also reported shorter AGD in infertile men (Eisenberg et al 2011). This series of studies demonstrates that early prenatal exposure to anti-androgen phthalates is linked, via shortened AGD in infancy, to decreased semen quality in adulthood, suggesting that these endocrine disrupting chemicals contribute to the declining sperm counts they reported in 2017 (Levine et al).

Shanna H. Swan, PhD
Professor, Environmental Medicine & Public Health

Key Select References:

Levine H, Jørgensen N, Martino-Andrade A, Mendiola J, Weksler-Derri D, Mindlis I, Pinotti R, **Swan SH.** Temporal trends in sperm count: a systematic review and meta-regression analysis . *Hum Reprod Update.* 2017 Nov 1;23(6):646-659.

Swan SH, Sathyanarayana S, Barrett ES, Janssen S, Liu F, Nguyen RH, Redmon JB; TIDES Study Team. First trimester phthalate exposure and anogenital distance in newborns . *Hum Reprod.* 2015 Apr;30(4):963-72.

Mendiola J, Jørgensen N, Andersson AM, Stahlhut RW, Liu F, **Swan SH.** Reproductive parameters in young men living in Rochester, New York . *Fertil Steril.* 2014 Apr;101(4):1064-71.

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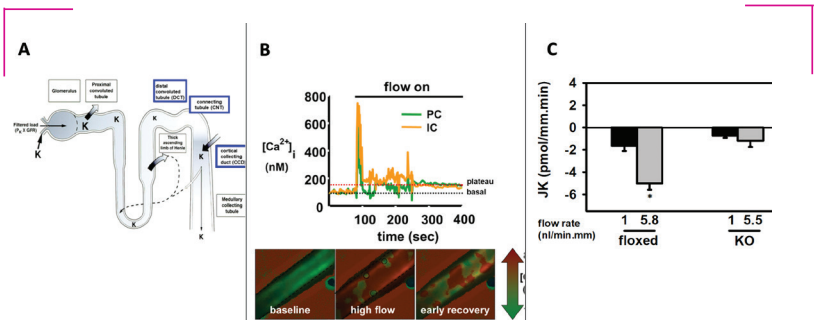
The Renal BK Channel Rediscovered – Clues From Development and Disease

Maintenance of K^+ homeostasis in adults, critical for diverse physiologic functions, requires that the kidney excrete >90% of K+ ingested daily. Children, in contrast, must retain dietary K^+ for somatic growth.

Renal K^+ excretion is mediated by K^+ secretion in the distal nephron (DN; Fig. A), comprised of principal cells (PCs), which absorb Na^+ and secrete K^+, and intercalated cells (ICs), which are specialized for acid/base transport but may reabsorb K^+ under certain conditions. K^+ secretion requires Na^+ diffusion from the urinary fluid across the apical Na^+ channel ENaC in PCs, and its electrogenic basolateral extrusion by Na-K-ATPase. Cell K^+ then passively diffuses into the lumen down a favorable electrochemical gradient through K^+ secretory channels. Of the two apical K^+-selective channels in the DN, the stretch-/Ca^+2-activated BK channel was the first described in both PCs and ICs. However, its vanishingly low open probability (Po) at the resting membrane potential led to the assumption that this channel does not participate in K^+ secretion. The prevalence of the ROMK channel, its high P_o, and selective localization to PCs led to the premise that this channel mediates K^+ secretion in the DN.



(A) Overview of K^+ handling in the nephron. K^+ is freely filtered at the glomerulus. >90% of the filtered load of K^+ is reabsorbed by the beginning of the distal tubule, regardless of total body K^+ balance. Net urinary K^+ excretion is determined, in the adult, by K^+ secretion by the aldosterone-sensitive DN (ASDN). (B) An increase in luminal fluid flow rate in the microperfused rabbit CCD, loaded with the $Ca2^+$-sensitive fluorescent indicator dye fura-2, increases tubule diameter by ~15% and $[Ca2^+]_i$. (C) Targeted disruption of BKα in ICs inhibits flow-induced K^+ secretion (JK) (but not Na^+ absorption) in microperfused CCDs from male IC-BKα-KO mice fed a high K^+ diet.

However, cumulative observations suggested that ROMK was not the only physiologically relevant K^+ secretory channel. First, patients with Bartter syndrome due to loss-of-function mutations in ROMK are not limited in their ability to secrete K^+. Second, an increase in luminal flow rate in the in vitro microperfused cortical collecting duct (CCD), a segment within the DN, of the weanling rabbit fails to stimulate K^+ secretion, as consistently observed in the adult, although conducting ROMK channels are abundant.

In microperfused rodent CCDs, Satlin *et al* observed that an increase in luminal flow rate, as might be induced *in vivo* by volume expansion or diuretics, led to circumferential stretch and an increase in cell $Ca2^+$ concentration (Fig. B), both known activators of the BK channel. Subsequent observations in the CCD that (i) flow-induced K^+ secretion (FIKS) was inhibited by the BK channel blocker iberiotoxin, and (ii) the developmental appearance of FIKS paralleled that of immunodetectable BKα, the pore-containing subunit of the BK channel, suggested a role of the BK channel in FIKS.

Lisa M. Satlin, MD
Professor and System Chair, Pediatrics
Professor, Medicine

Still unresolved was whether ICs and/or PCs mediate BK channel-mediated FIKS. Given that dietary K^+ loading of rabbits led to an increase in ICs of immunodetectable apical BKα and L-WNK1, a kinase that increases BKα abundance and functional channel expression in heterologous expression systems, Satlin and colleagues generated and characterized a mouse with IC-specific targeted disruption of BKα (IC-BKα-KO). Electrophysiologic analyses revealed BK channel-mediated whole cell K^+ currents in ICs of control but not IC-BKα-KO mice. When fed a high K^+ diet, blood $[K^+]$ was greater in IC-BKα-KO mice vs. controls in males only. FIKS was present in microperfused CCDs isolated from controls, but was absent in IC-BKα-KO CCDs of both sexes (Fig. C). These results provide compelling evidence that ICs, cells not traditionally considered to secrete K^+, are capable of secreting K^+ into the urinary fluid under conditions of dietary K^+ loading and high flow, and that sex-specific differences exist in the renal regulation of K^+ excretion.

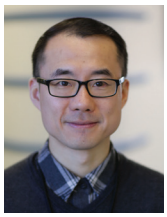
Pilot Project: 2021 Awardees

Project Title: Brain Activity Landscape With Cellular Mapping: Probing Circuitry Mechanism in a Mouse Model of Intellectual Disability

Principal Investigators: *Silvia De Rubeis, PhD (communicating PI) and Zhuhao Wu, PhD (co-PI)*

Abstract: Intellectual disability (ID) is a prevalent neurodevelopmental disorder with no effective pharmacological treatment. Mutations in the DDX3X gene are one of the most common causes of ID. DDX3X function in neurodevelopment is just beginning to emerge. *Understanding the role of DDX3X in the formation of brain circuits might offer a new key to decipher ID*. This proposal seeks to address this unmet need by putting together complementary expertise and innovative tools, applied to the first mouse model with construct validity for DDX3X mutations. Based on findings in this new model, we hypothesize that Ddx3x mutant mice have changes in neuronal populations that map onto specific circuits subserving brain function and behavior. We will: 1) *Map whole-brain neuronal populations in Ddx3x mutant mice.* To understand brain-wide neural defects, we will perform three-dimensional profiling of both general and specific neural populations using an advanced clearing method (iDISCO). 2) *Examine brain activity after behavior in Ddx3x mutant mice*. We will monitor whole brain activity landscape at cellular resolution after a simple naturalistic behavioral task called open field test. Expression of the immediate early gene cFos combined with iDISCO will be used as a proxy for neuronal activity. This proposal seeks to *transform our understanding of the mechanisms of ID by reaching whole-brain, circuitry-level resolution,* while propelling the development of a robust platform to probe convergences across many ID mouse models and thus decipher the complexity of ID. The proposal closely aligns with the *mission of MCHDI Pilot Program: 1)* it stems from a new collaboration between two early-stage investigators; *2)* it aims at establishing a new line of research that has the ultimate goal of paving the way for precision medicine approaches to ID, thus improving children's health; and *3)* will be instrumental to generate additional preliminary data for a NIH R01 application.

Silvia De Rubeis, PhD (communicating PI)
Assistant Professor, Department of Psychiatry
Seaver Autism Center for Research and Treatment
Mindich Institute for Child Health and Development
Friedman Brain Institute



Zhuhao Wu, PhD (co-PI)
Assistant Professor, Department of Cell, Developmental & Regenerative Biology &
Department of Neuroscience
Friedman Brain Institute

Project Title: Gene Expression in Endocervix During Pregnancy – Novel Biomarkers of Neonatal Outcomes

Principal Investigators: Magdalena Janecka, PhD (communicating PI), Lisa Eiland, MD, FAAP (co-PI) and Ernest Turro, PhD (co-PI)

Abstract: Neonatal adversity is associated with a host of short- and long-term health complications. Although these complications may be highly debilitating, and affect all body systems, our understanding of the mechanisms affecting neonatal adversity, and ability to predict it early in pregnancy, remain limited. Our project will address this knowledge gap by exploring the potential of using gene expression data from endocervical samples collected during routine examination of pregnant women. We will assess transcript abundance using RNA-seq, allowing us to quantify both coding and non-coding populations of RNAs. We will then test the associations between gene expression – both at a single RNA, and pathway levels – and subsequent birth outcomes (including gestational age and size for gestational age; APGAR score; NICU admission). The availability of rich demographic and health information for all women in the sample will further allow us to adjust for a host of maternal factors that could contribute to the observed associations. All RNA-seq data will be made available to other Mount Sinai researchers on terms consistent with the IRB – availability of full MSHS EHRs will allow to further enhance the impact and utility of our data. Upon successful completion of the proposed project, this study will offer new insights into the associations between transcript abundance in the endocervix during pregnancy and neonatal outcomes, thus advancing identification of high-risk pregnancies early in gestation, prevention of neonatal adversity and understanding of the mechanisms underlying neonatal outcomes. This pilot project is intended to seed a future R01 grant. Our expert team, operating within a unique MSHS setting – allowing integration of clinical care and research – will be well-positioned to extend the project to include a larger number of samples, a more diverse population, additional time-points of cervical swab collection and comparison with RNA-seq data obtained from blood and vaginal swabs.

Magdalena Janecka, PhD (communicating PI)
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Ernest Turro, PhD (co-PI)
Associate Professor,
Department of Genetics and Genomic Sciences
Mindich Child Health and Development Institute

Lisa Eiland, MD, FAAP (co-PI)
Associate Professor,
Department of Pediatrics
Division of Newborn Medicine
Mindich Child Health and Development Institute

Pilot Project: 2021 Awardees-Continue

Project Title: A Family-Mediated Intervention to Improve Outcomes in Minority Children with Autism Spectrum Disorder a ected by the COVID-19 Pandemic

Principal Investigators: M. Pilar Trelles, MD (communicating PI) and Jennifer Foss-Feig, PhD (co-PI)

Abstract: Social distancing measures introduced to mitigate the spread of SARS-CoV-2 have dramatically impacted the utility of treatment programs for children with ASD. As a consequence, there has been a dramatic increase in need for psychiatric services and hospitalizations. In minority groups, these effects are further by existing racial and ethnic healthcare disparities. In the wake of the COVID-19 pandemic, the Center Ann Sullivan of Peru (CASP) developed a 16-week virtual, parent- mediated curriculum designed to support families during this period, building on an evidence-based program dating back to 1984. The intervention empowers caregivers to help their children build on functional skills considered essential to promote independence, facilitate academic learning, and enable inclusion in all aspects of life. Making it ideal for immediate implementation, the program draws from its global experience working in high-stress environments with unstable political, financial and social climates, with emphasis placed on feasible interventions that are compatible with family life, and applicable beyond the current crisis. The proposed project aims to evaluate the utility of the CASP online curriculum, compared to treatment as usual, in n=40 underserved minority youth with ASD in New York City. *Our central hypothesis is that program participation will improve the well-being of children with ASD during the COVID-19 pandemic by ameliorating disruptive behaviors, improving adaptive behavior, reducing caregiver strain, and enhancing family quality of life.* Successful completion of our aims will (1) validate the effectiveness of integrating the CASP virtual model with traditional care delivery; (2) mitigate the impact of COVID-19 related disruptions for families raising children with ASD; (3) evaluate the utility of a caregiver-mediated interventions in the treatment of children with ASD affected by the COVID-19 pandemic; (4) solidify communities ties to diversify research participation in future studies; (5) provide pilot data for larger-scale NIH funding.

M. Pilar Trelles, MD (communicating PI)
Assistant Professor, Department of Psychiatry
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Jennifer Foss-Feig, PhD (co-PI)
Assistant Professor, Department of Psychiatry
Seaver Autism Center for Research and Treatment
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New Intramural Faculty

Jennifer Foss-Feig, PhD

Jennifer Foss-Feig, PhD, is an Assistant Professor in the Department of Psychiatry and at the Seaver Autism Center for Research and Treatment. Dr. Foss-Feig's research interests are in sensory and perceptual processing, particularly as they relate to possible underlying mechanisms of and biomarker development for autism spectrum disorder (ASD). She studies auditory, visual, and multisensory processing, with a particular interest in temporal processing and testing for markers of excitatory/inhibitory imbalance. More recent work also examines the neurocomputational basis of proactive social behavior deficits in ASD. Dr. Foss-Feig also examines the overlap between autism and other neurodevelopmental disorders, including schizophrenia, and works in monogenic disorders that confer risk for ASD but have known etiology that maps to testable and targetable circuit-based alterations. Dr. Foss-Feig's research combines EEG, fMRI, and psychophysical approaches with clinical and behavioral assessments to explore brain-behavior relations and identify underlying alterations that may be targets for intervention. Her work testing neural mechanisms across ASD and schizophrenia seeks to identify shared versus dissociable markers that could be useful as stratification tools for predictingpsychosis risk in ASD populations. Across her research, Dr. Foss-Feig seeks to use paradigms that map to known biological alterations, are objective and reliable, and

Continue

Key Publications:

McLaughlin C, Guillory SB, Isenstein EL, Grosman HE, Trelles MP, Siper PM, Kolevzon, A, Wang AT, **Foss-Feig JH.** Domain general differences in visual engagement and disengagement in autism spectrum disorder. *Autism*. [In Press]

Jutla A, Donohue MR, Veenstra-VanderWeele J, **Foss-Feig JH.** Reported autism diagnosis is associated with psychotic-like symptoms in the Adolescent Brain Cognitive Development cohort. *Eur Child Adolesc Psychiatry.* 2021 Mar 1.

Foss-Feig JH, Guillory SB, Roach BJ, Velthorst E, Hamilton H, Bachman P, Belger A, Carrion R, Duncan E, Johannesen J, Light GA, Niznikiewicz M, Addington JM, Cadenhead KS, Cannon TD, Cornblatt B, McGlashan T, Perkins D, Seidman LJ, Stone WS, Tsuang M, Walker EF, Woods S, Bearden CE, Mathalon DH. Abnormally Large Baseline P300 Amplitude Is Associated With Conversion to Psychosis in Clinical High Risk Individuals With a History of Autism: A Pilot Study. *Front Psychiatry.* 2021 Feb 9;12:591127.

Siper PM, Layton C, Levy T, Lurie S, Benrey N, Zweifach J, Rowe M, Tang L, Guillory S, Halpern D, Giserman-Kiss I, Del Pilar Trelles M, **Foss-Feig JH,** De Rubeis S, Tavassoli T, Buxbaum JD, Kolevzon A. Sensory Reactivity Symptoms Are a Core Feature of ADNP Syndrome Irrespective of Autism Diagnosis. *Genes (Basel).* 2021 Feb 27;12(3):351.

Gorenstein M, Giserman-Kiss I, Feldman E, Isenstein EL, Donnelly L, Wang AT, **Foss-Feig JH.** Brief Report: A Job-Based Social Skills Program (JOBSS) for Adults with Autism Spectrum Disorder: A Pilot Randomized Controlled Trial. *J Autism Dev Disord.* 2020 Dec;50(12):4527-4534.

are feasible or adaptable for individuals with neurodevelopmental disorders across the functioning spectrum. Dr. Foss-Feig has received funding from NIMH, Autism Speaks, Autism Science Foundation, the Marino Autism Research Institute, and the Brain and Behavior Research Foundation. She is a licensed clinical psychologist in the state of New York.

Jennifer Foss-Feig, PhD
Assistant Professor, Psychiatry
Mindich Child Health and Development Institute

Kaustav Mukherjee, PhD

Kaustav Mukherjee, PhD is an Instructor in Dr. James Bieker's lab of the Cell, Developmental and Regenerative Biology department at Icahn School of Medicine at Mount Sinai. He is also a member of the Black Family Stem Cell Institute and the Mindich Child Health and Development Institute.

A geneticist by training, he worked on yeast RNA biology during his PhD before completing his postdoctoral work at Dr. James Bieker's lab. Currently, he focuses on the biology of in vivo and in vitro erythropoiesis and associated red cell disorders such as anemia. He uses both animal models as well as stem cells to investigate the transcriptional regulation of gene expression during erythropoiesis by the transcription factor EKLF/Klf1 in erythroid cells, as well as macrophages that constitute the erythroblast island niche. He also uses patient-derived stem cells of congenital erythroid disorders affecting young children to study dyserythropoiesis leading to severe anemia. He extensively uses next generation sequencing, epigenetics, and single cell RNA-seq in his research, and is skilled in the use of Bioinformatics and computational biology for analysis of NGS and sc-seq data. His research is supported by a pilot grant from the Black Family Stem Cell Institute titled "Investigating transcription regulation during erythroid differentiation of human induced pluripotent stem cells".

Key Publications:

Mukherjee K, Xue L, Planutis A, Gnanapragasam MN, Chess A, Bieker JJ. EKLF/KLF1 expression defines a unique macrophage subset during mouse erythropoiesis. *Elife.* 2021 Feb 11;10:e61070.

Dr. Mukherjee has completed his undergraduate degree from Osmania University, India, his Master's in Biotechnology from Madurai Kamaraj University,

Kaustav Mukherjee, PhD
Instructor, Cell, Developmental and Regenerative Biology

Praveen Raju, MD, PhD

Praveen Raju, MD, PhD is a pediatric neurologist and Associate Professor of Neurology and Pediatrics at the Icahn School of Medicine at Mount Sinai.

Dr. Raju completed his MD in 2001 at the University of Pennsylvania School of Medicine, where he also completed his PhD in Cell and Molecular Biology / Genetics as an NIH-funded MSTP Fellow. He served as a Pediatrics resident at Babies & Children's Hospital of New York / Columbia-Presbyterian Medical Center and subsequently finished his Pediatric Neurology Fellowship training at Boston Children's Hospital / Harvard Medical School in 2006 where he served as Chief Fellow during his final year. Prior to joining ISMMS, he served on the faculty at Weill Cornell Medicine for 10 years where he was the Caryl & Israel A. Englander Clinical Scholar in Children's Health and also cared for children with neurological issues at Memorial Sloan Kettering Cancer Center.

At Mount Sinai Kravis Children's Hospital, Dr. Raju's primary clinical focus in pediatric onco-neurology treats patients with neurological complications of cancer including seizures, headaches, and neuropathy. His clinic also has a special interest in pediatric neurofibromatosis (NF) and neurogenetic disorders.

In addition to his clinical responsibilities, Dr. Raju directs the Laboratory for Pediatric Brain Tumor Research at ISMMS and studies the developmental origins of pediatric brain tumors with a particular translational focus on identifying improved therapies for medulloblastoma. He is also the Director of the Mount Sinai Pediatric Neurology Residency Program and Associate Director of the Mount Sinai Medical Scientist Training Program (MSTP).

Key Publications:

Pranzatelli MR, Tate ED, Swan JA, Travelstead AL, Colliver JA, Verhulst SJ, Crosley CJ, Graf WD, Joseph SA, Kelfer HM, **Raju GP.** B cell depletion therapy for new-onset opsoclonus-myoclonus. *Mov Disord.* 2010 Jan 30;25(2):238-42.

Takenouchi T, **Raju GP.** Germinal matrix hemorrhage in Zellweger syndrome. *J Child Neurol.* 2010 Nov;25(11):1398-400.

Lao Z, **Raju GP,** Bai CB, Joyner AL. MASTR: a technique for mosaic mutant analysis with spatial and temporal control of recombination using conditional floxed alleles in mice. *Cell Rep.* 2012 Aug 30;2(2):386-96.

Suero-Abreu GA, **Raju GP,** Aristizábal O, Volkova E, Wojcinski A, Houston EJ, Pham D, Szulc KU, Colon D, Joyner AL, Turnbull DH. In vivo Mn-enhanced MRI for early tumor detection and growth rate analysis in a mouse medulloblastoma model. *Neoplasia.* 2014 Dec;16(12):993-1006.

Hoshino A, Kim HS, Bojmar L, Gyan KE, Cioffi M, Hernandez J, ... **Raju GP,** ... Lyden D. Extracellular Vesicle and Particle Biomarkers Define Multiple Human Cancers. *Cell.* 2020 Aug 20;182(4):1044-1061.e18.

Praveen Raju, MD, PhD
Associate Professor, Neurology
Associate Professor, Pediatrics
Director, Laboratory for Pediatric Brain Tumor Research
Director, Pediatric Onco-Neurology &
Neurofibromatosis Clinic
Director, Pediatric Neurology Residency Program
Associate Director, Medical Scientist
Training Program (MSTP)

Christopher M. Sturgeon, PhD

Christopher M. Sturgeon, PhD is an Associate Professor at the Icahn School of Medicine at Mount Sinai. Chris's lab studies the development of the human hematopoietic system, using the *in vitro* differentiation of human pluripotent stem cells (hPSC) as a model system. The ability to differentiate hPSC towards a *bona fide* hematopoietic stem cell (HSC) would be a major step forward for the treatment of patients in need of a suitable donor match. Similarly, hPSCs offer unprecedented access to early embryonic hematopoietic lineages, which may have untapped clinical potential. To harness these possibilities, it is essential to be able to direct the differentiation of hPSCs in a controlled fashion. To that end, Chris's research has developed defined media approaches, coupled with staged addition of recombinant morphogens such as BMP, WNT, and RA, to recapitulate these early embryonic developmental stages.

Ongoing research interests include:

- Investigating the molecular mechanisms of hematopoietic development and the immediate precursor to the HSC, hemogenic endothelium
- Characterizing the translational potential of HSC-independent immune lineages
- Identifying the developmental trajectory of nascent mesoderm as it differentiates towards blood
- Understanding the role of RNA splicing in embryonic hematopoiesis

Christopher M. Sturgeon, PhD
Associate Professor, Cell, Developmental & Regenerative Biology
Associate Professor, Medicine

Key Publications:

Dege C, Fegan KH, Creamer JP, Berrien-Elliott MM, Luff SA, Kim D, Wagner JA, Kingsley PD, McGrath KE, Fehniger TA, Palis J, **Sturgeon CM.** Potently Cytotoxic Natural Killer Cells Initially Emerge from Erythro-Myeloid Progenitors during Mammalian Development. *Dev Cell.* 2020 Apr 20;53(2):229-239.e7.

Dege C, **Sturgeon CM.** Directed Differentiation of Primitive and Definitive Hematopoietic Progenitors from Human Pluripotent Stem Cells. *J Vis Exp.* 2017 Nov 1;(129):55196.

Fok WC, Niero ELO, Dege C, Brenner KA, **Sturgeon CM,** Batista LFZ. p53 Mediates Failure of Human Definitive Hematopoiesis in Dyskeratosis Congenita. *Stem Cell Reports.* 2017 Aug 8;9(2):409-418.

Creamer JP, Dege C, Ren Q, Ho JTK, Valentine MC, Druley TE, **Sturgeon CM.** Human definitive hematopoietic specification from pluripotent stem cells is regulated by mesodermal expression of CDX4. *Blood.* 2017 Jun 1;129(22):2988-2992.

Sturgeon CM, Ditadi A, Awong G, Kennedy M, Keller G. Wnt signaling controls the specification of definitive and primitive hematopoiesis from human pluripotent stem cells. *Nat Biotechnol.* 2014 Jun;32(6):554-61.

Nita Vangeepuram, MD, MPH

Nita Vangeepuram, MD, MPH has had years of clinical training and experience as a general pediatrician with a special interest in management of pediatric obesity and its complications. Her research focuses on pediatric obesity and diabetes prevention and treatment, community-engaged research, and health disparities/health equity research. She completed a joint fellowship in General Academic Pediatrics and Environmental Pediatrics and is an Assistant Professor in the Departments of Pediatrics and Population Health Science and Policy and Engagement Core Lead of Mount Sinai's Institute for Health Equity Research. Her recent Career Development Award (K23) and current R03 support a study using community-based participatory research, in addition to other novel methods (peer education and mobile health technologies), to develop models for the prevention of Type 2 diabetes among at-risk East Harlem youth. Her long-term career goal is to lead national efforts to prevent and treat conditions disproportionately impacting ethnic-minority youth and their families, by leveraging the assets of community-academic partnerships. This goal represents a synergy of her clinical training, community outreach and advocacy work, and public health research training. She has specific experience in collaborating with community stakeholders to develop and implement health surveys, interventions, and disease prevention programs, and in conducting quantitative and qualitative research. She has led studies to examine barriers and facilitators for recruitment and retention of diverse populations for community-based and other types of research. Dr. Vangeepuram's primary interest is to ensure that patient/family perspectives guide our research through participatory approaches and that our research is representative of the diverse populations we serve.

Key Publications:

Vangeepuram N, Angeles J, Lopez-Belin P, Arniella G, Horowitz CR. Youth Peer Led Lifestyle Modification Interventions: A Narrative Literature Review. *Eval Program Plann.* 2020 Dec;83:101871.

Mayer VL, **Vangeepuram N,** Fei K, Hanlen-Rosado EA, Arniella G, Negron R, Fox A, Lorig K, Horowitz CR. Outcomes of a Weight Loss Intervention to Prevent Diabetes Among Low-Income Residents of East Harlem, New York. *Health Educ Behav.* 2019 Dec;46(6):1073-1082.

Vangeepuram N, Williams N, Constable J, Waldman L, Lopez-Belin P, Phelps-Waldropt L, Horowitz CR. TEEN HEED: Design of a clinical-community youth diabetes prevention intervention. *Contemp Clin Trials.* 2017 Jun;57:23-28.

Vangeepuram N, Townsend K, Arniella G, Goytia C, Horowitz CR. Recruitment in Clinical Versus Community-Based Sites for a Pilot Youth Diabetes Prevention Program, East Harlem, New York, 2011-2012. *Prev Chronic Dis.* 2016 Jan 28;13:E14.

Vangeepuram N, Carmona J, Arniella G, Horowitz CR, Burnet D. Use of Focus Groups to Inform a Youth Diabetes Prevention Model. *J Nutr Educ Behav.* 2015 Nov-Dec;47(6):532-539.e1.

Nita Vangeepuram MD, MPH
Assistant Professor, Pediatrics
Assistant Professor, Environmental Medicine & Public Health
Assistant Professor, Population Health Science and Policy

Faculty Grants/Awards/Honors

Brian Brown, PhD, NIH, NIH Director's Transformative Research Award, "Development of a Platform for Spatial Functional Genomics"

Supinda Bunyavanich, MD, MPH, Paula Busse, MD, Juan Wisnivesky, MD, PhD, NIH/IAID, U01, "Childhood Asthma in Urban Settings"

Silvia De Rubeis, PhD, NIH/NICHD, R01, "Cellular and Molecular Determinants of DDX3X Syndrome"

Silvia De Rubeis, PhD, Icahn School of Medicine at Mount Sinai, 2021 Distinguished Scholar Award

Adolfo Garcia-Ocaña, PhD and Sarah A. Stanley, MBBCh, PhD, NIH/NIDDK, R01, "Neural Control of Pancreatic Endocrine Function in Obesity and Diabetes"

Magdalena Janecka, PhD, NIH/NIMH, R01, " Maternal Health in Pregnancy and Autism Risk - Genetic and Non-Genetic Mechanisms"

Amy R. Kontorovich, MD, PhD, NIH/NHLBI, R01, "Uncovering Early Signals of Hereditary Ttr Amyloidosis in Minority Populations at High Genetic Risk"

Andrew Sharp, PhD, NIH/NINDS, R03, "Investigating Tandem Repeat Expansions as a Cause of Schizophrenia"

Andrew Sharp, PhD, NIH/NIA, R03, "Investigating Tandem Repeat Variation as a Cause of Alzheimer's Disease From Exome Sequencing Data"

Trainee Grants/Awards/Honors

Marta Garcia-Forn, PhD, Fundación Alfonso Martín Escudero, Research Fellowship, "Cellular and Molecular Mechanisms of DDX3X Syndrome"

Geming Lu, MD, Einstein-Sinai Diabetes Research Center, Immuno-Technology Core Microgrant, "Single-Cell Mass Cytometry Analysis of Human Islet Cells Following Harmine and Exendin-4 Treatment"

Geming Lu, MD, Einstein-Sinai Diabetes Research Center, Pilot & Feasibility Grant, "Combination Therapy for Type 1 Diabetes"

Adele Mossa, PhD, DDX3X Foundation, Uplifting Athletes Young Investigator Draft Grant, "Cortico-Cerebellar Communication in DDX3X Syndrome"

Adele Mossa, PhD, Beatrice and Samuel A. Seaver Foundation, "The Role of Cerebellar Development in DDX3X Syndrome"

Bhavana Shewale, MBBS MS, NYSTEM, Trainee Scholar, "Role of Protein Assembly and Tension Sensing in De Novo Sarcomerogenesis"

Alejandro Martin Trujillo, PhD, NIH/NHLBI, BioData Catalyst Fellows Program, "Development of Robust and Efficient Pipelines for Identifying Tandem Repeat Expansions in Whole Genome Sequencing Data, and Its Application to Congenital Heart Defects"

Faculty Highlights

Publications

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