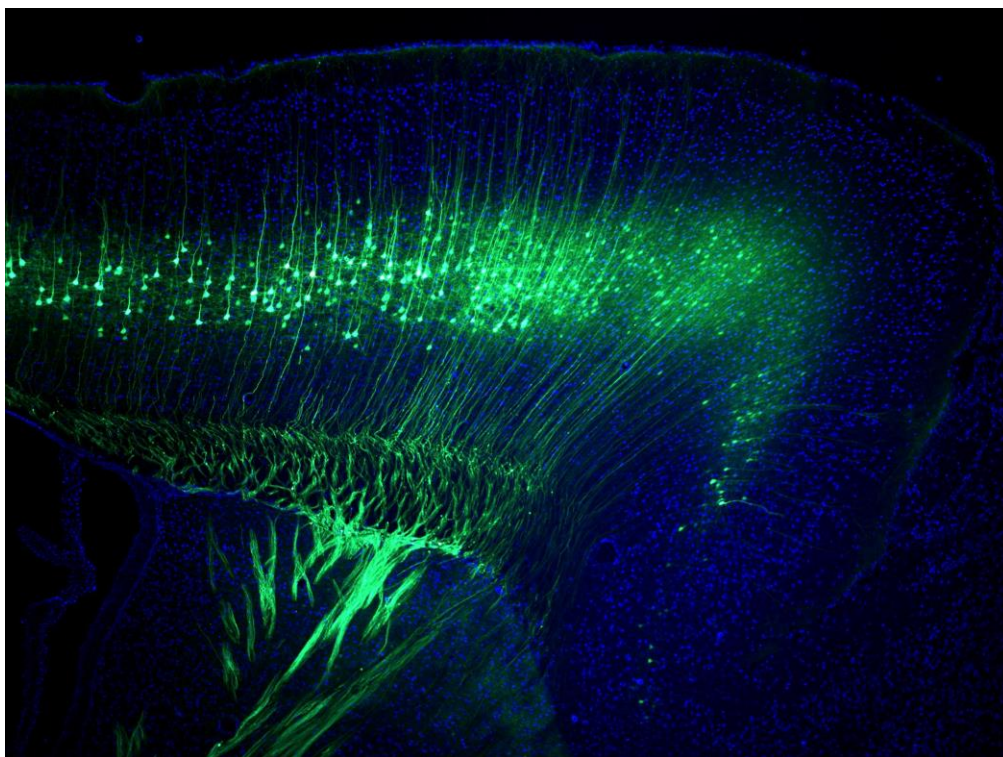


21st Annual Child Health Research Day

Sponsored by

The Jack and Lucy Clark Department of Pediatrics
The Mindich Child Health and Development Institute
The Department of Environmental Medicine and Public Health

Program and Abstracts



Corticopontine connections. Retrograde labeling of cortical neurons projecting to the pons in the mouse brain. This technique uses viral particles that are competent for retrograde transport. The viral particles are injected in the pons and transported to cortical neurons innervating the pons. (Image Courtesy of Dévina Ung, PhD, and Andrea Boitnott, De Rubeis Lab, Seaver Autism Center for Research and Treatment)

April 11, 2019
Hatch Auditorium & Guggenheim Pavilion

A Program of
The Jack and Lucy Clark Department of Pediatrics
The Mindich Child Health and Development Institute
The Department of Environmental Medicine and Public Health

Keynote Speaker:

Benjamin L. Shneider, M.D.

George Peterkin Endowed Chair, Professor of Pediatrics
and Head of Section, Pediatric Gastroenterology, Hepatology and Nutrition,
Baylor College of Medicine, Chief of Service, Pediatric Gastroenterology, Hepatology
and Nutrition, Texas Children's Hospital

Child Health Research Day

Steering Committee:

Jaime Chu, MD, Chair
Dusan Bogunovic, PhD
John Bucuvalas, MD
Silvia De Rubeis, PhD
David Dunkin, MD
Bian Lu, PhD
Lauren Petrick, PhD
Alison Sanders, PhD
Rebecca Trachtman, MD

Advisory Committee:

Bruce D. Gelb, MD
Lisa M. Satlin, MD
Karen Wilson, MD, MPH
Robert Wright, MD, MPH
Rosalind Wright, MD, MPH

Administrator: Carla Monaco

**Breakfast is courtesy of the
Dr. Howard Rappaport Memorial Lectureship Fund**

Icahn School of Medicine at Mount Sinai

21st Annual Child Health Research Day

Schedule of Events

April 11, 2019 – Hatch Auditorium

7:45-8:00 a.m.	<u>Coffee and Tea</u>
8:00-8:10 a.m.	<u>Welcome and Introduction</u> Lisa M. Satlin, MD, Chair, The Jack and Lucy Clark Department of Pediatrics
8:10-9:05 a.m.	<u>Grand Rounds: The Dr. Howard Rappaport Memorial Lecture</u> “Clinical Investigation in Pediatric Liver Diseases - Trials and Tribulations” Benjamin L. Shneider, MD George Peterkin Endowed Chair, Professor of Pediatrics and Head of Section, Pediatric Gastroenterology, Hepatology and Nutrition, Baylor College of Medicine, Chief of Service, Pediatric Gastroenterology, Hepatology and Nutrition, Texas Children’s Hospital
9:05-9:30 a.m.	<u>Breakfast</u>
9:30-11:45 a.m.	<u>Plenary Presentations – Hatch Auditorium</u> Moderators: Dusan Bogunovic, PhD and Alison Sanders, PhD
9:30-9:45 a.m.	A Randomized Control Trial to Reduce Food Allergy Anxiety about Casual Exposure by Holding the Allergen: TOUCH Study Christopher Knight, <u>Tamar Weinberger</u> , Rachel Annunziato, Eric Riklin, Eyal Shemesh, Scott Sicherer
9:45-10:00 a.m.	Zebrafish Modeling of Intestinal Injury and Therapeutics Defines in Vivo Epithelial Responses Relevant to Pediatric Inflammatory Bowel Disease <u>Joshua Morrison</u> , Joshua Morrison, Ling-shiang Chuang, Nai-yun Hsu, Philippe Ronel Labrias, Shikha Nayar, Jaime Chu, Judy H. Cho
10:00-10:15 a.m.	Exploring Sex-Specific Associations between Prenatal Air Pollution and Early Childhood Wheeze Phenotypes <u>Nicholas Zirn</u> , Alison G. Lee, Whitney J. Cowell, Rosalind J. Wright
10:15-10:30 a.m.	Bronchiolitis Hospitalization Association with Delivery Method and Birth Season <u>Lindsey C. Douglas</u> , Maya Leventer-Roberts, Ohad Levikron, Karen M. Wilson
10:30-10:45 a.m.	Autoinflammation, but Not Viral Disease, in Human TBK1 Deficiency <u>Justin Taft</u> , Diana Legarda, Sofija Buta, Iris Hollink, Adrian Ting, Dusan Bogunovic
10:45-11:00 a.m.	Early Prenatal Exposure to Endocrine Disrupting Chemicals is Associated with Child Neurodevelopment at Age Seven <u>Eva M. Tanner</u> , Carl-Gustaf Bornehag, Chris Gennings
11:00-11:15 a.m.	Correlation of Preterm Infant Salivary Cortisol Levels with Scores on the Neonatal Infant Stressor Scale <u>Shalizi Pourkaviani</u> , Xueying Zhang Emily Spear, Rebecca Satty, Madiline D’Agostino, Annemarie Stroustrup
11:15-11:30 a.m.	Maternal Prenatal Stress “Mixture” Predicts Infant Temperament in the PRISM Study: Race-Specific Effects <u>Rebecca Campbell</u> , Rebecca K. Campbell, Paul Curtin, Julie D. Flom, Michelle Bosquet Enlow, Kelly J. Brunst, Rosalind J. Wright
11:30-11:45 a.m.	An Examination of the Pediatric Precision Medicine Undiagnosed Diseases Program at Mount Sinai <u>Bryn Webb</u> , Gilad D. Evrony, Brittany Wenger, Dusan Bogunovic, Neal A.L. Cody, George A. Diaz, Bruce D. Gelb
11:45 a.m.-12:45 p.m.	<u>Poster Session and Lunch</u> Guggenheim Pavilion Atrium
12:45-1:00 p.m.	<u>Poster Presentation Awards Ceremony</u> Presented by Jaime Chu, MD

Icahn School of Medicine at Mount Sinai
Nineteenth Annual Child Health Research Day
April 11, 2019

WELCOME

We welcome you to the 21st Annual Child Health Research Day at Mount Sinai! This event aims to highlight the outstanding research activities of students, housestaff, clinical and research post-doctoral fellows, research staff, social workers, nurses and junior faculty in the Department of Pediatrics, the Mindich Child Health and Development Institute (MCHDI) and the Department of Environmental Medicine and Public Health (EMPH). The breadth of research, broadly related to the health and welfare of infants, children and adolescents, presented in today's plenary and poster sessions, exemplifies the commitment to scientific discovery and scholarship central to our academic mission. The event provides a unique opportunity for junior investigators in the Department of Pediatrics and EMPH and MCHDI to share the results of their research with colleagues, and thereby discover new applications for their work or identify potential future areas for collaboration. We thank you for attending and congratulate all the participants on their accomplishments!

Lisa M. Satlin, MD
Chair, The Jack and Lucy Clark Department of Pediatrics

Bruce D. Gelb, MD
Director, The Mindich Child Health and Development Institute

Robert O. Wright, MD, MPH
Chair, The Department of Environmental Medicine and Public Health

ORAL PRESENTATION INDEX

ABSTRACTS

PAGE #

A Randomized Control Trial to Reduce Food Allergy Anxiety about Casual Exposure by Holding the Allergen: TOUCH Study <i>Christopher Knight, Tamar Weinberger, Rachel Annunziato, Eric Riklin, Eyal Shemesh, Scott Sicherer</i>	1
Zebrafish Modeling of Intestinal Injury and Therapeutics Defines in Vivo Epithelial Responses Relevant to Pediatric Inflammatory Bowel Disease <i>Joshua Morrison, Ling-shiang Chuang, Nai-yun Hsu, Philippe Ronel Labrias, Shikha Nayar, Jaime Chu, Judy H. Cho</i>	2
Exploring Sex-Specific Associations between Prenatal Air Pollution and Early Childhood Wheeze Phenotypes <i>Nicholas Zirn, Alison G. Lee, Whitney J. Cowell, Rosalind J. Wright</i>	3
Bronchiolitis Hospitalization Association with Delivery Method and Birth Season <i>Lindsey C. Douglas, Maya Leventer-Roberts, Ohad Levikron, Karen M. Wilson</i>	4
Autoinflammation, but Not Viral Disease, in Human TBK1 Deficiency <i>Justin Taft, Diana Legarda, Sofija Buta, Iris Hollink, Adrian Ting, Dusan Bogunovic</i>	5
Early Prenatal Exposure to Endocrine Disrupting Chemicals is Associated with Child Neurodevelopment at Age Seven <i>Eva M. Tanner, Carl-Gustaf Bornehag, Chris Gennings</i>	6
Correlation of Preterm Infant Salivary Cortisol Levels with Scores on the Neonatal Infant Stressor Scale <i>Shaliz Pourkaviani, Xueying Zhang, Emily Spear, Rebecca Satty, Madiline D'Agostino, Annemarie Stroustrup</i>	7
Maternal Prenatal Stress “Mixture” Predicts Infant Temperament in the PRISM Study: Race-Specific Effects <i>Rebecca K. Campbell, Paul Curtin, Julie D. Flom, Michelle Bosquet Enlow, Kelly J. Brunst, Rosalind J. Wright</i>	8
An Examination of the Pediatric Precision Medicine Undiagnosed Diseases Program at Mount Sinai <i>Bryn D. Webb, Gilad D. Evrony, Brittany Wenger, Dusan Bogunovic, Neal A.L. Cody, George A. Diaz, Bruce D. Gelb</i>	9

POSTER GUIDE & PRESENTATION INDEX

(To view all abstracts visit <https://icahn.mssm.edu/about/departments/pediatrics/programs>)

ABSTRACTS

(In alphabetical order)

PAGE & POSTER #

Developing and Integrating a Social Determinants of Health Screener into an Outpatient Pediatric Clinic in East Harlem, NYC <i>Jennifer Acevedo, Lauren Zajac, Rebecca Rinehart, Rebecca Kann, Leora Mogilner</i>	10
A Quality Improvement Project To Increase Compliance in the Provision of Asthma Home Management Plan of Care Documentation <i>Mofolasayo Adeniyi, Cynthia Angeles, Lin Lin Kin</i>	11
Long-term Tolerance of Egg in Allergic Patients Undergoing Baked Egg Food Challenges <i>Amanda Agyemang, Jay Liberman, Anna Nowak-Węgrzyn</i>	12
Repeat Third Trimester Syphilis Screening in a Low-Risk Population <i>Monica K. Amoo-Achampong, Frank Ma, Lisa Eiland</i>	13
Disparities in Exposure to Ambient Air Neurotoxicants and Neurodevelopment in Children by Maternal Nativity Status <i>Faven Araya, Jeanette A. Stingone, Luz Claudio</i>	14
Comparison of Length of Stay Between Children Admitted to an Observation Versus Inpatient Unit <i>Jennifer Bailey, C. Anthoney Lim, Julie Oh, Erick Eiting, Ethan Cowan, Barbara Barnett, Yvette Calderon</i>	15
Chart Review to Evaluate Co-Sensitization Between Cashew and Sesame <i>Mary Grace Baker, Sarah Saf, Travis Sifers, Christopher S. Knight, Anna Nowak-Węgrzyn</i>	16
Parental Satisfaction Post Dental Treatment Under Intravenous Sedation versus General Anesthesia in Pediatric Patients <i>Michelle Baptiste, Laurence Hyacinthe</i>	17
Effectiveness of Shared Reading Program on Neurodevelopmental Outcomes and Maternal Stress in the Neonatal Intensive Care Unit (NICU) <i>Stephanie L. Bernard, Olivia Janssen, Camille Hebert, Cynthia Katz, Leora Mogilner, Jennifer B. Bragg</i>	18
Debriefing Implementation Program in the Pediatric Intensive Care Unit <i>Caroline Black, Christopher Strother, Iris Mandell, Jennifer Kero, Lauren Zinns</i>	19
Screening for Maternal Family Planning Needs During the Pediatric Well Visit: A Needs Assessment <i>Aisha A. Bobb-Semple, Megan R. D'Andrea, Natasha Ramsey, John Rowland, Leora Mogilner</i>	20
Assessing Pediatric Resident Readiness to Screen Mothers of Young Children for Family Planning Needs <i>Aisha A. Bobb-Semple, Natasha Ramsey, Megan R. D'Andrea, John Rowland, Leora Mogilner</i>	21
Changing the Prognosis; Liver Transplantation for Infantile Pyruvate Carboxylase Deficiency <i>Margo Sheck Breilyn, Brandon Stauffer, Ronen Arnon, Pankaj Prasun, George A. Diaz, and Kimihiko Oishi</i>	22
Neural Circuit and Brain System Alterations Underlying Social Recognition Memory Deficits	23

Angiogenic Profile Identifies Pulmonary Hypertension in Children with Down Syndrome <i>Douglas Bush, Csaba Galambos, Kristine Wolter-Warmerdam, Brandie D. Wagner, Dunbar Ivy, Steven H Abman, Deven McMorrow, Fran Hickey</i>	24
Piezo1 Channels Contribute to Flow Induced Intracellular Calcium Transients in Mouse Cortical Collecting Duct (CCD) <i>Rolando Carrisoza-Gaytan, Daniel Flores, Marianela Dalghi, Gerard Apodaca, Thomas Kleyman, Lisa Satlin</i>	25
Papillary Thyroid Carcinoma in a Pediatric Patient with Beta Thalassemia <i>Lauryn Choleva, Christopher Romero, Elizabeth Wallach, Mabel Yau, Jasmine Gujral, Swathi Sethuram, Robert Rapaport, Meredith Wilkes</i>	26
Identifying And Helping Patients with Food Insecurity Through a Quality Improvement Initiative <i>Liliana Cruz, Sri Nikhita Chimatapu, Ekaterina Golovko, Surkhay Bebiya, Jacqueline Maya, Anandini Suri, Rakesh Donthula, Charmin Gohel, Sebastian Proano, Lisa Forman, Daniel Ibanez, Gwen Raphan</i>	27
Optimizing Approaches to Drug Allergies in the Pediatric Inpatient Setting <i>Nessy Dahan, Roxanne C. Oriel</i>	28
Improving Rates of HPV Vaccination in the Pediatric Outpatient Setting <i>Nessy Dahan, Erina Lie, Camille Almada, Vanessa Eddy, Shane Goda, Brian Park, Amanda Reich, Ann Symonds, Van Don Williams II, Cynthia Katz</i>	29
Early-life Dentine Manganese Concentrations and Intrinsic Functional Brain Connectivity in Adolescents and Young Adults <i>Erik de Water, Demetrios Papazaharias, Claudia Ambrosi, Lorella Mascaro, Yuri Levin-Schwartz, Elza Rechtman, Giuseppa Cagna, Daniele Corbo, Roberto Gasparotti, Roberto G. Lucchini, Manuela Oppini, Donatella Placidi, Christine Austin, Manish Arora, Cheuk Y. Tang, Donald R. Smith, Robert O. Wright, Megan K. Horton</i>	30
Student Ambassador Program to Increase HPV Vaccination Coverage Rates among Adolescents <i>Grisselle DeFrank, Melanie Arnold, Martha Arden, Janet Lee</i>	31
Combined effects of BTEX and material hardship on ADHD-suggestive behaviors among a nationally-representative sample of U.S. children <i>Kayla Dellefratte, Jeanette A. Stingone, Luz Claudio</i>	32
A novel role for Mannose phosphate isomerase in the response to radiation and DNA damage <i>Charles DeRossi, Nataly Shtraizent, Joshua Morrison, Geoffrey Girnun, Jaime Chu</i>	33
Differential Transcript Usage in the Placenta is Associated with Aberrant Fetal Growth <i>Maya A. Deyssenroth, Shouneng Peng, Ke Hao, Carmen J. Marsit, Jia Chen</i>	34
Network analysis of nasal transcriptome profiles reveals causal key driver genes of severe asthma in children <i>Anh.Do, Y. Chun, J. Andrade, G. Grishina, A. Grishin, A. Vicencio, E. Schadt, S. Bunyavanich</i>	35
Characterizing the prenatal inflammatory milieu associated with maternal asthma: A proteomics approach <i>Julie D. Flom, Megan Niedzwiecki, Elena Colicino, Anu Joshi, Whitney Cowell, Alison Lee, Cecilia Berin, Rosalind J. Wright</i>	36
Tooth and hair analysis of children with kidney and hypertensive disease <i>Smita Goodman, Jeffrey Saland, Manish Arora</i>	37

Why are children drinking too much cow's milk? Identifying gaps in caregiver education. <i>Irina Gorbounova, Rachel Levantovsky, Melissa Hill, Emma Loebel, John C. Rowland, Leora Mogilner</i>	38
Implementing the Solutions for Patient Safety (SPS) Unplanned Extubations Bundle: Improved Education with the Help of RN and RT Champions <i>Glynda Guia-Johnson, Kelvin Ziegler, Ellen Black, Yong Sing Da Silva, Juliana Guiney, Kelsey Mulhern, Ali Rosenberg, Lindsey Schroy</i>	39
Undervirilized male infant with features of atypical CAH following prenatal exposure to Nystatin <i>Jasmine Gujral, Divya Khurana, Gertrude Costin, Swathi Sethuram, Christopher Romero, Lauryn Choleva, Meredith Wilkes, Elizabeth Wallach, Mabel Yau, Robert Rapaport</i>	40
Effects of Growth Hormone Stimulation on the Immunologic Cellular Landscape in Pediatric Patients <i>Jasmine Gujral, Brian A. Kidd, Hao-Chih Lee, Eddy A. Golden, Mabel Yau, Joel T. Dudley, Robert Rapaport</i>	41
Multi-omic analysis of acute response to growth hormone stimulation in pediatric patients <i>Jasmine Gujral, Loukia N. Lili, Eddy A. Golden, Mabel Yau, Brian A. Kidd, Joel T. Dudley, Robert Rapaport</i>	42
Identifying critical windows of prenatal particulate air pollution exposure and children's blood pressure at age 4-6 <i>Gleicy M. Hair, Maria José Rosa, Allan C. Just, Itai Kloog, Katherine Svensson, Maria Luisa Pizano-Zarate, Ivan Pantic, Lourdes Schnaas, Marcela Tamayo-Ortiz, Andrea A. Baccarelli, Itai Kloog, Martha M. Tellez-Rojo, Robert O. Wright, Alison P. Sanders</i>	43
Genetic testing in children with Autism Spectrum Disorder <i>Shatha Hajja, Samhita Jain, Jorge Revelo, Amanda Santos, Than Win, Bassam Al-Jomard, Larissa Polanco, Tania Lopez</i>	44
Keystones of Development Online Residency Curriculum: Weaving Attachment, Autonomy, and Executive Function into Well-Child Visits, Birth to Age 5 <i>Blair Hammond, Aliza Pressman, Mariel Benjamin, Carrie Quinn</i>	45
NYC Health Department Perinatal Hepatitis C Outreach Program Pilot <i>Emily Harrison, Alison Whitney, Perminder Khosa, Angelica Bocour, Ann Winters</i>	46
Current Use of Point-of-Care Ultrasound and Education in Pediatric Hospital Medicine Fellowship Programs in the United States <i>Emily H. Hertzberg, Valeriy Chorny</i>	47
Refined Carrier Frequencies of Citrin Deficiency in Various Populations <i>Eri Imagawa, Luca Fierro, George A. Diaz, Kimihiko Oishi</i>	48
A New Pediatric Disease Cluster: High Frequency of Chiari Malformation Associated with Tethered Cord, Mast Cell Activation Syndrome, Ehlers-Danlos Syndrome, and Postural Orthostatic Tachycardia Syndrome <i>Twana Jackson, Lincoln Ferguson, Paolo Bolognese, Anne Maitland, Warren Rosenfeld</i>	49
Lower Airway Pneumocystis jirovecii and Severe Asthma in Childhood <i>Megan N. Januska, Victoria Ribeiro, Alfin G. Vicencio</i>	50
De Novo Crohn's Disease of the Pouch in Children Undergoing Ileal Pouch-Anal Anastomosis for Ulcerative Colitis: A Tertiary Care Inflammatory Bowel Disease Center Experience <i>Lauren A. Jarchin, Elizabeth A. Spencer, Sergey Khaitov, Alexander Greenstein, Jacqueline Jossen, Joanne Lai, David Dunkin, Nanci Pittman, Keith Benkov, Marla Dubinsky</i>	51
Case report of Ovotesticular 46,XY Disorders of Sex Development: Preliminary Diagnosis and Gender	52

dysphoria in young adulthood <i>Gregory G. Jebian B., Mabel Yau, Mithra L. Narasimhan, Anne Macdonald, Lakshmi Mehta, Heino Meyer-Bahlburg, Maria I. New</i>	
Queer Medical Pipeline: A Model for Promoting Careers in Healthcare for Urban LGBTQ Youth <i>Scott K. Jelinek, Don Nguyen, Barbara Warren</i>	53
Oral Health in Pediatric Oncology Patients: A Survey Aiming at Quality Improvement <i>Shikha Kanotra, Laurence Hyacinthe</i>	54
Exploring Interventions: Managing Compassion Fatigue (CF) And Burnout (BO) In Pediatric Subspecialty Providers <i>Samuel M. Kase, Jeanie L. Gribben, Elisha D. Waldman, Andrea S. Weintraub</i>	55
The Efficacy and Stability of Food Allergy Herbal Formula 2 and Its' Extracts Over Time <i>Monica Kim, Kamal Srivastava, Nan Yang, Xiu-Min Li, David Dunkin</i>	56
Laboratory Screening for the Newly Arrived Immigrant Child <i>Cassandra Koid-Jia-Shin, Mirtha G. Gayoso-Liviac, Juan C. Fierro, Rasmita Budhathoki, Thach Nguyen, April W. J. Tan, Ian Gampon, Daniel Ibanez, Edward Forman, Joselito Sanchez, Christina Cuccia</i>	57
Physician and Parent Comfort, Awareness, Barriers, and Implementation of the Guidelines for the Prevention of Peanut Allergy <i>Michelle Lai, Scott H. Sicherer</i>	58
Effect of Glucocorticoid Therapy on Fertility in Females with Non Classical Steroid 21-hydroxylase deficiency <i>Mithra Lakshmi Narasimhan, Mabel Yau, Gregory Jebian, Anne Macdonald, Maria I New</i>	59
Integrative Analysis of Methylation Quantitative Trait Loci (meQTLs) in Human Placenta <i>Corina Lesseur, Maya Deyssenroth, Shouneng Peng, Ke Hao, Carmen J. Marsit, Jia Chen</i>	60
Prevalence of smoking cessation counseling conducted by pediatric dentists at the Institute of Family Health of Harlem <i>Wing Yin Maggie Leung, Laurence Hyacinthe, John Pfail</i>	61
Prenatal cortisol modifies the association between maternal lifetime traumatic stress and body mass index in 6 month-olds <i>Lianna R. Lipton, Kelly J. Brunst, Julie D. Flom, Farida Nentin, Nastasia Pollas, Brent A. Coull, Michelle Bosquet Enlow, Rosalind J. Wright</i>	62
Risk Factors and Behaviors of Adolescent E-Cigarette in NYC <i>Yuki Low, Elizabeth Garland</i>	63
Fluoride Exposure and Kidney and Liver Function among Adolescents in the United States: NHANES, 2013-2016 <i>Ashley J. Malin, Corina Lesseur, Stefanie A. Busgang, Paul Curtin, Robert O. Wright, Alison P. Sanders</i>	64
Type I Interferon Dysregulation in Down Syndrome <i>Louise Malle, Dusan Bogunovic</i>	65
Vertical Rotation of the Orbital Bandeau in Unilateral Coronal Synostosis <i>Ilana G. Margulies, Paymon Sanati-Mehrizy, Pedram Goel, Peter M. Som, Mark M. Urata, Peter J. Taub</i>	66
3 Cases of Peanut-related Food Protein-induced Enterocolitis Syndrome; Will Early Introduction Lead to More? <i>Joao Pedro Matias Lopes, Jacob Kattan</i>	67

Prevalence and Risk Factors of Polycystic Ovarian Syndrome Among an Ethnically Diverse Overweight/Obese Adolescent Population <i>Jacqueline Maya, Janet Siegel, Tina Q. Cheng, Thaina Rousseau-Pierre</i>	68
Unraveling the neurobiology of DDX3X Syndrome, a recently identified form of Intellectual Disability <i>Danielle Mendonca, Andrea Boitnott, Dévina Ung, Kristi Niblo, Elodie Drapeau, Silvia De Rubeis</i>	69
ASD and Symptoms of ADHD Predict Abnormal Neuronal Preparation for Motor Activity <i>Marta Migó, Sylvia Guillory, Christopher McLaughlin, Emily Isenstein, Hannah Grosman, Christina Layton, Katharine N Thakkar, Francisco X. Castellanos, Jennifer H. Foss-Feig</i>	70
Integrating Primary Care and Behavioral Health Through a Collaborative Care (CC) model: Feasibility and Outcome of a Universal Screening <i>Maya Mihic, Thaina Rousseau-Pierre</i>	71
Crouzon Syndrome with Acanthosis Nigricans (CAN) without Craniosynostosis <i>Cassie S. Mintz, Ayuko Iverson, Saadi Ghatan, Aldo Londino, Puneet Pawha, Ethylin Jabs, Lakshmi Mehta</i>	72
Teaching Social Determinants via Service Learning and Community Exploration <i>Leora Mogilner, Ashley Stevens, Cynthia Katz</i>	73
Diagnostic Accuracy of Fetal Echocardiography in Major Congenital Heart Disease <i>Namrita Mozumdar, John Rowland, Hari Rajagopal, Miwa Geiger, Shubhika Srivastava, Kenan Stern</i>	74
Defects in RNA Polymerase II pausing and elongation control results in severe hemolytic anemia in the EKLF/Klf1 Nan mutant <i>Kaustav Mukherjee, Chrysoula Deligianni, James J. Bieker</i>	75
Assessment of Metal Exposures in Deciduous Teeth of Patients with Inflammatory Bowel Disease <i>Nilendra Nair, Christine Austin, Manel Rocha, Catarina Gouveia, Paul Curtin, Caroline Eisele, Jean-Frédéric Colombel, Joana Torres, Inga Peter, Manish Arora</i>	76
Investigating environmental exposure combinations and early age at menarche among U.S. adolescent girls <i>Sabine Oskar, Mary S. Wolff, Susan L. Teitelbaum, Jeanette A. Stingone</i>	77
Point-of-Care Ultrasound Diagnosis of Small Bowel-Small-Bowel vs Ileocolic Intussusception <i>Brian L. Park, Joni E. Rabiner, James W. Tsung</i>	78
Development of an Adolescent-Friendly Pregnancy & Parenting Curriculum at a Teaching and Referral Hospital in Western Kenya <i>Natasha Ramsey, Priscah Onkware, Maureen Chepkoech, Vincent Kibet, Julie Thorne</i>	79
Epidemiology and Outcomes of Emergency Department Visits for Hypersensitivity in Infants at a Tertiary Care Center <i>Nicole Ramsey, Hsi-en Ho, Julie Wang</i>	80
Sex-specific associations between exposure to multiple metals and visuospatial memory skills in children <i>Elza Rechtman, Demetrios Papazaharias, Stefano Renzetti, Giuseppa Cagna, Elena Colicino, Danielle Hazeltine, Marco Peli, Donatella Placidi, Donald R Smith, Robert O. Wright, Roberto G Lucchini, Megan Horton</i>	81
Generation and Initial Characterization of 3D Cortical Collecting Ducts (CCDs)-on-a-Chip <i>Joshua L. Rein, Szilvia Heja, Daniel Flores, Rolando Carrasquillo-Gaytan, Neil Lin, Kimberly A. Homan, Jennifer</i>	82

Dissecting de novo indel biology with long-read technology <i>Felix Richter, Allison H. Seiden, Nihir Patel, Andrew P. Sharp, Bruce D. Gelb</i>	83
Investigating the etiology of multifocal atrial tachycardia in Costello syndrome via iPSC disease modeling <i>Nelson A. Rodriguez, Rafael Dariolli, Nihir Patel, Jingqi Gong, Tara Keshavarz-Shirazi, Rebecca Josowitz, Simon Ng, Xianming Lin, Glenn Fishman, Eric Sobie, Bruce D. Gelb</i>	84
Implementation of an Asynchronous Curriculum to Replace Traditional Didactics: Initial Observations and Challenges <i>Priya S. Rolfes, Carolyn Rosen, John Rowland, Leora Mogilner</i>	85
The Stability and Anti-inflammatory Effects of Berberine/Heparin/Chitosan Nanoparticles <i>Mollie Shear, Kamal Srivastava, Nan Yang, Xiu-Min Li, David Dunkin</i>	86
Precocious puberty in a female with a suprasellar dermoid cyst and multiple pituitary hormone deficiencies <i>Swathi Sethuram, Meredith Wilkes, Jasmine Gujral, Lauryn Choleva, Mabel Yau, Elizabeth Wallach, Christopher J. Romero, Robert Rapaport</i>	87
Improving maternal postpartum depression screening: A Primary Care Clinic QI project. <i>Alice Shajan, Farhan Zakri, Mostafa Khalid, Barbara Porrello, Diego Craik, Parvathy Krishnan, Xiang Le Ng, Ananya Manchikalapati, Mohamed Ahmed, Iya Chikvashvili, Felicia Fojas, Pilar Gonzalez</i>	88
Vision Screening in a Primary Care Clinic: a Quality Improvement Initiative to Improve Early Detection of Amblyopia <i>Alice Shajan, Gwen Raphan</i>	89
Quality Improvement Intervention in a Pediatric Emergency Department to Improve Culture Turnaround Time <i>Alice Shajan, Horton Lee</i>	90
Towards Disease in Interception: Contribution of Polygenic Risk Scores Towards Age-Dependent Risk of IBD in a High-Risk Population <i>Elizabeth A. Spencer, Kyle Gettler, Drew Helmus, Shannon Telesco, Amy Hart, Marla C. Dubinsky, Judy Cho</i>	91
Proteomic Markers in Pediatric IBD <i>Elizabeth A. Spencer</i>	92
In vitro comparison of the temperatures of refrigerated, room temperature and warmed formula at point of delivery into the newborn <i>Jeffrey V. Suell, Liandra Presser, Sonali Tatapudy, Robert Green</i>	93
Identification of Outcome Measures for Clinical Trials in Rare Diseases Associated with Neurodevelopmental Disorders <i>Lara Tang, Mikaela Rowe, Stacey Lurie, Ivy Giserman-Kiss, Jessica Zweifach, Danielle Halpern, Pilar Trelles, Jennifer Foss-Feig, Maureen Mulhern, Alexander Kolevzon, Paige M. Siper</i>	94
Home Visiting Program for NICU Graduates: Feasibility and Potential for Impact <i>Candace Tannis, Katrina Leung, John Rowland, Anna Cushing, Maureen Braun, Karen Wilson, Elaine Lin</i>	95
Comparison of Body Measures, Lifestyle Behaviors, and Behavior Change Determinants in Male and Female Adolescents in a Pilot Diabetes Prevention Study <i>Nita Vangeepuram, Candace Tannis, Tanya Braune, John Rowland, Carol R. Horowitz</i>	96
One-year morbidity and mortality of infants diagnosed with birth asphyxia (BA) or low birth weight (LBW) admitted to the Newborn Unit (NBU) at Moi Teaching and Referral Hospital in Eldoret, Kenya: a prospective	97

cohort study

Carolyn Wagner, Laura Ruhl, Winston Nyandiko, Eric Ngetich, Vincent Kibet, Astrid Christoffersen-Deb, Julia Songok

Developing and Validating an Augmented Reality Smartphone Application for Weight Estimation and Dosing in Children **98**

Temima Waltuch, Kevin Munjal, George Loo, Czer Anthoney Lim

Dental Caries in Pediatric Dental Patients Treated with Dental Rehabilitation under General Anesthesia or Intravenous Sedation at The Mount Sinai Hospital **99**

Joshua Woodruff, Laurence Hyacinthe

Ankylosis of the Temporomandibular Joint in Pediatric Patients: A Systematic Review and Analysis of 227 Joints **100**

Hope Xu, Collin Rozanski, Kasey Wood, Paymon Sanati-Mehrizy, Peter J. Taub

Dexmedetomidine For Postoperative Sedation Following Stereotactic Lead Placement: A Case Report **101**

Hope Xu, Anthony Bui, Maite La Vega-Talbott, Sheemon P. Zackai

Internal Cranial Expansion for Management of Refractory Idiopathic Intracranial Hypertension: A Case Report **102**

Hope Xu, Collin Rozanski, Peter J. Taub

The association of prenatal exposure to intensive traffic with neurobehavioral development as reflected by the NICU Network Neurobehavioral Scale (NNNS). **103**

Xueying Zhang, Emily Spear, Chris Gennings, Paul C. Curtin, Allan C. Just, Robert O. Wright, Annemarie Stroustrup

Perceptual Processing and Prediction Error Bias in Autism Spectrum Disorder **104**

Yian Zhang, Sylvia Guillory, Jennifer H. Foss-Feig

POSTER GUIDE & PRESENTATION INDEX

ABSTRACTS

(In order by affiliation)

PAGE & POSTER #

Adolescent Medicine	31, 68, 71, 79
Allergy & Immunology	1, 12, 16, 28, 35, 58, 65, 67, 80
Cardiology	74
Cell, Developmental, & Regenerative Biology	75
Child & Adolescent Psychiatry	23, 69, 70, 94, 104
Critical Care Medicine	3, 39, 101
Dental Medicine	17, 54, 61, 99
Diversity & Inclusion	53
Emergency Medicine	15, 19, 78, 90, 98
Endocrinology & Diabetes	26, 40, 41, 42, 52, 59, 87
Environmental Medicine & Public Health	3, 6, 8, 10, 14, 30, 32, 34, 36, 43, 60, 62, 63, 64, 76, 77, 81, 95, 103
Gastroenterology & Hepatology	33, 51, 56, 76, 86, 91, 92
General Pediatrics	4, 11, 20, 21, 27, 29, 38, 44, 45, 47, 49, 57, 73, 85, 88, 89, 95, 96
Genetics & Genomic Sciences	2, 9, 22, 35, 42, 48, 60, 72, 76, 83, 84
Global Health	79
Infectious Diseases	46
The Mindich Child Health & Development Institute	2, 5, 9, 23, 30, 33, 34, 35, 56, 60, 64, 65, 69, 75, 82, 83, 84, 86, 103
Medical Education	10
Microbiology	5
Nephrology	25, 37, 43, 64, 82
Newborn Medicine	7, 13, 18, 55, 93, 97
Pulmonology	3, 24, 50
Radiology	66
Surgery	66, 100, 102

A Randomized Control Trial to Reduce Food Allergy Anxiety about Casual Exposure by Holding the Allergen: TOUCH Study

Authors Names: Christopher Knight, Tamar Weinberger, Rachel Annunziato, Eric Riklin, Eyal Shemesh, Scott Sicherer

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Patients with nut allergies often fear casual exposure to allergens, which is unlikely to cause a severe reaction. The study assessed if a behavioral exposure intervention, whereby patients were guided to touch their allergen, along with education, results in a reduction of patient and parent worry about casual exposure compared with education alone.

Hypothesis: We hypothesized that this behavioral intervention would result in a reduction of patient and parent worry about casual exposure compared with education alone.

Methods: Enrollment criteria included patients aged 9 to 17.5 years with peanut or tree nut allergy who endorsed worry about casual exposure to their allergen. Participants were randomized to the touch condition plus education about the risks of casual exposure (n=30, intervention) or to receive education only (n=30, controls). The primary outcome was the difference in patient-reported worry from pre- to immediately post-intervention between the conditions. Secondary outcomes included improvement in patient- and parent-reported worry within groups and improvement in quality of life (QoL) one month after study participation.

Results: There was no greater improvement in patient worry in the intervention group compared to the control group from pre- to immediately post-intervention ($P=0.12$). Rather, both groups experienced a statistically significant decrease in patient- and parent-reported worry ($P<0.001$). Both study arms had improvement in QoL one month after the visit (P -value 0.03 and 0.01, respectively), but the intervention was not superior to the control ($P=0.76$).

Conclusion: Supervised contact with the nut is not superior to education alone. Education alone about the risks of casual exposure can decrease worry and improve QoL, an effect that was sustained even one month after the visit.

Zebrafish Modeling of Intestinal Injury and Therapeutics Defines *in vivo* Epithelial Responses Relevant to Pediatric Inflammatory Bowel Disease

Author Names: Joshua Morrison*, Ling-shiang Chuang*, Nai-yun Hsu, Philippe Ronel Labrias, Shikha Nayar, Jaime Chu, Judy H Cho

Department: Genetics and Genomic Sciences

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute; The Charles Bronfman Institute for Personalized Medicine

Introduction: The incidence of pediatric inflammatory bowel disease (IBD) has markedly increased over recent decades and highlights the need for novel and improved therapeutics. There is a critical demand for new high-throughput *in vivo* systems to rapidly evaluate emerging therapeutic agents and their efficacy.

Hypothesis: A dextran sodium sulfate (DSS)-induced intestinal injury model in zebrafish would enable quicker and higher-throughput screening of IBD therapeutic agents and their *in vivo* effects.

Methods: One- and three-course DSS treatments were used to induce intestinal injury in zebrafish larvae. In live fish, we used neutral red and CytoID staining to quantify lysosomes and autophagosomes, respectively, to investigate intestinal epithelial barrier function. Zebrafish were treated with lipid mediator and IBD therapeutic agents Prostaglandin E2 (PGE2), Mesalamine (5-ASA), and Mercaptopurine (6-MP) in combination with intestinal injury.

Results: Neutral red and CytoID largely stain specialized lysosome-rich enterocytes (LREs) in the intestine. Single treatments of DSS are able to induce intestinal inflammation with minimal extra-intestinal inflammation. LREs are able to recover after single DSS treatments, but the ability to recover is lost after three DSS treatments. 5-ASA shows a dose-dependent prevention of DSS injury, while, in contrast, PGE2 and 5-ASA are statistically effective only after the removal of DSS and during recovery of injury.

Conclusions: DSS-induced injury in zebrafish induces targeted and acute intestinal inflammation and can serve as a model for both acute and chronic injury. Our findings establish the zebrafish DSS-model as a powerful tool to rapidly test both temporal and dose-dependent effectiveness of new and current IBD therapeutics.

Exploring Sex-Specific Associations between Prenatal Air Pollution and Early Childhood Wheeze Phenotypes

Author Names: Nicholas Zirn, Alison G. Lee, Whitney J. Cowell, Rosalind J. Wright

Department: Environmental Medicine and Public Health, Pediatrics

Division: Pulmonary, Critical Care and Sleep Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Prior studies have linked exposure to outdoor particulate air pollution in mid-gestation (16-25 weeks) and asthma development in school-aged children. No prior study has examined associations between ($PM_{2.5}$) and early childhood wheeze trajectories accounting for sex-specific effects.

Hypothesis: Children exposed to increased prenatal ambient air pollution will have an increased risk of wheezing respiratory illness by age 36 months. Sex-specific associations were explored.

Methods: Leveraging an ethnically-mixed, prospective pregnancy cohort, we examined time-varying associations between daily prenatal $PM_{2.5}$ and childhood wheeze trajectories (N=1224). Group-based trajectory modeling was used to empirically estimate distinct temporal patterns of wheeze. We employed distributed lag models (DLMs) to identify vulnerable windows of exposure to prenatal $PM_{2.5}$, adjusting for child sex, ethnicity, maternal age, education and wheeze trajectory probability.

Results: Longitudinal trajectory modeling identified two wheeze phenotypes: Never/Infrequent wheeze (N=1011, 83%) vs Early repeated wheeze (N=213, 17%). In all children, DLMs identified a statistically significant sensitive window of $PM_{2.5}$ exposure at 26-34 weeks gestation with increased risk of early repeated wheeze (Figure 1a). In sex-stratified analyses, this pattern was evident only in girls (Figure 1b).

Conclusions: Children born to mothers with higher prenatal $PM_{2.5}$ exposure in later pregnancy (26-34 weeks) were more likely to have 2 or more episodes of wheeze by age 36 months. Girls were particularly impacted. These data suggest that effects of *in utero* $PM_{2.5}$ exposure on early wheeze phenotypes vs. asthma may be operating through different mechanisms given variable sensitive windows for exposure effects and observed sex differences emerging in the literature.

Bronchiolitis Hospitalization Association with Delivery Method and Birth Season

Author Names: Lindsey C. Douglas, Maya Leventer-Roberts, Ohad Levikron, Karen M. Wilson

Department: Pediatrics

Division: General Pediatrics, Hospital Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institution Affiliation: Clalit Research Institute

Introduction: Caesarian section birth reduces exposure to the maternal vaginal microbiome causing impaired immunity. Impaired immunity has been implicated as one of the causes of infant bronchiolitis.

Hypothesis: We hypothesized an association between Caesarian birth and hospitalization for bronchiolitis.

Methods: This was a retrospective cohort study of all infants born from 2008-2010 utilizing the Clalit Health Services Database that covers over 50% of the Israeli population. We extracted demographic and clinical characteristics at birth and bronchiolitis admissions in the first two years of life. A multivariable logistic regression model was generated with birth characteristics as predictors and bronchiolitis admission as the primary outcome. We repeated the analysis using machine learning gradient boosted decision trees.

Results: There were 124,526 infants born the time period studied and 5,102 (4.1%) were admitted for bronchiolitis. Our model found an interaction between birth season and delivery method. In a stratified model by birth season, the odds ratio (OR) for bronchiolitis hospitalization after scheduled Caesarian section as compared to spontaneous vaginal delivery were 1.28 (97.5% Confidence Interval (CI): 1.11-1.48), 1.40 (97.5% CI: 1.15-1.71), 1.46 (97.5% CI: 1.25-1.70), and 1.14 (97.55% CI: 1.00-1.29), for winter, spring, summer, and fall birth dates, respectively. Analysis with gradient boosted decision trees confirmed these findings and multiple features derived from birth month emerged as important predictors.

Conclusions: When compared to spontaneous vaginal delivery, scheduled Caesarian section birth is associated with an increased odds of bronchiolitis admission in the first two years of life, and the effect size differs by birth season.

Autoinflammation, but Not Viral Disease, in Human TBK1 Deficiency

Author Name(s): Justin Taft¹, Diana Legarda², Sofija Buta¹, Iris Hollink⁵, Adrian Ting², & Dusan Bogunovic^{1,3,4}

Departments: ¹Department of Microbiology, ³Department of Pediatrics, ⁵Department of Clinical Genetics,

Institution Affiliation: Icahn School of Medicine at Mount Sinai, Erasmus Medical Center

Institute Affiliation: ²Immunology Institute, ⁴Mindich Child Health and Development Institute

Introduction: TBK1 is ubiquitously expressed and involved in multiple cellular pathways, including the regulation of IFN-I, NF- κ B, and RIPK1-dependent apoptosis (RDA). In mice, biallelic loss of TBK1 is lethal by E14.5. Despite this murine lethality, two humans in their mid-20s were discovered with a homozygous loss-of-function mutation in TBK1. A brother (P1) and sister (P2) from a consanguineous family, both suffer from chronic autoinflammation, and multiple sclerosis in the case of P1.

Hypothesis: In the absence of TBK1, a hypomorphic ability to activate innate immunity protects these individuals from viral infection, while unchecked RDA drives autoinflammation.

Methods: We used a combination of molecular biology techniques coupled with CyTOF.

Results: Unexpectedly, neither sibling has shown hypersusceptibility to viral infection, nor was any detected in patient-derived dermal fibroblasts. To explain this, we show that while the TLR3 signaling axis is destroyed, cytosolic PRRs MDA5, RIG-I, and cGAS retain a hypomorphic ability to signal in the absence of TBK1. Immune cell profiling via CyTOF identified expanded, active CD4 and CD8 T cell populations that indicate self-recognition and implicate the negative regulatory function of TBK1 on RDA as possible fuel for the autoinflammation. Fibroblasts from P1 and P2 exhibit caspase 3 activation at baseline and were markedly more sensitive to TNF α -induced RDA than cells from healthy controls.

Conclusions: These findings accentuate the limitations of non-human models, suggest a compensatory mode of innate immune activation in the absence of TBK1, highlight the extreme sensitivity of the IFN-I response, and link RDA to chronic autoinflammation.

Early Prenatal Exposure to Endocrine Disrupting Chemicals is Associated with Child Neurodevelopment at Age Seven

Author Name(s): Eva M. Tanner¹, Carl-Gustaf Bornehag^{1,2}, Chris Gennings¹

Department: Environmental Medicine & Public Health

Division: Biostatistics

Institution Affiliation: ¹Icahn School of Medicine at Mount Sinai, ²Karlstad University, Karlstad, Sweden

Institute Affiliation: N/A

Introduction: Endocrine disrupting chemicals (EDCs) are xenobiotics capable of interfering with hormone action. Prior studies link numerous EDCs, such as bisphenols, to adverse neurodevelopmental outcomes, however most assessed one chemical at a time. Our objective was to evaluate the overall EDC mixture impact, and identify bad actors.

Hypothesis: Prenatal EDC exposure is associated with lower levels of neurodevelopment among school-age children.

Methods: Among 770 mother-child pairs from the Swedish SELMA study, we used Weighted Quantile Sum (WQS) regression to assess the association between 41 EDCs measured in 1st trimester urine and blood, with Wechsler Intelligence Scale for Children (IV) scores administered at age seven. Linear models were adjusted for sex, gestational age, mother's education, IQ, smoking, and weight.

Results: An inter-quartile-range change in the WQS index was nonsignificantly associated with a 0.48-point decline in full scale IQ scores among all children (90% CI=-1.54, 0.46), and a significant 1.4-point decline among boys (90% CI=-1.82, -0.22). Processing speed and working memory were the most impacted subdomains, particularly for boys. Bisphenol A and F (BPA, BPF), triclosan, and chlorpyrifos were consistently identified as bad actors, with BPF contributing the largest weights to the WQS index.

Conclusions: Early prenatal exposure to EDCs is related to cognitive deficits at age 7, with cognitive profiles similar to those observed in autism spectrum and attention-deficit disorders. The BPA replacement analogue, BPF, may not be any safer for children. Future studies are needed to confirm negative impacts of replacement analogues and further evaluate EDC exposure related cognitive profiles.

Correlation of Preterm Infant Salivary Cortisol Levels with Scores on the Neonatal Infant Stressor Scale

Author Names: Shaliz Pourkaviani, Xueying Zhang Emily Spear, Rebecca Satty, Madiline D'Agostino, Annemarie Stroustrup

Department: Pediatrics

Division: Newborn Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

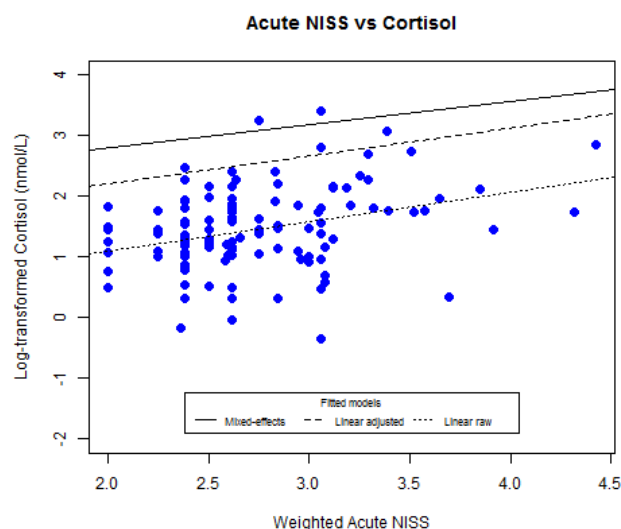
Introduction: Repeated stress during the early period of infant development is hypothesized to produce long-lasting effects on cognitive, behavioral, and somatic development. Despite efforts to nurture preterm infant development during the life-saving birth hospitalization, the neonatal intensive care unit (NICU) is a stressful environment. Variation in cortisol levels reflects multiple stressors during the neonatal period, potentially impacting development of immature hypothalamic, pituitary and adrenal axes. The Neonatal Infant Stressor Scale (NISS) is one tool used to quantify neonatal stress for clinical and research purposes.

Hypothesis: We hypothesized that our results suggest that the Neonatal Infant Stressor Scale may provide an accurate noninvasive measure of neonatal stress.

Methods: In this study, we compared acute and chronic NISS scores to an accepted biomarker of infant stress response, salivary cortisol, in a cohort premature infants born 28-0/7 - 32- 6/7 weeks gestation through the course of the NICU hospitalization.

Results: We examined 143 salivary specimens from 68 patients. Using a Pearson correlation analysis and mixed-effects model we concluded that both acute and chronic NISS scores were significantly correlated with salivary cortisol (P-values <0.001). The mixed-effects model with random coefficients for infant and family revealed significant association between the weighted acute NISS score and salivary cortisol levels (adjusted $\beta = 0.41$, P-value= 0.02).

Conclusions: Our results suggest that the Neonatal Infant Stressor Scale may provide an accurate noninvasive measure of neonatal stress. NISS scores may be used to monitor and reduce stress levels for premature infants.



The plots show the NISS (x) and log-transformed cortisol concentrations (y). The three lines show three fitted models. The 'Linear raw' is the unadjusted linear regression model, 'Linear adjusted' is adjusted for PMA, and 'Mixed-effects' is adjusted for PMA as well as the random effect of subjects/families

Maternal prenatal stress “mixture” predicts infant temperament in the PRISM study: Race-specific effects

Author Name(s): Rebecca K. Campbell, Paul Curtin, Julie D. Flom, Michelle Bosquet Enlow, Kelly J. Brunst, Rosalind J. Wright

Department(s): Pediatrics (RKC, JF, RJW); Environmental Medicine and Public Health (PC, RJW);

Institution Affiliation(s) (if not ISSM): Department of Psychiatry, Boston Children’s Hospital (MBE); Department of Psychiatry, Harvard Medical School (MBE); Department of Environmental Health, University of Cincinnati College of Medicine (KB)

Institute Affiliation: Institute for Exposomic Research

Introduction: Prenatal stress and psychological dysfunction are linked with fetal neurodevelopment. Effects of specific stressor type (lifetime trauma vs. current negative life events, NLEs) and psychological dysfunction (depressive vs. posttraumatic stress symptoms) may vary by vulnerable subgroups.

Hypothesis: Considering stress as a complex mixture can elucidate the relative impact of specific stressor types and psychological symptoms on infant negative affectivity (NA) by race/ethnicity.

Methods: Pregnant women (N=535) completed the Life Stressor Checklist-Revised, Crisis in Families System survey, Edinburgh Postnatal Depression Scale, and PTSD Checklist-Civilian version. NA in 6-month-olds was assessed with the Infant Behavior Questionnaire-Revised. First, linear regression was used to examine associations between each stress measure and infant NA. Weighted Quantile Sum (WQS) regression was then used to model effects of a composite stress “mixture” of the four stress domains on NA and identify which domain(s) had the strongest association with NA in the full sample and in race-stratified models.

Results: Higher stress and psychological dysfunction were associated with increased NA in linear regressions adjusted for maternal race/ethnicity, education and child sex. WQS regression revealed PTSD, depression and trauma contributed similarly to the association with NA in the sample overall (WQS weights: 0.35, 0.35 and 0.29, respectively). In Blacks, depression accounted for 50% of the association with NA; in Hispanics, PTSD explained 66% of the association, and PTSD, NLEs and trauma each contributed ~30% in whites.

Conclusions: Understanding racial/ethnic differences in the psychological sequelae associated with chronic stress may better elucidate the influence of stress on maternal-fetal programming of neurodevelopment.

An Examination of the Pediatric Precision Medicine Undiagnosed Diseases Program at Mount Sinai

Author Names: Bryn D. Webb, Gilad D. Evrony, Brittany Wenger, Dusan Bogunovic, Neal A.L. Cody, George A. Diaz, Bruce D. Gelb

Departments: Pediatrics, Genetics & Genomic Sciences

Division: N/A

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: The Undiagnosed Diseases Program (UDP) aims to select pediatric patients at Mount Sinai who suffer from diseases of unknown etiology and provide additional investigation with the aim to identify an underlying genetic etiology for the patient's rare symptoms.

Hypothesis: The use of next generation sequencing methodologies will establish a diagnosis in a meaningful proportion of undiagnosed disease cases.

Methods: Stepwise investigation includes detailed chart review, state-of-the-art molecular studies, and return of results, with appropriate genetic counseling, to the participating families and referring physicians. >80 cases have been referred to the UDP (of which ~75% were deemed appropriate referrals), and 34 families have enrolled. Molecular studies employed include whole exome sequencing, whole genome sequencing, and other 'omic' methodologies. In some cases, functional studies have been arranged to further investigate suspected pathogenic variants.

Results: The UDP has established a genetic diagnosis in 27% of cases, in line with the expected percentage positive rate using these technologies. Furthermore, in 43% of solved cases, additional treatment options were identified based on establishing the molecular diagnosis. Ultra-rare and novel findings have also been found including identifying the sixth reported family in the world with a severe neurodevelopmental disorder caused by recessive variants in *DPH1* (in press) and identifying a novel disease caused by biallelic mutations in *MADD* presenting with severe developmental delay, hypopituitarism, and pancreatic insufficiency, which we are now collaborating with an international consortium to report.

Conclusions: The MCHDI UDP has been successful in establishing molecular diagnoses in patients with undiagnosed disease.

Developing and Integrating a Social Determinants of Health Screener into an Outpatient Pediatric Clinic in East Harlem, NYC

Author Names: Jennifer Acevedo¹, Lauren Zajac¹, Rebecca Rinehart², Rebecca Kann³, Leora Mogilner⁴

Departments: 1-Environmental Medicine and Public Health, 2-Medical Education, 4-Pediatrics

Institution Affiliations: Icahn School of Medicine at Mount Sinai, 3-Barnard College

Introduction: Screening for and addressing social determinants of health (SDH) are critical to providing effective pediatric primary care. Optimal SDH screening must consider the specific needs of the population and the resources available, while easily integrating into clinic workflow.

Hypothesis: An SDH screener in an urban resident continuity practice in East Harlem will identify common SDH and provide effective resources and referrals for families.

Methods: We designed an electronic SDH screening tool for 8 social needs based on literature review, and input from focus groups with parents and healthcare providers. Trained personnel used a secure online platform to screen a convenience sample of caregivers of patients 0-21 years presenting for care. Caregivers who screened positive for social needs were given referrals to community-based organizations and/or educational materials. Descriptive statistics were generated using SAS.

Results: 437 caregivers were screened over a 9-month period. 59% of caretakers screened positive for at least one social need, and an average of 1.7 unmet needs were identified per family. Home environmental concerns (pests or mold), were the most common issues cited (37%), followed by tobacco smoke exposure (27%), and food insecurity (17%). The majority of caretakers with an identified social need accepted at least one referral (79%).

Conclusions: Determining how to best assess SDH in clinical practice is critical, particularly in low resource settings. A new SDH screening tool was easy to use and identified a high prevalence of social needs. A majority of patients accepted referrals for resources, and future research will explore factors associated with successful referral completion.

A Quality Improvement Project to Increase Compliance in the Provision of Asthma Home Management Plan of Care Documentation

Author Name(s): Mofolasayo Adeniyi, Cynthia Angeles, Lin Lin Kin

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai; Bronx Care Health System

Introduction: Asthma is a major cause of morbidity and hospitalization, with a prevalence of 12.5% in children under the age of 18. The Joint Commission (JC) and the Children's Asthma Care (CAC) advisory committee recommend that pediatric inpatient asthma management should include the provision of the Home Management Plan of Care (HMPC) document upon discharge.

Objective: To increase the compliance of provision of the HMPC in the pediatric inpatient unit.

Methods: An internal benchmark of 90% compliance was set. Quarterly data collection and analysis were done from 2009 -2018. Interventions initially focused on resident education and sending reminders periodically. In 2016, an EMR hard stop was introduced, requiring the presence of HMPC document before an electronic discharge order could be placed. Hard stops were also placed to ensure that all HMPC document fields were completed before the document could be saved. HMPC was considered complete if it included all five sections: dose, method and timing of rescue medications, use of controllers and follow-up appointment information.

Results: A total of 16 PDSA cycles were completed. Initial compliance in HMPC documentation had increased from 59% to 72% between 2009 and 2014 secondary to behavior-based interventions. Compliance increased to 96% in 2018, after introduction of EMR modification.

Conclusions: Resident training increased compliance rates of HMPC documentation, however utilization of EMR modifications with hard stops further yielded sustained increase. EMR based interventions may result in sustained improvement in clinical practice among health workers, compared to education and behavior-based interventions alone.

Long-term Tolerance of Egg in Allergic Patients Undergoing Baked Egg Food Challenges

Authors names: Amanda Agyemang, Jay Liberman, Anna Nowak-Węgrzyn

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Jaffe Food Allergy Institute

Introduction: Baked egg (BE) is tolerated by approximately 70% of egg- allergic children. In this study, we follow-up patients who tolerated baked egg oral food challenges (BE-OFC) and characterize their tolerance to non-baked, lightly cooked egg.

Methods: We performed retrospective chart reviews of egg allergic patients who visited Mount Sinai pediatric allergy clinic from January 2008 to December 2017 and passed BE-OFCs to BE muffin.

Hypothesis: We hypothesize that most children will progress to lightly cooked egg tolerance with minimal adverse effects during home introduction.

Results: Among 101 patients, 65% were male with a median age of 4.9 years (IQR: 2.9-3.1) at the time of the BE-OFC. 67% tolerated the baked egg challenge. 44 (85%) continued to tolerate baked egg products by the first follow up appointment (median: 9 months; IQR: 6-16) and subsequently 1 year after passing BE-OFC. Eight patients (17%) discontinued BE within 1 year, due to symptoms with ingestion at home (50%) or fear of home introduction (50%). Symptoms reported by patients with home introduction were mostly mild, such as oral itching or abdominal pain, however there was one report of anaphylaxis. 80% of subjects reporting symptoms with home introduction did not continue BE. Thirty-six patients (82%) eating BE ultimately progressed to tolerating non-baked egg over a median of 14 months (IQR: 9-23).

Conclusions: The majority of subjects passing BE-OFC added BE to regular diet at home. Tolerance is also sustained long-term in patients who ingested baked egg products.

Repeat Third Trimester Syphilis Screening in a Low-Risk Population

Author Name(s): Amoo-Achampong, Monica K.¹; Ma, Frank²; Eiland, Lisa²

Department: Pediatrics

Division: Newborn Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai¹; Mount Sinai West²

Introduction: Congenital syphilis occurs when *Treponema* spirochetes are transmitted from a pregnant woman to her fetus. In NYC, previously stable rates of infection are rising in women ages 15-44. NYS law requires screening of pregnant women at the 1st prenatal visit and at delivery. Studies show universal 3rd trimester syphilis screening is cost effective in communities with seroconversion rates $\geq 0.017\%$.

Hypothesis: Even in a low-risk population, repeat intrapartum screening for syphilis enhances the detection of maternal and congenital syphilis.

Methods: Data from patients delivering at Mount Sinai West (MSW) from June 2017-June 2018 were reviewed. Serologic RPR results at initial prenatal visit were compared to L&D admission results. Patients were screened with RPR followed by confirmatory fluorescent treponemal antibody absorption (FTA-ABS), if RPR was reactive. Primary outcome was incidence of seroconversion. Secondary outcomes included reactive RPR on admission, false positive RPR on admission and detection of congenital syphilis.

Results: From June 2017-June 2018, 4239 pregnant women were admitted to MSW for delivery. On admission, 14 women had reactive RPR results: 10 women had falsely positive RPR, 4 had reactive FTA-ABS with one woman seroconverting during pregnancy and the other 3 women had a history of syphilis prior to pregnancy. Seroconversion rate was 0.027%.

Conclusions: Our study identified a seroconversion rate of 0.027% in a presumed low-risk population, confirming benefits of repeat syphilis screening in pregnancy. Repeat screening allowed detection of maternal syphilis in a mother whose initial screen was negative and treatment of her RPR non-reactive exposed infant.

Disparities in Exposure to Ambient Air Neurotoxicants and Neurodevelopment in Children by Maternal Nativity Status

Author Name(s): Faven Araya, Jeanette A. Stingone, Luz Claudio

Department: Environmental Medicine and Public Health

Division: N/A

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: Exposure levels to environmental pollutants vary significantly among different populations. Exposure to different levels of hazardous air pollution (HAP) may contribute to disparities in neurodevelopment observed in children. HAP exposure levels could also interact with other factors to cause disparities in health outcomes. The aim of this study was to determine if exposure to HAP varies by maternal nativity status, and if those exposures are associated with neurodevelopmental measures in young children.

Hypothesis: We hypothesize that maternal nativity is an important, but often overlooked factor when examining neurodevelopmental disparities caused or aggravated by exposure differences to air pollution during childhood.

Methods: Data from the Early Childhood Longitudinal Study-Birth cohort was linked to the 2002 National Air Toxics Assessment using residential location at nine months of age. Participants were assigned exposures to ten HAPs with suspected neurotoxic effects. Multiple linear regression models were used to assess the joint effect of maternal nativity status and HAP exposure on neurodevelopment.

Results: Children of foreign-born mothers were observed to have higher exposure to high levels of HAPs. Isophorone exposure, a marker of industrial pollution, and maternal nativity status were independently associated with lower standardized BSF-R mental scores in children. Interaction between nativity status and isophorone was not statistically significant, but the change in mental scores associated with isophorone exposure was greater in children of foreign-born mothers.

Conclusions: Exposure to high levels of HAPs was more commonly found among children of foreign-born mothers. This exposure may negatively contribute to their neurodevelopment.

Comparison of Length of Stay Between Children Admitted to an Observation Versus Inpatient Unit

Author Names: Jennifer Bailey, C. Anthony Lim, Julie Oh, Erick Eiting, Ethan Cowan, Barbara Barnett, Yvette Calderon

Departments: Pediatrics

Division: Emergency Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai; Mount Sinai Beth Israel

Introduction: Many children requiring admission have self-limited illnesses and are discharged within 48 hours, making them ideal candidates for care in a pediatric observation unit (POU). Differences in patient outcomes for admission to a POU versus a pediatric inpatient unit (PIU) have not been thoroughly evaluated.

Hypothesis: Children admitted to a POU have a shorter length of stay (LOS) compared to children admitted to a PIU.

Methods: Admission data from the POU was compared to inpatient pediatric data from two sites (PIU16 and PIU17) to control for seasonal and hospital variation in admissions. Information regarding the current illness, past medical history, and emergency department, POU or PIU course were collected. The LOS was analyzed as the primary outcome. Changes in level of care were collected as secondary outcomes.

Results: 195 admissions were analyzed, 92 from POU, 53 from PIU16, and 50 from PIU17. There were no significant differences in patient or illness characteristics between the groups. The mean LOS was 24.4 hours (SD 13.1) for the POU, 62.0 hours (SD 39.7) for PIU16, and 47.5 hours (SD 29.5) for PIU17. The LOS for the POU group was significantly less compared to PIU16 ($p < 0.0001$) and PIU17 ($p < 0.0001$). 5 POU patients were converted to PIU. 4 patients from POU, 1 from PIU16, and 5 from PIU17 were transferred to the intensive care unit.

Conclusions: Children admitted to the POU have a significantly shorter LOS compared to those admitted to a PIU. POU's may provide the means toward efficient, patient-centered care for children requiring brief hospitalizations.

Chart Review to Evaluate Co-Sensitization Between Cashew and Sesame

Author Names: Mary Grace Baker¹; Sarah Saf^{1,2}; Travis Sifers¹; Christopher S. Knight¹; Anna Nowak-Wegrzyn¹

Department: Pediatrics

Division: Allergy & Immunology

Institution Affiliations: ¹Icahn School of Medicine at Mount Sinai ²; Hôpital d'Enfants Armand Trousseau

Institute Affiliation: Elliot and Roslyn Jaffe Food Allergy Institute

Introduction: The diagnosis of tree nut and seed allergy has increased in recent years. An association between tree nut and sesame allergy has been observed, and we noticed a particular association between cashew and sesame allergy. We sought to investigate this further.

Hypothesis: We hypothesized that there would be a significant number of patients sensitized to both cashew and sesame.

Methods: We reviewed electronic medical records for office visits between July 1-August 31, 2016 at our practice. Patients with evidence of cashew sensitization, defined as skin prick test (SPT) wheal 3 mm greater than the negative control or cashew-specific serum IgE >0.35 kU_A/L (ssIgE), who were avoiding cashew were identified. Demographic, clinical, and laboratory data were compiled.

Results: We reviewed 673 visits and identified 382 patients (57%) with cashew sensitization including 60% males, median age 8.1 years. A history of a convincing allergic reaction to cashew was documented in 62 patients (16%). The median cashew-ssIgE was 5.11 kU_A/L, and the median SPT wheal diameter was 6 mm. One hundred fifty three (40%) cashew-sensitized patients were avoiding sesame; of those, 57 (37%) had a history of a convincing allergic reaction to sesame. The median sesame-ssIgE was 11 kU_A/L (range: 0.38->100 kU_A/L); the median SPT was 6 mm (range: 0-23 mm). Patients with higher cashew-ssIgE were more likely to avoid sesame ($P=.0014$).

Conclusions: Cashew allergy is common in our practice, and 57% of patients evaluated demonstrate sensitization to cashew. There is a high rate of allergic co-sensitization between cashew and sesame.

Parental Satisfaction Post Dental Treatment Under Intravenous Sedation versus General Anesthesia in Pediatric Patients

Author Name(s): Michelle Baptiste, Laurence Hyacinthe

Department: Dentistry

Division: Pediatric Dentistry

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: Dental treatment with either general anesthesia or intravenous sedation may be indicated for pediatric patients in order to complete necessary dental treatment safely and efficiently. Indications for these treatment types include age and extent of treatment, uncooperative behavior, medical history, or acute infection. However, for numerous reasons including negative outcomes of these procedures publicized through the media, parents are apprehensive and become stressed before deciding to proceed with treatment. Nevertheless, the number of successful treatments outnumbers these unfortunate events. Past research reveals that surveyed parents are highly satisfied and they observe an improved quality of life following comprehensive dental rehabilitation under general anesthesia. At the same time, there is a paucity of research that measures parental satisfaction after treatment with intravenous sedation. Therefore, this study will contribute to this lack of research by surveying parents of patients treated at Mount Sinai with general anesthesia as well as intravenous sedation.

Hypothesis: We hypothesize that there will be parental acceptance of dental treatment with general anesthesia or intravenous sedation after having gone through the experience.

Methods: Patients treated at Mount Sinai Hospital for dental treatment under intravenous sedation or under general anesthesia from the February 1, 2018- February 1, 2019 will be identified by reviewing the OR and Sedation schedule. Parents of these patients will be invited to participate in a telephone survey. A 10 question multiple choice survey will be conducted by the main investigator to assess concerns prior to procedure as well as satisfaction and experience after their child's procedure. Descriptive statistical analysis will then be used.

Results: Pending

Conclusions: Pending

Effectiveness of Shared Reading Program on Neurodevelopmental Outcomes and Maternal Stress in the Neonatal Intensive Care Unit (NICU)

Author Names: Stephanie L. Bernard, Olivia Janssen, Camille Hebert, Cynthia Katz, Leora Mogilner, Jennifer B. Bragg

Department: Pediatrics

Division: Newborn Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Shared reading programs have been shown to be effective in increasing literacy in older children but are infrequently found in the NICU.

Objectives: To assess the impact of a shared reading program in the NICU on maternal stress, frequency of reading post discharge, language performance at age 2.

Methods: Families of infants 28-33 weeks gestation are eligible for enrollment. They will periodically be given books and education on importance of reading while in the NICU. Demographics and NICU variables will be recorded. Surveys assessing socioeconomic status, home reading environment and maternal stress will be administered. Bayley's performance at age 2 will be recorded. A retrospective control group will be recruited that did not receive literacy education. Unpaired t-tests and multi-variable logistic regression will be performed to assess treatment impact on outcomes.

Results: To date, 7 families have been enrolled in the study and enrollment is ongoing. Data analysis is ongoing.

Conclusions: A shared reading program in the NICU is an important tool to empower families while in the NICU and post-discharge. We hypothesize that families who participate in this program will report higher likelihood to read at home, improved maternal stress and language performance at age 2. Future directions include continued enrollment, standardization of NICU literacy curriculum, staff training and continued assessment of impact through long-term neurodevelopmental follow-up.

Debriefing Implementation Program in the Pediatric Intensive Care Unit

Author Name(s): Caroline Black, Christopher Strother, Iris Mandell, Jennifer Kero, Lauren Zinns

Department: Pediatrics

Division: Emergency Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Clinical event debriefing (CED) is a brief discussion that occurs after a patient event (resuscitation, procedure). CED allows teams to discuss the case, what went well, identify areas for improvement, and promote communication. There are many challenges to implementing CEDs, such as unpredictability of when events occur and finding time to conduct them.

Hypothesis: We hypothesize that implementation of a CED curriculum in the pediatric intensive care unit (PICU) improves team performance along with earlier debrief occurrence.

Methods: A prospective quality improvement study was conducted in the PICU September 2017 to October 2018. Participants included PICU faculty and nurses. Debriefing champions (1 attending, 2 nurses) underwent a 8-hour training course. Remaining faculty and 23 nurses underwent shorter sessions. A survey was distributed prior to curriculum implementation to participants. Following completion of the study, a post-survey was distributed. Surveys included the TeamSTEPPS Teamwork Perceptions Questionnaire (T-TPQ).

Results: 81 surveys were analyzed. Nurses reported increased use of CED from 42% a quarter of the time to 53% all of the time. CED occurrence immediately after critical events improved from 22% to 35%; majority occurring within 6 hours. Faculty led CEDs increased from 85% to 100%. In the 5 teamwork domains of T-TPQ, nursing responses were generally positive with largest improvement in leadership and situation monitoring. Attending responses showed no significant changes.

Conclusion: CED training can be used to encourage increased debriefing and improve some team behaviors. While a larger study is needed, we believe such training can improve patient safety.

Screening for Maternal Family Planning Needs During the Pediatric Well Visit: A Needs Assessment

Author Names: Aisha A. Bobb-Semple, Megan R. D’Andrea, Natasha Ramsey, John Rowland, Leora Mogilner

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Almost half of all pregnancies in the United States are unintended, and thus can be associated with negative outcomes for both the mother and infant. The postpartum period is a particularly high-risk time for unintended pregnancy; as such, the AAP recommends that pediatricians discuss family planning by the two-month preventive visit. To our knowledge, there has been limited research investigating mothers’ comfort with discussing family planning needs in the pediatric setting.

Objective: To evaluate mothers’ interest in family planning referrals provided through their child’s pediatrician in an inner city pediatric resident clinic.

Methods: An anonymous 22-question survey was administered to mothers of children under age two years presenting to the clinic for pediatric well visits, assessing mothers’ comfort in speaking with their child’s provider about their plans for future pregnancy, current contraception use, and their views on potential barriers to utilizing family planning referrals.

Results: 73 mothers completed the survey. 54% reported they would feel comfortable or very comfortable talking to their child’s doctor about their plans for having another child. 70% reported that they would be likely or very likely to accept a referral for family planning from their child’s doctor if they were offered one.

Conclusions: Half of mothers surveyed were comfortable having conversations with their child’s doctor about family planning, and an even greater percentage were interested in accepting referrals from their child’s doctor for family planning services. Future research should explore the effectiveness of such a referral program.

Assessing Pediatric Resident Readiness to Screen Mothers of Young Children for Family Planning Needs

Authors: Aisha A. Bobb-Semple, Natasha Ramsey, Megan R. D'Andrea, John Rowland, Leora Mogilner

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Background: Almost half of all pregnancies in the United States are unintended and women particularly are at high risk during the postpartum period. Effective contraception during that time is important, so the AAP advises pediatricians to discuss family planning by the two-month preventive visit. Previous work has shown that these discussions can be implemented in a pediatric resident clinic, with residents providing referrals for services as needed.

Objective: To assess resident knowledge, attitudes and confidence around discussing family planning with mothers of their young patients.

Methods: Pediatric residents in an urban pediatric residency training program completed anonymous surveys. Surveys were available in both paper and electronic formats.

Results: 39 residents completed the survey for a response rate of 65%. 55% of respondents felt knowledgeable about guidelines around providing contraceptive care for adolescents. These residents were more likely to be farther along in residency ($p=0.04$) and more likely to feel confident speaking with mothers about their contraceptive needs ($p < 0.001$). 90% of respondents were not aware of recommendations to screen mothers of infants for family planning needs.

Conclusions: Most pediatric residents surveyed were not aware of guidelines suggesting screening mothers of young children for family planning needs. Residents who felt more knowledgeable about guidelines for contraceptive care of adolescents were more likely to feel confident speaking with mothers about family planning. These data highlight resident willingness to screen mothers of infants for family planning needs and the opportunity to build on their current knowledge and practices.

Changing the Prognosis; Liver Transplantation for Infantile Pyruvate Carboxylase Deficiency

Author Name(s): Margo Sheck Breilyn^{1,2}, Brandon Stauffer¹, Ronen Arnon¹, Pankaj Prasun^{1,2}, George A. Diaz^{1,2}, and Kimihiko Oishi^{1,2}

Department: Genetics & Genomic Sciences¹; Pediatrics²

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Background: Pyruvate carboxylase deficiency (PCD) is a rare autosomal recessive disorder characterized by severe lactic acidosis, psychomotor retardation and seizures. Pyruvate carboxylase is an anaplerotic enzyme which catalyzes the synthesis of oxaloacetate from pyruvate. In the absence of pyruvate carboxylase activity, oxaloacetate becomes deficient resulting in dysfunction of Krebs cycle and energy metabolism. Anaplerotic therapies including citrate, aspartate, and triheptanoin supplementation can be attempted. However, the majority of patients with the severe neonatal and infantile forms die within the first year of life. Therapeutic orthotopic liver transplantation with a living related donor was reported previously in an infant with the neonatal form of PCD, however the clinical benefits remain unclear and have not been repeated. We report the case of a one-year old female with molecularly confirmed PCD who underwent liver transplantation for her intractable lactic acidosis. This is the first case of liver transplantation for PCD utilizing a cadaveric liver as a donor.

Objective: To evaluate the short-term outcome of a patient with infantile PCD who had liver transplant.

Methods: Retrospective chart review was performed. Pre- and post-transplant biochemical and clinical profiles were analyzed. Venous blood gas tests obtained during the pre-transplant period (Pre: one year prior to transplant) and post-transplant period (Post: three months post-transplant) were statistically analyzed. Written informed consent was obtained from the parents.

Results: Analysis of the patient's venous blood gas showed that liver transplant significantly improved the values of lactate (Pre; 8.07 ± 2.69 , Post; 3.46 ± 1.05 mM (mean \pm SD), $p=0.0001$), pCO₂ (Pre; 25.84 ± 7.88 , Post; 33.30 ± 4.65 mmHg (mean \pm SD), $p=0.0001$), HCO₃ (Pre; 16.04 ± 5.51 , Post; 21.30 ± 3.20 mEq/L (mean \pm SD), $p=0.0001$), and base excess (Pre; -7.62 ± 5.55 , Post; -2.64 ± 2.88 mM (mean \pm SD), $p=0.0001$). Clinically, the patient became more comfortable and active, with lower respiratory rates as reflected by venous pCO₂ values. The requirement of sodium citrate supplement was reduced from 16.7 to 5.8 mEq/kg/day and her plasma amino acid analyses showed complete normalization of the values of alanine and proline after the transplant.

Conclusions: The biochemical aberrations were significantly ameliorated with liver transplant. Liver transplant can improve the quality of life of patient with PCD and should be considered for treating this disorder.

Neural Circuit and Brain System Alterations Underlying Social Recognition Memory Deficits

Author Names: Natalie Burlant, Keerthi Thirtamara Rajamani, Erin Glennon, Lacin Koro, Kristi Niblo, Camille Goldman, Joseph Buxbaum, Valery Ginevich, Robert Froemke, Hala Harony-Nicolas

Department: Psychiatry

Division: N/A

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: The Seaver Autism Center, The Mindich Child Health and Development Institute, The Friedman Brain Institute

Introduction: Social deficits are a hallmark feature of several psychiatric and neurodevelopmental disorders, including autism spectrum disorder (ASD). To date, behavioral therapies are the primary intervention for treating impaired social behaviors, whereas pharmacological treatments have been ineffective at addressing these symptoms. To inform treatment targets, there is an urgent need to understand the pathophysiology underlying social deficits. Our research focuses on understanding the mechanisms by which mutations in an ASD high-risk gene, *SHANK3*, lead to the manifestation of behavioral deficits by studying their effect on (1) neural circuits that underlie social behavior, (2) the hypothalamic oxytocin system, which modulates several social behaviors, and (3) the interaction between both in health and disease.

Hypothesis: We hypothesized that *Shank3* mutation impacts the oxytocin system, leading to social memory deficits in the *Shank3*-deficient rat.

Methods: We use immunohistochemistry to study the morphology of oxytocin neurons in the paraventricular nucleus (PVN), brain microdialysis to quantify oxytocin levels during social behavior, fiber photometry to record activity of oxytocin neurons during social interaction, and chemogenetic tools to manipulate the oxytocin neural population *in vivo*.

Results: We found no differences in the number of oxytocin neurons in the PVN of wild-type and *Shank3* knock-out (KO) rats. However, we observed increased oxytocin labeling in the PVN and lateral septum of *Shank3* KO rats. These changes were accompanied by reduced neural activity in the PVN of *Shank3* KO rats.

Conclusions: These findings suggest an overall impairment in oxytocin function may underlie social memory deficits in our ASD rat model.

Angiogenic Profile Identifies Pulmonary Hypertension in Children with Down Syndrome

Author Names: Douglas Bush, Csaba Galambos^{1,2}, Kristine Wolter-Warmerdam³, Brandie D. Wagner⁴, Dunbar Ivy^{2,3}, Steven H Abman^{2,3}, Deven McMorrow², Fran Hickey²

Department: Pediatrics

Division: Pulmonology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Other Institution Affiliations: ¹Departments of Pathology and ²Pediatrics, University of Colorado School of Medicine; ³The Children's Hospital Colorado, Aurora CO; and ⁴Department of Biostatistics and Informatics, University of Colorado and The Children's Hospital Colorado.

Introduction: Past studies have shown that lung angiogenic signaling is dysregulated in children with Down syndrome (DS) possibly contributing to the development of pulmonary hypertension (PH).

Hypothesis: Abnormal angiogenic protein levels will identify children with DS who have PH.

Methods: A prospective study of 78 children from birth to 21 years of age, was conducted to evaluate clinical data, echocardiograms, and cardiac catheterizations. Four patient populations were enrolled, including: 1) children with DS and PH (DS+PH, n= 12); 2) control children without DS who have PH (C+PH, n= 15); 3) children with DS without known PH (DS-PH, n= 26); and 4) healthy non-DS controls (C-PH, n= 25). Blood samples were collected at enrollment (time of diagnosis via cardiac catheterization for those with PH) and concentrations for 11 proteins were evaluated by Luminex® angiogenesis assay. A classification tree was created to identify angiogenic peptide signals that may be associated with PH in children with DS compared with controls.

Results: Mean ages were similar across study groups: DS+PH (6.1 ± 6.3 years), C+PH (9.1 ± 5.8 years), DS-PH (6.2 ± 4.6 years), and C-PH (7.6 ± 5.4 years). The classification tree revealed that elevated endostatin levels ($>4.98 \log_{10} \text{ pg/mL}$) were associated with DS while lower angiogenin ($<5.428 \log_{10} \text{ pg/mL}$) levels further distinguished PH in those with DS from the other groups.

Conclusions: High endostatin coupled with low angiogenin levels identified children with DS who had PH suggesting these angiogenic factor levels may serve as biomarkers for PH in this population.

Piezo1 Channels Contribute to Flow Induced Intracellular Calcium Transients in Mouse Cortical Collecting Duct (CCD)

Authors: Rolando Carrisoza-Gaytan¹, Daniel Flores¹, Marianela Dalghi², Gerard Apodaca², Thomas Kleyman², Lisa Satlin¹

Department: Pediatrics

Division: Nephrology

Institution Affiliations: Icahn School of Medicine at Mount Sinai¹, University of Pittsburgh²

Introduction: Within the CCD, an acute increase in tubular fluid flow rate (TFFR) subjects principal (PC) and intercalated (IC) cells to an immediate increase in intracellular Ca^{2+} concentration ($[\text{Ca}^{2+}]_i$), followed by a decay to a plateau elevation in $[\text{Ca}^{2+}]_i$ that exceeds baseline, sustained by luminal Ca^{2+} entry. The increase in $[\text{Ca}^{2+}]_i$ is necessary for flow induced K^+ secretion (FIKS) in the microperfused mammalian CCD. The identity of a basolateral Ca^{2+} entry path is unknown.

Hypothesis: We hypothesize that Piezo channels, a family of mechanically-activated Ca^{2+} permeable channels, contribute to the basolateral Ca^{2+} entry.

Methods/Results: Immunofluorescence studies in mice expressing Piezo1-tdTomato construct revealed robust basolateral and faint apical Piezo1 localization in both PC and IC. Flow-induced $[\text{Ca}^{2+}]_i$ transients, were measured in microperfused mouse CCDs, loaded with the Ca^{2+} indicator fura-2, and subject to low or high TFFRs, in the absence or presence of GsMTx4 (piezo-blocker) or Yoda1 (piezo1-activator). IC in CCDs from male and female mice preincubated with luminal or bath GsMTx4 (5 μM) failed to exhibit a flow-induced increase in $[\text{Ca}^{2+}]_i$ in response to an acute increase in TFFR. Addition of luminal or bath Yoda 1 (1 μM) to CCDs perfused at a low TFFR led to an increase in $[\text{Ca}^{2+}]_i$ over 90 s in both PC and IC; addition of Yoda1 (10 μM) to the bath further increased $[\text{Ca}^{2+}]_i$.

Conclusions: Mechanosensitive Piezo1 channels, expressed on the basolateral>apical membranes of PC and IC in the mouse CCD, contribute to the increase in $[\text{Ca}^{2+}]_i$ triggered by an increase in TFFR.

Papillary Thyroid Carcinoma in a Pediatric Patient with Beta Thalassemia

Author Name(s): Lauryn Choleva, Christopher Romero, Elizabeth Wallach, Mabel Yau, Jasmine Gujral, Swathi Sethuram, Robert Rapaport, Meredith Wilkes

Department: Pediatrics

Division: Endocrinology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Beta thalassemia is characterized by the abnormal synthesis of β hemoglobin chains resulting in hemolytic anemia. Treatment involves frequent blood transfusions, which leads to deposition of iron in many organs. Iron overload has been associated with various malignancies, most notably liver and hematological. To date, 7 cases of papillary thyroid cancer in patients with beta thalassemia have been reported in the adult literature, but none in pediatrics.

Hypothesis: N/A

Methods: The patient is a 15 year 4 month old female with beta thalassemia who has required chronic red blood cell transfusions. She underwent a splenectomy at the age of 10 years and received chelating therapy with deferasirox and deferiprone. MRI revealed iron deposition in her pancreas, liver, kidneys, bone marrow and pituitary gland. On exam, her thyroid gland was asymmetric with the right lobe measuring 1cm larger than the left. The gland was firm in consistency with palpable lymph nodes along the right anterior cervical chain. A thyroid ultrasound revealed an enlarged right lobe containing 3 focal hypoechoic masses with calcific foci. Biopsy obtained was consistent with papillary thyroid carcinoma. She underwent total thyroidectomy and histological examination confirmed the diagnosis. Her postoperative course was uncomplicated and she was started on replacement therapy with levothyroxine.

Results: N/A

Conclusions: To our knowledge this is the first case of papillary thyroid carcinoma in a pediatric patient with beta thalassemia. The incidence of thyroid cancer in patients with beta thalassemia is currently unknown, however there may be utility in routine surveillance of this patient population.

Identifying And Helping Patients with Food Insecurity Through a Quality Improvement Initiative

Author Name(s): Liliana Cruz, Sri Nikhita Chimatapu, Ekaterina Golovko, Surkhay Bebiya, Jacqueline Maya, Anandini Suri, Rakesh Donthula, Charmin Gohel, Sebastian Proano, Lisa Forman, Daniel Ibanez, Gwen Raphan

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine Mount Sinai- Elmhurst Hospital.

Introduction: In the US more than 13 million children live in food insecure homes lacking consistent access to enough food. The AAP recommends screening at each health maintenance visit.

Hypothesis: Improved techniques of screening for food insecurity in our pediatric outpatient clinic would increase the reported rates and allow us to extend services to more families in need.

Methods: Starting October 2018 a paper questionnaire with 2 validated food security questions was distributed during health care visits. Providers addressed positive responses and distributed a list of food resources. 50 charts pre and post intervention were reviewed to assess the number of families reporting food insecurity with the question built into our EMR versus the paper questionnaire.

Results: Prior to our intervention, only 29% of providers said they always screened for food insecurity. Pre-intervention only 4% of families reported food insecurity as compared to 44% post-intervention. Post-intervention 18% of those with food insecurity reported frequently having trouble.

Conclusion(s): Our project revealed an incredibly higher rate of food insecure households compared to the prior assessment tool. This increased awareness of the severity of the issue and allowed us to extend help to those in need.

Optimizing Approaches to Drug Allergies in the Pediatric Inpatient Setting

Author Names: Nesy Dahan, Roxanne C. Oriel

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institution Affiliation: The Jaffe Food Allergy Institute

Introduction: Drug allergy labeling is often inaccurate and unconfirmed, leading to unnecessary drug substitutions and increased healthcare costs.

Hypothesis: Evaluation of the process for allergy reconciliation in the electronic medical record during hospital admissions will provide insight into the drug allergy labeling process at Mount Sinai Hospital.

Methods: A retrospective chart review of all patients admitted to one general pediatric inpatient unit was conducted over a 4-week period between December 2017 and January 2018. Data recorded included: demographics, drug allergy details (agent, reaction, and severity), provider responsible for and location of allergy listing, infection type and antibiotic prescribed if applicable, and admitting and discharge diagnoses.

Results: A total of 145 patients were admitted to the unit. The inpatient physician and registered nurse reviewed the allergy section for only 66/145 (45.5%) patients. Drug allergies were recorded for 21/145 (14.5%) patients, of which 9/21 (42.9%) were antibiotic allergies. Antibiotic allergy classes included 6/9 (66.7%) penicillin, 2/9 (22.2%) cephalosporin, and 1/9 (11.1%) sulfa. Antibiotic allergy reactions included “rash” (6/9, 66.7%), “unknown” (2/9, 22.2%), and “hives” (1/9, 11.1%). Notably, 2/145 (1.4%) patients had incorrectly labeled drug allergies in the emergency department amended during the admission process.

Conclusions: Drug allergies are inconsistently reviewed by physicians and nurses during the hospital admission process, which can lead to medical errors. To prevent errors, allergy reconciliation should be mandatory for all admissions to the inpatient unit, including transfers between units.

Improving Rates of HPV Vaccination in the Pediatric Outpatient Setting

Author Names: Nessy Dahan, Erina Lie, Camille Almada, Vanessa Eddy, Shane Goda, Brian Park, Amanda Reich, Ann Symonds, Van Don Williams II, Cynthia Katz

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Human papillomavirus (HPV) vaccination rates in the pediatric population are suboptimal despite proven benefits of cancer and wart prevention. Series completion can be optimized with early initiation at 9 years.

Hypothesis: An interdisciplinary quality improvement initiative involving physicians and nurses in the Pediatric Associates outpatient clinic will improve HPV vaccine initiation and series completion rates amongst patients ages 9-11 by 10% compared to baseline.

Methods: Phase one involved a retrospective chart review over a 6-week period between September and October 2018 to collect pre-intervention vaccination rates. Phase two involved creation of a HPV database from January 2015 through December 2018. Providers were also surveyed to investigate barriers to early initiation of vaccine series.

Results: 221 patients ages 9-11 were seen in the clinic across 6-weeks. Of these, 48% had ≥ 1 dose of HPV vaccine, and 32% completed the series. The three-year database revealed that between ages 9-21, 34% (19% male, 15% female) had not initiated HPV vaccine, and 52% (25% male, 27% female) had completed the series. Awareness versus practice for starting vaccination at age 9 varied based on post-graduate year (PGY-1: 75.0% vs. 37.5%, PGY-3: 84.6% vs. 46.2%, PGY-3: 91.7% vs. 63.6%).

Conclusions: HPV vaccination rates in our pediatric outpatient clinic are suboptimal, reflecting national trends. Planned PDSA cycles for improvement include: investigating reasons for missed vaccination, educating providers to address recommendations for early initiation of vaccine series and availability of nurse visits for HPV vaccination, creating EPIC reminders for incomplete vaccinations, and establishing nurse-driven patient recalls.

Early-life Dentine Manganese Concentrations and Intrinsic Functional Brain Connectivity in Adolescents and Young Adults

Author Name(s): Erik de Water, Demetrios Papazaharias, Claudia Ambrosi, Lorella Mascaro, Yuri Levin-Schwartz, Elza Rechtman, Giuseppa Cagna, Daniele Corbo, Roberto Gasparotti, Roberto G. Lucchini, Manuela Oppini, Donatella Placidi, Christine Austin, Manish Arora, Cheuk Y. Tang, Donald R. Smith, Robert O. Wright & Megan K. Horton

Department: Environmental Medicine and Public Health

Division: Environmental Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai, University of Brescia, UC Santa Cruz

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Manganese (Mn) is an essential nutrient that is neurotoxic at high levels of exposure. Early-life Mn exposure is associated with impaired cognitive and motor control and changes in the underlying neural circuitry (i.e., prefrontal cortex and basal ganglia) in adolescents. The aim of this study was to examine associations between early-life Mn concentrations and intrinsic functional connectivity (iFC) of the brain in adolescents and young adults, focusing on the prefrontal cortex (PFC) and basal ganglia. We measured Mn concentrations in dentine of deciduous teeth, allowing us to directly measure fetal exposure and to identify windows of vulnerability from pregnancy until childhood.

Hypothesis: We hypothesized that adolescents with higher early-life Mn concentrations would show altered iFC of the basal ganglia and PFC.

Methods: 60 participants (33 girls; 15-23 years) from Northern Italy (Public Health Impact of Manganese Exposure study; PHIME) completed a 10-minute resting state functional Magnetic Resonance Imaging (fMRI) scan, and provided deciduous teeth. We used laser ablation inductively coupled plasma mass spectroscopy (LA-ICP-MS) to determine prenatal, early postnatal (0-1 years) and childhood (1-6 years) dentine Mn concentrations. We performed seed-based correlation analyses (FDR-corrected $p < 0.05$) to examine associations between natural log-transformed Mn concentrations and iFC of 6 basal ganglia seed regions (left and right putamen, caudate, pallidum) and 1 PFC seed region (bilateral middle frontal gyrus) from Harvard-Oxford Structural Atlases. We included SES, age and IQ as covariates.

Results: Participants with higher prenatal Mn concentrations showed: 1) reduced connectivity between the middle frontal gyrus and superior parietal lobule ($\beta = -0.21$, FDR-corrected $p = 0.001$); 2) Increased connectivity between the right putamen and cerebellum ($\beta = 0.18$, FDR-corrected $p = 0.013$). Participants with higher early postnatal Mn concentrations showed: 1) reduced connectivity between the middle frontal gyrus and medial prefrontal cortex ($\beta = -0.24$, FDR-corrected $p = 0.009$); 2) increased connectivity between the left pallidum and precuneus ($\beta = 0.13$, FDR-corrected $p < 0.001$).

Conclusions: Higher prenatal and early postnatal Mn concentrations are associated with altered iFC of brain areas involved in cognitive (i.e., PFC, parietal cortex) and motor control (i.e., putamen, pallidum, cerebellum) in adolescents and young adults.

Student Ambassador Program to Increase HPV Vaccination Coverage Rates among Adolescents

Author Names: Grisselle DeFrank, Melanie Arnold, Martha Arden, Janet Lee

Department: Pediatrics

Division: Adolescent Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: Human Papillomavirus (HPV) exposure during can lead to several types of cancer in adulthood. In 2017, New York State health regulations changed to allow health care providers to administer HPV vaccine to at-risk minors without the consent of parents or guardians. We developed a peer-educator program led by “Student Ambassadors” to provide and disseminate information about the HPV vaccine to adolescents in a high school setting with the goal of increasing HPV immunization rates in a School-Based Health Center (SBHC).

Hypothesis: We hypothesized that we would be able to increase the number of HPV vaccines given to students by 20% over 5 months by implementing student initiated projects.

Methods: Baseline HPV vaccination coverage rates for one SBHC were collected in January 2019. Student Ambassadors, selected from interested volunteers, developed 3 multimedia, youth-friendly interventions to increase HPV vaccine awareness. Student Ambassador interventions were implemented on January 16, 2019. The data collection period will be April 2019 until June 2019.

Results: We expect to find a 20% increase in the number of HPV vaccines given to students at the SBHC over a 5-month period from January 2019 to June 2019. Results are pending as the project is still ongoing.

Conclusions: We hope to find that Student Ambassadors are effective at developing peer-led HPV outreach projects that successfully increase vaccination rates in a NYC urban high school. Our next steps will include implementing the peer-led HPV outreach campaign for the entire 2019-2020 school year, and disseminating this intervention in other high schools.

Combined effects of BTEX and material hardship on ADHD-suggestive behaviors among a nationally-representative sample of U.S. children

Author Name(s): Kayla Dellefratte, Jeanette A. Stingone, Luz Claudio

Department: Environmental Medicine and Public Health

Division: International Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation:

Introduction: Both environmental and social factors may contribute to the development of attention deficit hyperactivity disorder (ADHD).

Hypothesis: The purpose of this study was to determine the relationship between early-life exposure to common urban air pollutants (benzene, toluene, ethylbenzene, and xylene, BTEX), household material hardship (a measure of socioeconomic status), and ADHD-suggestive behaviors in kindergarten-age children in a subset (N=4650) of the Early Childhood Longitudinal-Study Birth Cohort.

Methods: Pollutant exposure was estimated by linking the 2002 National Air Toxics Assessment to each child's residential ZIP code at enrollment. Material hardship was assigned as a composite score of access to food, health care, and housing. Teachers rated children on ADHD-suggestive behaviors using a 5-point Likert scale. Children with summary scores in the bottom decile were classified as displaying behaviors suggestive of ADHD. Logistic regression models were constructed to estimate the effect of both BTEX and material hardship on ADHD-suggestive behaviors.

Results: The odds of ADHD behaviors were greater in children with combined high level exposure to BTEX and in those experiencing material hardship (Odds Ratio 1.54 [1.12, 2.11] and 2.12 [1.25, 3.59], respectively), adjusting for confounders. These associations were stronger when restricting the study population to urban areas. There was no statistical evidence of interaction between BTEX exposure and material hardship, although the effects of BTEX were slightly greater in magnitude among those with higher material hardship scores.

Conclusions: These results are congruent with recent studies and add to growing literature on how environmental and socioeconomic factors affect neurodevelopment.

A Novel Role for Mannose Phosphate Isomerase in the Response to Radiation and DNA Damage

Author Names: Charles DeRossi, Nataly Shtraizent, Joshua Morrison, Geoffrey Girnun, and Jaime Chu

Department: Pediatrics

Division: Hepatology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Radiation exposure leads to repetitive DNA damage. The cell responds by upregulating metabolic pathways to synthesize new nucleotides and combat DNA damage. Mannose phosphate isomerase (MPI) is an enzyme most studied for its role in a congenital disorder of glycosylation, but we found that MPI regulates other metabolic pathways, distinct from glycosylation, to drive cell survival in development and cancer. However, the effects of MPI on DNA repair mechanisms are unknown.

Hypothesis: MPI loss leads to defective pentose phosphate pathway (PPP) function and increased susceptibility to DNA damage.

Methods: We probed the PPP through enzymatic analysis and LC/MS. MPI was depleted using morpholino oligonucleotides and siRNA. Zebrafish and MEFs were exposed to DNA damaging radiation and analyzed for cellular responses including phenotype, enzymatic assay, and metabolite analysis.

Results: Knockdown of MPI in zebrafish and MEFs led to a reduction in glucose-6-phosphate dehydrogenase, the first step of the PPP. Targeted metabolomics analysis identified ribose-5-phosphate, an intermediate of the PPP and important nucleotide precursor for DNA repair, to be substantially reduced following MPI depletion. Loss of MPI in zebrafish and MEFs produced a susceptibility to DNA damaging radiation, whereas MPI overexpression was protective. Notably, a high molecular weight, nuclear form of MPI was detected in both zebrafish and MEFs following irradiation in a dose- and time-dependent manner, suggesting a novel function.

Conclusion: Our results reveal MPI to be necessary for maintenance of the PPP, and to play an important role in response to DNA damaging agents.

Differential Transcript Usage in the Placenta is Associated with Aberrant Fetal Growth

Author Name(s): Maya A. Deyssenroth, Shouneng Peng, Ke Hao, Carmen J. Marsit, Jia Chen

Department: Environmental Medicine and Public Health

Division: Environmental Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: The role of the placenta in dictating fetal growth is well recognized and supported by reported differences in gene expression patterns. However, most studies focus on summarizing transcriptional activity at the gene level and do not capture alternative splicing, a major driver in development that enables adaptive responses to the rapidly changing physiological conditions experienced early in life. In this study, we conducted a genome-wide analysis of the placental transcriptome to evaluate alternative splicing in deviations of appropriate fetal growth.

Hypothesis: We hypothesized that alternative transcript usage in the placenta is associated with deviations in fetal growth.

Methods: RNA sequencing of placenta samples was conducted in the Rhode Island Child Health Study (RICHS, n=200). Differential transcript usage (DTU) was assessed using the DRIMSeq R package. Available SNP genotyping information was incorporated to assess genetic drivers of splicing using the sQTLseeker R package.

Results: We identified 223 genes demonstrating SGA-DTU and 31 genes demonstrating LGA-DTU. Furthermore, we identified 150 splicing quantitative trait loci (sQTLs), out of which 14 overlapped with SGA-DTU genes and none overlapped LGA-DTU genes. The SNPs driving sQTLs have been previously associated in GWAS with health outcomes, including cardiometabolic traits, autoimmune disorders and neurologic conditions.

Conclusions: We report the first genome-wide characterization of placental transcript usage and associations with aberrant fetal growth. Our findings reveal novel placental biomarkers of growth and development and further suggest a potential pathway by which perturbations in placental processes early in life may heighten susceptibility to disease traits later in life.

Network analysis of nasal transcriptome profiles reveals causal key driver genes of severe asthma in children

Author Name(s): A. N. Do¹, Y. Chun¹, J. Andrade¹, G. Grishina², A. Grishin², A. Vicencio², E. Schadt¹, S. Bunyavanich^{1,2,3}

Department: ¹Genetics and Genomic Sciences, ²Pediatrics

Division: ²Allergy & Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: ³The Mindich Child and Health Development Institute

Introduction: Severe asthma remains a major source of asthma burden, especially in children.

Objective: Identification of key driver genes of severe asthma could highlight novel mechanistic pathways of disease.

Methods: We recruited 305 subjects with asthma and controls for nasal brushing and RNA sequencing. Severe asthma genes were identified by differential expression analysis. To characterize the biologic context for these genes, a weighted gene co-expression network was constructed and examined for gene ontology enrichments. Member genes of enriched modules were used to build probabilistic causal networks and to identify causal key drivers. The findings were then tested in an independent cohort (n=190).

Results: We identified a severe asthma signature comprised of 373 genes (absolute log₂-fold-change ≥1, FDR≤0.05), of which 256 were over-expressed and 117 were under-expressed in severe asthmatic children vs. controls. This signature was significantly over- and under-represented in co-expression modules for cilium assembly and inflammatory response, respectively. Probabilistic causal network construction and key driver analysis identified nineteen causal key driver genes of the cilium assembly module and nine of the inflammatory response module. Testing of the findings in an independent cohort revealed 68% of the severe asthma signature was common to asthma more broadly, and also cilium assembly and inflammatory response co-expression modules trigger asthma. Six causal key drivers (*LRRC48*, *PACRG*, *LRRC23*, *TMEM231*, *FAM166B*, and *CCDC17*) of the cilium assembly module and two causal key drivers (*MNDA* and *SRGN*) of the inflammatory response module were confirmed broadly enriched in asthma.

Conclusion: Causal key driver genes modulating cilia activity and inflammatory process represent high-yield therapeutic targets for asthma.

Characterizing the Prenatal Inflammatory Milieu Associated with Maternal Asthma: A Proteomics Approach

Author Names: Julie D. Flom^a, Megan Niedzwiecki^{b,c}, Elena Colicino^{b,c}, Anu Joshi^c, Whitney Cowell^{a,b}, Alison Lee^b, Cecilia Berin^a, Rosalind J. Wright^{a,b,c}

Departments: Pediatrics^a, Institute for Exposomic Research^b, Environmental Medicine and Public Health^c

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Institute for Exposomic Research

Introduction: Maternal asthma constitutes a pro-inflammatory state in pregnancy that contributes to offspring asthma risk. The prenatal inflammatory milieu is a consequence of shifts in interacting systems including neuroendocrine and immune. High-throughput technologies enabling system-wide profiling of the inflammatory response may be particularly informative.

Methods: Analyses included n=449 mothers in the PRogramming of Intergenerational Stress Mechanisms cohort. Women reported active asthma (physician/ED visit/inpatient admission or use of bronchodilator/inhaled corticosteroid/systemic corticosteroid during pregnancy). We characterized 92 inflammatory markers in maternal sera obtained at 30.0±5.8 weeks gestation using the Olink multiplex inflammation panel. Associations between each inflammatory marker and asthma were first estimated using multivariable logistic regression with adjustment for education, pre-pregnancy body mass index (BMI), race/ethnicity, age, prenatal smoke exposure and fetal sex; key inflammatory markers were identified as those with the greatest magnitude or strongest statistical significance. We next used weighted quantile sum (WQS) regression to assess associations between the mixture of key inflammatory markers and asthma in the overall sample and stratified by pre-pregnancy overweight/obese versus normal/underweight.

Results: The sample was 32.1% Black and 32.8% Hispanic; 15.4% had active asthma. Higher levels of interleukin (IL)-15R α and IL-10R β were associated with active maternal asthma. In WQS analyses, the association between key inflammatory markers and maternal asthma was only significant among overweight/obese mothers.

Conclusions: Identified biomarkers have been implicated in the regulatory immune response and may elucidate mechanisms of fetal programming of asthma. We will employ network modeling to examine whether these factors mediate associations between maternal and children's asthma risk.

Tooth and Hair Analysis of Children with Kidney and Hypertensive Disease

Author Name(s): Smita Goodman, Jeffrey Saland, Manish Arora

Department: Pediatrics

Division: Nephrology and Hypertension

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Chronic kidney disease (CKD) affects children of all ages and is a major risk for progression to End Stage Renal Disease which is in turn associated with significantly reduced life expectancy. Some etiologies have been established, but potential environmental risk factors for *onset* as well as *progression* from earlier or milder forms of CKD remain unknown. Other etiologies have distinct and dramatic clinical characteristics yet poorly understood triggers for these often abrupt clinical changes. Tooth-matrix biomarkers can retrospectively reconstruct the history of exposure to multiple classes of analytes spanning the prenatal and childhood periods and define the time of their deposition. Hair assays allow for approximate daily measures of elements and we can examine changes prior to, during and after episodes of illness.

Hypothesis: Teeth from children with kidney diseases or with hypertension contain time-sequenced layers of chemicals that were present in a child during the formation of the tooth and we aim to identify which of these chemicals can modify the risk of developing these conditions, the frequency and severity of relapse/remission cycles, or the risk and rate of disease progression. Elemental metabolic signatures detected in hair assays during remission differ from those during episodes illness and we aim to uncover additional detail about abrupt relapses of nephrotic syndrome or other changes in renal function such as transplant rejection.

Methods: Teeth and hair are collected from patients with pediatric CKD or hypertension from the clinical population served by our division and screened for chemical exposures in a range of biological matrices.

Results: Collection of teeth (25 patients) and hair (12 patients) is ongoing.

Conclusions: Pending

Why are Children Drinking too Much Cow's Milk? Identifying Gaps in Caregiver Education

Author Name(s): Irina Gorbounova, Rachel Levantovsky, Melissa Hill, Emma Loebel, John C. Rowland, Leora Mogilner

Department(s): Pediatrics

Division(s): General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Consumption of more than 24 oz of cow's milk is a risk factor for iron deficiency anemia. There is limited data on children's overconsumption of cow's milk (defined as >24 oz daily) and what factors are associated with milk overconsumption.

Hypothesis: Children overconsume milk more frequently if caregivers have poor knowledge of nutrition guidelines.

Methods: A convenience sample of caregivers of children 2-4 years old attending two pediatric practices was surveyed. The survey included demographic factors, socioeconomic status, caregiver knowledge of nutrition guidelines regarding milk consumption, and beliefs about benefits of cow's milk. Group comparisons were made using Chi square and Fisher's exact test.

Results: 153 surveys met inclusion criteria. 59.1% were Medicaid recipients, 37.3% were WIC recipients, and 22.5% were food insecure. 80% of WIC beneficiaries knew cow's milk consumption guidelines, compared to 57.3% of non-WIC caregivers ($p=0.005$). However, 49.1% of WIC beneficiaries believed more milk is better for children's health, compared to 22.8% of non-WIC caregivers ($p=0.001$). Caregivers who had this belief were more likely to have children who overconsume milk ($p=0.002$). Children overconsumed milk at a similar frequency despite WIC status (16.4% vs. 12.6% for WIC and non-WIC beneficiaries, respectively; $p=0.53$).

Conclusions: Despite WIC beneficiaries' superior knowledge of cow's milk consumption guidelines, they were more likely to believe that more milk is better for their children, and their children overconsumed cow's milk to a similar degree as children of non-WIC caregivers. This gap between knowledge and practice presents an opportunity to educate families of risks associated with cow's milk overconsumption.

Implementing the Solutions for Patient Safety (SPS) Unplanned Extubations Bundle: Improved Education with the Help of RN and RT Champions

Author Name(s): Glynda Guia-Johnson, Kelvin Ziegler, Ellen Black, Yong Sing Da Silva, Juliana Guiney, Kelsey Mulhern, Ali Rosenberg, Lindsey Schroy

Department: Pediatrics

Division: PICU

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: None

Introduction: Unplanned extubations (UE) can cause significant harm in pediatric populations in addition to increased costs and lengths of stay, and decreased patient satisfaction. Solutions for Patient Safety (SPS), a national network of pediatric hospitals, developed an UE prevention bundle to reduce the rate of UEs in pediatric patients.

Hypothesis: We hypothesize that a multidisciplinary peer-led training program utilizing nurse (RN) and respiratory therapy (RT) trainers will effectively spread the SPS UE bundle elements and sustain these practices through a peer-to-peer auditing system, effectively reducing UEs in the PICU over the next year.

Methods: The PICU Nurse Manager and Pediatrics Nurse Educator trained RN and RT champions on the use of a new endotracheal tube (ETT) taping product and method of securement, as well as introduced a re-taping checklist and protocol for scenarios with high risk of UEs. Champions then trained their RN and RT peers for more efficient dissemination of the curriculum.

Results: Nearly 90% of nurses were trained over a two-month period. Champions provided hands on teaching and support to RN and RT staff to ensure compliance with bundle elements. Just-in-time coaching and auditing of the taping process and protocol are used to monitor implementation. This work has led to a preliminary reduction in the expected number of UEs.

Conclusions: A multidisciplinary, peer-driven education curriculum utilizing RN and RT champions has been effective in implementing the SPS UE bundle in our PICU. We will continue to track the long-term impact of this intervention on our PICU's UE rate.

Undervirilized Male Infant with Features of Atypical CAH Following Prenatal Exposure to Nystatin

Author Name(s): Jasmine Gujral, Divya Khurana, Gertrude Costin, Swathi Sethuram, Christopher Romero, Lauryn Choleva, Meredith Wilkes, Elizabeth Wallach, Mabel Yau, Robert Rapaport

Department: Pediatrics

Division: Pediatric Endocrinology and Diabetes

Institution Affiliation: Icahn School of Medicine at Mount Sinai & Jamaica Hospital

Introduction: Ketoconazole reversibly suppresses steroidogenesis by inhibiting cytochrome P450 enzymes. Intrapartum Nystatin has rarely been associated with hypospadias. We report a male infant born to a mother with intrapartum nystatin use, who presented with undervirilization and transient adrenal corticosteroid abnormalities.

Hypothesis: N/A

Methods: The patient is a 31-week boy born to a mother with intermittent vaginal discharge during pregnancy for which she received up to 60 vaginal Polygynax capsules containing Nystatin (100,000 IU) starting from first trimester. The infant had micropenis, chordee, perineoscrotal hypospadias and bifid scrotum with bilaterally palpable gonads.

Results: 17 OHP on newborn screening was 304 ng/ml (nl <35), Karyotype 46 XY, SRY+ on FISH and no Mullerian structures were seen on ultrasound. Elevation of adrenal corticosteroids did not indicate a specific enzymatic defect (Table 1). Hydrocortisone was started on day of life (DOL) 5, fludrocortisone and NaCl on DOL 12. Genetic testing for 16 genes was negative (Table 2). Over the next few months all medications were successfully discontinued. Retesting with Cosyntropin and hCG at 8 months indicated normal baseline and stimulated adrenal steroids and T/DHT.

Conclusions: To our knowledge, this is the first report of undervirilization in a 46XY infant with neonatal abnormalities in P450Cyp21 pathway and clinical evidence of disruption in fetal P450Cyp19 pathway. Since neonatal T and DHT were normal it is possible that fetal undervirilization resulted from insufficient T and DHT production/action during sexual differentiation. We postulate that intrapartum nystatin use leads to severe but transient defects in corticoid and androgen synthesis and/or action.

Table 1: Laboratory values (DOL- Day of life, B- Baseline, S1- Stimulated with ACTH, S2- Stimulated with HCG)

	Reference Range	DOL 2	DOL 3-4	DOL 10	8 months (off HC)	15 months (off HC)
ACTH (pg/ml)	7.2-63		135 (B)		11 (B)	7.4 (B)
Cortisol (mcg/dl)	3-23		6.5 (B) 10 (S1)		19 (B) 29 (S1)	4.5 (12 pm)
Sodium (mmol/L)	134-144	142	144	134	140	141
Potassium (mmol/L)	3.8-5.3	6.3	5.8	6.9	5.7	4.9
Bicarbonate (mmol/L)	15-25	18	19	23	21	19
Plasma Renin Activity (ng/ml/hr)	Premature: 1 – 7 Days: 11 – 167 1– 11 Months: 2.35 – 37			174.1	7.8	3.1
Aldosterone (ng/dl)	Premature: 12-736 1-6 months: 2-76			100	11	
17 Hydroxyprogesterone (ng/dl)	186-472 (B) 334-1725 (S)	516 (B)	4853 (S1)		57 (B) 182 (S1)	22 (B)
17 Hydroxypregnenelone (ng/dl)	559-2906 (B) 831-9760 (S)		> 4000(S1)		528 (B) 1150 (S1)	34 (B)
Dehydroepiandrosterone Sulfate (mcg/dl)	122-710 (B)		2415 (B)			
11 Deoxycortisol (ng/ml)	< Or = 235 (B)		1431 (B)		54 (B) 107 (S1)	
Deoxycorticosterone (ng/dl)	28-78		65			
Corticosterone (ng/dl)	201-5030 (B)		992			
Testosterone (ng/dl)	31- 35 weeks Day 4 37-198	383 (B)			3.6 (B) 587 (S2)	
Dihydrotestosterone (ng/dl)	Premature: 10-53		23 (B)		1.4 (B) 111 (S2)	
Testosterone : Dihydrotestosterone ratio	1.5-15.5 (S2)				5.3 (S2)	

CAH 21 Hydroxylase (CYP21A2)	Negative
3betahydroxysteroid dehydrogenase deficiency (HSD3B2)	Negative
Androgen receptor (AR)	Negative
Aristaless related Homeobox (ARX)	Negative
Cytochrome P450 Oxidoreductase (POR)	Negative
Chromobox protein homolog 2 (CBX2)	Negative
Hydroxysteroid 17beta dehydrogenase 3 (HSD17B3)	Negative
Cytochrome P450 Family 11 (CYP11A1)	Negative
Chromobox protein homolog 2 (CBX2)	Negative
Mastermindlike DomainContaining protein 1 (MAMLD1)	Negative
Luteinising Hormone/Choriogonadotropin Receptor (LHCGR)	Negative
Aldoketo Reductase Family 1 Member C4 (AKR1C4)	Negative
Aldoketo Reductase Family 1 Member C2 (AKR1C2)	Negative
Mitogen Activated Protein Kinase 1 (MAP3K1)	Negative
Wingless-type MMTV Integration site family member 4 (WNT4)	Negative
Wilms Tumor 1 (WT1)	Negative
Zinc Finger Protein, Multitype 2 (ZFPM2)	Negative

Table 2: XomeDxSlice results of gene panel tested (GeneDx)

Effects of Growth Hormone Stimulation on the Immunologic Cellular Landscape in Pediatric Patients

Author Name(s): Jasmine Gujral, Brian A. Kidd, Hao-Chih Lee, Eddy A. Golden, Mabel Yau, Joel T. Dudley, Robert Rapaport

Department: Pediatrics; Genetics and Genomic Sciences

Division: Pediatric Endocrinology and Diabetes

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Institute for Next Generation Healthcare

Introduction: Multiple interactions exist between growth hormone (GH) and the immune system. Data about acute effects of GH on the immune system are lacking.

Hypothesis: GH is a stimulus to the immune system and GHD and GHS children respond differently to GH stimulation.

Methods: Prospective study in pediatric patients undergoing GH stimulation testing. Immunologic markers- complete blood count (CBC) and time of flight mass flow cytometry (CyTOF), were collected at the beginning (T0) and end (T3) of the test. Differences in patients by time point (T0 and T3) and GH response (GH sufficient {GHS} versus GHD) were calculated using a two-way ANOVA test.

Results: 22 out of 54 (39 boys, 15 girls, 5-18 years) had peak GH level <10 ng/ml (GHD). Absolute lymphocyte count, lymphocyte percentage, absolute eosinophil count, absolute monocyte count and platelet count demonstrated significant ($p<0.05$) decrease from T0 to T3. CyTOF analysis showed a decrease ($p<0.05$) from T0 to T3 in percentages of B cells, monocytes, plasmacytoid dendritic cells, T helper cells and a subset of cytotoxic T cells with low CD57 expression. Total white blood cell count, absolute neutrophil count and neutrophil percentage demonstrated increase ($P<0.05$) from T0 to T3, as did granulocyte percentage by CyTOF. No significant differences were found between CBC or CyTOF measurements and GH status.

Conclusions: The study provides the first high-resolution map of acute changes in the immune system with GH stimulation. CBC and CyTOF analyses showed significant changes. No significant differences were observed between the GHD and GHS groups.

Multi-omic analysis of acute response to growth hormone stimulation in pediatric patients

Author Name(s): Jasmine Gujral, Loukia N. Lili, Eddy A. Golden, Mabel Yau, Brian A. Kidd, Joel T. Dudley, Robert Rapaport

Department(s): Pediatrics, Genetics and Genomic Sciences

Division: Endocrinology and Diabetes,

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Institute for Next Generation Healthcare

Introduction: Although growth hormone (GH) has multiple complex interactions with the immune system, the acute effects of GH at the global molecular scale remain unknown. A multi-omic approach offers the opportunity to characterize these across the molecular landscape.

Hypothesis: GH is a stimulus to the immune system and GH deficient (GHD) and GH sufficient (GHS) children respond differently to GH stimulation.

Methods: Prospective study of pediatric patients with short stature undergoing a 3-hour GH stimulation test. Paired blood samples were obtained at T0 and at T3 for transcriptomics, proteomics and metabolomics. Molecular signatures were determined using a linear model adjusting for paired samples, response to GH stimulation, sex, BMI and batch (FDR <0.1). Integrative functional analysis was performed with the Gene Set Enrichment Analysis (GSEA) and MetaboAnalyst tools.

Results: 22 out of 54 (39 boys, 15 girls, 5-18 years) had peak GH level <10 ng/ml (GHD). 3,119 genes, 42 inflammatory proteins and 590 metabolites changed expression significantly with time, regardless of GH status. The majority of significant genes (98%) were upregulated, whereas the majority of significant metabolites (74%) were downregulated. Enriched pathways of significant genes and proteins included activated cytokine, chemokine, interleukin and interferon signaling, and metabolic pathways of cholesterol, fatty acid and insulin response. Concordantly, significant metabolites were enriched for amino acid and lipid metabolism.

Conclusions: This study provides a molecular systems overview of acute GH stimulation, a broad perspective on immune system activation, metabolic alterations and insulin response, and new avenues for differentiating between GHD and GHS patients.

Identifying critical windows of prenatal particulate air pollution exposure and children's blood pressure at age 4-6

Author Name(s): Gleicy M. Hair, Maria José Rosa, Allan C. Just, Itai Kloog, Katherine Svensson, Maria Luisa Pizano-Zarate, Ivan Pantic, Lourdes Schnaas, Marcela Tamayo-Ortiz, Andrea A. Baccarelli, Itai Kloog, Martha M. Tellez-Rojo, Robert O. Wright, Alison P. Sanders

Department: Department of Pediatrics/Department of Environmental Medicine and Public Health

Division: Nephrology/Environmental Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Exposure to air pollution is associated with increased blood pressure (BP) in adults and children. Limited evidence suggests that air pollution exposure during the prenatal period may contribute to adverse cardiorenal health later in life. Here we apply a distributed lag model (DLM) approach to identify potential critical windows that may underlie the association between prenatal particulate matter $\leq 2.5 \mu\text{m}$ in diameter (PM_{2.5}) and children's BP at 4-6 years.

Hypothesis: We hypothesized that children's exposure to PM_{2.5} during gestation are associated to a significant increase in BP later in life.

Methods: Participants included 537 mother-child dyads enrolled in the longitudinal cohort study Programming Research in Obesity, GRowth, Environment, and Social Stress (PROGRESS) based in Mexico City. Prenatal daily PM_{2.5} exposure was estimated using a validated satellite-based spatio-temporal model and BP was measured using the automated Spacelabs system with a sized cuff. We used DLMs to examine associations between daily PM_{2.5} exposure and systolic BP (SBP), taking in to account child's age, sex and BMI, as well as maternal education status and indoor smoking report during the second and third trimester.

Results: We found that PM_{2.5} exposure between weeks 17 to 35 of gestation (days 115 to 244) was associated with children's increased SBP at 4-6 years. A constant exposure to 10 $\mu\text{g}/\text{m}^3$ increase in PM_{2.5} sustained throughout this critical window resulted in a cumulative effect of 1.4 mmHg (CI:0.3,2.5) increase in SBP. In a stratified analysis by sex, this association persisted in boys but not in girls.

Conclusions: Our findings suggest that second and third trimester PM_{2.5} exposure may increase BP as early as 4-6 years of life. Long-term follow-up will increase our understanding of whether these subtle early life changes influence later life cardiorenal trajectories and risk for adult disease.

Genetic Testing in Children with Autism Spectrum Disorder

Author Name(s): Shatha Hajja, Samhita Jain, Jorge Revelo, Amanda Santos, Than Win, Bassam Al-Jomard, Larissa Polanco, Tania Lopez.

Department: Pediatrics

Division: Primary Care Department

Institution Affiliation: Icahn School of Medicine at Mount Sinai; Elmhurst Hospital Center

Background: Per AAP guidelines, genetic testing should be performed in all children with autistic spectrum disorder (ASD). Genetic screening includes specifically high-resolution chromosome studies (karyotype) and DNA analysis for Fragile X. However, in our Primary Pediatric Care clinic compliance to these guidelines was noted to be poor.

Goals: To bring awareness to all of our clinicians of the current AAP guidelines regarding genetic screening; To increase percentage of genetic screening done in all patients in our clinic with ASD.

Methods: We retrospectively reviewed 139 patient charts with ASD in PPC clinic looking for genetic screening tests specifically; chromosome analysis, Fragile X, high resolution chromosome microarray and genetics referral. Our intervention included an educational email sent to all Primary Pediatric Physicians in our Clinic, encompassing detailed steps regarding genetic screening/ referral and reinforcing its importance. We then assessed for compliance amongst our physicians to the AAP guidelines, pre and post intervention.

Results: In the pre-intervention cohort of children seen in our PPC clinic and diagnosed with Autism Spectrum Disorder, there were 24 females and 115 males, average age of the cohort was 4.9 years. In the post intervention cohort, there were 15 girls and 79 boys, average of the cohort was 5.7 years. Of the 139 charts reviewed in the pre-intervention period, genetic testing or referral was done for 37 (26.6%) patients. After the intervention, 50 (53%) of the 94 charts reviewed had documented Genetic testing or referral. The intervention of circulating an email with AAP guideline on Genetic testing for children with Autism Spectrum Disorder, increased the compliance to these guidelines by 26.4%.

Conclusions: Our compliance with AAP recommendations regarding genetic screening for ASD patients was sub optimal in the PPC clinic. After appropriate intervention of circulating an email, reinforcing the AAP guidelines on genetic screening of children with ASD, compliance had dramatically improved.

Keystones of Development Online Residency Curriculum: Weaving Attachment, Autonomy, and Executive Function into Well-Child Visits, Birth to Age 5

Author Names: Blair Hammond, Aliza Pressman, Mariel Benjamin, Carrie Quinn

Department: Pediatrics

Division: General Pediatrics, Developmental and Behavioral Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Mount Sinai Parenting Center

Introduction: Research has identified specific parenting behaviors that promote children's cognitive, social and emotional health. Per the AAP, pediatricians are supposed to counsel on these behaviors in primary care, but they may be less likely to do so without formal training during residency. An e-learning parenting curriculum would allow easy dissemination to multiple residency programs with little demand on faculty while providing a flexible learning experience for residents.

Objective: To assess the effectiveness and likeability of an online, animated curriculum which teaches residents to promote positive parenting behaviors that foster optimal child development during well-child visits.

Methods: Pediatric residents at 5 institutions completed the *Keystones of Development*, an online 13-module curriculum, in their behavior and development rotation during our pilot study year. We used a single-arm pre-posttest study design to assess intervention effect of self-reported behavioral outcomes (discussing, modeling, and praising positive parenting practices) and predictors of behavior (knowledge, perceived barriers and attitudes; and self-efficacy which was assessed retrospectively) with paired t-tests. Likeability and use were assessed at posttest.

Results: 50 pediatric residents (mean age=28.5yrs;77% female) participated. Within one-month post-intervention, there was a large statistically significant increase in behaviors that promote positive parenting, specifically through discussion ($p<0.01$; $d=1.09$) and praise ($p<0.05$; $d=0.77$). Significant changes in the predictor, perceived barriers, ($p<0.01$) was seen; knowledge and attitudes were not significant but changed in the desired direction. Self-efficacy to perform each respective behavior increased significantly ($p<0.01$). Residents liked the curriculum (4.2/5) and found it useful in practice (4.3/5).

Conclusions: The *Keystones of Development* online curriculum was well received by pediatric residents and resulted in increased discussion and praise of positive parenting behaviors during well-child visits. This study yields promising results that suggest benefit of a free, online curriculum to the field through wider dissemination, use, and refinement for other subgroups and contexts.

NYC Health Department Perinatal Hepatitis C Outreach Program Pilot

Author Name(s): Emily Harrison, Alison Whitney, Perminder Khosa, Angelica Bocour, Ann Winters

Department: Pediatrics

Division: Infectious Disease

Institutional Affiliation: Icahn School of Medicine at Mount Sinai; New York City Department of Health and Mental Hygiene

Introduction: Nationally, there has been an increase in hepatitis C (HCV) in reproductive-age women, raising concern for increased perinatal HCV exposure. The New York City (NYC) Health Department piloted an outreach program to link mothers with HCV to care and recommend screening of their children.

Hypothesis: Outreach to mothers with HCV will promote maternal linkage to care and screening of children.

Methods: Women reported to the Health Department with HCV were matched to mothers on birth certificates from January 2013–December 2015 and January–June 2018 to identify perinatally exposed infants; 40 women were randomly selected for outreach. Information on HCV testing and care, and social factors were obtained. Mothers were offered linkage to care and advised on pediatric screening via telephone.

Results: Thirty-five women were eligible for outreach. 31% were treated and 31% were linked to care but untreated. Half had a history of drug use. Half of pediatricians were aware of maternal HCV. 80% of women in the 2018 cohort were reached, compared to 20% in the 2015 cohort. From the 2018 cohort, 3 women were unaware of their HCV status and 2 women accepted linkage services. Pediatric screening recommendations were disseminated to 1 in the 2015 cohort and 10 in the 2018 cohort.

Conclusion: Outreach to women with recent deliveries was more successful than to those with remote deliveries. Outreach to ensure that women are aware of their HCV status, to provide linkage to care, and to encourage childhood testing can promote maternal HCV care and screening of children.

Current Use of Point-of-Care Ultrasound and Education in Pediatric Hospital Medicine Fellowship Programs in the United States

Author Names: Emily H. Hertzberg¹, Valeriy Chorny²

Department: Pediatrics

Division: Hospital Medicine; Ambulatory Care & General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai¹; SUNY Downstate Medical Center / Kings County Medical Center²

Introduction: Point-of-care ultrasound(POCUS) helps pediatricians care for children in a safe, efficient manner by reducing exposure to radiation and facilitating timely and accurate diagnoses. Little research has been done on the use of POCUS by Pediatric Hospitalists and training of Pediatric Hospital Medicine(PHM) fellows. As PHM fellowships are awaiting ACGME accreditation, we conducted a survey to assess fellowship directors' attitudes regarding POCUS.

Hypothesis: The goal is to understand the current practices in use and education of POCUS by PHM fellowship directors across the United States.

Methods: This is a cross-sectional IRB-exempt survey of all PHM fellowship programs across the US and Canada. The web-based survey was distributed by list-serve to all directors. The survey consisted of 19 multiple-choice and open-ended questions developed by the authors.

Results: A total of 31 fellowship directors responded to the survey(68%). Of the 31 programs represented, a majority of the programs consider themselves large, academic programs. Currently, 77% of programs do not train their fellows in POCUS. Of the programs that reported yes or not sure (86%) to interest in incorporating POCUS training, they were most interested in teaching procedural support for PIV(88%) and lumbar punctures(88%) followed by ultrasound evaluation of skin and soft tissue (84%), bladder (76%), and lung(64%).

Conclusions: Our survey demonstrated that most PHM fellowship programs do not currently train their fellows on POCUS and most hospitalists do not use POCUS. While there is significant interest from fellowship directors to include POCUS in training, there are a number of barriers identified.

Refined Carrier Frequencies of Citrin Deficiency in Various Populations

Author Name(s): Eri Imagawa, Luca Fierro, George A. Diaz and Kimihiko Oishi

Department: Genetics and Genomic Sciences

Division: Genetics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation:

Introduction: Citrin deficiency (CD) is an autosomal recessive urea cycle disorder. It is caused by variants in the *SLC25A13* gene that encodes a calcium-dependent mitochondrial aspartate/glutamate carrier. Previously, the majority of patients had been reported from Asian populations, particularly from Japan with an estimated carrier frequency of 1/69. Recently, an increasing number of CD cases have been reported from non-Asian countries, suggesting that CD is a pan-ethnic disease. To date, accurate carrier frequencies of CD have not been well characterized.

Hypothesis: We hypothesized that the frequency of CD may be variable between the populations.

Methods: We reviewed minor allele frequencies of pathogenic *SLC25A13* variants in samples of various populations using large-scale genomic databases including one private (BioMe at Mount Sinai Medical Center) and three publicly available databases.

Results: The estimated carrier frequency of CD in each population was 1/62-126 in Asian, 1/266 in Ashkenazi Jewish, 1/680-790 in European, 1/966-998 in Hispanic, 1/467-1,060 in African-American, and 1/411-1,458 in the others. Updated estimates of carrier frequency in the Japanese population were 1/35-42.

Conclusions: The frequencies of *SLC25A13* pathogenic variants are variable between the populations with a unique variant profile. The newly estimated CD carrier frequency in the Japanese population was higher than that of previously reported. It is possible that many individuals with CD are still undiagnosed in Japan. Ethnic background can be an important factor for diagnosing CD.

A New Pediatric Disease Cluster: High Frequency of Chiari Malformation Associated with Tethered Cord, Mast Cell Activation Syndrome, Ehlers-Danlos Syndrome, and Postural Orthostatic Tachycardia Syndrome

Author Name(s): Twana Jackson, Lincoln Ferguson, Paolo Bolognese, Anne Maitland, Warren Rosenfeld

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: South Nassau Communities Hospital, Icahn School of Medicine at Mount Sinai

Institute Affiliation: None

Introduction: We conducted a retrospective chart review following the observation that inpatients with CM-1 had a high incidence of several co-morbidities, specifically: Mast Cell Activation Syndrome (MCAS), Postural Orthostatic Tachycardia Syndrome (POTS), Ehlers-Danlos Syndrome (EDS) and Tethered Cord Syndrome (TCS).

Hypothesis: The presence of ≥ 3 comorbid conditions plus CM-I may represent a new clinical disease association, which we have termed MATCHED.

Methods: Children ≤ 18 years of age admitted to the Mount Sinai – South Nassau Medical Center, Chiari Center with a diagnosis of CM-I, from April 2016 through October 2018, were studied to determine the incidence of comorbidities in these patients.

Results: Fifteen patients met criteria for entry in our study. 10 patients in our cohort had CM-I and ≥ 3 associated conditions which met the criteria of our unique disease association. Patients ages ranged from 9-18 years and there was a 4:1 female predominance. EDS was present in 93.3% of patients, followed by POTS and MCAS which was present in 60% of our cohort, and lastly TCS was present in 46.7% of patients.

Patient	Sex	Age	CMI	Co-morbid Medical Conditions			
				TCS	EDS	POTS	MCAS
1	M	10	*	*	*	*	
2	F	12	*	*	*	*	
3	F	15	*	*	*		*
4	F	16	*	*	*	*	*
5	F	18	*				
6*	F	15	*		*	*	
7*	F	14	*	*	*		
8*	F	9	*		*		*
9	F	15	*		*		
10	F	18	*		*	*	*
11**	F	17	*	*	*		*
12**	M	15	*	*	*	*	*
13**	F	15	*		*	*	*
14**	M	12	*		*	*	*
15	F	17	*		*	*	*

Co-morbid Medical Conditions:
Chiari Type I Malformation (CM-I), Tethered Cord Syndrome (TCS), Ehlers-Danlos Syndrome (EDS), Postural Orthostatic Tachycardia Syndrome (POTS) Mast Cell Activation Syndrome (MCAS)

Familial cohorts:
* Three females with a familial association with 3 of the following: CM-I, POTS, EDS, and Syringomyelia
** Two males and two females, who also met MATCHED criteria

Conclusions: CM-I patients demonstrated a high incidence of co-morbidities. In our cohort of 15 patients, 67% of patients had ≥ 3 conditions plus CM-I and met the criteria for our proposed clinical disease association; which we have termed MATCHED (M = MCAS; A = autonomic dysfunction i.e. POTS; T = TCS; CH = CM-I; ED = EDS). Further investigation of CM-1 patients with MATCHED may be helpful in defining this association and understanding the underlying etiology.

Lower Airway *Pneumocystis jirovecii* and Severe Asthma in Childhood

Author Names: Megan N. Januska, Victoria Ribeiro, Alfin G. Vicencio

Department: Pediatrics

Division: Pulmonology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: Asthma is one of the most common chronic diseases of childhood. The diagnosis of asthma, however, encompasses a heterogeneous group of physiologically distinct diseases, many of which we are still learning. We previously noted increased representation of *Pneumocystis jirovecii*, an atypical fungus not previously associated with asthma, in the lower airways of children with severe asthma. Given the novelty of this finding as well as its implications for tailored asthma therapy, we sought to validate our previous findings using a commercially-available assay.

Hypothesis: We hypothesized that children with severe asthma have an increased representation of *Pneumocystis jirovecii* in their lower airways.

Methods: We analyzed concentrated bronchoalveolar lavage (BAL) fluid samples collected during clinically-indicated flexible bronchoscopy for the presence of *Pneumocystis jirovecii* via direct fluorescent antibody (DFA) and quantitative polymerase chain reaction (qPCR) testing.

Results: We tested concentrated BAL fluid samples from 27 children, including 18 children with severe asthma, three children with mild to moderate asthma, three children with airway autoimmune inflammatory response (AAIR) syndrome, and one child each with habit cough, bronchomalacia, and bronchiectasis, for the presence of *Pneumocystis jirovecii*. Four children with severe asthma tested positive for *Pneumocystis jirovecii* (22.2%), while none of the other children did ($p=0.27$).

Conclusions: Children with severe asthma have an increased representation of *Pneumocystis jirovecii* in their lower airways as demonstrated through clinically applicable assays. Future studies will focus on the role of such point-of-care testing in the therapeutic management of children with severe asthma.

De Novo Crohn's Disease of the Pouch in Children Undergoing Ileal Pouch-Anal Anastomosis for Ulcerative Colitis: A Tertiary Care Inflammatory Bowel Disease Center Experience

Author Name(s): Lauren A. Jarchin, Elizabeth A. Spencer, Sergey Khaitov, Alexander Greenstein, Jacqueline Jossen, Joanne Lai, David Dunkin, Nanci Pittman, Keith Benkov, Marla Dubinsky

Department: Pediatrics

Division: Gastroenterology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Approximately 10% of children undergo colectomy with ileal pouch-anal anastomosis (IPAA) for ulcerative colitis (UC). We aimed to describe the post-operative outcomes, with an emphasis on chronic pouch inflammation including de novo Crohn's disease (CD) at a tertiary care inflammatory bowel disease center.

Hypothesis: We hypothesized that an underestimated number of children diagnosed with ulcerative colitis develop Crohn's disease after IPAA.

Methods: Electronic medical records of all children who underwent colectomy ≤ 18 years between 2008-2017 were reviewed. Clinical and laboratory data were recorded. Primary outcome was frequency of chronic pouch inflammation including de novo CD. Secondary outcomes included early (≤ 30 days from index surgery) and late (> 30 day) post-operative complications. Descriptive statistics summarized the data and univariate analysis tested associations with outcomes.

Results: Fifty-eight underwent colectomy and 56 completed IPAA. Median age at diagnosis was 14 years [12-16.2] and at colectomy 16.2 years [14.2-17.7] with median follow-up of 13 months [5-43]. 78% were biologic exposed. Eleven had chronic pouchitis, 73% antibiotic refractory and 25% met criteria for de novo CD by median of 19 months [9-41]. 21% and 50% experienced early and late surgical complications, most commonly ileus and recurrent IPAA stricture. The pouch failure rate was 3.6%. Chronic pouch inflammation was associated with a later diagnosis of de novo CD ($P=0.0025$).

Conclusions: In pediatric UC, CD is not uncommon after IPAA. Chronic pouch inflammation often precedes a diagnosis of de novo CD. Families should be informed of the short and long-term outcomes in children prior to UC surgery.

Case report of Ovotesticular 46,XY Disorders of Sex Development: Preliminary Diagnosis and Gender Dysphoria in Young Adulthood

Author Names: Gregory G. Jebian B., Mabel Yau, Mithra L. Narasimhan, Anne Macdonald, Lakshmi Mehta, Heino Meyer-Bahlburg, Maria I. New

Department: Pediatrics

Division: Endocrinology - Adrenal Steroid Disorders

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Disorders of Sex Development (DSD) comprise a spectrum of conditions resulting in genetic-gonadal discordance. Ovotesticular DSD (OT-DSD) is defined by the presence of both ovarian and testicular tissue. Herein we report a 26 year old with presumed OT-DSD, born with ambiguous genitalia and assigned female at birth. Patient underwent clitorrectomy at 3 months old and gonadectomy at 9 months old; histology reportedly showed one testis and one ovary. Patient did not receive hormone replacement therapy prior to presenting to our clinic.

Methods: Hormone serum levels, genetic karyotyping, array comparative genomic hybridization, pelvic sonography, psychological, psychiatrist and developmental psychoendocrinologic evaluations.

Results: The patient was found to have no uterus or ovaries on ultrasound and endocrine laboratories consistent with bilateral gonadectomy and normal adrenal function. Normal male karyotype (46,XY, *SRYx1*, *DYZx1*) was found. Microarray testing was negative for mutations in genes related to 46,XY DSD including *AR*, *DHH*, *MAP3K1*, *NR0B1*, *NR5A1*, *SRY*, *SRD5A1* and *WT1*. Psychoendocrinologic evaluation was consistent with Gender Dysphoria, Depression and ADHD.

Conclusions: The genetic etiology of OT-DSD remains unknown for a large number of patients and many DSD-causative genes remain to be identified. Early bilateral gonadectomy made hormonal evaluation ineffective, hence genetic testing became the next diagnostic recourse. In the absence of known variants, whole genome sequencing is required to understand the pathophysiology. Testosterone replacement, necessary for bone protection and psychosexual development, was delayed while the patient identified as “agender”.

**Queer Medical Pipeline:
A Model for Promoting Careers in Healthcare for Urban LGBTQ Youth**

Author Name(s): Scott K. Jelinek, Don Nguyen, Barbara Warren

Department: Pediatrics; Office for Diversity and Inclusion

Division: LGBT Programs and Policies

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: There is an underrepresentation of LGBTQ healthcare providers in the medical workforce and consequently, a limited number of LGBTQ identifying mentors to offer support and guidance.

Hypothesis: A pipeline program that exposes LGBTQ high school-aged youth in the New York City area to careers in medicine and healthcare would increase the number of LGBTQ youth interested in entering the medical work force.

Methods: The Icahn School of Medicine at Mount Sinai created LGBT YQUTH MED (Young Queer Urban Teens for Health in MEDicine), a pipeline program with structured science and medicine-related enrichment experiences, including interactive outreach presentations, “Saturdays at Sinai” immersion experiences, workforce development, and health-related internship opportunities.

Results: A total of 51 participants experienced the interactive outreach presentations and six students attended the Saturday at Sinai event. The majority of the youth found the program interesting and engaging, rating it an average 4.73 out of 5 on the Likert scale. When asked how helpful it was to hear other LGBTQ student's personal stories, the response was 4.76 out of 5, and most of the youth thought it was very important to have an LGBTQ role model/mentor, giving it a rating of 4.52 out of 5. The youth also responded they were very much interested in a career in medicine and public health after participating in the program, responding on average 4.04 out of 5.

Conclusions: The pipeline program provided visibility, mentorship and guidance necessary to encourage LGBTQ youth to pursue careers in medicine and science and increase the representation of queer healthcare providers in the medical workforce.

Oral Health in Pediatric Oncology Patients: A Survey Aiming at Quality Improvement

Author Name (s): Shikha Kanotra, Laurence Hyacinthe

Department: Dentistry

Division: Pediatric Dentistry

Institution Affiliation: Icahn School Of Medicine at Mount Sinai

Introduction: It is estimated that 160,000 children worldwide are diagnosed with cancer every year (WHO, 2009). Children continue to experience adverse effects of their cancer five years after their diagnosis and treatment (Santacroce et al., 2010).

Most pediatric Oncology patients are treated with a combination of chemotherapy and radiation therapy. Both the therapies individually or in combination have shown to affect oral mucosa and leading to various side effects including but not limited to mucositis, alteration in taste to more serious side effects of dental decay due to dry mouth. Such symptoms can lead to decreased oral intake due to inability to eat, weight loss, low self-esteem, poor performance in school, frequent absences from school and thereby causing psychological dysfunction in the long run.

It is plausible that educating patients and early recognition of these side effects and involvement of an oral health care provider can improve outcomes. The survey will help determine what measures are being taken by the Pediatric Hematologists and Oncologists for oral care needs of such patient. This survey will also determine the importance of having a pediatric dentist in the team for care for pediatric oncology patients.

Hypothesis: We hypothesize that pediatric oncology patient will benefit by having a pediatric dentist in the team for their comprehensive care.

Method: A 10 question quality of life survey (see below) to be sent out to Pediatric Hematology-Oncology and Pediatricians at Mount Sinai Health System. Once the data has been collected it will be analyzed to evaluate the degree of awareness amongst Pediatric Hematologists- Oncologists and Pediatricians and measures taken by them for these patients. Statistical analyses will comprise of descriptive statistics, proportions, median and mean values. The data would be used to develop action plans to provide education to Pediatric Hematologists - Oncologists and Pediatricians, build tools for patient education and empower patients and providers to be able to request and seek early oral health care measures.

Results: Pending

Conclusion: Pending

Exploring Interventions: Managing Compassion Fatigue (CF) And Burnout (BO) In Pediatric Subspecialty Providers

Author Name(s): Samuel M. Kase, Jeanie L. Gribben, Elisha D. Waldman, Andrea S. Weintraub

Department: Pediatrics

Division: Newborn Medicine

Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: n/a

Introduction: While it is essential for physicians to actively engage in self-care activities outside the workplace, it is of equal importance for institutions to offer structured support for physician well-being.

Hypothesis: To compare institutional wellness offerings to offset CF and BO with (1) real-time physician participation in such programming and (2) with what physicians themselves wish could be implemented.

Methods: A survey of personal and professional characteristics was distributed to pediatric subspecialty providers nationally. Descriptive statistics, bivariate analyses using chi square tests or independent t-tests, multivariable logistic regression modeling, and qualitative content analysis were performed using survey responses.

Results: Almost 60% of respondents participated in some institutional offering for self-care. Participants' "wish lists" for supports to improve well-being were coded into 4 categories: social/emotional support (SES), improved leadership/mentorship (LM), organizational change (ORG), and improved physical work environment (PHYS). Spiritual practice, "sympathetic/empathic" personality trait, and junior faculty level were each significant predictors of desire for SES ($p<0.03$). Desire for improved LM was significantly associated with current use of "debriefs" ($p=0.015$) and/or Schwartz Rounds ($p=0.033$). Spiritual practice and "conscientious/in control" personality trait were independent predictors of desire for ORG ($p<0.05$). There were no significant associations with desire for PHYS.

Conclusions: While there is universal need for wellness offerings to offset CF and BO, a "one-size-fits-all" approach is unlikely to succeed for pediatric subspecialists. Lack of awareness of what providers truly want and barriers to participation perpetuate underutilized interventions. As pediatric leadership design new wellness initiatives, "individualized wellness plans" may well be warranted.

The Efficacy and Stability of Food Allergy Herbal Formula 2 and Its' Extracts Over Time

Author Name(s): Monica Kim, Kamal Srivastava, Nan Yang, Xiu-Min Li, David Dunkin

Department: Pediatrics

Division: Gastroenterology and Nutrition

Institution Affiliation: Icahn School of Medicine at Mount Sinai; New York Medical College

Institute Affiliation: Mindich Child Health and Development Institute

Introduction: Atopic and immune-mediated diseases including food allergies and inflammatory bowel diseases are increasing in incidence in the US and current available therapies are limited. Food allergy herbal formula 2 (FAHF-2) and its' extracts have shown safety and efficacy in reducing allergic symptoms and abrogating colitis through a series of experiments as well as high safety and immunomodulatory effects in clinical trials. Therefore, we decided to study the efficacy and stability of FAHF-2 and its' refined products between different batches as well as over time.

Hypothesis: We hypothesized that FAHF-2, B-FAHF-2, and E-B-FAHF-2 will stay consistent in terms of their components and effects on cytokine, antibody and histamine release from cells after treatment between different batches and as time elapses.

Methods: We identified compounds in each formula through high performance liquid chromatography (HPLC) and utilized enzyme-linked immunosorbent assays (ELISA) to measure levels of TNF- α and IgE released from treated RAW cells (mouse macrophage cell line) and U266 cells (human lymphocyte cell line), respectively.

Results: HPLC showed that the active compounds in the various batches of FAHF-2 consist of the same compounds at similar retention times. In addition, RAW cells have shown similar dose-dependent responses to treatment with B-FAHF-2 and different batches of FAHF-2 and E-B-FAHF-2 as time has elapsed.

Conclusions: For the first time, we reported the longest stability of FAHF-2 products. The active compounds between batches of FAHF-2 manufactured at different time points are consistent, along with their biological effects on TNF- α release in RAW cells after treatment. Studies are being continued on the effects of B-FAHF-2 on TNF- α release in RAW cells and are also being conducted on the effects of FAHF-2 and its' extracts on IgE release in U266 cells as well as histamine release in a different cell line.

Laboratory Screening for the Newly Arrived Immigrant Child

Author names: Cassandra Koid-Jia-Shin, Mirtha G. Gayoso-Liviac, Juan C. Fierro, Rasmita Budhathoki, Thach Nguyen, April W. J. Tan, Ian Gampon, Daniel Ibanez, Edward Forman, Joselito Sanchez, Christina Cuccia.

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai, Elmhurst Hospital.

Introduction: Our outpatient clinic population comprises a significant number of immigrant children from diverse backgrounds. It is imperative that our providers are familiar with the current recommendations in caring for these children.

Hypothesis: A quality improvement project can improve laboratory screening for the newly arrived immigrant child at their initial health maintenance visit.

Methods: Baseline data was collected for this cross-sectional study involving 50 children from birth to age 12 years between October 2017 and December 2017 for their initial health maintenance visit. We instructed physicians on the Immigrant Health Tool Kit laboratory screening recommendations. A table including standard and optional laboratory screening based on existing recommendations was developed and distributed. A lab favorite list was created and shared on the electronic records. Post-intervention chart review was performed in 50 patients.

Results: A total of 100 children were included, who represented a total of 24 countries with a majority of children from Colombia (24%). After comparison of pre and post intervention, adherence to ordering screening labs increased for the following: Lead (96% vs 98%, $p=0.562$) repeat lead (0% vs 43%; $p<0.001$), Hepatitis BsAg (66% vs 80%; $p=0.117$), TB screening (72% vs 96%; $p<0.0008$), TSH (0% vs 43%; $p=0.0114$), HIV (0% vs 71%; $p<0.00001$) and Syphilis (0 vs 60%; $p<0.00001$).

Conclusions: In a short period of time our QI project improved compliance with the recommendations of the AAP Immigrant Health Toolkit for appropriate laboratory screening for the newly arrived immigrant child.

Physician and Parent Comfort, Awareness, Barriers, and Implementation of the Guidelines for the Prevention of Peanut Allergy

Author Names: Michelle Lai, Scott H. Sicherer

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Jaffe Food Allergy Institute

Introduction: Utilization of the NIAID-sponsored Expert Panel Guidelines for preventing peanut allergy is understudied, so we explored stakeholder views.

Hypothesis: We hypothesized that stakeholder implementation of guidelines would be low.

Methods: Surveys were administered to a convenience sample of physicians (50 [90% pediatricians]) and parents (100) of infants under age 1 year in multiple practice settings in NYC. Surveys described the guidelines.

Results: Most physicians (84%) were guideline-aware, most commonly via colleagues and the AAP newsletter. Physicians' mean comfort was 4.1 (5-point Likert scale, very comfortable to very uncomfortable). Among Guideline-aware physicians, 60% followed them as written; the remainder modified the approach regarding testing, referrals, introduction time, or patient selection. The greatest physician-perceived implementation barriers were parental acceptance (60%), fear of giving peanut early (46%), and access to allergists (24%). Physicians identified patient handouts (78%) and more infant-safe forms of peanut (52%) as needed resources. For parents, 58% were Guideline-aware, and 90% indicated comfort with early introduction. Pediatricians, internet and friends were the most common sources of information for parents. The greatest parent-identified barriers were fear of reaction (36%), choking (11%), and lack of infant-safe forms (6%). Parents identified a need for more physician advice (44%), brochures (24%) and allergist access (18%).

Conclusions: Guideline awareness and comfort was high among physicians in this cohort, but many modified the approach. Physicians perceived parental acceptance as a major barrier, yet almost all parents reported comfort. Physician advice and written materials were highlighted as needed resources.

Effect of Glucocorticoid Therapy on Fertility in Females with Non Classical Steroid 21-hydroxylase Deficiency

Author Names: Mithra Lakshmi Narasimhan, Mabel Yau, Gregory Jebian, Anne Macdonald, Maria I New

Department: Pediatrics

Division: Endocrinology /Adrenal Steroid Disorders

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Congenital Adrenal Hyperplasia (CAH) due to 21-hydroxylase deficiency is one of the most common monogenic autosomal recessive endocrine disorder. The nonclassical form of CAH (NCCAH) is associated with a mild deficiency of the 21 hydroxylase enzyme. Females with NCCAH present with hyperandrogenic symptoms such as acne, hirsutism, hair loss, early pubarche, menstrual irregularities and infertility. The excess male hormones cause disruption of hypothalamo-pituitary-ovarian axis thereby interfering with the process of ovulation. Here, we evaluate the efficacy of glucocorticoid treatment in improving the fertility in NCCAH females in our clinic population who had symptoms of hyperandrogenemia.

Hypothesis: We hypothesized that females with NCCAH had improved fertility on treatment with glucocorticoids.

Methods: We evaluated the fertility status of patients by reviewing their charts retrospectively. We defined infertility as failure to conceive even after 2 years of having unprotected sexual intercourse. The hormonal panel includes serum concentrations of the following hormones: 17 hydroxyprogesterone (17-OHP), androstenedione, testosterone, FSH and LH. Serum 17-OHP and androstenedione were measured using tandem mass spectrometry and serum testosterone and FSH and LH were measured using electrochemiluminescence immunoassay.

Results: We found improved fertility in the treated group (74%) when compared to the untreated group (66%).

Conclusion: Females with Nonclassical 21-hydroxylase deficiency show improved fertility when treated with glucocorticoids in contrast to the untreated group. This suggests the efficacy of glucocorticoid therapy in improving the fertility by lowering the serum androgen concentration in females with NCCAH.

Integrative Analysis of Methylation Quantitative Trait Loci (meQTLs) in Human Placenta

Author Name(s): Corina Lesseur¹, Maya Deyssenroth¹, Shouneng Peng², Ke Hao², Carmen J. Marsit³, Jia Chen¹

Departments:

1. Department of Environmental Medicine and Public Health, Icahn School of Medicine at Mount Sinai, New York, NY,
2. Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY.
3. Department of Environmental Health, Emory University, Atlanta, GA.

Institution Affiliation: Icahn School of Medicine at Mount Sinai; Emory University

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Inter-individual variation in DNA methylation has been related to age, sex, environmental exposures, and inherited genetic variants such as single nucleotide polymorphisms (SNPs). The influence of individual's genotype on DNA methylation is known as methylation quantitative trait loci (meQTL). The placenta is a crucial organ not only for fetal growth and development but also for fetal programming of adult metabolic disease.

Hypothesis: We hypothesized that meQTLs effects are widespread in the placental methylome and relate to genetic variants associated to weight-related phenotypes through GWAS.

Methods: Using genome-wide DNA methylation (Infinium MethylationEPIC BeadChip BeadArray) and SNP genotypes (MegaEX array), we performed a genome-wide meQTL analysis in 158 placentas from the Rhode Island Child Health Study (RICHS).

Results: We identified 1,264,466 meQTLs (SNP-CpG pairs, FDR 5%); overall 17% (120,754) of the interrogated CpG methylation sites (703,330) were influenced by SNPs. The majority (~97%) of these placental meQTLs related to cis-acting SNPs (117,177 CpGs), whereas only 3% are trans-meQTLs (3,577 CpGs). SNPs that influence placental DNA methylation (meSNPs) are enriched for placental expression quantitative-trait loci (eQTL) eSNPs from the same individuals (enrichment OR= 14.5, $P < 2e-16$) with 154,208 SNPs influencing both methylation and gene expression. meSNPs are also enriched for variants associated with birth weight (OR= 3.5, $P = 6.4 \times 10^{-201}$), childhood obesity (OR= 6, $P = 9.5 \times 10^{-40}$) and childhood BMI (OR= 4.13, $P = 3 \times 10^{-52}$) through GWAS studies.

Conclusions: An important amount of genetic variants influence placental DNA methylation, suggesting a significant heritable component; methylation at these sites could play a role in later-life metabolic diseases.

Prevalence of Smoking Cessation Counseling Conducted by Pediatric Dentists at the Institute of Family Health of Harlem

Author Name(s): Wing Yin Maggie Leung; Laurence Hyacinthe; John Pfail.

Department(s): Dentistry

Division(s): Pediatric Dentistry

Institute Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Institute of Family Health of Harlem

Introduction: According to the National Youth Tobacco Survey published in 2014 by the Centers for Disease Control and Prevention, one in every four high school students is current tobacco user and one in every thirteen middle school students has used tobacco products. Concurrently, there is also a rapid increase in electronic cigarettes use among this adolescent population. Given the alarming new trend, proper awareness and smoking cessation counseling programs should be implemented in order to educate and disseminate accurate information to these youths. According to the guideline set forth by the American Academy of Pediatric Dentistry, oral health professionals should document patients' tobacco use status, as well as to provide prevention and smoking cessation services to children related to tobacco use and nicotine addiction. The goal of this study is to evaluate the prevalence of smoking cessation counseling conducted by pediatric dentists at the Institute of Family Health of Harlem

Hypothesis: I hypothesize that the majority of the pediatric population being served at the Institute of Family Health of Harlem are not being counseled for smoking cessation during their dental visits.

Methods: All charts at the Institute of Family Health between July 2017 and June 2018 will be reviewed. Data will be collected from Dentrix—a dental software used by pediatric dentists at the Institute of Family Health of Harlem. Patients between the age of ten and eighteen at the time of their dental appointment will be included in the study. Comparisons will be performed between two groups—middle school students between the age of ten and thirteen and high school students between the age of fourteen and eighteen. By using a keyword mining approach, the following seven keywords—‘smoking’, ‘vaping’, ‘cigarettes’, ‘e-cigarettes’, ‘e-cig’, ‘juul’ and ‘tobacco’—will be searched in each charts. Quantitative analysis will be performed after chart review. Based on the result, the percentage of pediatric dental appointments that have address smoking will be calculated; the number of smoking cessation referrals that were provided will be tallied and subsequent visits that address smoking cessation after a patient verbalized that he/she is an active/former smoker will be evaluated.

Results: Pending

Conclusions: Pending

Prenatal Cortisol Modifies the Association between Maternal Lifetime Traumatic Stress and Body Mass Index in 6 Month-Olds

Author Names: Lianna R. Lipton¹, Kelly J. Brunst², Julie D. Flom¹, Farida Nentin¹, Nastasia Pollas¹, Brent A. Coull³, Michelle Bosquet Enlow⁴, Rosalind J. Wright¹

Department: Pediatrics; Environmental Medicine & Public Health

Institution Affiliations: Icahn School of Medicine at Mount Sinai¹; University of Cincinnati College of Medicine²; Harvard School of Public Health³; Harvard Medical School⁴

Institute Affiliations: Institute for Exposomic Research

Introduction: Recent evidence underscores the need to consider complex interactions among maternal trauma, disrupted cortisol production, and fetal sex to elucidate intergenerational health effects of maternal traumatic stress.

Hypotheses: We examined associations between maternal trauma and infant body mass index (BMI) in 374 mother-child dyads. We hypothesized that children born to women with greater trauma and higher cortisol production would have higher BMIs with males being more vulnerable.

Methods: Pregnant women completed the Life Stressor Checklist-Revised to ascertain lifetime traumatic events. Cortisol secretion over the third trimester was determined using maternal hair. Infant length and weight were measured using a standardized protocol. BMI z-scores were calculated using WHO growth charts. Multivariable regression was used to assess associations between maternal trauma and child BMI adjusting for maternal age, education, race/ethnicity, and prenatal cortisol, and child sex and birthweight. Interaction terms were used to assess effect modification by cortisol exposure and child sex.

Results: Women were primarily minorities (38% Hispanic, 31% black). Lifetime trauma was not independently associated with infant BMI ($\beta = -0.1$, $p = 0.23$). Children born to mothers reporting greater trauma and higher prenatal cortisol had higher BMI z-scores ($\beta_{\text{int}} = 0.186$, $p_{\text{int}} = 0.045$). A 3-way interaction with maternal trauma, prenatal cortisol, and child sex approached significance ($p_{\text{int}} = 0.091$) with boys being most impacted.

Conclusions: These findings are relevant given the link between higher weight-for-length in infancy and later obesity. Analyses will be extended to consider associations among these factors and body composition trajectories as we follow these children prospectively.

Risk Factors and Behaviors of Adolescent E-Cigarette in NYC

Author Name(s): Yuki Low¹, Elizabeth Garland²

Department: ¹ Bronx High School of Science; ² Environmental Medicine and Public Health

Division: Preventive Medicine and Community Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: none

Introduction: Although, adolescent combustible smoking rates are decreasing, adolescent use of electronic cigarettes (e-cigarettes) is increasing. E-cigarettes contain nicotine and volatile chemicals that cause cognitive impairment and addiction in adolescents. There are no studies actually looking at the trends in adolescent e-cigarettes and the risk factors for initiation within NYC.

Hypothesis: There are many risk factors associated with e-cigarette use in NYC adolescents.

Methods: 2015 data from the Youth Risk Behavior Surveillance System (YRBS) was compared to information on 2017 NYC Department of Health and Mental Hygiene Epi-query portal. Socio-demographics and e-cigarette usage was analyzed using SPSS for multivariate analysis.

Results: In YRBS 19,000 (14.8%) males and 22,000 (16.9%) females used e-cigarettes in the past 30 days compared to 24,000 (18.3%) and 20,000 (15.5%) in NYC. White non-Latino and Latino students were more likely than Asian and Black to self-report usage. Factors associated with e-cigarette usage were: cigarette use, marijuana use, binge drinking, unstable housing situation (kicked out, ran away or abandoned), e-bullying, depression, carrying a weapon, less than 8 hours of sleep and sexual intercourse. Trends for NYC are increasing compared to national data.

Conclusions: Results from this study are similar to the literature. It is clear that adolescents with high-risk behaviors cluster together. Future interventions can target adolescents who may engage in these risk behaviors.

Fluoride Exposure and Kidney and Liver Function among Adolescents in the United States: NHANES, 2013-2016

Author Name(s): Ashley J. Malin, Corina Lesseur, Stefanie A. Busgang, Paul Curtin, Robert O. Wright, Alison P. Sanders

Departments: Environmental Medicine and Public Health; Pediatrics

Division: Environmental Health/Nephrology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Exposomics, MCHDI

Introduction: Hepato- and nephrotoxicity of fluoride have been demonstrated in animals, but few studies have examined the potential for these effects in humans. Therefore, we conducted the first study to examine the relationship between fluoride exposure and kidney and liver parameters among adolescents in the United States (U.S.).

Hypothesis: We hypothesized that greater fluoride exposure would be associated with altered kidney and liver parameters in this population.

Methods: There were 1983 and 1742 adolescents in NHANES 2013-2016 who had plasma and household tap water fluoride concentration measurements respectively, as well as serum and urinary kidney and liver parameter measurements. We employed survey-weighted linear regression to examine relationships between fluoride exposure and kidney and liver parameters after covariate adjustment. A Holm-Bonferroni correction accounted for multiple comparisons.

Results: A 1 $\mu\text{mol/L}$ increase in plasma fluoride was associated with a 10.36 mL/min/1.73 m² lower estimated glomerular filtration rate (95% CI: -15.93, -4.79; $p=0.002$), a 0.29 mg/dL higher serum uric acid level (95% CI: 0.08, 0.50; $p=0.04$), and a 1.29 mg/dL lower blood urea nitrogen level (95% CI: -1.87, -0.70; $p<0.001$). A 1 mg/L increase in water fluoride was associated with a 0.93 mg/dL lower blood urea nitrogen level (95% CI: -1.39, -0.47; $p=0.001$) and a 0.06 g/dL lower serum albumin level (95% CI: -0.10, -0.02; $p=0.05$).

Conclusions: Fluoride exposure may contribute to complex changes in kidney and liver related parameters among U.S. adolescents. Additionally, poorer kidney or liver function may contribute to increased bodily fluoride absorption.

Type I Interferon Dysregulation in Down Syndrome

Author Name(s): Louise Malle, Dusan Bogunovic

Department(s): Department of Microbiology

Division(s): Microbiology, Immunology

Institution Affiliation(s): ISMMS

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Down syndrome (DS) is the most common genetic cause of intellectual and developmental disabilities in children and young adults. DS individuals also present with increased susceptibility to skin inflammation, autoimmune features and basal ganglia calcifications – these features are reminiscent of patients of other diseases caused by chronic exposure to Type I IFN, the so-called “Type I Interferonopathies.” Interestingly, the genes encoding the Type I IFN receptor, IFNAR1 and IFNAR2, are located on Chromosome 21, suggestive of possible gene dosage effects in DS.

Hypothesis: Increased signaling caused by carrying additional copies of IFNAR1 and IFNAR2 in DS individuals alters IFN-I responsiveness, contributing to DS pathology.

Methods: We studied the responsiveness of DS and healthy control (HC)-derived fibroblasts to Type I IFN stimulation via measurement of STAT phosphorylation in vitro. We also performed ex vivo studies on DS and HC whole blood, performing deep immunophenotyping via CyTOF technology and assessing the response of DS and HC immune cells to IFN stimulation and JAK inhibitors.

Results: We found that DS patient cells have increased sensitivity to Type I IFN stimulation compared to HCs.

Conclusions: Our preliminary data indicates that Type I IFN might play a role in DS pathology, which provides therapeutic potential to be explored in future studies.

Vertical Rotation of the Orbital Bandeau in Unilateral Coronal Synostosis

Author Names: Ilana G. Margulies, Paymon Sanati-Mehrzy, Pedram Goel, Peter M. Som, Mark M. Urata, Peter J. Taub

Department: Surgery, Radiology

Division: Plastic and Reconstructive Surgery

Institution Affiliations: Icahn School of Medicine at Mount Sinai, Keck School of Medicine at the University of Southern California

Introduction: Unilateral coronal synostosis (UCS) results in anterior plagiocephaly characterized by many dysmorphic changes; however, rotation of the ipsilateral orbital bar along the horizontal axis has not been investigated. Thus, the authors sought to characterize the rotation of the ipsilateral orbital bar in UCS.

Hypothesis: The ipsilateral orbital bar in UCS patients will be rotated along the horizontal axis.

Methods: Craniometric analysis was performed on CT images of 35 non-syndromic UCS patients (0-18 months) and 16 control patients (0-24 months, 32 orbits).

Results: The angles of the supraorbital bar to 0° vertical and to the floor of the middle cranial fossa were higher in UCS vs. control patients in the majority of age matched groups ($p < 0.05$). While the angle of the supraorbital bar to the orbital roof and the angle of the posterior aspect of the orbit to 0° horizontal were not different in UCS vs. control patients ($p > 0.05$), the angle of the orbital roof to 0° horizontal was lower in UCS vs. control patients in all age matched groups ($p < 0.05$).

Conclusions: Rotation of the supraorbital bar around the horizontal axis in UCS is confirmed by craniometric analysis. The concurrent flattening of the orbital roof with no change in the angulation of the supraorbital margin or posterior aspect of the orbit suggests that the apex of this rotation possibly lies at the posterior orbital roof. The novel characterization and quantification of this deformity will better inform the operative approach and enable a more accurate surgical correction.

3 Cases of Peanut-related Food Protein-induced Enterocolitis Syndrome; Will Early Introduction Lead to More?

Author Names: Joao Pedro Lopes, Jacob Kattan

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Jaffe Food Allergy Institute

Introduction: Early dietary introduction of peanut has been shown to reduce the frequency of development of peanut allergy in high risk infants. Families have been recommended to add peanut to the diet early in life. While peanut has not been reported as a common trigger of food protein-induced enterocolitis syndrome (FPIES), it is possible that earlier introduction could lead to an increase in peanut-related FPIES (prFPIES). We report 3 cases of prFPIES seen in the past 8 months.

Hypothesis: Earlier introduction could lead to an increase in prFPIES.

Methods: Retrospective chart review of 3 patients with dietary peanut introduction before 8 months of age, diagnosed with prFPIES.

Results: Case 1: 5-month-old male, who developed repetitive vomiting 2-3 hours after his second and third peanut ingestions, with positive SPT for peanut (6mm). Case 2: 6-month-old female who, on day 2 of peanut introduction, started vomiting 90 minutes after ingestion. Skin prick test (SPT) was negative to peanut. Case 3: 7-month-old female who became pale, lethargic, and vomited two hours after peanut introduction. SPT and serum immunoglobulin E were negative. A peanut oral food challenge (OFC) was scheduled (it was unclear if the trigger was peanut butter or the wheat bread it was spread on), and 2 hours after peanut intake she started vomiting.

Conclusions: Although prFPIES has been described, peanut is not a common trigger of FPIES. These 3 cases of prFPIES in our practice raise the question of whether early dietary peanut introduction will increase the incidence of prFPIES.

Prevalence and Risk Factors of Polycystic Ovarian Syndrome Among an Ethnically Diverse Overweight/Obese Adolescent Population

Author Name(s): Jacqueline Maya, Janet Siegel, Tina Q. Cheng, Thaina Rousseau-Pierre

Department(s): Pediatrics

Division(s): Adolescent Medicine

Institution Affiliation(s): Icahn School of Medicine at Mount Sinai; Elmhurst Hospital Center Adolescent Clinic

Introduction: More remains to be known about Polycystic Ovary Syndrome (PCOS) among overweight/obese adolescents across different ethnicity and its association to mental health illness.

Hypothesis: To determine the prevalence of PCOS among overweight/obese adolescents, evaluate known risk factors for PCOS in a diverse population, and discern cardiovascular risk and mental health comorbidity.

Methods: EMR of overweight/obese adolescents from April 1, 2016 to July 30, 2018 were reviewed to identify the presence of PCOS using NIH criteria, race/ethnicity, known risk factors for PCOS (Lipid, BMI, HA1c, BP) and mental health conditions (anxiety/depression and self-harm/suicidal ideation).

Results: Patients with PCOS were more likely to be hyperlipidemic (19.6% vs 9.9%, $p=0.05$), obese (67.4% vs 50.9%, $p=0.03$), and have acanthosis (68.9% vs 28.2%, $p<0.001$) than those without. Hispanic Ethnicity was not more common among adolescents with PCOS (57.8% vs 77.9%, $p=0.03$). Despite a high % of depression/anxiety; depression/anxiety (37% vs 33%, $p=.590$) as well as self-harm/suicidal ideation (17% vs 17%, $p=.96$) were not more common among those with PCOS vs those without. In a logistic regression model, after adjusting for demographics and clinical features of interest, ethnicity, acanthosis and BMI were associated with PCOS.

Conclusions: Patients with PCOS are more likely to be obese, hyperlipidemic, have acanthosis and be Non-Hispanic. There was no difference in the prevalence of depression/anxiety and self harm/suicidal ideation.

Acknowledgments: Statistical Support Provided by the Department of Pediatrics-Icahn School of Medicine at Mount Sinai

Unraveling the neurobiology of DDX3X Syndrome, a recently identified form of Intellectual Disability

Author Name(s): Danielle Mendonca, Andrea Boitnott, Dévina Ung, Kristi Niblo, Elodie Drapeau, Silvia De Rubeis

Department: Psychiatry

Division:

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Seaver Autism Center for Research and Treatment, The Mindich Child and Health Development Institute

Introduction: DDX3X syndrome is an Intellectual Disability (ID) caused by mutations in the X-linked gene *DDX3X*, which encodes an RNA helicase implicated in mRNA translation. Affected individuals can have behavioral problems, including autism, movement disorders and cortical abnormalities. Most affected individuals are females with *de novo* mutations in *DDX3X* while the few known affected males inherit their mutations from asymptomatic mothers. The molecular and cellular functions of DDX3X during brain development are unknown.

Hypothesis: We hypothesize that DDX3X is a sex-specific regulator of synaptic translation, cortical development and synaptogenesis.

Methods: We have generated a novel mouse modeling DDX3X haploinsufficiency (*Ddx3x*^{+/-}) we are using to examine cortical development and physical, sensory and motor development during early postnatal life. To dissect the molecular mechanisms underlying DDX3X syndrome, we combine biochemical assays to isolate DDX3X complexes and viral-based Translating Ribosome Affinity Purification followed by RNA sequencing to identify DDX3X target genes. We also study how DDX3X haploinsufficiency or loss impacts synaptogenesis in female and male single-embryo neuronal cultures.

Results: We observe that *Ddx3x* null male mice die *in utero* and *Ddx3x* haploinsufficient females have specific physical, sensory and motor delays. Our preliminary cortical lamination data indicates *Ddx3x* expression occurs mostly in deep layer neurons in the developing primary motor cortex. Additionally, we identified that wild-type females have more *Ddx3x* than males and *Ddx3x* participation in the eIF4F translational complex in cortical neurons.

Conclusions: We have generated a mouse model with construct validity for DDX3X syndrome. Using our approaches, we expect to expose the core neurobiology of DDX3X syndrome.

ASD and Symptoms of ADHD Predict Abnormal Neuronal Preparation for Motor Activity

Author Name(s): Marta Migó, Sylvia Guillory, Christopher McLaughlin, Emily Isenstein, Hannah Grosman, Christina Layton, Katharine N Thakkar, Francisco X. Castellanos, Jennifer H. Foss-Feig

Department: Psychiatry

Division: n/a

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Seaver Autism Center

Introduction: Certain psychiatric patients display abnormal neuronal predictive processes— such as abnormal Lateralized Readiness Potentials (LRP)— prior to onset of voluntary motor activity. While LRPs have not been tested in autism spectrum disorder (ASD), affected patients display abnormal predictive processes. Impairment in motor skill preparation and execution have also been observed in Attention-Deficit/Hyperactivity Disorder (ADHD). Additionally, ASD and ADHD are frequently comorbid. However, the effects of ASD and ADHD comorbidity on neuronal motor predictive processes have not been studied. The present study examines neurophysiological preparation of voluntary motor activity in patients with ASD displaying a range of ADHD symptomatology.

Hypothesis: We predict abnormal LRP amplitudes in ASD, and that ADHD severity will correlate with longer preparation periods, hence, earlier LRP onset latencies.

Methods: Electroencephalographic (EEG) data were collected from 16 typically-developing (TD) and 16 ASD) participants (ages 8-17) with varying ADHD symptomatology. Participants pressed a button every 2-3 seconds which, depending on condition, triggered a tone played in their headphones (Action-to-Effect condition; ATE) or not (Action-to-No-Effect condition, ATNE).

Results: LRP amplitude was significantly larger in ASD than TD in the ATE condition ($t(30)=2.15$, $p=0.04$). Within the ASD group, ADHD symptoms, measured by the “Attention/Hyperactivity Problems” subscale from the Child Behavior Checklist, significantly correlated with LRP onset latency (ATE, $R^2=0.47$, $p=0.003$; ATNE, $R^2=0.32$, $p=0.023$).

Conclusions: Our results suggest that ASD is associated with atypical LRP response, and that ADHD symptoms predict longer motor preparation periods. Future research could reveal LRP to be effective in subtyping ASD and ADHD comorbidity.

Integrating Primary Care and Behavioral Health Through a Collaborative Care (CC) model: Feasibility and Outcome of a Universal Screening

Author Names: Maya Mihic, Thaina Rousseau-Pierre

Department: Pediatrics

Division: Adolescent Medicine

Institution Affiliation: Elmhurst Hospital Center

Introduction: AAP recommends integrating behavioral health in primary care to improve diagnosis and management of depression. Through a Collaborative Care Model, we implemented the integration of behavioral health in our clinic.

Methods: We initiated a QI Project implementing depression screening in the adolescent clinic using PHQ9 in our EMR along with staff-wide training and the creation of a care-flow. We reviewed charts from January 2016 to January 2019 with PHQ9 score ≥ 5 and collected the highest PHQ9, evaluation by a Social Worker (SW), referral and enrollment in treatment.

Results: 397 charts had PHQ9 score ≥ 5 with 31% scoring 10-27. Our SW evaluated 60% of those scoring ≥ 10 . Reasons for no evaluation: parental refusal, enrolled in care, not depressed, referral made to behavioral health care (BHC) by a PCP and no show. 74% of the patients evaluated by our SW were referred for BHC (47% scoring 10-14, 50% scoring 15-19 and 50% scoring 20-27). Of the patients who were referred to therapy, 64% were enrolled in BHC (49% of those scoring 10-14, 57% scoring 15-19 and 70% scoring 20-27).

Conclusions: Our QI demonstrated the feasibility of routinely screening all adolescents within a primary care setting for depression with a validated tool, the PHQ-9. The availability of a SW within our primary care clinic resulted in over 60% of patients scoring ≥ 10 to have an evaluation by a SW and most of all 49% to 70% enrollment in BHC.

Crouzon Syndrome with Acanthosis Nigricans (CAN) without Craniosynostosis

Author Names: Cassie S. Mintz, Ayuko Iverson, Saadi Ghatan, Aldo Londino, Puneet Pawha, Ethylin Jabs, Lakshmi Mehta

Department: Genetics & Genomic Sciences

Division: Medical Genetics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Craniosynostosis is a cardinal feature of Crouzon (CS) and other related syndromes and is a common reason to consider molecular testing to confirm the specific diagnosis. Characteristic facial features of CS include proptosis due to shallow orbits, ocular hypertelorism, midface hypoplasia, small beaked nose and prognathism. These features are considered to be secondary to abnormal skull fusion, but may involve more complex mechanisms. Crouzon syndrome with acanthosis nigricans (CAN) is considered a separate entity from classic CS as patients show distinct findings, including aqueductal stenosis and choanal atresia. Additionally, the recurrent mutation is in fibroblast growth factor receptor 3, *FGFR3*, while classic CS is associated with mutations in *FGFR2*. Patients with CAN and CS do share clinical features of craniosynostosis and the facial features described above.

Case Presentation: We present a 3-month-old boy with unilateral choanal stenosis, bulging fontanel, hydrocephalus due to aqueductal stenosis and failure to thrive. At initial evaluation, head circumference was 42 cm (50%), which was increased from previous at 22%. CT head showed obstructive hydrocephalus and brain MRI showed a Chiari 1 malformation. He underwent successful endoscopic third ventriculostomy. Exam included proptosis with shallow orbits and midface hypoplasia leading to suspicion of CS. Notably, there was no craniosynostosis on CT. Genetic testing revealed a *de novo* mutation in *FGFR3*, p. A391E, the common mutation associated with CAN. The patient has no skin changes at this time, which may develop later. Other reported cases with CAN presented with craniosynostosis, making this patient's presentation particularly unique.

Conclusions: Our patient's presentation provides evidence to support the hypothesis that the typical facial features of CS and CAN are part of a developmental field defect due to abnormal FGFR signaling, rather than a consequence of craniosynostosis alone.

Teaching Social Determinants via Service Learning and Community Exploration

Author Name(s): Leora Mogilner¹, Ashley Stevens², Cynthia Katz¹

Department: Pediatrics

Division: General Pediatrics

Institution Affiliations: Icahn School of Medicine at Mount Sinai¹; New York Presbyterian Hospital (Columbia Campus)²

Introduction: Residents must be familiar with their community and understand the impact of social determinants of health (SDH). Service learning combines community service with reflection to achieve community engagement and understanding of SDH. An intern orientation program engaged interns in conversations about SDH and advocacy through service learning and a team building experience.

Hypothesis: A community tour and volunteer experience will impact knowledge of local community, attitudes around SDH screening, and intern bonding.

Methods: The intervention included a didactic session on SDH; walking tour of East Harlem (EH); and visit to a community-based organization where residents engaged in a volunteer activity (packing meals). Residents completed a mixed methods survey with retrospective pre/post questions to reflect on the impact of the activity.

Results: 35 residents were surveyed in 2017-2018. The program was effective in increasing interns' understanding of demographics and health issues affecting EH children ($p=0.001$) and increasing comfort discussing food insecurity with patients ($p=0.001$). All respondents reported a future plan to screen patients for food security. 97% agreed the activity helped them get to know their fellow interns. When asked for favorite aspects of the day, "volunteering at the pantry and giving back to the community," "learning more about the EH community" and "getting to know our co-residents," were identified.

Conclusions: A volunteer experience followed by a walking tour gave interns the opportunity to learn about EH while performing community service and bonding with their co-interns. Future research should ascertain whether this experience sustains a program culture of advocacy.

Diagnostic Accuracy of Fetal Echocardiography in Major Congenital Heart Disease

Author Name(s): Namrita Mozumdar, John Rowland, Hari Rajagopal, Miwa Geiger, Shubhika Srivastava, Kenan Stern

Department: Pediatrics

Division: Cardiology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Accuracy of fetal echocardiography (FE) is not well defined, and reporting of diagnostic discrepancies (DD) is not standardized.

Hypothesis: Prenatal diagnosis will be discordant in 20% of cases, and that majority of discrepancies will be minor in severity.

Methods: Retrospective single center study of prenatally diagnosed major congenital heart disease (CHD), defined as expected need for intervention within first year of life. DD between pre and postnatal findings were reviewed and categorized.

Results: From 2009-2017, 222 fetuses with median gestational age at first FE of 24 weeks were included. The most common indication was suspected/known CHD (79.7%). There were 30 DD (13.5%), and majority were false negative (56.7%). Most were minor in severity, with one severe. The majority were possibly preventable (90.0%), with the most common contributing factor being technical limitations (43.3%). The most common anatomic segment involved was the ventricular septum (27.3%), primarily missed septal defects. Those with DD were more likely to have high anatomic complexity (16.7% vs 3.6%, $p=0.02$), maternal comorbidities (40.0% vs. 22.1%, $p=0.03$) and a younger maternal age (median of 27 vs. 30 years, $p=0.02$). They were also more likely to have a later gestation at initial FE (median of 29.5 vs 24 weeks, $p=0.003$) and fewer total FE (median of 2 vs. 3, $p=0.002$), as well as have a fellow as the initial sonographer (36.7% vs. 16.7%, $p=0.03$).

Conclusions: FE had a DD rate of 13.5%, mostly minor and moderate in severity. Factors associated with DD included higher anatomic complexity, maternal comorbidities, fewer total FE, and fellow involvement.

Defects in RNA Polymerase II Pausing and Elongation Control Results in Severe Hemolytic Anemia in the EKLF/Klf1 Nan Mutant

Author Name(s): Kaustav Mukherjee, Chrysoula Deligianni, James J. Bieker

Department(s): Cell, Developmental and Regenerative Biology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Mindich Child Health and Development Institute; Tisch Cancer Institute; Black Family Stem Cell Institute

Introduction: EKLF/Klf1 is an erythroid zinc-finger transcription factor that activates transcription of genes and circuits required for erythroid development. An E325K substitution in the EKLF Zn-finger leads to Congenital Dyserythropoietic Anemia, CDA IV, in humans. An autosomal dominant mutation in murine EKLF at the same amino acid, E339D, is called Nan (Neonatal anemia) in mice. Nan/Nan mice show severe anemia and embryonic lethality, whereas Nan/+ heterozygotes survive but have severe hemolytic anemia, like CDA IV patients. Nan-EKLF has altered DNA binding specificity resulting in the downregulation of some EKLF target genes even in Nan/+ heterozygotes.

Hypothesis: Since EKLF interacts with transcriptional co-activator CBP/p300, which recruits transcription elongation factors, gene downregulation in Nan/+ is possibly due to loss of CBP/p300 and subsequent reduction in productive transcription elongation.

Methods: We used ChIP-Seq to assay the global occupancies of RNA Polymerase II phospho-Ser5 (initiating) and RNA Pol II phospho-Ser2 (elongating) forms, as well as the occupancies of critical elongation factors P-TEFb and NELF. We also used ATAC-Seq to assay for the levels of open chromatin in WT and Nan/+.

Results: We found that a subset of genes downregulated in Nan/+ have sufficient levels of phospho-Ser5 RNA Pol II at the transcription start site (TSS), but reduced levels of phospho-Ser2 RNA Pol II in the gene body. This subset of genes included those encoding critical factors necessary for erythropoiesis. We also found that there were comparable levels of open chromatin at these promoters in WT and Nan/+, and the occupancy of negative elongation factor (NELF) was unchanged in WT and Nan/+. However, the occupancy of positive elongation factor (P-TEFb) was drastically reduced at the promoters of this subset of genes.

Conclusions: In Nan/+ heterozygote, the altered binding of EKLF results in gene downregulation due to defects in successful release of RNA Polymerase II from paused to productive elongation. This defective pause-release is due to reduced recruitment of elongation factor P-TEFb.

Assessment of Metal Exposures in Deciduous Teeth of Patients with Inflammatory Bowel Disease

Author Name(s): Nilendra Nair, Christine Austin, Manel Rocha, Catarina Gouveia, Paul Curtin, Caroline Eisele, Jean-Frédéric Colombel, Joana Torres, Inga Peter, Manish Arora

Departments: Genetics and Genomic Sciences; Environmental Medicine and Public Health; Medicine

Division: Gastroenterology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Environmental exposures during early life play a role in the pathogenesis of Inflammatory Bowel Disease (IBD). Teeth develop incrementally, storing environmental information such as metals as we age. The study of deciduous (baby) teeth-matrix biomarkers allows assessment of cumulative exposures, starting as early as the second trimester of prenatal development, and continuing into early childhood until teeth shedding.

Hypothesis: By studying deciduous teeth, we aimed to identify whether critical exposures during early development may be associated with IBD diagnosis later in life.

Methods: Thirty baby teeth were obtained from 14 Portuguese adult IBD patients and 16 from unaffected controls (3 from unaffected siblings of IBD patients). Laser ablation-inductively coupled mass spectrometry analysis was used to create temporal metal exposure profiles from the second trimester of pregnancy through the first 6 months of life. Data were analyzed using distributed lag models by estimating the time-lagged association of exposures with IBD diagnosis while accounting for correlated exposures.

Results: We found divergences in metal uptake in the teeth of individuals who eventually developed IBD as compared to controls in a time-dependent manner. Lead exposure, a known inflammatory toxicant, was significantly higher during intra-uterine and the first 6 months of life ($p < 0.05$). In IBD patients, copper and chromium levels were significantly higher up to 15 weeks post-natally and from 10 to 15 weeks before birth, respectively (both $p < 0.05$).

Conclusions: These data suggest that deregulation in metal uptake during a critical window in early-life is a feature of IBD, prior to the emergence of any clinical symptoms.

Investigating Environmental Exposure Combinations and Early Age at Menarche Among U.S. Adolescent Girls

Author Name(s): Sabine Oskar, Mary S. Wolff, Susan L. Teitelbaum, Jeanette A. Stingone

Department: Environmental Medicine & Public Health

Division: N/A

Institution Affiliation(s): Icahn School of Medicine at Mount Sinai and Mailman School of Public Health at Columbia University

Introduction: In the U.S. the average age of menarche has decreased over time. Early menarche in girls has been linked to adverse health outcomes later in life including breast cancer, type 2 diabetes and cardiovascular disease. Increasing evidence supports that exposure to hormonally active environmental compounds may be associated with adverse reproductive outcomes. Previous studies on hormonally active agents and pubertal timing often examine exposures in isolation, not accounting for the “joint effect” of multiple high-dimension exposures. Our aim was to use a two-step data driven approach to generate hypotheses about combinations of exposures associated with early menarche.

Hypothesis: We hypothesized that tree-based analytical methods could help identify combinations of exposures associated with earlier menarche.

Methods: Data from 253 girls (12-16 years) from National Health and Nutrition Examination Survey with 41 exposure biomarker measures across six classes of hormonal agents were analyzed. First, we used random forest classifier, a machine learning analytical method, to identify exposure profiles among adolescent girls with earlier menarche. We then used traditional epidemiological methods to quantify the magnitude of effect in both unadjusted and adjusted analyses.

Results: Mono-(2-ethyl)-hexyl phthalate (MEHP), with a mean depth of 2.64 was identified as the most important predictor based on mean decrease in accuracy and decrease in Gini coefficient. Later menarche was found to be more likely in girls with higher urinary MEHP concentrations, consistent with previous research. The 2 next important single predictors were 2,4-dichlorophenol and mono-(3-carboxypropyl) phthalate, which is also in agreement with earlier reports. The most frequent interaction pair was MEHP and blood lead. Consistent with studies that have used other data analysis methods, the order of predictors in overweight girls differed when stratifying random forest models by BMI; however, differences may also be due to small sample size (42% overweight). Regression analyses showed that MEHP was independently associated with later menarche (PR= -0.56, 95% CI: -0.99 to -0.126). The association between MEHP and menarche remained after adjusting for confounders (PR= -0.52, 95% CI: -0.95 to -0.10). There was no evidence of multiplicative interaction between MEHP and blood lead.

Conclusions: Tree-based analytics identified exposures relevant to early menarche that were consistent with previous literature. These results illustrate how a data-driven approach can generate hypotheses within high-dimensional exposure data.

Point-of-Care Ultrasound Diagnosis of Small Bowel-Small-Bowel vs Ileocolic Intussusception

Author Name(s): Brian L. Park¹, Joni E. Rabiner², James W. Tsung¹

Department: Pediatrics

Division: Emergency Medicine

Institution Affiliation(s): ¹Icahn School of Medicine at Mount Sinai, ²Columbia University Medical Center

Introduction: Identification of intussusception is feasible with emergency department (ED) point-of-care ultrasound (PoCUS) due to its ease-of-use and high accuracy. Little is known about the clinical characteristics and outcomes of small bowel-small bowel intussusception (SB-SBI) relative to ileocolic intussusception (ICI) when identified by PoCUS.

Methods: We conducted a retrospective cohort study at a single, tertiary-care, urban pediatric ED of intussusception identified by PoCUS. Demographic information, clinical data, and outcomes, including clinical course, intussusception characteristics, recurrence rates, and interobserver agreement (Cohen's kappa), were evaluated.

Results: ED PoCUS identified thirty-six patients with intussusception over a 4-year period. Twenty patients (56%) identified were SB-SBI. The median age was 52 months (IQR 31.8-76.8 months) for SB-SBI and 8 months (IQR 5.8-13.5 months) for ICI. The mean diameter was 1.71 cm (SD 0.52 cm) for SB-SBI and 2.74 cm (SD 0.44 cm) for ICI ($p < 0.05$). Two of 20 (10%) SB-SBI subjects required surgical intervention, while the rest spontaneously reduced. Fourteen of 16 (88%) ICI subjects required intervention. There were two (10%) recurrences of SB-SBI and three (19%) recurrence of ICI confirmed on PoCUS. Cohen's kappa was 0.85 (95% CI 0.68-1).

Conclusions: SB-SBI may be more common than previously thought when screened with ED PoCUS. PoCUS can differentiate between variants of intussusception that range from a surgical emergency to a transient source of abdominal pain that may be recurrent and otherwise unexplained, allowing clinicians to better manage these patients accordingly.

Development of an Adolescent-Friendly Pregnancy & Parenting Curriculum at a Teaching and Referral Hospital in Western Kenya

Author Name(s): Natasha Ramsey, Priscah Onkware, Maureen Chepkoech, Vincent Kibet, Julie Thorne

Department: Pediatrics

Division: Global Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Arnhold Institute for Global Health

Introduction: Adolescent pregnancy is a problem across the world and is associated with poor maternal and neonatal outcomes, lower educational and employment attainment. In Kenya, 50% of women have begun childbearing by age 20 although adolescent reproductive health has been prioritized by the Kenyan Ministry of Health. A study using human centered design (HCD) conducted at Moi Teaching and Referral Hospital led to prototypes for implementation of an adolescent centered pregnancy care program including peer support groups and improved healthy sexuality and pregnancy education.

Hypothesis: Adolescents are interested in an adolescent-focused pregnancy curriculum that includes sexual health, pregnancy, and parenting information. Engaging adolescents in the planning phase will lead to an improved product.

Methods: A participatory approach was used to develop the adolescent pregnancy curriculum. Raw data from the HCD study was reviewed and stakeholder and consultant meetings were held to develop curriculum content. The curriculum draft was pitched in a workshop of adolescents.

Results: A 3-part curriculum was assembled, including sexual health, pregnancy care, and childcare. Participants included four pregnant and parenting adolescents and one community health volunteer. Participants wanted weekly sessions led by an experienced woman, not a peer. They provided many suggestions on topics they wanted and when they should be taught in the curriculum. A session was sampled with teens and highlighted the low levels of health literacy among participants.

Conclusion: There is significant interest from adolescents in a sexual health and pregnancy curriculum. Using a participatory approach is important in designing a final product. Next steps include piloting the material through support groups for refinement.

Epidemiology and Outcomes of Emergency Department Visits for Hypersensitivity in Infants at a Tertiary Care Center

Author Name(s): Nicole Ramsey, Hsi-en Ho, Julie Wang

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Anaphylaxis is a life-threatening, multi-system, rapid-onset hypersensitivity reaction. There are limited reports on anaphylaxis in infants. This study characterizes hypersensitivity visits of patients <1 year to an emergency department (ED) at Mount Sinai over a five-year period.

Methods: This is a retrospective chart review of ED visits for children under one year old from 2013-2018. Inclusion criteria were pediatric cases with anaphylaxis, allergic reaction or urticaria as the primary diagnosis or where epinephrine was prescribed or administered. Descriptive statistics were used to analyze the data. This study was approved by the Mount Sinai Institutional Review Board with a waiver of consent.

Results: Of the 98 cases (1.6% of all ED cases), median age was 9 months and 39 (40%) were girls. Twenty-three percent were African-American, 16% Caucasian, and 4% Asian. Visits ranged from 10 to 19 per year. Fourteen percent had a known food allergy and 41% percent had a known atopic condition. Most (54%) had only skin involvement; a combination of skin, GI and respiratory symptoms was the next common presentation (13%). Common triggers were egg (43%), peanut (24%), and fish (11%). Forty-six percent of children were given an epinephrine auto-injector (EAI) at discharge. Rate of EAI prescription and referral to allergy specialists increased with severity of reaction. Time to follow-up decreased with severity of reaction.

Conclusions: Infants with anaphylaxis are not often evaluated in the ED. Egg is the most common identified trigger in the ED. Management and time to specialist follow-up vary with hypersensitivity grade.

Sex-specific associations between Exposure to Multiple Metals and Visuospatial Memory Skills in Children

Author Names: Elza Rechtman, Demetrios Papazaharias, Stefano Renzetti, Giuseppa Cagna, Elena Colicino, Danielle Hazeltine, Marco Peli, Donatella Placidi, Donald R Smith, Robert O. Wright, Roberto G Lucchini, Megan Horton

Department: Environmental Medicine and Public Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Mount Sinai Transdisciplinary Center on Health Effects of Early Environmental Exposures, Institute for Exposomic Research

Introduction: Early life exposure to metals may have lifelong impacts on neurodevelopment. In rodents, visuospatial memory deficits following early life metal exposure have been measured with an 8-arm radial maze (RAM) task. In humans, the virtual radial arm maze (VRAM), is the computerized analogue of the RAM task developed to assess visuospatial memory children. The goal of this study was to examine cross-sectional associations between exposure to metals and performance on the VRAM among 11-15-year-old children.

Hypothesis: We hypothesized that exposure to metals would be associated with poorer visuospatial memory skills.

Methods: Manganese (Mn), lead (Pb), chromium (Cr) and copper (Cu) were measured in blood, urine, hair, nails and saliva of 184 participants residing near a ferroalloy plant in Italy. Concurrently, we administered the VRAM to all participants. Performance is based on the time required to complete the VRAM. Using generalized weighted quartile sum (gWQS) regression we investigated associations between a mixture of 4 metals in 5 matrices and VRAM performance, controlling for covariates. We estimated metal mixture weights and effects for the whole cohort and for each sex individually.

Results: A higher metal mixture index was associated with poorer VRAM performance (β : 0.28; $p=0.02$). Upon stratification, this association was observed in girls (β : 0.65; $p<0.001$) and not boys.

Conclusions: Exposure to metals during development may disturb visuospatial skills acquisition in females. These findings highlight the importance of assessing exposure to metals mixtures over the evaluation of a single metal which may underestimate the true effects of exposure.

Generation and Initial Characterization of 3D Cortical Collecting Ducts (CCDs)-on-a-Chip

Author Names: Joshua L. Rein¹, Szilvia Heja², Daniel Flores², Rolando Carrisoza-Gaytan², Neil Lin³, Kimberly A. Homan³, Jennifer A. Lewis³, Lisa M. Satlin²

Departments: ¹Medicine, ²Pediatrics, ³School of Engineering and Applied Sciences, Wyss Institute for Biologically Inspired Engineering

Division: Nephrology

Institution Affiliations: ^{1,2}Icahn School of Medicine at Mount Sinai; ³Harvard University

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Renal tubular epithelia grown in their native 3D geometry provide a more physiologically relevant environment. Cell doming is unique to epithelial cell monolayers and may reflect cellular differentiation and vectorial transport of salt and water. We describe the doming characteristics of cells grown in a novel in vitro 3D microphysiological model that recapitulates the in vivo geometry of the CCD.

Hypothesis: ENaC mediated Na⁺ absorption contributes to doming in mpkCCD cells grown in the 3D geometry of the CCD.

Methods: 3D CCD-like tubules were generated using a pin pullout technique through a fibrin/gelatin extracellular matrix enclosed within a polydimethylsiloxane scaffold. mpkCCD cells were cultured in the lumen under continuous perfusion via a peristaltic pump in a closed loop circuit. Epithelial integrity and doming characteristics in the absence (control) or presence of 10 μ M benzamil (BZ), an inhibitor of ENaC, were studied with widefield microscopy.

Results: Diffusional permeability of luminal 3-5 kDa FITC dextran was significantly lower with cells vs. without cells (52.2 ± 13.1 vs. 1.9 ± 0.9 μ m/min, $P=0.01$). Mean dome volume was $\sim 3.1 \pm 0.5$ nl in 3 tubules in the absence vs. $\sim 0.04 \pm 0.01$ nl in the 1 tubule that demonstrated doming in the presence of BZ ($P=0.01$). Tubule sections expressed immunodetectable ENaC, Na-K-ATPase and laminin.

Conclusion: We generated and initially characterized a 3D CCD-on-a-chip that promises to provide a relevant model for studying transport physiology and addressing a variety of clinically important questions. The mechanism(s) underlying dome formation remains to be determined.

Dissecting *de novo* indel biology with long-read technology

Author Name(s): Felix Richter[†], Allison H. Seiden[†], Nihir Patel, Andrew P. Sharp, Bruce D. Gelb

[†] Equal contributions

Department(s): Pediatrics, Genetics and Genomic Sciences

Division(s):

Institution Affiliation(s): Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Mechanisms for insertion/deletion (indel) mutagenesis, like polymerase slippage, have been hypothesized but not well-characterized in the human genome.

Hypothesis: The association of *de novo* indels with parental age at conception will vary by subtype.

Methods: We phased parent-of-origin for *de novo* indels with low-coverage long-read whole genome sequencing, achieving better phasing compared to short-read sequencing (medians of 84% and 23%, respectively). We wrote an application programming interface to classify indels into three subtypes by sequence context. With these methods and cohorts phased with short- (N=305) and long-read (N=10) tracing or three-generation haplotyping (N=225), we characterized associations between *de novo* indels and parental age. Fisher's method was used to meta-analyze Pearson's correlation coefficients and p-values.

Results: One *de novo* indel subtype (change in copy count, CCC) was significantly correlated with paternal ($P=9.1 \times 10^{-5}$) but not maternal ($P=0.12$) age at conception. The other two indel subtypes appeared unrelated to parental age. We replicated this effect in three unphased cohorts ($N_{\text{trios}}=3391$, $P_{\text{father}}=1.0 \times 10^{-9}$, $P_{\text{mother}}=0.78$). While the percentage of the variance explained by paternal age was low, it was stronger for *de novo* CCC indels in repetitive regions ($R^2=0.023$, $P=0.0039$) than those outside of repeats ($R^2=0.010$, $P=0.043$). Although these results are consistent with polymerase slippage during spermatogenesis, we did not find an association with replication timing.

Conclusions: Our results suggest that spermatogenesis-specific events have a minor role in CCC indel mutagenesis, one not observed for other indel subtypes or for maternal age in general. These results have implications for indel modeling in evolution and disease.

Investigating the Etiology of Multifocal Atrial Tachycardia in Costello Syndrome via iPSC Disease Modeling

Author Name (s): Nelson A. Rodriguez, Rafael Dariolli, Nihar Patel, Jingqi Gong, Tara Keshavarz-Shirazi, Rebecca Josowitz, Simon Ng, Xianming Lin, Glenn Fishman, Eric Sobie, Bruce D. Gelb

Department: Pediatrics; Genetics & Genomic Sciences

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Patients with Costello syndrome (CS) have *HRAS* gain-of-function mutations (G12S 80%, G12A 7.2%). During early childhood, 50% develop multifocal atrial tachycardia (MAT), a difficult-to-treat tachyarrhythmia, of unknown pathogenesis.

Hypothesis: Overactive *HRAS* activity causes abnormal calcium handling, triggering atrial arrhythmogenesis in CS patients.

Methods: *HRAS* Gly12 mutations were introduced into a human induced pluripotent stem cell (iPSC) line with an atrial cardiomyocyte (CM) reporter. Calcium transients and electrophysiology were assessed in iPSC-derived CMs with calcium/voltage-sensitive dyes and patch clamping. HCN inhibitors targeted pacemaker-like activity in mutant RedHigh CMs. Transcriptomic data were analyzed via differential gene expression and gene ontology.

Results: Compared to the isogenic wild-type (WT) control [78 ± 3 bpm], mutant atrial-like CMs displayed high beat rates [G12A 101 ± 3 bpm, G12S 106 ± 4 bpm; $p < 0.0001$], shorter calcium decay times, [WT 416 ± 30 ms, G12A 253 ± 11 ms; ($p < 0.0001$), G12S 345 ± 43 ms (not significant)], increased calcium peak amplitudes [WT 0.7 ± 0.1 A.U.; G12S 1.5 ± 0.2 A.U. ($p < 0.01$); G12A 1.2 ± 0.2 A.U.; ($p < 0.05$)] and irregularity. Spontaneously beating mutants exhibited increased action potential duration [WT 252 ± 13 ms, G12A 342 ± 15 ms, G12S 360 ± 21 ms; $p < 0.0001$]. Mutant CMs demonstrated increased expression of genes for calcium regulation and transport, positive regulation of heart rate, and negative regulation of RAS. Mutants exhibited a nodal CM gene profile. HCN inhibitors ivabradine and crizotinib decreased beating rates.

Conclusion: CS-associated *HRAS*^{G12} mutations engendered electrophysiological abnormalities and transcriptional changes in atrial CMs. This is the first MAT model highlighting the complex role of mutations in the dysregulation of calcium transients, and its promotion of atrial remodeling and arrhythmogenesis.

Implementation of an Asynchronous Curriculum to Replace Traditional Didactics: Initial Observations and Challenges

Author Name(s): Priya S. Rolfes, Carolyn Rosen, John Rowland, Leora Mogilner

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: n/a

Introduction: Clinical demands have made it challenging to find protected time for resident didactics. The Physician Education & Assessment Center (PEAC) provides a pediatric ambulatory online curriculum used by many residency programs. No studies have been published that assess the feasibility and efficacy of an asynchronous pediatric curriculum.

Hypothesis: Use of an asynchronous online curriculum would be more accessible than our previous in-person (IP) didactic format.

Methods: A pre-intervention survey was sent to residents who had exposure to the IP curriculum. Questions assessed attendance and satisfaction with the IP curriculum, expectations of the new PEAC curriculum, and personal preference for curriculum type. Data was collected on PEAC module completion.

Results: Although residents were satisfied with the IP curricular content, they estimated that they had poor attendance at lectures. Residents anticipated completing 70% of PEAC modules over the year, however, after 6 months they only completed 6% of modules. 65% of residents stated they would prefer an asynchronous curriculum to an IP curriculum. There was no association between learning tool preference and module completion ($p=0.60$). Correlation analysis showed no relationship between an individual's projected and actual modules completed ($r=0.11$; $p=0.55$).

Conclusions: Though the concept of an asynchronous curriculum was well received, most residents did not come close to completing the projected number of modules over 6 months. Barriers to participation must be addressed in order to improve resident engagement. Once participation has improved, future steps will include comparing knowledge outcomes between curricula.

The Stability and Anti-inflammatory Effects of Berberine/Heparin/Chitosan Nanoparticles

Author Name(s): Mollie Schear, Kamal Srivastava, Nan Yang, Xiu-Min Li, David Dunkin

Department: Pediatrics

Division: Gastroenterology and Nutrition

Institution Affiliation: Icahn School of Medicine at Mount Sinai and New York Medical College

Institute Affiliation: Mindich Child Health and Development Institute

Introduction: Food allergy affects 8% of children and 3% of adults in the US. IgE-mediated anaphylaxis can be serious and sometimes fatal. There is no cure for food allergy and standard care includes food avoidance and rescue epinephrine. We found that the natural compound Berberine (BBR) suppresses IgE *in vitro*. However, BBR is poorly bioavailable. To address this we developed a BBR/Heparin/Chitosan nanoparticle (BHC) which significantly increased BBR bioavailability when tested in mice. To determine whether BHC can be efficiently translated for scaled-up clinical use, we evaluated structure, stability and anti-inflammatory effects of BHC

Hypothesis: We hypothesize that BHC is stable over time, and will effectively lower proinflammatory cytokines

Methods: We examined the structure and stability of BHC by examining nanoparticle size, zeta potential, and cryoprotection over time. Ability of BHC to suppress IL-1 β stimulated IL-8 production in CACO2 cells was tested. Unencapsulated BBR was included as control. IL-8 levels were measured by ELISA.

Results: We found that BHC is stable over a period of 8 months when stored in solution at 4°C. BHC cryopreserved with sucrose retained stability. In vitro, BHC suppressed IL-8 production in CACO2 cells however effect was similar to unencapsulated BBR.

Conclusions: BHC nanoparticles are stable and consistent over months and are amenable to cryoprotection suggesting good feasibility for clinical drug development. BHC demonstrated potent IL-8 suppression ($p < 0.001$) in CACO2 cells however lack of significant differences between BBR and BHC in this regard suggests that increased bioavailability of BHC is not a factor in this in vitro system.

Precocious Puberty in a Female with a Suprasellar Dermoid Cyst and Multiple Pituitary Hormone Deficiencies

Author Names: Swathi Sethuram, Meredith Wilkes, Jasmine Gujral, Lauryn Choleva, Mabel Yau, Elizabeth Wallach, Christopher J. Romero, Robert Rapaport

Department: Pediatrics

Division: Endocrinology and Diabetes

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: Dermoid cysts account for 0.3-0.5% of all intracranial tumors. Premature gonadotropin activation in the setting of multiple pituitary hormone deficiencies (MPHD) has not been described in patients with dermoid cysts.

Hypothesis: N/A

Methods: We report a 6 years 8 months old female who developed precocious puberty and multiple pituitary hormone deficiencies following suprasellar surgery for a dermoid cyst.

Results: The patient presented at 6years&8months of age with two months of headaches and vomiting. Initial evaluation showed bilateral optic nerve edema without visual field defects. She was pre-pubertal with weight at 5th percentile and height at 10th percentile, appropriate for family. Her MRI brain showed a 2.7x2.1x4.3cm fat containing, minimally enhancing mass within the suprasellar region and third ventricle with severe hydrocephalus. A complete mass resection revealed a dermoid cyst. Post-operatively, she developed persistent diabetes insipidus, central hypothyroidism and adrenal insufficiency. Three months post-operatively, she developed thelarche. Central precocious puberty confirmed by hormonal testing(see Table I).

Conclusion: Tumors and surgery in the suprasellar region are associated with pituitary hormone deficiencies including hypogonadotropic hypogonadism. MPHD with precocious puberty has rarely been described, more commonly with intracranial radiotherapy. There has been no such description in patients with dermoid cysts. We hypothesize that pituitary stalk manipulation following surgery could result in defective pituitary hormone production with selective disinhibition of the gonadotropins and prolactin, as with our patient. Physicians must be aware of the rare association of precocious puberty with MPHD in patients with suprasellar tumors pre or post operatively. Continued pubertal evaluation is important.

TABLE 1

	PITUITARY HORMONE EVALUATION	
	PRE-OPERATIVELY	POST-OPERATIVELY, prior to medications
IGF-1 ng/mL (42-240)	67	75
Free T4 ng/dL (0.8-1.5)	1.48	0.72
TSH uIU/ml (0.4-4.2)	0.695	Not available
8am CORTISOL mcg/dL (6.7-22.6)	11	2
FSH mIU/mL (1-4.2)	Not available	2.2
LH mIU/mL (0.02-0.3)	Not available	1
ESTRADIOL pg/mL (<15)	Not available	27
PROLACTIN ng/mL (1.4-24)	4.3	89.5

Post operative hormones drawn at 8am. IGF-1 = Insulin like growth factor 1; Free T4 = Free thyroxine;

TSH= Thyroid stimulating hormone; FSH = Follicle stimulating hormone; LH = Luteinizing hormone

Improving Maternal Postpartum Depression Screening: A Primary Care Clinic QI Project

Authors Name(s): Alice Shajan, Farhan Zakri, Mostafa Khalid, Barbara Porrello, Diego Craik, Parvathy Krishnan, Xiang Le Ng, Ananya Manchikalapati, Mohamed Ahmed, Iya Chikvashvili, Felicia Fojas, Pilar Gonzalez

Department: Pediatrics

Division: Pediatric Ambulatory Care

Institution Affiliation: Elmhurst Hospital Center / Icahn School of Medicine at Mount Sinai

Introduction: Maternal depression affects the well-being of the entire family and has an effect on newborn morbidity and mortality. The AAP recommends that all mothers be screened during routine well- child visits for risk of developing postpartum depression.

Hypothesis: We hypothesized that the rates of post-partum maternal depression screening in the PPC clinic can be improved by utilizing patient health questionnaire (PHQ9).

Methods: A chart review was done on well baby visits to assess the rates of provider adherence to maternal PHQ screening. PDSA cycle 1 included in-servicing clinic providers on the AAP recommendations and email reminders to all providers regarding documentation of maternal PHQ2/9. For PDSA cycle 2, the PHQ-9 questionnaire was handed to mothers to complete in the waiting room. The questionnaires were reviewed by PCAs who documented it. During the clinic visit, providers review the PHQ-9 score and referred mothers to the pediatric clinic social worker as needed for further assessment and intervention.

Results: In PDSA cycle 1, 86 charts from 2-month-old well baby visits were reviewed. Our baseline provider adherence rate was 13.9%. After PDSA 1, our adherence rate increased to 39.4%. After initiating PDSA cycle 2, there was a significant increase in the adherence rate to 90.8%. Interestingly, the number of positive PHQ-9 screen that required social work referral was also increased from 66.67% to 82.54%.

Conclusions: We were able to improving maternal depression screening by utilizing the PHQ-2/9 after two cycles of intervention from a baseline of 13.9% to 82.54%.

Vision Screening in a Primary Care Clinic: a Quality Improvement Initiative to Improve Early Detection of Amblyopia

Author Name(s): Alice Shajan, Gwen Raphan

Department: Pediatrics

Division: Pediatric Ambulatory Care

Institution Affiliation: Elmhurst Hospital Center / Icahn School of Medicine at Mount Sinai

Introduction: Amblyopia is caused by abnormal visual experience resulting from strabismus, refractory error or stimulus deprivation. Visual outcomes for the affected patients are better with earlier detection. Provider-based visual acuity testing is an effective tool for the early detection of risk factors that can lead to amblyopia in the ambulatory setting.

Hypothesis: Improving provider knowledge of vision screening based on AAPOS/AAP guidelines can promote prompt referral.

Methods: Charts were reviewed of all 5-7 year old patients who underwent vision screening at the clinic during a health maintenance visit (HMC) 1 month prior and 1-month post-intervention. An educational session on the importance of vision screening and latest criteria for referral was conducted for providers in the PPC clinic in October 2018. Signs were posted in patient exam rooms regarding the referral criteria. Charts were reviewed to assess appropriate referral to pediatric ophthalmology.

Results: Prior to intervention, a total of 44 patients had failed vision screening in the HMC and only 9 were referred to pediatric ophthalmology. After the intervention, out of 28 patients had failed vision screening in the HMC, 17 were referred. Our SMART aim of improving the referral rates increased from 20.5% to 60.7%.

Conclusions: Provider education regarding the updated guidelines as per AAPOS/AAP for vision screen based on visual acuity identified more patients who had failed vision screen and thus improved referral rates to pediatric ophthalmology. With time, the hope is that appropriate referral will lead to earlier detection and prevention of amblyopia.

Quality Improvement Intervention in a Pediatric Emergency Department to Improve Culture Turnaround Time

Author Name(s): Alice Shajan, Horton Lee

Department: Pediatrics

Division: Pediatric Emergency Medicine

Institution Affiliation: Elmhurst Hospital Center / Icahn School of Medicine at Mount Sinai

Introduction: The rapid identification of culture isolate and susceptibility plays a critical role for the optimal antibiotic treatment of patients. Delays in culture turnaround time affects patient care in ways such as the time to report true positives, altering antibiotics, and the duration of hospitalization.

Hypothesis: We hypothesized that the culture turnaround time can be improved by decreasing the time interval between collection and incubation.

Methods: Prior to the intervention, the culture specimens were placed in a bin to be collected by a lab messenger. The intervention we proposed is to send cultures via the hospital pneumatic tube system directly to the lab. An in-service was performed to all providers to send cultures promptly after collection. Signs were posted as a reminder. The time differences between collection and incubation were recorded. Data was collected for 4 weeks prior to intervention and for 4 week post-intervention.

Results: The average time between specimen collection and incubation was 8 hours and 52 minutes in the pre-intervention and 6 hours and 42 minutes in the post-intervention period with 201 and 143 specimens pre and post intervention. There was an improvement of 2 hours and 10 minutes between pre- and post-intervention, which was statistically significant with a Z of 3.39, $p=0.001$.

Conclusions: We were able to significantly improve culture turnaround time by decreasing the delay from specimen collection to incubation by around 2 hours by a simple intervention.

Towards Disease Interception: Contribution of Polygenic Risk Scores Towards Age-Dependent Risk of IBD in a High-Risk Population

Author(s): Elizabeth A. Spencer, Kyle Gettler, Drew Helmus, Shannon Telesco, Amy Hart, Marla C. Dubinsky, Judy Cho

Department: Pediatrics

Division: Gastroenterology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: We sought to explore polygenic risk scores (PRS) in predicting disease onset within inflammatory bowel disease (IBD) using a high-risk Ashkenazi Jewish (AJ) population.

Hypothesis: Within multiplex families, the PRS in the affected will be increased with younger age of diagnosis, and the PRS in unaffected will decrease with age.

Methods: Multiplex AJ families with IBD were enrolled. PRS were calculated using imputed IBD association statistics from the GWAS by Liu, J. Z., et al. Nat Genet (2015) as well as affected GSA data. P-value and linkage disequilibrium-based pruning and thresholding was performed as described in Khera et al. Nat Genet (2018).

Results: A total of 88 multiplex families (N=620) and 6 control Jewish families (N=39) were enrolled. GSA chip was performed on a preliminary cohort of 282 individuals (77 affected, 191 unaffected family members, 14 controls). The most significant PRS model at separating cases from controls and unaffected individuals was using p-values < 0.0005 and an R² value of 0.4 as seen in Figure 1. No unaffected individuals above the age of 25 years had a score in the top 10% (Figure 2). PRS did not correlate with age at diagnosis ($\rho = 0.26$, $p=0.1$).

Conclusions: A substantive fraction of AJ familial risk is driven by the composite of common variation from the over 200 other IBD loci that are present in non-AJ IBD. With no unaffected >25 yo with a PRS in the top 10%, it suggests that children carrying a top-10% PRS should be carefully monitored for disease inception.

Figure 1: Affected/Unaffected/Control Status by PRS

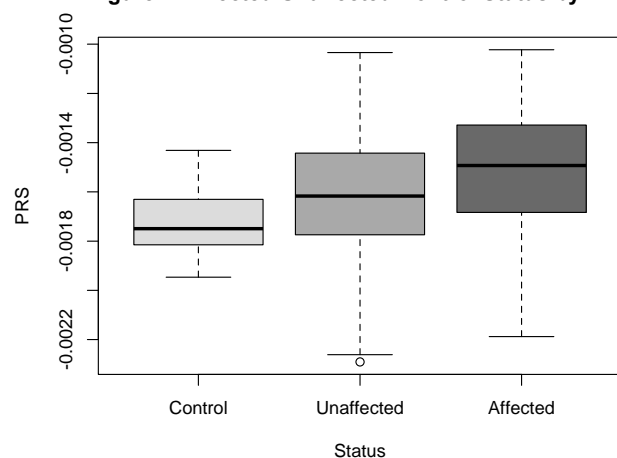
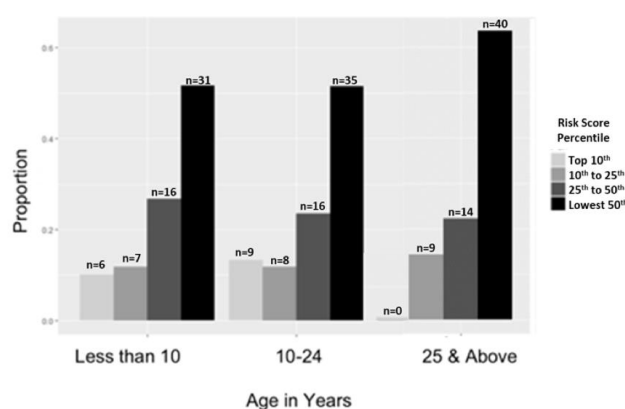


Figure 2: Risk Percentile by Age in Unaffected



Proteomic Markers in Pediatric IBD

Author(s): Elizabeth A. Spencer

Department: Pediatrics

Division: Gastroenterology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Inflammatory bowel disease (IBD) is a disease classically diagnosed after symptom onset via an invasive procedure or imaging study. We sought to identify serum protein markers in pediatric IBD that could be used to identify those at risk for IBD.

Hypothesis: Patients with IBD will have a unique proteomic profile as compared to healthy controls.

Methods: Pediatric (<18 years old) patients from three groups receiving colonoscopies were prospectively enrolled: 1. Healthy controls receiving a colonoscopy for functional abdominal pain 2. Pre-IBD-diagnosis 3. Post-IBD-diagnosis and naïve to therapy. Serum was collected on each patient. The O-link Proseek Inflammatory Panel, a protein extension assay which tests for 92 different inflammatory proteins in serum, was run on a preliminary cohort of 90 of these patients. The 92 markers were compared between those with IBD and controls using univariate analysis by t-test with false discovery rate correction.

Results: Of the 90 patients, 48 patients have IBD (38 Crohn's (CD), 10 Ulcerative colitis (UC)) and 42 patients are healthy controls (See Table 1 for patient characteristics). Forty-three percent of controls have a family history of IBD. By univariate analysis, there were 28 proteins identified with significantly different abundances between cases and controls (See Table 2).

Conclusions: There are numerous protein markers that are present in significantly different quantities in IBD patients versus healthy controls. There is strong potential for using the serum proteome in IBD as a diagnostic biomarker panel.

Table 1:

	Crohn's disease	Ulcerative colitis
	n=38	n=10
Male sex	24 (63%)	7 (70%)
Median age at diagnosis (years)	13.6	14.3
Disease location (Montreal)		
Ileal (L1)	7 (18%)	
Colonic (L2)	11 (29%)	
Ileocolonic (L3)	20 (53%)	
Upper tract (L4)	5 (13%)	
Disease behavior (Montreal)		
Non-stricturing, non-penetrating (B1)	32 (84%)	
Stricturing (B2)	3 (8%)	
Penetrating (B3)	3 (8%)	
Perianal disease (p)	4 (11%)	
Extent of disease (Montreal)		
Proctitis (E1)		2 (20%)
Left-sided (E2)		0
Extensive (E3)		0
Pancolitis (E4)		8 (80%)
Family History of IBD	18 (47%)	

Table 2:

Protein	Symbol	q-value
Oncostatin-M	OSM	2.6E-07
Interleukin-8	IL8	2.8E-06
Interleukin-6	IL6	2.8E-06
Interleukin-17A	IL17A	1.4E-05
Matrix metalloproteinase-1	MMP1	1.4E-05
Transforming growth factor alpha	TGFalpha	2.1E-05
Hepatocyte growth factor	HGF	3.3E-05
C-X-C motif chemokine 9	CXCL9	4.1E-05
Tumor necrosis factor ligand superfamily member 14	TNFSF14	0.0002
C-C motif chemokine 3	CCL3	0.0003
Latency-associated peptide transforming growth factor beta-1	LAPTGFbeta1	0.0003
Interleukin-24	IL24	0.0011
Vascular endothelial growth factor A	VEGFA	0.0012
T-cell surface glycoprotein CD5	CD5	0.0012

Monocyte chemotactic protein 3	MCP3	0.0012
Protein S100-A12	ENRAGE	0.0017
Interleukin-12 subunit beta	IL12B	0.0042
Interleukin-17C	IL17C	0.0042
CUB domain-containing protein 1	CDCP1	0.0042
C-C motif chemokine 4	CCL4	0.0051
C-C motif chemokine 20	CCL20	0.0051
Interleukin-15 receptor alpha	IL15RA	0.0089
Caspase-8	CASP8	0.0102
Matrix metalloproteinase-10	MMP10	0.0104
Signaling lymphocytic activation molecule	SLAMF1	0.0114
C-X-C motif chemokine 10	CXCL10	0.0177
Stem cell factor	SCF	0.0177
C-X-C motif chemokine 1	CXCL1	0.0177

In Vitro Comparison of the Temperatures of Refrigerated, Room Temperature and Warmed Formula at Point of Delivery into the Newborn

AUTHORS: Jeffrey V. Suell, Liandra Presser, Sonali Tatapudy, Robert Green

DEPARTMENT: Pediatrics

DIVISION: Neonatal and Perinatal Medicine

INSTITUTION AFFILIATION: Icahn School of Medicine at Mount Sinai

INTRODUCTION: Premature newborns are fed with expressed breast milk or formula (EBM/FOR) delivered by syringe pump via an orogastric tube. The EBM/FOR is often warmed prior to feeding.

HYPOTHESIS: We hypothesized that heat exchange between the tubing carrying the EBM/FOR and the atmosphere would cause the temperature of the feed to approach that of the surrounding environment.

METHODS: A syringe was oriented with tubing entering the incubator 62cm from its attachment to the syringe. Infusions of 20ml were carried out over 3, 15 and 30 minutes. Incubator temperature was either 31 or 37°C. A thermocoupler measured formula temperature. We compared (ANOVA) the effects of the source of the formula in the syringe being refrigerated (cold), room temp (RT) or warmed formula.

RESULTS: At the conclusion of the 3 min infusion mimicking OGT gavage feeding, into either a 31°C or 37°C incubator, there was a significant ($p < 0.001$) difference in the three formula sources' TPOE for cold, RT and warmed formula. For the 15 minute and 30 minute infusions into a 37°C incubator, there was a no significant difference in TPOE for the three formula sources either 1/3 of the way through ($p = 0.15$ and 0.2 respectively) or at the conclusion of the infusion ($p = 0.7$ and 0.3).

CONCLUSIONS: These results suggest that for gavage feeding, the source temperature significantly effects the temperature of formula delivered. However, in slower infusions, differences are insignificant. Therefore, time and money spent warming slow feeds represents provider inefficiency that forces time away from bedside and unneeded expenditures on bottle warmers and their accessories.

Identification of Outcome Measures for Clinical Trials in Rare Diseases Associated with Neurodevelopmental Disorders

Author Name(s): Lara Tang, Mikaela Rowe, Stacey Lurie, Ivy Giserman-Kiss, Jessica Zweifach, Danielle Halpern, Pilar Trelles, Jennifer Foss-Feig, Maureen Mulhern, Alexander Kolevzon, Paige M. Siper

Department: Psychiatry

Division: Child and Adolescent Psychiatry

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Seaver Autism Center for Research and Treatment

Introduction: Current clinical trials for patients with neurodevelopmental disorders are limited by a lack of standardized measures appropriate for severely affected populations. This study sought to identify potential outcome measures across cognitive, language, and sensory domains in two single-gene causes of autism spectrum disorder and intellectual disability.

Hypothesis: We hypothesize feasibility of standardized assessments will vary based on syndrome-specific phenotypes.

Methods: Twelve children with ADNP syndrome (3-12 years) and twelve with FOXP1 syndrome (3-16 years) were evaluated using standardized cognitive or developmental assessments. Expressive and receptive language measures included the Expressive Vocabulary Test (EVT-2) and the Peabody Picture Vocabulary Test (PPVT-4). Sensory reactivity was measured using the Sensory Assessment for Neurodevelopmental Disorders (SAND).

Results: Thirty-three percent of participants with ADNP syndrome and 83% with FOXP1 completed standardized cognitive assessments. All other participants completed a developmental assessment, resulting in a developmental quotient rather than a standardized IQ score. The PPVT-4 was obtained from 75% of participants with ADNP syndrome and 100% of those with FOXP1. The EVT-2 was obtained from 50% of participants with ADNP syndrome and 100% of those with FOXP1. The SAND was successfully obtained from all participants.

Conclusions: These findings suggest a broader array of outcome measures may be suitable for clinical trials in FOXP1 syndrome compared to those in ADNP. Sensory reactivity represents a core domain that was successfully measured across syndromes. Future studies must examine whether these measures adequately capture treatment response.

Home Visiting Program for NICU Graduates: Feasibility and Potential for Impact

Author Name(s): Candace Tannis¹, Katrina Leung², John Rowland³, Anna Cushing², Maureen Braun², Karen Wilson², Elaine Lin²

Department (s): Department of Environmental Medicine and Public Health¹, Department of Pediatrics, General Pediatrics², Department of Population Health³

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: The number of infants admitted to the neonatal intensive care unit (NICU) has been steadily increasing. Home visiting services have long been a part of caring for high-risk children and families with limited resources. There is little data on the effectiveness of physician-led home visiting programs on post- NICU health outcomes within the first year of life. The Mount Sinai Pediatric Visiting Doctors (PVD) program was started in 2013 to care for high risk children including those who are post NICU admission.

Hypothesis: We hypothesized that PVD program participants' utilization of acute care would be less than their matched hospital clinic counterparts.

Methods: Our study compares the patient characteristics and acute care utilization of the PVD program participants with those of the hospital-based outpatient clinic.

Results: We found that PVD group participants were of lower GA and BW, had a higher prevalence of respiratory sequelae and longer NICU stay than non-PVD group participants; and were more likely to attend a development clinic appointment. We found no statistically significant differences in ED use, hospitalization and ICU care after adjusting for BW, GA and other unbalanced factors, when comparing our two groups.

Conclusions: Physician led clinic-based home visiting services is a feasible way of providing clinical care to this population. Additional research is needed on medical and psychosocial risk factors that impact the efficacy of these programs on reducing acute care utilization.

Comparison of Body Measures, Lifestyle Behaviors, and Behavior Change Determinants in Male and Female Adolescents in a Pilot Diabetes Prevention Study

Author Name(s): Nita Vangeepuram, Candace Tannis, Tanya Braune, John Rowland, Carol R. Horowitz

Department: Pediatrics, Population Health Science and Policy

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: N/A

Introduction: There are limited studies on sex differences in youth diet and physical activity behaviors.

Hypothesis: There are differences in body measures, lifestyle behaviors, and behavioral determinants between male and female adolescents at-risk for Type 2 diabetes.

Methods: We screened 149 overweight/obese adolescents for pre-diabetes and obtained body measurements and administered a health/lifestyle survey as part of a community-based youth diabetes prevention program. We analyzed data using descriptive statistics and bivariate analyses (chi-square and t-tests).

Results: Survey data were available from 92 females and 57 males ages 13-19 years (32% Black, 69% Hispanic). There were no demographic differences between males and females. Average BMI z-score and waist circumference did not vary by sex, but percent body fat was higher in females than males (42% vs. 32%), based on published sex-specific norms. Compared to boys, girls had poorer self-perceived health status, reported less success with weight loss, and worried more about getting diabetes (Table 1). Girls were less confident in their ability to eat healthy when hungry after school or when bored, and to exercise when feeling bad about their bodies. Girls also had more perceived healthy lifestyle barriers, less positive social influences, more negative emotions and poorer body image. Girls generally had less healthy behaviors than boys with some exceptions (Table 2).

Conclusions: Girls were prone to less healthy behaviors and had relatively higher body fat percentage in this sample of overweight/obese teens. Future research will further examine sex differences in weight related behaviors and program effectiveness.

Table 1: Sex Differences in Reported Diet and Physical Activity Behavioral Determinants (baseline survey data from participants enrolled in youth diabetes prevention program, East Harlem, NY)

Variable	Males (n=57)	Females (n=92)	P value
General Perceptions about Weight and Health	N (%)	N (%)	
Health is fair or poor	22 (39%)	54 (59%)	0.017
Very or extremely successful when tried to lose weight in the past	23 (40%)	19 (21%)	0.009
Worried about getting diabetes	41 (72%)	86 (93%)	<0.001
Healthy Lifestyle Self-Efficacy (confidence level rating of 4 or 5 on five point scale)			
I can eat healthy foods when I am hungry after school	34 (60%)	40 (43%)	0.055
I can eat healthy when I'm bored	27 (47%)	25 (27%)	0.012
I can exercise when I feel bad about my body	48 (84%)	53 (58%)	<0.001
Perceived Barriers to Healthy Eating and Active Living (Agree/Strongly Agree)			
I eat unhealthy when I'm in a bad mood	17 (30%)	44 (48%)	0.030
Unhealthy foods are too tempting	25 (44%)	62 (67%)	0.005
Healthy foods are too expensive	15 (26%)	47 (51%)	0.003
I'm too rushed in the morning to eat a healthy breakfast	21 (37%)	65 (71%)	<0.001
It's hard to find time to fit physical activity into my schedule	13 (23%)	38 (41%)	0.021
I get embarrassed if other kids see me being physically active	6 (11%)	34 (37%)	<0.001
Exercise is too hard	3 (5%)	18 (20%)	0.016
I'm not motivated to exercise	7 (12%)	26 (28%)	0.022
I don't feel like exercising when I'm in a bad mood	18 (32%)	50 (54%)	0.007
It's too expensive to exercise	2 (4%)	13 (14%)	0.049
Social Influences			
Friends are somewhat or very physically active	42 (74%)	50 (54%)	0.018
Family members make comments about other people's weight	15 (26%)	38 (41%)	0.063
Emotions and Body Image			
Sometimes or very much bothered or troubled by feeling unhappy, sad or depressed	21 (37%)	53 (58%)	0.014
Describe themselves as very overweight	11 (19%)	30 (33%)	0.077
Would like to weigh less	47 (82%)	90 (98%)	0.001
Compare bodies to those of TV/movie stars, music artists, people who appear in magazines/social media	6 (11%)	31 (34%)	0.002

Table 2: Sex Differences in Reported Diet and Physical Activity Behaviors (baseline survey data from participants enrolled in youth diabetes prevention program, East Harlem, NY)

Variable	Males (n=57)	Females (n=92)	P value
Dietary Behaviors			
Buy bagel at a food stand on the way to or from school	4 (7%)	19 (21%)	0.034
Buy candy or chocolate at bodegas/convenience stores on the way to or from school	3 (5%)	17 (18%)	0.026
Buy coffee or tea at bodegas/convenience stores on the way to or from school	3 (5%)	17 (18%)	0.026
Buy salad at fast food places on the way to or from school*	0 (0%)	12 (13%)	0.004
Eat breakfast everyday	17 (30%)	16 (17%)	0.076
Physical Activity Behaviors			
Less active or much less active compared to most people their age	9 (16%)	35 (38%)	0.004
Exercised in order to lose weight or keep from gaining	54 (95%)	67 (73%)	<0.001

weight in the past month			
≥3 hours strenuous exercise (heart beats rapidly) in the past week (e.g. biking fast, dancing fast, running, jogging, swimming laps, rollerblading, soccer, basketball)	27 (47%)	16 (17%)	<0.001
20 or more blocks on bike, skateboard, scooter or rollerblade in a day	12 (21%)	5 (5%)	0.007
≥3 hours of unscheduled physical activity in the past week (activities such as riding a bike, dancing, playing outside or other similar activities; not including scheduled activities done in school, practices, games, classes or walking)	22 (39%)	14 (15%)	0.001
“Dancing” as one of the most common unscheduled physical activities done in the past week*	1 (2%)	27 (29%)	<0.001
“Playing outdoor sports (basketball, soccer, football, or others)” as one of the most common unscheduled physical activities done in the past week	32 (56%)	19 (21%)	<0.001
“Going to a gym” as one of the most common unscheduled physical activities done in the past week	13 (23%)	7 (8%)	0.008
≥3 hours walking alone or with others in the past week*	16 (28%)	39 (42%)	0.078
≥3 hours spent playing video games on an average weekday*	31 (54%)	7 (8%)	<0.001
≥3 hours spent playing video games on an average weekend day*	38 (67%)	7 (8%)	<0.001

*indicates that girls reported healthier behavior than boys

One-Year Morbidity and Mortality of Infants Diagnosed with Birth Asphyxia (BA) or Low Birth Weight (LBW) admitted to the Newborn Unit (NBU) at Moi Teaching and Referral Hospital in Eldoret, Kenya: a Prospective Cohort Study

Author Names: Carolyn Wagner⁵, Laura Ruhl^{2,5}, Winston Nyandiko⁴, Eric Ngetich³, Vincent Kibet³, Astrid Christoffersen-Deb^{4,6}, Julia Songok⁴

Department: Pediatrics

Division: Newborn Medicine

Institution Affiliation: ¹Faculty of Medicine, University of Toronto, Canada, ²Department of Pediatrics, Indiana University, USA, ³Moi Teaching and Referral Hospital (MTRH), Eldoret, Kenya, ⁴Moi University School of Medicine, Eldoret, Kenya, ⁵Ichan School of Medicine at Mount Sinai, USA, ⁶University of British Columbia, Vancouver, Canada

Introduction: MTRH is the referral hospital for Western Kenya and supports a volume of 200-250 admissions each month to its NBU. Morbidity and mortality statistics reveal that BA and LBW make up a majority of admission diagnoses, and that mortality averages are 40% and 35% respectively. Furthermore, for babies who survive the NBU, very little is known about their post-discharge course. There is no record of attrition rates for follow-up or any long-term data on these babies growth, development, and mortality.

Objective (Hypothesis): We aim to determine the one-year morbidity and mortality rate for infants admitted to MTRH NBU with a diagnosis of BA or LBW.

Methods: We will perform a prospective cohort study of 500 newborns admitted to MTRH NBU with a diagnosis of BA or LBW. Data collection will occur throughout hospital stay and follow-up clinic by chart review and parent interview. To develop our study protocol, we performed an assessment of inpatient and outpatient clinical flow and processes. We then developed a flow chart for data collection. Chart reviews will provide inpatient and outpatient data. At months 6 and 9, we will obtain information regarding neurodevelopmental milestones using the Developmental Screening Questionnaire. At the one-year end point, a final telephone interview will be conducted to determine mortality, weight and neurodevelopmental concerns.

Results: We adapted our study protocol including development of data collection tools, a REDcap database, clinical flow algorithms, and an adapted Sarnat staging tool for clinical care in the NBU.

Conclusion: Our study is ready to begin recruiting patients and perform ongoing data collection. We anticipate continuing the study over the next 18 months.

Developing and Validating an Augmented Reality Smartphone Application for Weight Estimation and Dosing in Children

Author Name(s): Temima Waltuch, Kevin Munjal, George Loo, Czer Anthony Lim

Department: Pediatrics

Division: Emergency Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai; Mount Sinai Beth Israel

Introduction: Inaccurate weight estimation is a contributing factor to medical error in pediatric emergencies. Current AHA guidelines recommend use of length-based weight estimation using tools such as Broselow® tape. We developed AiRDose smartphone application that uses augmented reality to provide length-based weight estimates and dosing recommendations.

Hypothesis: Measurements and estimates obtained using AiRDose are equivalent to standard methods.

Methods: AiRDose was developed with assistance from RxHealth using Apple's ARKit. Lengths were measured for children presenting to two emergency departments using AiRDose, Broselow®, and tape-measure; actual weight was obtained from the patient chart. The primary outcome was to compare the length estimated by AiRDose to actual length obtained by tape-measure. Secondary outcome compares estimated weights from AiRDose and Broselow® methods to actual weight. We defined an acceptable difference of 20% between AiRDose and standard measurements as clinically relevant.

Results: 130 children (mean age=59.3±37.3mo) were recruited. Median AiRDose length was 106.7cm *IQR*(32.6) and median tape-measure length was 108.6cm *IQR*(33.5). 100% of AiRDose lengths were within a 10% difference and 96% were within a 5% difference of tape-measure lengths. There was a significant correlation between AiRDose and tape-measure length measurements ($r=0.994$, $p<0.0001$). AiRDose weights significantly correlated with Broselow® weights ($r=0.988$, $p<0.0001$). Both AiRDose and Broselow® weights correlated significantly with the actual weight ($r=0.908$, $p<0.0001$; $r=0.905$, $p<0.0001$).

Conclusions: In this prospective validation study, our preliminary data shows that AiRDose estimated length is very strongly correlated with actual length and that AiRDose estimated weight is very strongly correlated with Broselow® estimated weight and actual weight.

Dental Caries in Pediatric Dental Patients Treated with Dental Rehabilitation under General Anesthesia or Intravenous Sedation at The Mount Sinai Hospital

Author Names: Joshua Woodruff, Laurence Hyacinthe

Department: Dentistry

Division: Pediatric Dentistry

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Dental Caries is the most common chronic childhood disease in the United States. Children requiring dental treatment under general anesthesia/ intravenous sedation are at high risk for the development of new and recurrent dental caries. Previous studies have noted the development of new and recurrent dental caries to be between 37%-52% of patients 6 months after the treatment with general anesthesia. The aim of this chart review is to determine the percentage of pediatric dental patients treated with general anesthesia/intravenous sedation at The Mount Sinai Hospital with new or recurrent dental caries.

Hypothesis: We hypothesize that pediatric dental patients treated at The Mount Sinai Hospital for dental rehabilitation with general anesthesia or intravenous sedation experience a high rate of new or recurrent caries.

Methods: A chart review of pediatric dental patients treated for dental rehabilitation under general anesthesia and intravenous sedation at The Mount Sinai Hospital was conducted by using Dentrix Software patient chart entries for patients treated from 01/01/2015 to 12/31/2017. Patient chart entries reviewed for post-operative examinations completed at: 1 week, 3 weeks, 3 months, 6 months, and 1 year. Statistical analyses will comprise of descriptive statistics and proportions. Data will be reviewed to determine the percentage of patients with new or recurrent caries based on the following criteria: general anesthesia, intravenous sedation and at which post-operative visit caries was noted for general anesthesia and intravenous sedation, respectively.

Results: Pending

Conclusions: Pending

Ankylosis of the Temporomandibular Joint in Pediatric Patients: A Systematic Review and Analysis of 227 Joints

Author Names: Hope Xu, Collin Rozanski, Kasey Wood, Paymon Sanati-Mehrizy, Peter J. Taub

Department: Surgery

Division: Plastic and Reconstructive

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: While surgical interventions for temporomandibular joint (TMJ) ankylosis are well-documented, there is lack of consensus regarding the ideal approach in pediatric patients. Surgical interventions include gap arthroplasty, interpositional arthroplasty, or total joint reconstruction.

Hypothesis: We hypothesized improved outcomes after gap or interpositional arthroplasty in pediatric patients with TMJ ankylosis.

Methods: A systematic review of PubMed and Scopus (Jan 1, 1990-Jan 1, 2017) was performed and included studies in English with at least one patient under the age of 18 diagnosed with TMJ ankylosis who underwent surgical correction. Primary outcomes included surgical modality, preoperative MIO (MIO_{preop}), postoperative MIO (MIO_{postop}), ΔMIO ($MIO_{postop} - MIO_{preop}$), and complications.

Results: 24 case series/reports with 176 patients and 227 joints were included. By independent sample t-tests MIO_{postop} (mm) was greater for gap arthroplasty (30.18) compared to reconstruction (27.47) ($t=4.9$, $p=0.043$), interpositional arthroplasty (32.87) compared to reconstruction ($t=3.25$, $p=0.002$), but not for gap compared to interpositional ($t=-1.9$, $p=0.054$). ΔMIO (mm) was greater for gap arthroplasty (28.67) compared to reconstruction (22.24) ($t=4.2$, $p=0.001$), interpositional arthroplasty (28.33) compared to reconstruction ($t=3.27$, $p=0.002$), but not for interpositional compared to gap ($t=0.29$, $p=0.33$). Weighted-average follow-up time was 28.37 months ($N=164$). 4 of 176 (2.27%) patients reported development of re-ankylosis. There was no significant difference in occurrence of re-ankylosis between interventions.

Conclusions: Given the technical ease of gap arthroplasty and nonsignificant differences in ΔMIO , MIO_{postop} , or re-ankylosis between gap and interpositional arthroplasty, gap arthroplasty should be considered for primary ankylosis repair in pediatric patients, with emphasis on postoperative physiotherapy to prevent recurrence.

Dexmedetomidine For Postoperative Sedation Following Stereotactic Lead Placement: A Case Report

Author Names: Hope Xu, Anthony Bui, Maite La Vega-Talbott, Sheemon P. Zackai

Department: Pediatrics

Division: Pediatric Critical Care

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Dexmedetomidine has become a popular alternative to benzodiazepines and opioids as a safe agent for sedation and pain control, especially in pediatric populations. We present a case of postoperative use of dexmedetomidine in a pediatric patient with a known history of behavioral difficulties who underwent stereotactic lead placement for medically refractory epilepsy.

Hypothesis: We hypothesized safe and effective sedation with dexmedetomidine in a pediatric patient following stereotactic lead placement.

Methods: Chart review was conducted for a 16-year-old male with a history of medically refractory epilepsy and severe autism that was admitted to the pediatric ICU status post stereotactic EEG lead placement. Prior attempts at video EEG monitoring were complicated by patient agitation and required physical restraint and lead replacement. Due to this precedent, the patient was administered a low-dose infusion of dexmedetomidine (0.5 mcg/kg/min) upon extubation following lead placement.

Results: Postoperatively, the patient remained hemodynamically stable and sedated on low-dose dexmedetomidine with grossly unremarkable neurologic exam. Dexmedetomidine did not appear to induce non-stereotypic seizures. Multiple focal electrographic seizures were successfully captured on EEG and the leads were removed five days later without complication. Over the course of admission, the patient was weaned off dexmedetomidine and discharged home in stable condition a day after lead removal.

Conclusions: This appears to be the first case report describing the successful use of dexmedetomidine in a pediatric patient undergoing stereotactic lead placement. Dexmedetomidine may be a safe and viable sedative alternative for children, especially in the presence of significant behavioral difficulties.

Internal Cranial Expansion for Management of Refractory Idiopathic Intracranial Hypertension: A Case Report

Author Names: Hope Xu, Collin Rozanski, Peter J. Taub

Department: Surgery

Division: Plastic and Reconstructive

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Internal cranial expansion (ICE) is a recently developed technique for treating idiopathic intracranial hypertension (IIH) that involves removing and shaving down the skull's inner calvarial table and cancellous bone to increase intracranial volume and reduce intracranial pressure. We present a never before reported case of a pediatric patient with a history of refractory IIH who underwent ICE for symptomatic management.

Hypothesis: We hypothesized symptomatic improvement in a pediatric patient with medically refractory IIH following ICE.

Methods: Chart review was conducted for a 6-year-old male with a history of polycythemia vera who presented with vision changes and headaches. The patient began wearing glasses at age 2 and required three prescription changes within three weeks at age 5. He was diagnosed with papilledema and IIH, and treated with Diamox 125mg BID increased to 750 BID without improvement. He underwent two procedures for optic nerve fenestration, but complained of increasing headache frequency and severity. Preoperative MRI imaging was significant for mild bulging of the optic nerve heads and increased fluid within the optic nerve sheaths bilaterally. At age 6, the patient underwent craniectomies for internal cranial expansion skull remodeling.

Results: No complications were noted at one year follow-up, and the patient reported stabilization of his headaches and vision after ICE. At eighteen months, the patient reported complete resolution of his pre-operative symptoms.

Conclusions: ICE is a safe procedure that can provide symptomatic relief in young children. ICE may have a role in a multidisciplinary management approach to refractory IIH for pediatric patients.

The Association of Prenatal Exposure to Intensive Traffic with Neurobehavioral Development as Reflected by the NICU Network Neurobehavioral Scale (NNNS)

Author Name(s): Xueying Zhang, Emily Spear, Chris Gennings, Paul C. Curtin, Allan C. Just, Robert O. Wright, Annemarie Stroustrup

Department: Pediatrics, Environmental Medicine and Public Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Traffic-related air pollution has been shown to be neurotoxic to the developing fetus in term pregnancies. It is unknown whether there is an increased risk of adverse neurobehavioral outcome in preterm infants exposed to higher levels of air pollution during the fetal period.

Hypothesis: We hypothesize that prenatal exposure to traffic-related air pollution is associated with adverse neurobehavioral outcomes in preterm infants.

Methods: We use residential address during pregnancy proximity to major roads and traffic load as a proxy for traffic-related air pollution exposure. We examined the relationship between proximity to traffic and performance on the NICU Network Behavioral Scale (NNNS), a measure of neurobehavioral outcome in infancy, for 197 preterm neonates enrolled in the Hospital Exposures and Long-Term Health cohort. Air pollution exposure was estimated by density of major roads or vehicles at multiple buffering areas around the residential address. The NNNS was assessed before NICU discharge. We used latent profile analysis (LPA) to classify infants into distinct NNNS performance profiles. Logistic regression analysis was conducted between exposure and LPA groups. Covariates adjusted in models included gestational age at birth, birth weight, post-menstrual age at NNNS assessment, socioeconomic status, race, and maternal smoking status.

Results: We identified three LPA profiles in our study population. High density of major roads within 300 meters of the residential address during pregnancy was significantly associated with one of the less favorable profiles ($P < 0.05$).

Conclusions: Prenatal exposure to intensive air pollution emitted from major roads may impact the early neurodevelopment of preterm infants.

Perceptual Processing and Prediction Error Bias in Autism Spectrum Disorder

Author Names: Yian Zhang, Sylvia Guillory, Jennifer H. Foss-Feig

Department: Psychiatry

Institution Affiliations: Icahn School of Medicine at Mount Sinai; New York University

Institute Affiliation: Seaver Autism Center

Introduction: Autism spectrum disorder (ASD) is defined by social deficits and restricted, repetitive behaviors. Underlying these symptoms are impairments in perceptual processing, which occur in parallel along bottom-up and top-down pathways. Prediction error is the extent to which these two pathways disagree and helps us learn by refining previously-made generalizations. Prior research supports the notion that prediction error bias is present in ASD, and that people with ASD rely more on incoming stimuli than do typically-developing (TD) individuals.

Hypothesis: ASD participants will have a bias for sensory input and therefore, attenuated N1 response to subthreshold tones compared to TD participants.

Methods: Participants included 5 ASD adults (Mean=26.8 years, 4 male), and 5 TD adults (Mean=22 years, 4 male). Starting with three practice blocks, participants heard tones paired with a checkerboard visual and responded whether or not they heard the sounds. Participants then underwent an individual auditory thresholding task to establish four auditory stimuli conditions: 0%, 25%, 50%, and 75% of threshold. During the four main blocks, EEG was recorded while participants responded whether they heard tones on trials that evolved from initially more 75% threshold stimuli to more 0% and 25% trials in later blocks. N1 event-related potential amplitude was measured.

Results: All participants exhibit “conditioned hallucinations,” hearing subthreshold tones when paired with visual stimuli. N1 response did not differ between ASD and TD.

Conclusions: Altered N1 to subthreshold stimuli was not identified in ASD; more data is needed to further test neural correlates of prediction error bias.