Eighteenth 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 Preventive Medicine

Program and Abstracts

Image Courtesy of Sara Benede, PhD (Department of Pediatrics, Division of Allergy and Immunology): “Serotonin-producing cells in the gastrointestinal tract of allergic mice after oral challenge with ovalbumin.”

April 21, 2016
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 Preventive Medicine

Keynote Speaker:
Louis J. Muglia, M.D., Ph.D.
Co-Director, Perinatal Institute and Director, Center for Prevention of Preterm Birth, Cincinnati Children’s Hospital,
Professor of Pediatrics, University of Cincinnati College of Medicine

Child Health Research Day
Steering Committee:

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Lisa M. Satlin, MD
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
Eighteenth Annual Child Health Research Day
Schedule of Events
April 21, 2016 – Hatch Auditorium

7:45-8:00 a.m.  Coffee and Tea

8:00-8:10 a.m.  Welcome and Introduction  
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 
Dr. Robert Wright, MD, MPH, Chair, The Department of Preventive Medicine

8:10-9:05 a.m.  Grand Rounds: The Dr. Howard Rappaport Memorial Lecture
“Preventing Prematurity: Using Human Genomics to Understand Birth Timing”
Louis J. Muglia, MD, PhD
Co-Director, Perinatal Institute and Director, Center for Prevention of Preterm Birth, Cincinnati Children’s Hospital, Professor of Pediatrics, University of Cincinnati College of Medicine

9:05-9:30 a.m.  Breakfast

9:30-11:45 a.m.  Plenary Presentations – Hatch Auditorium  
Moderators: Allan Just, PhD and Anna Nowak-Wegryzn, MD

9:30-9:45 a.m.  Mycobiome Analysis of Lower Airway Secretions from Children with Fungal-Sensitized Severe-Persistent Asthma
Evelyne Magali St. John Sutton, Angela Tsuang, Shankar Viswanathan, Robert Burke, David L. Goldman, Alfin Vicencio

9:45-10:00 a.m.  Modeling Bacterial Mixtures in the Cervix during Pregnancy and the Association with microRNA Expression and Subsequent Gestational Age
Alison P. Sanders, Chris Gennings, Katherine Svensson, Valeria Motta, Adriana Mercado-Garcia Maritsa Solano, Andrea A. Baccarelli, Martha M. Tellez-Rojo, Robert O. Wright, Heather H. Burris

10:00-10:15 a.m.  Early-Life Exposure to Inorganic Arsenic Exacerbates Development of Fatty Liver Disease
Kathryn Bambino, Kirsten C. Sadler, Jaime Chu

10:15-10:30 a.m.  Early-Life Exposure to Ambient Benzene, Toluene, Ethylbenzene and Xylene and ADHD-Related Behaviors and Diagnosis among a Nationally-Representative Sample of Kindergarten Children
Jeanette A. Stingone, Luz Claudio

10:30-10:45 a.m.  Electrophysiological Assessment of Idiopathic ASD
Julia George-Jones, Alexander Kolevzon, Joseph Buxbaum, Paige Siper

10:45-11:00 a.m.  Epigenetic Landscape of a Large Cohort of Children with Neurodevelopmental Disorders
Mafalda Barbosa, Ricky Joshi, Paras Garg, Nihar Patel, Corey Watson, William Gibson, Fatima Lopes, Lisenha Vissers, Silvia de Rubeis, Jennifer Reichert, Patricia Maciel, Tijtske Kleefstra, Han G. Brunner, Joseph D. Buxbaum, Andrew J. Sharp

11:00-11:15 a.m.  Emotion Dysregulation and Gender Differences in Development of Eating Pathology in Adolescents
Melanie Brown, Patrycja Klimek, Nadia Micali

11:15-11:30 a.m.  Investigating Genetic Pathways in Phenotype-Specific Congenital Heart Disease Pathogenesis
Jonathan J. Edwards, Andrew Rouillard, Nicolas Fernandez, Sunita Shankaran, Martin Tristani-Firouzi, H. Joseph Yost, Avi Ma’ayan, Bruce D. Gelb, PCGC Investigators

11:30-11:45 a.m.  Evaluating Home and Office Based Multidisciplinary Programs for Children with Special Health Care Needs
Elaine Lin, Maureen Braun, Nicole Tennermann, Kanasha Seales, Sanite Theophile, Alissa Pierre-Charles, Joseph Truglio

11:45 a.m.-12:45 p.m.  Poster Session and Lunch
Guggenheim Pavilion Atrium

12:45-1:00 p.m.  Poster Presentation Awards Ceremony
Presented by Jaime Chu, MD
Icahn School of Medicine at Mount Sinai
Eighteenth Annual Child Health Research Day
April 21, 2016

WELCOME

We welcome you to the 18th 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 at Mount Sinai and our affiliates, the Mindich Child Health and Development Institute (MCHDI) and the Department of Preventive Medicine. 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 young investigators in the Department of Pediatrics, MCHDI and Department of Preventive Medicine 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 Preventive Medicine
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Mycobiome Analysis of Lower Airway Secretions from Children with Fungal-Sensitized Severe-Persistent Asthma

Author Names: Magali St. John Sutton, Angela Tsuang, Shankar Viswanathan, Robert Burke, David L. Goldman, Alfin G. Vicencio

Department: Pediatrics
Division: Pulmonology
Institution Affiliations: Icahn School of Medicine at Mount Sinai; Albert Einstein College of Medicine of Yeshiva University

Background: Prior data suggest that select patients with severe-persistent asthma and serologic evidence of fungal sensitization may benefit from anti-microbial therapy. Importantly, it is unclear by serologic studies alone whether sensitization is the result of active infection in the lower airways, or repeated but transient exposure to environmental fungal allergens. As such, obtaining evidence of lower airway fungal infection is crucial for accurate diagnosis, and may help dictate which patients might truly benefit from anti-microbial therapy.

Methods: We maintained an IRB-approved protocol to bank bronchoalveolar lavage (BAL) samples from children undergoing flexible bronchoscopy. Over 1.5 years, 93 samples were collected. From this cohort, we sought to identify 3 specific groups of patients for comparative analysis: 1) severe-persistent asthma with fungal sensitization, 2) severe-persistent asthma with no evidence of fungal sensitization, and 3) patients with anatomic/structural airway abnormalities as non-asthma controls (i.e. tracheomalacia, airway stenosis, tracheostomy). DNA was isolated from the BAL pellet, and mycobiome analysis was performed using PCR against pan-fungal rRNA.

Results: BAL fluid from 6 fungal-sensitized asthmatics, 9 non-sensitized asthmatics and 11 controls were analyzed. Species corresponding to serology patterns of sensitized patients (including Aspergillus, Alternaria, Cladosporium, Penicillium) were not detected. Interestingly, Pneumocystis jirovecii (PJ) was found in 6/6 (100%) fungal-sensitized asthmatics, 5/9 (55%) non-sensitized asthmatics (one of whom seroconverted a year later), and 1/11 (9%) controls. Compared with controls, children with severe asthma (both fungal-sensitized and non-sensitized) had significantly higher rates of PJ detection (73 vs 9%, P = 0.002 OR 27.5, CI: 2.6 – 289.3; adjusted Wilcoxon P = 0.026). In addition to PJ, Microbotryomycetes and Rhodotorula species were also highly prevalent in asthma patients compared to controls (adjusted Wilcoxon P = 0.026 and 0.032, respectively). Differences between fungal-sensitized and non-sensitized asthma patients did not reach statistical significance. Lastly, Sterigmatomyces and Cryptococcus species were highly prevalent in non-asthma controls compared to asthmatics (adjusted Wilcoxin P = 0.026).

Conclusions: We found significant differences in lower airway mycobiomes of severe asthmatics compared with controls, with 3 species present primarily in asthma patients, and 2 species present primarily in non-asthma controls. Although the number of patients studied limits interpretation of our findings, the results suggest a role for lower airway mycobiome alterations in severe asthma. Further studies are required to determine the clinical significance of our findings, whether targeted anti-microbial therapy would benefit affected patients, and whether select fungal species protect individuals from an asthma phenotype.
Modeling Bacterial Mixtures in the Cervix during Pregnancy and the Association with microRNA Expression and Subsequent Gestational Age

Author Names: Alison P. Sanders, Chris Gennings, Katherine Svensson, Valeria Motta, Adriana Mercado-Garcia, Maritsa Solano, Andrea A. Baccarelli, Martha M. Tellez-Rojo, Robert O. Wright, Heather H. Burris

Department: Preventive Medicine

Division: Environmental Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health Development Institute

Introduction: Preterm birth (PTB) affects 12% of births and is associated with increased childhood mortality and morbidity. Bacterial vaginosis (BV) is a strong predictor of spontaneous PTB; however, treating BV does not reduce the risk of PTB. Local tissue programming by epigenetic mechanisms may be responsible for this paradox. MicroRNAs (miRNAs) post-transcriptionally control gene expression and play an important role in the host response to microbes.

Hypothesis: We hypothesized that bacterial mixtures in the cervix during pregnancy are associated with shorter gestations and altered miRNA expression.

Methods: We obtained cervical swabs from 80 women 18-20 weeks gestation. Expression of bacterial 16S rRNA from five candidate organisms and 800 human and viral miRNAs were quantified using the NanoString platform. To model the bacteria as a mixture, we applied a weighted quantile sum (WQS) approach and then performed linear regression to examine the associations with gestational age and miRNA expression adjusting for women’s age, parity, education, and environmental tobacco smoke.

Results: Per each decile increment of the WQS bacterial mixture, gestations were 1.4-days shorter (p=0.006). The mixture was also associated with altered expression of five miRNAs including miR-494, 371a, 888, 223, and 23a (p<0.05; q<0.3). The associations with miRNA expression were largely driven by the presence of U. urealyticum, G. vaginalis, and mycoplasma.

Conclusions: Previously, we have shown that miR-223 expression is associated with shorter gestations. Taken together, our findings suggest that subclinical bacterial colonization in the cervix is predictive of PTB and that miRNAs in the cervix may mediate this relationship.
Early-life Exposure to Inorganic Arsenic Exacerbates Development of Fatty Liver Disease

Author Names: Kathryn Bambino, Kirsten C. Sadler, Jaime Chu

Departments: Pediatrics, Preventive Medicine, Developmental & Regenerative Biology

Division: Gastroenterology and Hepatology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Fatty Liver Disease (FLD) is the most common liver pathology in the world. It can be caused by alcohol abuse, diet, viral infection and toxicants. Arsenic ranks as the #1 toxin on the American Toxic Substances and Disease Registry (ATSDR) watch list. Prenatal and childhood exposure to inorganic arsenic leads to increased risk of disease and mortality in childhood, including liver disease. However, the mechanism underlying its effects remains largely unknown and few studies have examined specific outcomes associated with arsenic exposure in a whole animal system.

Hypothesis: Early-life exposure to inorganic arsenic modifies the incidence and progression of fatty liver disease by exacerbating the unfolded protein response (UPR).

Methods: We used qRT-PCR to analyze gene expression. The detection of neutral lipids by oil red O staining was used to determine the incidence of steatosis in zebrafish larvae.

Results: We have established a zebrafish model to determine the mechanism of arsenic toxicity. Gene expression analysis of livers from arsenic-exposed embryos revealed induction of genes involved in the UPR. Consistent with this data, embryos co-treated with inorganic arsenic and ethanol, an inducer of UPR, showed increased lethality compared to embryos treated with either agent alone. Arsenic treated embryos also develop steatosis, a known sequelae of UPR and potential precursor to advanced liver disease.

Conclusions: Early-life exposure to inorganic arsenic leads to induction of the UPR, and results in fatty liver disease. Further studies will elucidate the mechanism by which early life exposure to arsenic leads to progressive liver disease.
Early-life Exposure to Ambient Benzene, Toluene, Ethylbenzene and Xylene and ADHD-Related Behaviors and Diagnosis among a Nationally-Representative Sample of Kindergarten Children

Author Names: Jeanette A Stingone, Luz Claudio

Department: Preventive Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Benzene, toluene, ethyl benzene and xylene (BTEX) are a group of volatile organic compounds previously linked to adverse birth outcomes and cognitive development.

Hypothesis: We hypothesized that greater exposure to BTEX in the ambient air was associated with behaviors suggestive of attention-deficit and hyperactivity disorder (ADHD).

Methods: Using residential ZIP Code at 9 months of age, annual average estimates of BTEX compounds from the 2002 U.S. EPA’s National Air Toxics Assessment were linked to parent and teacher interviews for 4,900 nationally representative children who were born in 2001 and followed from 9 months of age through kindergarten. Kindergarten teachers and parents rated children on six behaviors related to ADHD symptomology using a 5-point likert scale. Children with summary scores in the bottom decile were classified as displaying behaviors suggestive of ADHD symptomology. Multivariable logistic regression models, accounting for study design and adjusted for maternal race, age, marital status, household language, socioeconomic status, neighborhood deprivation index, and home environment were constructed.

Results: Children exposed to the upper quartile of benzene had 1.45 times the odds of displaying behaviors suggestive of ADHD-related symptomology than children with lower levels of exposure (95% confidence interval, CI, 1.11, 1.89). No associations were observed with other BTEX compounds or with parent-reported behaviors. Children exposed to the highest quartile for all BTEX compounds had 1.88 times the odds of having an ADHD diagnosis than other children (95% CI 1.03, 3.42).

Conclusions: These findings add to a growing literature that air pollution may impact children’s neurobehavioral development.
Electrophysiological Assessment of Idiopathic ASD

Author Names: Julia George-Jones, Alexander Kolevzon, Joseph Buxbaum, Paige Siper

Departments: Pediatrics, Psychiatry

Division: Child and Adolescent Psychiatry

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: There is a critical need to identify biomarkers and objective outcome measures that can be used to understand underlying neural mechanisms in autism spectrum disorder (ASD). Visual evoked potentials (VEPs) offer a noninvasive technique to evaluate the functional integrity of visual pathways while probing for disease pathophysiology.

Hypothesis: We expect children with ASD will display weaker responses than typically developing (TD) controls.

Methods: Transient VEPs (tVEPs) were obtained from 32 children with ASD and 32 TD children. A conventional contrast-reversing checkerboard condition was compared to a novel short-duration condition, which was developed to enable objective data collection from severely affected populations who are often excluded from electroencephalographic (EEG) studies.

Results: Children with ASD showed significantly smaller amplitudes compared to TD children at two of the earliest critical VEP components, $P_{60}-N_{75}$ and $N_{75}-P_{100}$, which reflect primarily excitatory and inhibitory postsynaptic activity, respectively. There were no group differences in response latency among groups. All time-domain analyses persisted when the ASD group was divided by IQ. Findings were consistent across both stimulus conditions. Ninety percent of children with ASD completed the short-duration condition compared to 70% for the standard condition.

Conclusions: The current study establishes the utility of a short-duration tVEP test for use in children at varying levels of functioning and describes neural abnormalities in children with idiopathic ASD.

Funding: This study was funded by the Seaver Foundation and Autism Speaks (PI: Siper).
Introduction: Neurodevelopmental Disorders (ND) encompass a spectrum of neurological disease caused by disruption of brain development. Even when using state of the art genetic tests to uncover the etiology of ND – namely microarray and whole genome sequencing (WGS) - pathogenic mutations cannot be identified in 30% of the children. Patients with idiopathic ND cannot benefit from tailored prognosis and follow up and their families are not offered accurate genetic counseling or prenatal diagnosis.

Hypothesis: A fraction of ND patients have deregulation of dosage sensitive genes due to constitutive epigenetic defects, which are missed by conventional molecular genetic methods.

Methods: We performed genome-wide DNA methylation profiling in 498 ND patients who had no pathogenic findings on microarray and WES/WGS. To identify epigenetic defects we compared the methylation profiles of our patients with 1,427 controls. Validation entailed bisulfite sequencing of both child and parents.

Results: We identified 129 epigenetic defects in 101 children. In three patients the epigenetic defect encompassed a known imprinted gene: hypomethylation of MEG3 in the context of matUPD14; hypomethylation of the imprinted locus NAA60/ZNF597, likely representing a novel imprinting disorder; hypomethylation of L3MBTL1 which is currently an autism candidate gene. Recurrent epigenetic defects – i.e. observed in at least two independent ND patients – were detected in six loci.

Conclusions: Overall, our study indicates that pathogenic epigenetic defects likely are the cause of ND in up to 15% of the patients who were refractory to conventional mutation screening approaches, opening up a new area in the study of human disease genetics.
Emotion Dysregulation and Gender Differences in Development of Eating Pathology in Adolescents

Author Names: Melanie Brown, Patrycja Klimek, Nadia Micali

Department: Psychiatry

Division: Child and Adolescent Psychiatry

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Emotion dysregulation has been identified as a transdiagnostic risk factor for general and eating disorder-specific psychopathology in adolescents. Gender may play a critical role in the development of eating pathology. Onset of disordered eating (DE) and eating disorders (ED) typically occurs in puberty for females, when hormonal changes affect emotional lability and eating behaviors; whereas onset in males appears to occur later in adolescence and young adulthood. Few longitudinal studies have investigated gender-specific patterns in the development of DE behaviors in youth. We examined the prospective association between emotional instability and DE in 3800 adolescents from a large-scale, prospective study (ALSPAC).

Hypothesis: We hypothesized that low emotional stability (LES) at age 13.5 years would predict DE behaviors across genders at 16 years.

Methods: We measured emotional stability using a subscale of the International Personality Item Pool (IPIP) and DE from youth self-report. Logistic regression models examined longitudinal associations.

Results: LES at 13.5 years predicted both binge-eating and purging at age 16 for females (p<0.01); females with LES had a two-fold increased odds of binge-eating or purging compared to girls without LES. For males, LES was prospectively associated with binge-eating but not purging (OR=2.96, p<0.01).

Conclusions: DE is common and leads to adverse outcomes in youth. Our results suggest a differential risk pathway from emotional stability to DE in female versus male youth. This has important implications for prevention.
Investigating Genetic Pathways in Phenotype-Specific Congenital Heart Disease Pathogenesis

Author Names: Jonathan J. Edwards, Andrew Rouillard, Nicolas Fernandez, Sunita Shankaran, Martin Tristani-Firouzi, H. Joseph Yost, Avi Ma’ayan, Bruce D. Gelb, PCGC Investigators

Department: Pediatrics

Division: Cardiology

Institution: Icahn School of Medicine at Mount Sinai

Institute: The Mindich Child Health and Development Institute

Introduction: Despite genetic defects being the primary driver of congenital heart disease (CHD) pathogenesis, most of the molecular mechanisms of different CHD phenotypes remains unknown.

Hypothesis: A phenotype-driven gene-set enrichment analysis—focusing on left ventricular outflow tract obstruction (LVOTO), conotruncal defects (CTD), and heterotaxy (HTX)—will identify novel phenotype-specific genetic mechanisms.

Methods: Whole exome sequencing results from 1,228 sporadic CHD trios enrolled in Pediatric Cardiac Genomics Consortium, of which were classified as LVOTO (n=432), CTD (n=444), and HTX (n=183) were compared to 900 control trios. All variants were filtered de novo and in silico. Genes were filtered for heart expression and, using Enrichr, were evaluated for protein complex (CORUM) and GO terms enrichment as determined by Benjamini corrected p ($Q$) < 0.05 and ranked by combined score. In an orthogonal analysis, genes were filtered by Mouse Genome Informatics-knockout phenotypes and evaluated for CORUM and GO enrichment using combined libraries. PPI networks, including intermediated genes, were extracted from reference data. CRISPR-mediated knockout of candidate genes was performed in G0 zebrafish embryos.

Results: Using the unbiased database, CORUM, we identified WAVE-2 enrichment in all cases, a signal driven by $ABI1$, $CYFIP1$, and $NCKAP1$ variants in LVOTOs ($Q = 0.0012$). Both orthogonal analyses also identified enrichment of actin cytoskeleton organization ($Q = 0.013$). An LVOTO-PPI network identified strong interaction between these three and two additional WAVE2 complex genes—$BRICK1$ and $WASF2$—with actin cytoskeleton organization genes, including $CDC42$, $CUL3$, and $RACGAP1$. CRISPR-mediated knockout of these eight genes identified high penetrance of cardiac defects including reversed cardiac looping and dilated ventricle.

Conclusions: Unbiased, orthogonal systems analyses implicate regulation of actin cytoskeleton organization, including WAVE2 complex, in LVOTO pathogenesis. These novel findings are preliminarily confirmed in zebrafish. Future work will explore the molecular mechanisms for these observed phenotypes, which may potentially link recent data connecting planar cell polarity pathway to cardiac development.
Evaluating Home and Office Based Multidisciplinary Programs for Children with Special Health Care Needs

Author Names: Elaine Lin, Maureen Braun, Nicole Tennermann, Kanasha Seales, Sanite Theophile, Alissa Pierre-Charles, Joseph Truglio

Department: Pediatrics
Division: General Pediatrics
Institute Affiliation: Icahn School of Medicine at Mount Sinai

Introductions: As Medicaid reimbursement models evolve there is increasing emphasis on value-based care, especially for high-utilizers of healthcare resources. The Pediatric Visiting Doctors Program at Mount Sinai is a novel program that provides physician-led comprehensive, multidisciplinary care in the home for CSHCN. A clinic-based component was added to this program to serve medically complex children who live outside the home visits catchment area but would benefit from enhanced medical, care coordination, and social work services. Data on outcomes of such comprehensive programs are needed.

Hypothesis: Home and clinic based multidisciplinary programs for medically complex children can decrease ER visits and hospitalizations.

Methods: We reviewed the charts of medically complex children who have been enrolled in the Pediatric Visiting Doctors Program and clinic-based program for at least three months and analyzed emergency room visits, hospitalizations, and lengths of stay. Utilization was compared across equal time periods before and after enrollment.

Results: Since July 2013, 29 medically complex children have been enrolled in the home visiting program and since July 2015, 22 patients have been enrolled in the clinic-based program. Within the home visits program, ED visits have reduced by 56%, hospitalizations by 45% and length of stay by 5 days. Within the clinic based program, ED visits have been reduced by 59%, hospitalizations by 67% and length of stay by 2 days. Further analysis and enrollment are ongoing.

Conclusions: Multidisciplinary care and care coordination delivered at home and in a clinic setting may decrease health care utilization.
Outcomes of 116 Consecutive Open Food Challenges (OFCs) to Extensively-Heated (Baked) Milk

Author Names: Amanda Agyemang, Elizabeth Feuille, Zara Atal, Jeanifer Poon, Ingrid Steinwandtdner, Hugh Sampson, Anna Nowak-Węgrzyn

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Milk allergy is one of the most common food allergies in children. The ingestion of baked milk appears to accelerate development of tolerance to unheated milk in allergic patients, in addition to improving the variety of nutrition.

Hypothesis: We hypothesize that children with milk allergy will tolerate extensively-heated milk open oral food challenges (OFCs) to muffin and pizza. We hypothesize that we can identify clinical markers to predict which patients are likely to tolerate baked milk OFCs.

Methods: We conducted a retrospective chart review of subjects who underwent milk and pizza OFCs. We analyzed the data for overall outcomes and clinically predictive characteristics for tolerance to baked milk.

Results: Eighty-four subjects underwent OFCs to muffin and 72% tolerated full servings. The median serum cow milk-specific IgE of subjects who passed and failed muffin OFCs were 2.8 kU/L and 5.2 kU/L, respectively (p <0.05). Casein-specific serum IgE was significantly lower in subjects who passed the muffin OFC (1.4 kU/L) than those who failed (9.4 kU/L). Thirty-two muffin-tolerant subjects underwent an OFC to pizza and 82% tolerated full servings. The median milk-specific IgEs of those who passed and failed pizza OFCs were not significantly different, at 3.3 kU/L and 3.1 kU/L respectively. The median skin prick wheal diameters were not significantly different in the muffin or pizza OFC.

Conclusion: The majority of milk-allergic children tolerate baked milk OFCs. Cow’s milk and casein serum IgE can help predict which patients will tolerate an OFC to baked milk in a form of muffin.
Circumcision Effect on Breastfeeding

**Author Names:** Ahmed A. Aly¹, Dalia Chehayeb Makarem², Mohamed Khalifa¹, Richa Verma¹, Adil Zaidi¹, Lawrence Noble¹, Daniel A. Rauch, MD¹

**Department:** ¹Pediatrics

**Division:** General Pediatrics

**Institution Affiliations:** Elmhurst Hospital Center; Icahn School of Medicine at Mount Sinai, Elmhurst, NY;²Icahn School of Medicine at Mount Sinai, New York, NY

**Background:** Little data exists on the effect of circumcision (circ) on breastfeeding (BF). A 2007 report from New Zealand reported no effect as did a 2015 PAS poster from California. However both reports were from high exclusive (exc) BF populations (both >60%) and the 2015 report only followed babies for 2 wks. There are no reports of the effect of circ or timing of circ on long term BF in a low excBF population.

**Objective:** To look at the effect of circ on long term BF.

**Design/Methods:** As part of an ongoing QI effort leading toward applying for Baby-Friendly status we looked at the effect of circ, including timing of circ, on excBF and anyBF (per mother's report at HCM visit) of healthy baby boys from our well baby nursery at time intervals of 1wk, 1m, 2m, 4m, and 6m in 2013. Circ is done on the day of d/c, usually D#2 (early), but later if the mother stays longer (later).

**Results:** Our circ rate was 30%. ExcBF rates were consistently low in both circ and uncirc babies with a high of 32% and 37% respectively at 1wk down to 19% and 29% at 6m with circ excBF rates significantly lower at 4m and 6m.

<table>
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<tr>
<th>ExcBF # (%)</th>
<th>hosp d/c</th>
<th>1wk</th>
<th>1m</th>
<th>2m</th>
<th>4m</th>
<th>6m</th>
</tr>
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<tr>
<td>circ</td>
<td>45/367 (12%)</td>
<td>108/334 (32%)</td>
<td>67/217 (31%)</td>
<td>42/172 (24%)</td>
<td>29/150 (19%)</td>
<td>24/136 (19%)</td>
</tr>
<tr>
<td>uncirc</td>
<td>126/845 (15%)</td>
<td>297/806 (37%)</td>
<td>165/550 (30%)</td>
<td>140/499 (28%)</td>
<td>122/408 (30%)</td>
<td>111/384 (29%)</td>
</tr>
<tr>
<td>p value</td>
<td>.223</td>
<td>.147</td>
<td>.812</td>
<td>.355</td>
<td>.013</td>
<td>.036</td>
</tr>
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When later circ babies are removed the excBF rates are much closer and not statistically different. The rates of anyBF are much higher for all babies but still in favor of uncirc starting at 1m.

<table>
<thead>
<tr>
<th>AnyBF # (%)</th>
<th>hosp d/c</th>
<th>1wk</th>
<th>1m</th>
<th>2m</th>
<th>4m</th>
<th>6m</th>
</tr>
</thead>
<tbody>
<tr>
<td>circ</td>
<td>357/367 (97%)</td>
<td>314/334 (94%)</td>
<td>174/217 (80%)</td>
<td>127/172 (74%)</td>
<td>87/150 (58%)</td>
<td>70/136 (56%)</td>
</tr>
<tr>
<td>uncirc</td>
<td>839/845 (99%)</td>
<td>759/806 (94%)</td>
<td>483/550 (88%)</td>
<td>415/499 (83%)</td>
<td>299/408 (73%)</td>
<td>249/384 (65%)</td>
</tr>
<tr>
<td>p value</td>
<td>.005</td>
<td>.918</td>
<td>.007</td>
<td>.007</td>
<td>.001</td>
<td>.062</td>
</tr>
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This remained true even when removing the later circ babies. Early circ trended to higher excBF and anyBF than later circ but was not statistically significant.

**Conclusions:** There was no difference in early BF in our population but circ is associated with lower excBF and anyBF rates starting at 1m, different from the 2007 report. Association does not show causality and circ may be a marker for BF beliefs among our parents who believe in circ. Timing of circ probably does not matter and later circ is likely a marker of maternal issues. Our data suffers from high drop-out rates. However it does help us identify a population that may need alternative or additional BF support.
Quality of Life and Feeding Difficulties Associated with Childhood FPIES and IgE-Mediated Food Allergies

Author Names: Zara Atal, Anna Nowak-Wegrzyn, Marion Groetch

Department: Pediatrics

Division: Allergy and Immunology

Institutional Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: There is little information about the impact of childhood FPIES on quality of life and feeding difficulties. The primary goal of this survey was to compare these outcomes in children with FPIES versus IgE-mediated food allergy.

Hypothesis: Quality of life will be lower and feeding difficulties will be greater for families with children with FPIES.

Methods: Anonymous surveys were administered online to the parents of children with FPIES and IgE-mediated food allergy. The quality of life survey (adapted from Cohen et al, 2004) included 17-questions that were scored from 1 (not troubled) to 7 (extremely troubled); maximum possible score was 119 per each family. The feeding survey (adapted from the Montreal Children’s Hospital feeding scale) included 14-questions and was scored from 1 (not difficult) to 7 (very difficult); maximum possible score was 98 per each affected child.

Results: Sixty-one responses to the FPIES survey and 131 responses to the IgE-mediated food allergy survey were analyzed. The median quality of life score for families with at least one child with FPIES was 78 compared to a median score of 70 for families with at least one child with IgE-mediated food allergy, p=.015. The median feeding difficulties score for children with FPIES (n=70; 9 families with 2 affected children) was 48 versus 31 for children with IgE-mediated food allergy (n=163; 26 families with 2 and 6 families with 3 children), p<.00001.

Conclusions: Parents of children with FPIES report significantly lower quality of life and greater feeding difficulties in their children compared to IgE-mediated food allergy.
Economic Impact of Childhood FPIES and IgE-Mediated Food Allergies

Author Names: Zara Atal, Anna Nowak-Wegryn

Department: Pediatrics

Division: Allergy and Immunology

Institutional Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: To date there has been no comprehensive assessment of the economic burden of FPIES.

Hypothesis: FPIES will have a greater economic impact than IgE-mediated food allergies (IgE-FA).

Methods: Anonymous survey was administered online to the parents of children with FPIES and IgE-FA. The economic impact survey (adapted from Gupta al., 2013), assessed direct medical, out-of-pocket, and indirect costs.

Results: Sixty-one responses to the FPIES survey and 131 responses to the IgE-mediated allergy survey were analyzed. In the past year, children with FPIES had 436 outpatient visits, 36 emergency room visits, and 18 hospitalizations versus 290 outpatient visits, 33 emergency room visits, and 2 hospitalizations for children with IgE mediated food allergies. Out-of-pocket costs were $7233 per child with FPIES, compared to $5029.8 per child with IgE-FA, p=.073. The largest expense for families with a child with FPIES were the costs associated with special diets, $2583.4 per child while for the families with a child with IgE-FA the largest expense was education and supervision, $1531 per child. In the past year, families with a child with FPIES missed an average 8.4 days of school and/or work versus average 4.8 days missed school and/or work days for families with a child with IgE-FA, .000243. However, four families of children with FPIES stated they had to give up their job in order to look after their child. The combined salary loss for these four families totaled $300,000.

Conclusions: Childhood FPIES and IgE-mediated food allergies result in significant direct and indirect costs for healthcare systems and families.
Desensitization of Mast Cell Subsets Associated with Different Manifestations of Food Allergy

Author Names: Sara Benede, Cecilia Berin

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Mast cells are key effector cells of allergic reactions, and are classified into mucosal (MMC) and connective tissue (CTMC) mast cells. The function of these different mast cell subsets in food allergy remains unknown.

Hypothesis: We hypothesized that MMC and CTMC could contribute to different clinical manifestations of food allergy, and underlie route-dependent desensitization to foods.

Methods: We evaluated mast cell activation by protease release using two different mouse models of food allergy with gastrointestinal or anaphylaxis symptoms in response to oral challenge. Bone marrow-derived MMC and CTMC were used to study desensitization in vitro.

Results: Using sensitized mice and evaluating those with and without clinical symptoms, we observed that anaphylaxis was significantly associated with CTMC, but not MMC, activation. In contrast, diarrhea was associated with MMC and CTMC activation. Desensitization of anaphylactic mice with oral immunotherapy led to suppression of both MMC and CTMC activation. In vitro sensitization and activation of MMC and CTMC resulted in signaling (phospho-Syk), degranulation (LAMP-1) and cytokine release (IL-6, IL-13 and TNF-α). All activation parameters could be desensitized in both MMC and CTMC. Extent of desensitization was correlated with extent of initial activation when an antigen-specific system was used (monoclonal DNP-IgE or serum from sensitized mice), and no evidence of bystander desensitization was found. Desensitization of mast cells at a dose below that required for activation was only observed using anti-IgE, and not antigen.

Conclusions: Different manifestations of food allergy are mediated by subsets of mast cells with differing protease profiles. MMC and CTMC show similar potential to become desensitized in vitro, as well as in vivo by an oral route.

Authors: Sejal Makvana Bhavsar, Camille L. Hamula, Tanis C. Dingle

Departments: Pediatrics, Pathology

Division: Infectious Diseases

Institution Affiliated: Icahn School of Medicine at Mount Sinai

Introduction: Mount Sinai Hospital implemented MALDI-TOF MS directly from positive blood culture bottles in the clinical microbiology laboratory in February 2016. This technology provides rapid, accurate identification of bacteria with improved clinical outcomes in adult studies.

Hypothesis: We hypothesize that rapid pathogen identification from positive blood cultures coupled with antimicrobial stewardship will improve clinical outcomes for pediatric patients at our institution.

Methods: Retrospective data was collected for patients admitted to Kravis Children’s Hospital with positive blood cultures from July 1, 2014- June 30, 2015 prior to the initiation of MALDI technology. Prospective data is currently being collected over a one year time period since February 2016. Clinical outcome measures recorded include time from report of a positive blood culture until organism identification and susceptibilities, the duration of broad spectrum and inappropriate antibiotics, and length of stay.

Results: There are 210 patients in our pre-intervention group and thus far 7 patients in our post-intervention group. Average time to organism identification decreased from 41 hours to 11.1 hours. Time to narrowing of antibiotics decreased from 58 hours to 30.2 hours. Time to appropriate discontinuation of antibiotics decreased from 50 hours to 20 hours. Due to the small size of the post-intervention group, statistical significance of these results cannot yet be determined.

Conclusions: Interim data suggests that MALDI-TOF technology leads to more rapid pathogen identifications which decreases the time patients are on unnecessary broad spectrum antibiotics. Its impact on length of stay and mortality will be determined as we continue to collect data through February 2017.
A Rare Cause of Pancytopenia in an Exclusively Breastfed Infant

Author Names: Aisha A. Bobb-Semple¹, Ching See Lau¹, Julie Teruya-Feldstein², Birte Wistinghausen¹

Departments: Pediatrics,¹ Pathology²

Division: Hematology and Oncology

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

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: We describe a 5-month-old infant, born full-term without significant history presenting with lethargy, emesis, pancytopenia, hemolytic anemia and elevated LDH, with broad differential diagnosis including malignancy, infection, toxins, myelodysplastic syndrome, thrombotic thrombocytopenic purpura (TTP), and Wilson Disease, ultimately diagnosed with vitamin B12 deficiency due to occult maternal pernicious anemia masked by folate supplementation.

Case: A 5-month-old breastfed female presented with one month of vomiting and lethargy. A complete blood count and peripheral blood smear review demonstrated pancytopenia, a mean corpuscular volume (MCV) of 96.4 fl, hemolysis and absent reticulocytosis. Elevated LDH raised suspicion of leukemia, prompting bone marrow aspiration and biopsy, which revealed hypercellular marrow with erythroid dysplasia. Direct Coombs and ADAMTS13 activity were negative, discounting TTP. Copper and ceruloplasmin were normal. Lead, arsenic, and infectious agents were not detected. Pancytopenia progressed, including severe neutropenia and requiring transfusion of packed red blood cells. Ultimately, vitamin B12 level was undetectable. Lethargy resolved within 48 hours of intramuscular B12 injection, followed by rapid improvement in reticulocyte count and pancytopenia. On further investigation, patient’s mother had normal hemoglobin and MCV, but undetectable vitamin B12 level and antibodies to intrinsic factor, indicating pernicious anemia, masked by folate supplementation.

Conclusions: Previously reported in infants of mothers with insufficient dietary intake (vegan diet or malnutrition), vitamin B12 deficiency is an unusual cause of pancytopenia in infants and rarely presents in infants of asymptomatic mothers. Inherited errors of vitamin B12 metabolism are even less common. B12 deficiency must be investigated in infants with non-specific hematologic or neurologic symptoms.
Assessment of the Attitudes and Knowledge of Pediatric Medicine Residents towards the Oral Health Assessment and Needs of a High Caries Risk Pediatric Patient Population in East Harlem

Author Names: Anna Brostowitz, Raghbir Kaur

Department: Dental Medicine

Institution Affiliations: Icahn School of Medicine at Mount Sinai;
The Mount Sinai Hospital, Institute for Family Health, New York, NY

Introduction: Oral health is an integral component of overall health. The complex healthcare system benefits from multidisciplinary approaches and collaboration to improve outcomes. Educational seminars held by pediatric dental residents share oral health knowledge with our pediatric medicine colleagues. This residency training creates a framework for improved integration of oral health care into primary care.

Hypothesis: We hypothesized that pediatric medicine residents have had limited oral health training, but believe it is an important part of overall health. A seminar by a pediatric dental resident will increase medicine residents’ knowledge and improve their attitudes towards pediatric oral health.

Methods: Twenty-two pediatric medicine residents participated in the study. Pre-seminar surveys were given collecting demographic data, asking questions of pertinent oral health topics, and attitudes toward pediatric oral health. A 30 minute seminar was given by second year pediatric dental residents. The same survey was given after the seminar, to assess improvement and/or changes.

Results: There was an average of 59% (n=22) correct responses in the pre-seminar survey and 87% (n=18) in the post-seminar survey. Prevention and diagnosis question categories showed the greatest improvement. The residents also reported greater confidence in diagnosing dental caries and discussing oral health with children and families.

Conclusions: This pilot study demonstrates that an educational seminar and a collaborative inter-departmental relationship increases pediatric medicine residents’ knowledge and improves their attitudes toward pediatric oral health. Continuation of this approach will result in prevention of oral disease and improved overall health of our high risk pediatric population.
Ambient PM$_{2.5}$ Exposure in Pregnancy, Maternal Prenatal Antioxidant Intakes, and Infant Autonomic Response

**Author Names:** Kelly J. Brunst$^1$, Chris Gennings$^2$, Brent A. Coull$^3$, Michelle Bosquet Enlow$^{3,5}$, Sri Kannan$^6$, Harish B. Ganguri$^6$, Itai Kloog$^7$, Joel Schwartz$^3$, Robert O. Wright$^2$, Rosalind J. Wright$^{1,2,8}$

**Departments:** $^1$Pediatrics, $^2$Preventive Medicine

**Division:** Pulmonology

**Institution Affiliations:**
- $^3$Harvard School of Public Health, Boston, MA, USA;
- $^5$Boston Children’s Hospital;
- $^6$Southern Illinois University, Carbondale, IL;
- $^7$Ben-Gurion University of the Negev, Beer Sheva, Israel

**Institute Affiliation:** $^8$The Mindich Child Health and Development Institute

**Introduction:** Factors disrupting fetal oxidant balance impact autonomic nervous system (ANS) development.

**Hypothesis:** Prenatal fine particulate matter (PM$_{2.5}$) exposure and maternal antioxidant intakes will impact infant ANS reactivity.

**Methods:** Mother-infant dyads were enrolled prenatally (n=279). Daily PM$_{2.5}$ exposure was estimated using a spatio-temporal model. An index of prenatal antioxidant intakes (vitamins A, C, E, β-carotene, zinc, magnesium, selenium) from an FFQ was derived. Antioxidants and PM$_{2.5}$ exposure averaged over pregnancy were categorized as high/low based on median splits. Dyads completed the Repeated Still-Face Paradigm (SFP-R), a stressor protocol with continuous cardiorespiratory monitoring, when infants were age 6 months. Mixed effects ANOVAs, adjusted for infant sex, activity level over the SFP-R, maternal age, race, education, and prenatal stress, examined associations between PM$_{2.5}$ and antioxidants on respiratory sinus arrhythmia (RSA, parasympathetic response) and T-wave amplitude (TWA, sympathetic response) at baseline and across SFP-R episodes (episode × PM$_{2.5}$ or diet). Interactions between PM$_{2.5}$ and antioxidants were examined (episode × PM$_{2.5}$ × diet).

**Results:** High PM$_{2.5}$ was significantly associated with lower baseline RSA (p=0.04) and decreased RSA reactivity to stress (p$_{\text{episode} \times \text{PM}}=0.0001$); antioxidant intakes were not associated with baseline RSA (p=0.07) or RSA reactivity (p$_{\text{episode} \times \text{diet}}=0.16$). Infants of mothers with both high PM$_{2.5}$ and low antioxidants showed the least RSA reactivity (p$_{\text{episode} \times \text{PM} \times \text{diet}}=0.06$). No significant changes in TWA were found.

**Conclusions:** High prenatal PM$_{2.5}$ predicted decreased infant parasympathetic reactivity to stress. Infants exposed to high PM$_{2.5}$ and low antioxidants were most impacted. Effects on ANS programming may impact disease risk (e.g., cardiometabolic disease, psychopathology).
Adrenal Insufficiency (AI) Testing in X-linked Adrenoleukodystrophy (X-ALD) Identified by Newborn Screen (NBS)

Author Names: Elizabeth Burtman, Lindsey Waldman, Marina Goldis, Lisette Estrella, Melissa Wasserstein, Robert Rapaport, Molly Regelmann

Department: Pediatrics

Division: Endocrinology and Diabetes

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Background: X-ALD is the most common peroxisomal disorder and has a reported incidence in 1:20,000 males. The New York State (NYS NBS) implemented X-ALD screening in December 2013 to allow for early identification of AI and neuro-decompensation. Patients with increased very long chain fatty acids (VLCFA) due to mutation in ABCD1 are referred for endocrine evaluation. NYS guidelines recommend screening males for AI with ACTH and cortisol (F) starting at 6 months.

Objective: We report our experience of screening for AI in X-ALD identified by NYS NBS.

Methods: F was measured before and one hour after administration of cosyntropin 0.25 mg IM and ACTH was measured at baseline. Normal stimulated F at 60 min was defined ≥18 mcg/dL.

Results: 4 male newborns (MN) and 2 male siblings (MS) were tested. Baseline F range was 1.7-23.3 mcg/dl. F at T=60 min was low in 1/4 M. ACTH was elevated in 2/2 tested MN; MN1 had F < 18 mcg/dL and MN4 had normal stimulated F. One of 2 MS had AI.
High Cholesterol Diet (HCD) Impairs K⁺ Secretion in the Rabbit Cortical Collecting Duct (CCD)

Author Names: Rolando Carrisoza-Gaytán, Flores Daniel, Lisa M. Satlin

Department: Pediatrics

Division: Nephrology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: The apical BK channel in the CCD, the nephron segment responsible in the adult for the final regulation of salt and water homeostasis, mediates flow-induced K⁺ secretion (FIKS). The subunit of the BK channels possesses multiple cholesterol recognition/interaction motifs and, in non-renal cells, are inhibited by enhanced cholesterol content in the lipid membrane and hypercholesterolemia. Cholesterol determines membrane fluidity, and changes in membrane cholesterol are expected during postnatal development and in response to HCD.

Hypothesis: HCD, via enhanced incorporation of cholesterol into the plasma membrane (PM), inhibits BK channel-mediated FIKS in the CCD.

Methods: NZW rabbits were randomized after weaning to receive either a standard or a cholesterol enriched diet (HCD; 0.3%) for 4-5 wks, at which time the animals were sacrificed. Kidneys were dissected for (i) isolation of renal cortical membranes for immunoblotting of BKα, ROMK and ENaCβ, and (ii) microperfusion of isolated CCDs to measure net Na⁺ absorption (JNa) and net K⁺ secretion (JK).

Results: Rabbits fed HCD (n=5) vs. control (n=4) showed lower plasma [Na] (130.7±0.7 vs. 137.1±0.7 mM; P<0.001), no difference in plasma [K], and greater serum cholesterol (30,491.6±253.6 vs. 1413.2±231.1 μg/mL; P<0.01). Immunoblotting demonstrated a greater abundance of ROMK and ENaC in total membranes and lower abundance of BK and ENaC in cortical PMs of HCD vs. control (n=2 in each group). In 5 HCD microperfused CCDs, JNa increased from 14.4±7.9 to 32.3±10.0 pmol/min.mm (P≤0.02) in response to an increase in luminal flow rate from 1 to 5 nl/min.mm; this flow-stimulated increase in JNa was less than observed in historical controls (13.2±2.1 to 72.2±10.3; n=13; P≤0.01). In HCD CCDs, JK increased from -4.8±1.6 to -12.4±2.5 pmol/min.mm (P≤0.05) in response to a 5-fold increase in flow rate; these transport rates are half those observed in historical controls (-10.4±2.2 to -22.7±5.6; n=13; P≤0.04).

Conclusions: Our results suggest that HCD blunts flow-stimulated but not basal JNa, and inhibits basal JK and FIKS, which may be due to a reduced abundance of ENaC and BK channels, respectively, in the CCD. Whether enhanced incorporation of cholesterol into the PM of the CCD underlies these changes in transport is currently under investigation.
Health Literacy in Patients with Chronic Kidney Disease

Author Names: Laura Castellanos, Helena Villalobos, Allison Parente, Jeffrey Saland, Rachel Annunziato

Department: Pediatrics

Divisions: Nephrology; Behavioral and Developmental

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Management of children with Chronic Kidney Disease (CKD), including post-transplant patients, can be challenging for nephrology teams and overwhelming for the patient/family. Transition to adult care is an especially vulnerable period and has grown in importance in the last years as more youth with chronic disease survive into adulthood. Health Literacy (HL) defined as “the degree to which individuals have the capacity to obtain, process, and understand basic health information and services needed to make appropriate health decisions” has been associated with poor clinical outcomes in adults but little is known about the relationship in the pediatric population and is an understudied contributor to difficulties during transition.

Hypothesis: We hypothesized that low levels of HL are correlated with poor disease management and transition readiness.

Methods: We are measuring HL with the S-TOFHLA (shortened version of The Test of Functional Health Literacy Assessment), Single Item Literacy Screen, the Brief Illness Perception Questionnaire (IPQ B) and the Developmentally Based Skills Checklist. Medical record review for clinical outcomes and assessment of transition readiness (based on a center-specific checklist).

Interim Results: To date, 75 patients are recruited. As measured by the S-TOFHLA, 95% have “adequate” HL. More variability is shown on the Single Item Literacy Screen (35.4% “more often have trouble reading medical instructions). S-TOFHLA score was significantly correlated with team-rated transition readiness, r = 0.51, p = 0.01.

Conclusions: Preliminary findings suggest that HL is correlated with transition readiness therefore future direction is the creation of a clinic-based intervention to improve HL.
Meeting Oral Health Needs in an Urban Elementary School

Authors Names: Jennifer Chase, Elizabeth Jamiołkowski, Kristin Oliver, Maan Dela-Cruz, Sharon Edwards

Departments: Pediatrics, Dental Medicine

Divisions: School Based Health, Dental Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Children from low income families, children in minority groups, and children with special health care needs suffer disproportionately from oral diseases and tooth decay. Data on dental need and neglect in New York City is limited. The school setting offers an opportunity to provide oral health screening and preventive treatments to at risk children.

Objective: To establish the prevalence of dental disease and determine the feasibility and uptake of a dental screening and fluoride varnish application program in an urban elementary school.

Methods: A team of Dental Residents and School Based Health Pediatricians developed educational materials and screening questionnaire to identify students with dental pain and those who had not been to a dentist in the last 12 months. Materials, questionnaire and consents were backpacked home. Several screening days were scheduled at the school. Oral exams and fluoride varnish application were performed.

Results: A total of 578 students (65%) participated in the oral screening. 413 students, 61% of those participating, had fluoride varnish applied. A total of 283 students (60%) had no caries and routine dental care was recommended, 77 students (16%) had findings of early decay and 117 students (24%) had frank caries or signs of advanced decay. Of students who returned a completed questionnaire 64% reported seeing a dentist in the last 12 months. Never the less, of these students, 15% had signs of early decay and 19% had extensive decay.

Conclusions: Dental screening and fluoride varnish application was widely accepted by families and school administration in an urban public school. Students had high rates of dental disease, despite having seen a dentist in the past year.
Functional Capacity of Regulatory T Cells in Peanut Allergic Children

Author Names: David Chiang¹, Hugh A. Sampson², M. Cecilia Berin²

Department: Pediatrics

Division: ²Allergy and Immunology

Institution Affiliations: ¹Graduate School of Biomedical Sciences; ²Icahn School of Medicine at Mount Sinai, New York, NY

Institute Affiliation: The Mindich Child Health and Development Institute

Rationale: The existence and function of food-allergen specific regulatory T cells in humans remains poorly understood.

Methods: Blood was obtained from peanut allergic (n=20) or control (n=20) subjects. PBMCs were stimulated with crude peanut extract (CPE), evaluated by flow cytometry, and the activation marker CD154 was used to identify peanut-specific T cells. CD25+ cells were removed by magnetic depletion or Tregs were selectively depleted by FACS, and cells were stimulated with peanut extract or staphylococcal enterotoxin B (SEB). Measured outcomes included activation (CD154, t=18hrs) and cytokine secretion (t=5 days). Statistics were conducted using Wilcoxon matched-pairs tests.

Results: Stimulation of PBMCs from allergic but not control subjects with CPE resulted in upregulation of CD154 on Foxp3⁺CD25⁺CD127low cells. CD25 depletion prior to peanut stimulation abolished the appearance of CD154+ Tregs, indicating that these cells were derived from the CD25+ population at baseline. CD25 depletion led to significant (p<0.05) increases in peanut-induced IFNγ, TNFα, IL-21, and IL-6, whereas surprisingly there were significant decreases in peanut-induced Th2 cytokines: IL-5, IL-9, and IL-13. Similar patterns were observed with SEB stimulation. Immune profiling of CD25-depleted PBMCs demonstrated depletion of basophils as well as Tregs. Selective Treg-depletion by FACS led to significantly increased Th1 and Th2 cytokine responses to peanut.

Conclusions: Our data demonstrate the presence of functional peanut-responsive Tregs in peripheral blood of peanut allergic subjects, but not healthy controls. Our data also demonstrate that basophils or other CD25+ cells, but not Tregs, contribute to the production of Th2 cytokines central to the pathogenesis of food allergy.
Prenatal Exposure to Particulate Air Pollution and Anthropometry in Urban Children: Sensitive Windows and Sex Difference

Author Names: Yueh-Hsiu Mathilda Chiu1,2, Hsiao-Hsien Leon Hsu2, Ander Wilson3, Brent A. Coull3,4, Itai Kloog5, Joel Schwartz4, Robert O. Wright2,3, Rosalind J. Wright1,2,3

Departments: 1Pediatrics, 2Preventive Medicine, 2,4Environmental Health, 3Biostatistics, 5Geography and Environmental Development

Divisions: 1Pulmonology, Environmental Health

Institution Affiliations: 1Icahn School of Medicine at Mount Sinai; 2,4Environmental Health; 3Biostatistics; 5Geography and Environmental Development

Institute Affiliation: 3The Mindich Child Health and Development Institute

Introduction: Prenatal air pollution exposure may be associated with childhood obesity. Timing of exposure may play an important role in these health effects but has not been extensively studied.

Hypothesis: We hypothesized that sex-specific sensitive prenatal windows of exposure to fine particulate matter (PM2.5) on anthropometry in children can be identified using an innovative analytic method.

Methods: Analyses included 240 full-term (≥37 weeks) children. Mothers' prenatal daily PM2.5 exposure was estimated using a validated satellite-based spatio-temporal resolved model. Body mass index z-score (BMI-z), fat mass, fat free mass, % body fat, tricep & subcapular skinfold, waist & hip circumference were assessed at age 4.0±0.7 years. Using Bayesian distributed lag interaction models (BDLIMs), we examined sensitive windows of weekly averaged PM2.5 levels and these measures, and effect modification by sex.

Results: Mothers were ethnically mixed (57% Hispanic, 25% Black) and most had ≤12 years of education (67%). BDLIMs adjusting for child age, sex, maternal age, education, race, and pre-pregnancy BMI showed significant positive relationships of PM2.5 exposure at 8-17 weeks gestation with BMI-z and exposure at 16-22 weeks with fat mass only in boys, and exposure at 10-13 weeks with waist circumference in both boys and girls. PM2.5 at 10-29 weeks gestation was significantly positively associated with waist-to-hip ratio in girls. The estimated cumulative effects across pregnancy were \( \beta = 0.21 \) (per unit increase in PM2.5; 95%CI=0.03-.37) for BMI-z and \( \beta = 0.36 \) (95%CI=-.12-.68) for fat mass in boys, and \( \beta = 0.02 \) (95%CI=.01-.03) for waist-to-hip ratio in girls.

Conclusions: PM2.5 exposure in early-to-mid pregnancy was associated with BMI-z and fat mass in boys, and across a wider window with waist-to-hip ratio in girls. Methods to better characterize vulnerable windows can inform sex differences and provide insight into underlying mechanisms.
Caregiver Satisfaction with Food Allergy Education Approaches

Author Names: Niti Y. Chokshi, 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: Education about avoidance and emergency treatment is integral to the management of children with food allergy.

Hypothesis: We developed a tablet-based kiosk containing educational videos and additional materials and evaluated caregiver satisfaction.

Methods: An anonymous questionnaire was administered to parents/caregivers in rooms with and without the kiosk. Data were evaluated with Chi-square and T-tests.

Results: Eight-eight surveys were completed, 40 from rooms with and 48 without the kiosk. Time spent using the kiosk was: <5 minutes (28%), 5-10 minutes (67%), and >10 minutes (6%). On a 1-7 scale (negative to positive, median score), respondents regarded the kiosk modality: poor-excellent (6), organization (6), boring-interesting (5), useless-useful (6), and difficult-easy to navigate (6). The scores given for the kiosk’s educational material scored (1-7 scale, median): poor-excellent (5.5), confusing-straightforward (6), boring-interesting (6), useless-useful (6), and not applicable-applicable (7). On a 1-7 scale (median), patients would recommend both the kiosk educational material (score 6) as well as the device (score 6) to others. In kiosk containing rooms, the most helpful education reported was MD education (66%) followed by kiosk education (24%). There was no difference between the two groups for perceived waiting time (p=0.775) or visit satisfaction (p=0.304). There was no significant difference in comfort level of managing food allergy when comparing kiosk-containing to control rooms.

Conclusions: Patients overall felt positively regarding the food allergy education kiosk and most would recommend this device to other parents. However, compared to standard education, the kiosk did not significantly change patient visit satisfaction, comfort with food allergy or perceived waiting time.
Introduction: Preterm infants spend the “third trimester” in a stressful hospital environment. Salivary cortisol levels have been used to evaluate stress response in children and adults and may allow objective assessment of fluctuations in newborn stress state. Little is known about infant cortisol level during the NICU hospitalization.

Hypothesis: Parent visitation reduces stress measured by salivary cortisol in hospitalized preterm infants.

Methods: Infants born at gestational age 28-32 weeks were enrolled upon NICU admission. Morning and afternoon saliva collection was attempted weekly. Associations between demographic and clinical factors and salivary cortisol level were evaluated by univariabl

Results: 83 cortisol measurements from 19 infants were analyzed. 50 measurements from 17 infants were paired. There was no diurnal cycle to infant cortisol level. Gestational age, chronological age, handling, suctioning, respiratory support, and heel stick were associated with salivary cortisol level. The mean decrement in salivary cortisol level following parent visitation was 0.75 nmol/L and the mean increase in salivary cortisol level for infants not experiencing a parent visit was 0.49 nmol/L (p > 0.05).

Conclusions: There may be a relationship between parent visitation and decreased salivary cortisol level among hospitalized preterm infants.
trappc11 is Required for Protein Glycosylation in Zebrafish and Humans


Department: Pediatrics
Division: Gastroenterology and Hepatology
Institution Affiliation: Icahn School of Medicine at Mount Sinai
Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Fatty liver disease (FLD) is a condition that can potentially lead to liver fibrosis, cirrhosis and cancer. The prevalence of FLD is increasing and is considered to be a public health issue, so any insight into its pathomechanism is invaluable to understanding this common disorder.

Methods: We utilized a zebrafish model for FLD who harbor a mutation in trappc11, encoding a member of the transport protein particle (TRAPP) trafficking complex. We analyzed trappc11 mutants and drug treated larvae, with qPCR for genes involved in the unfolded protein response (UPR), glycosylation, and terpenoid biosynthetic pathways. We also examined human cells after siRNA-mediated knockdown of TRAPP complex members, and assayed lipid accumulation in human patient cells with TRAPPC11 mutation.

Results: trappc11 mutant zebrafish have abnormal ER to Golgi trafficking, but this was not sufficient to induce a stressed UPR. Instead, defective protein glycosylation, reduced levels of lipid linked oligosaccharide (LLO), and upregulation of genes in the terpenoid biosynthetic pathway, was present. Inhibition of the terpenoid pathway phenocopied the stressed UPR, and was synthetically lethal with trappc11 mutation. In human samples, knockdown of trappc11 resulted in hypoglycosylation, and fibroblasts from humans with TRAPPC11 mutation accumulated lipid droplets.

Conclusions: Mutation of trappc11 in zebrafish leads to hypoglycosylation, likely via repression of the terpenoid biosynthetic pathway. Hypoglycosylation and lipid accumulation following TRAPPC11 perturbation in humans suggests a similar pathomechanism. These data identify a new, conserved role for TRAPPC11 in LLO biosynthesis and protein glycosylation, in addition to its established function in vesicle trafficking.
Coping with Patient Death on Pediatric Liver Transplant Teams

Author Names: Sarah E. Duncan, Ronen Arnon, Christie DiPietroantonio, Christopher S. Knight, Jaime Chu, Rachel A. Annunziato

Department: Pediatrics

Division: Behavioral and Developmental

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: The purpose of the present study is to determine a) how personnel on pediatric liver transplant teams cope with patient death and b) to offer recommendations for quality improvements.

Methods: Medical directors of 25 randomly selected pediatric liver transplant centers from different regions in the US were sent a Qualtrics survey link and requested to circulate the link to all physicians, nurses and support personnel on their team. The survey included questions about available resources, a needs assessment, and standardized measures of adjustment (Maslach Emotional Exhaustion Scale, EE, and the Bereavement Experiences Scale, BES).

Results: Completed surveys were received from 72 respondents (32 physicians, 32 nurses, 8 social workers). Most described having no formal training in coping with patient loss (83.3%); overwhelmingly (97.2%), respondents thought that formal debriefing procedures would be helpful; this was routine for just 50%. Mean scores on the EE (2.92) and the BES (1.38) were comparable to normative data, but nurses, 3.28 (SD=1.18), and social workers, 3.64 (SD=.08), reported significantly more EE than physicians, 2.39 (SD=1.02), F=7.20, p=< .01. Respondents who have formal debriefing procedures reported significantly less EE, 2.60 (SD=1.21), than those who do not, 3.24 (SD=1.05), t=-2.40, p=.02.

Conclusions: Although overall team members adjust well, findings suggest that provision of routine debriefing after loss is desired and may be associated with better coping. Further research is needed to determine if higher rates of emotional exhaustion among nurses and social workers is associated with loss and patient care responsibilities specific to these roles.
Treatment of Colitis by Epicutaneous Immunotherapy in a Murine Model

Author Names: David Dunkin, M. Cecilia Berin, Lucie Mondoulet, Zaruhi Hovhannisyan, Steven Tobar, Alina Iuga, Thibaut Larcher, Garabet Yeretssian, Pierre-Henri Benhamou, Hugh Sampson

Department: Pediatrics

Division: Gastroenterology and Hepatology

Institution Affiliations: Icahn School of Medicine at Mount Sinai, New York, NY; DBV Technologies, Bagneux, France; Columbia College of Physicians and Surgeons, New York, NY; National Veterinary School, Nantes, France

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Patients with Crohn’s disease have a defect in the induction of T-regulatory cells (Treg) via the gut. When Tregs are generated externally in response to food antigen and infused into patients, they suppress inflammation in Crohn’s via bystander suppression. We hypothesized that Tregs could be induced by applying antigen to intact skin using an epicutaneous delivery device, Viaskin®, and after their migration to the gut could abrogate colitis via bystander suppression.

Methods: C57BL/6 mice were exposed epicutaneously for 48 hours once a week to Viaskin patches containing ovalbumin (Viaskin-OVA). To determine if exposure blocked T-effector responses, mice were immunized with OVA, and cytokine production by draining lymph nodes (LN) was assessed by ELISA. Treg development in the MLN, spleen and intestines was analyzed. To determine whether Tregs from skin draining LNs could migrate to the gut to suppress colitis, Tregs from skin draining LNs or MLNs were co-transferred with CD45RBHI T cells in to RAG−/− mice. Mice were assessed for weight loss, colonic cytokine production and histology. Finally, to determine if epicutaneous tolerance induction could directly abrogate colitis, RAG−/− mice with colitis induced by the transfer of CD45RBHI T cells were epicutaneously exposed to Viaskin-OVA and then gavage fed OVA to activate Tregs and induce homing to the gut. Weight loss, colonic inflammatory cytokine production and histology were assessed.

Results: Epicutaneous exposure to OVA induced tolerance with suppression of OVA-specific IFN-γ from draining LNs. OVA exposure induced proliferation of OVA-specific Tregs in the spleen, MLN, and intestines. Tregs isolated from skin draining LNs were able to suppress the development of colitis as well as those isolated from MLNs. In this transfer model of colitis, 3 epicutaneous OVA exposures followed by 1 oral OVA feeding prevented weight loss (p<0.05), decreased colonic IFN-γ and IL-17 production (p<0.05), and abrogated histological colitis (p<0.05).

Conclusions: Epicutaneous exposure to OVA induces Tregs, which migrate to the gut and abrogate colitis via bystander suppression. Epicutaneous tolerance induction has potential as a treatment for Crohn’s disease and warrants further study.
Correct Use of Epinephrine Autoinjectors in Relation to Health Literacy in Patients with Food Allergies

Author Names: Maureen Egan, Julie Wang

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: Anaphylaxis occurs in an estimated 1.6% of the United States population with food allergies being the trigger in 31% of cases. Epinephrine Autoinjectors (EA) are essential in management of anaphylaxis. A national assessment of health literacy noted that 44% of adults have below or basic health literacy, which is associated with adverse health outcomes. No study to date has examined the correct use of EA in relation to health literacy.

Hypothesis: We hypothesized that there may be an association between health literacy and the ability to demonstrate appropriate use of an EA.

Methods: Parents of children with known food allergies who had previously been prescribed an EpiPen were included. Participants were administered the Newest Vital Sign (NVS), a validated instrument to assess health literacy. Additionally, subjects demonstrated use of an EpiPen that was evaluated based on a previously established 6 step checklist.

Results: Of the 70 parents included, 41.5% had a score of 0-3 on NVS indicating a possible/high likelihood of limited literacy while 58.5% participants had a score of 4-6 indicating likely adequate health literacy. Amongst patients with a NVS score of <4, 31% demonstrated correct use of an EpiPen (correct use defined as proper demonstration of all 6 steps). Among patients with a NVS of ≥4, 44% demonstrated correct use. The difference between the groups was not statistically significant (p=0.27).

Conclusions: Correct use of EpiPen was demonstrated in less than 50% of parents of children with known food allergies. The ability to correctly demonstrate use of EpiPen was not correlated with health literacy adequacy.
Partially Hydrolyzed Whey Formula for Cow’s Milk Allergic Patients

Author Names: Maureen Egan, Tricia Lee, Jade Andrade, Galina Grishina, Gustavo Gimenez, Hugh Sampson, Supinda Bunyavanich

Department: Pediatrics
Division: Allergy and Immunology
Institution Affiliation: Icahn School of Medicine at Mount Sinai
Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: Cow’s milk allergy (CMA) is the most common food allergy in children. For children with CMA, a hypoallergenic formula is recommended including extensively hydrolyzed or amino-acid based formulas. Partially hydrolyzed whey formula (pHWF) is not currently recommended for children with CMA. However, a prior publication identified a child able to tolerate pHWF, a more palatable and less costly formula, despite having CMA.

Hypothesis: There may be additional patients with CMA who are able to tolerate pHWF. We sought to better describe this phenotype.

Methods: Patients with CMA between the ages of 6 months to 18 years were screened. Inclusion criteria included a recent positive skin prick test or specific IgE level to cow’s milk and a clinical reaction to cow’s milk during an oral food challenge (OFC), among others. Enrolled participants underwent OFC to pHWF with additional skin, serum, and laboratory testing, including a Luminex-based peptide assay (LPA), to better characterize their CMA.

Results: 10 patients were included whose ages ranged from 11 months to 10.8 years. Milk-specific sIgE ranged from 1.16 kU/L to >100 kU/L, and casein-specific IgE ranged from <0.10 kU/L to >100 kU/L. All 10 patients failed the OFC to the pHWF (i.e. had a clinical reaction) indicating they were not able to tolerate the formula. Immunoblotting and LPA results supported the notion that these patients reacted to residual casein in pHWF.

Conclusions: pHWF should be avoided in patients with CMA. The initially described patient who was able to tolerate pHWF despite having CMA was likely an outlier.
Estimation of Pulmonary to Systemic Flow Ratio by Transthoracic Echocardiography is Poor in Hemodynamically Significant Left to Right Shunt

Author Names: Erin Faherty, Hari Rajagopal, Simon Lee, Barry Love, Shubhika Srivastava, Ira Parness, Santosh C. Uppu

Department: Pediatrics

Division: Cardiology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Transthoracic echocardiographic (TTE) estimation of the pulmonary to systemic flow ratio (Qp/Qs) was validated with oximetry in a small number of children and young adults. It is routinely used in clinical practice by noncongenital cardiologists as a noninvasive tool. We sought to assess its real world applicability with particular focus on hemodynamically significant shunt lesions.

Hypothesis: TTE derived Qp/Qs might not accurately detect hemodynamically significant shunt.

Methods: Retrospective single institutional review of TTE’s in patients with secundum atrial septal defect prior to cardiac catheterization (cath) from 2013-2015 was performed (n= 47). Qp/Qs was calculated from stored clips by previously described methods. These results were correlated with those obtained by oximetry. Patients were subdivided into two age groups <21 (Group 1) and ≥22 years (Group 2). Thirteen heart transplant patients without intracardiac shunt were used as controls for statistical analysis.

Results: Forty four subjects were evaluated (age range 4-69 yrs). Group 1 n=19; median age 10 yrs (range 4-21 yrs); Group 2 n=25; median age 49.5 yrs (range 22-69 yrs). Mean Qp/Qs derived by cath and TTE were 2.07±0.9 vs. 2.54±1.4 (p 0.01). Overall correlation was poor between the methods (r² = 0.29, p 0.0001) (figure). The correlation continued to be poor for Groups 1 & 2 (r² = 0.2, p 0.05 and r² = 0.4, p 0.0006 respectively). TTE derived Qp/Qs has poor sensitivity (58%) and PPV (63%) to identify hemodynamically significant shunt (Qp/Qs ≥2); the sensitivity (79%) and PPV (77%) improves to identify shunts ≥1.5 at the cost of specificity (65%) and NPV (68%).

Conclusion: TTE estimated pulmonary to systemic flow ratio although routinely utilized in clinical practice has poor correlation with oximetry derived Qp/Qs. The test performs poorly in all age groups and especially in detecting a hemodynamically significant shunt.
Participant’s Experience with Food Allergy Clinical Trials

Author Names: Jennifer Fishman, Jaime Ross, Sally Noone, Beth Strong, Zara Atal, Carly Ehritz, Jessica Gau, Julie Wang

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: Currently there are no treatments or cures for food allergies. Several treatment options are under investigation. This survey was done to examine why people with food allergies choose to participate in research clinical trials, to inquire where they learned about the trial, and to gain feedback from participants regarding their experiences in clinical trials.

Hypothesis: We hypothesized that families participated in clinical research trials to seek a cure for food allergy.

Methods: Anonymous online Qualtrics survey was emailed to our Food Allergy Institute email list.

Results: There were 81 respondents, of which 90% have participated in a clinical trial at Mount Sinai. 30% have a child who is currently participating in a trial. Main motivations of responders to participate in a study are the hope of a cure for themselves and/or their child (74%), and to help find a cure for others (60%). Participants learned of the study by physician referral (56%) and Mount Sinai newsletter/website (37%). From participating in clinical trials, approximately half have made changes in the way they manage their food allergies. They feel less limited in social activities, have expanded their child’s diet, and more frequently eat in restaurants. 90% would recommend participation in a clinical trial to others.

Conclusions: The majority of respondents participate in clinical research trials because they are hopeful for a cure for themselves and/or others with food allergy. They have had a positive experience and would recommend that others consider participating in a food allergy clinical trial.
Constructing a Comprehensive Map of Long Non-Coding RNAs in Brain Using Integrated Transcriptomic Approaches

**Author Names:** Nancy J. Francoeur\textsuperscript{1,2,4-7}, Xiao Xu\textsuperscript{1,2,4-7}, Brigham Hartley\textsuperscript{1,3,6}, Kristen Brennand\textsuperscript{1,3,6}, Harm van Bakel\textsuperscript{2,7}, Elizabeth Tseng\textsuperscript{8}, Robert Sebra\textsuperscript{2,7}, Dalila Pinto\textsuperscript{1,2,4-7}

**Departments:** Psychiatry\textsuperscript{1}, Genetics and Genomic Sciences\textsuperscript{2} and Neuroscience\textsuperscript{3}

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai;

**Institute Affiliations:** The Mindich Child Health and Development Institute\textsuperscript{4}; Seaver Autism Center\textsuperscript{5}; Friedman Brain Institute\textsuperscript{6}, Icahn Institute for Genomics and Multiscale Biology\textsuperscript{7}, Pacific Biosciences\textsuperscript{8}

**Introduction:** Long non-coding RNAs (lncRNAs) have been implicated in gene transcriptional regulation, and have been suggested to play a role in neurodevelopmental disorders (NDD). We have established a lncRNA enrichment approach that is used in combination with both short-read and long-read RNA-sequencing technologies.

**Hypothesis:** Our lncRNA capture-sequencing (SeqCap) technique allows us to identify splice variants and full-length isoforms of lncRNAs expressed in brain, which will substantially improve our ability to incorporate lncRNA expression profiles into the analysis of gene regulatory networks underlying NDDs.

**Methods:** We used lncRNA-SeqCap to specifically target and preferentially sequence lncRNA transcripts in each sample and applied our custom analysis pipeline to annotate and classify novel transcripts uncovered by our experimental technique to link isoforms to gene biotypes.

**Results:** We have generated short-read lncRNA-SeqCap for five prefrontal cortex post-mortem brain samples, as well as astrocytes, neural progenitor cells and neurons derived from iPSCs of a single individual. Capture efficiency of spiked-in external RNA controls indicated an average 1,000-fold enrichment of captured transcripts when comparing the pre- and post-capture libraries, as measured by qPCR. To identify full-length isoforms, we further combined long-read sequencing (IsoSeq) with lncRNA-SeqCap. Assessment of the IsoSeq-capture efficiency showed >100-fold enrichment of target expression levels post-capture. We have found many novel isoforms for lncRNA as well as protein-coding genes, and have uncovered evidence of interleaved transcripts arising from a combination of both gene biotypes.

**Conclusions:** Our protocol allows us to construct an improved map of lncRNA gene structures that will serve as valuable resource to the field.
Normal Left Ventricular Size in Premature Newborns by the Echocardiographic Bullet Method

Author Names: Geri Galotti, Ken Bayle, Jill Nielsen-Farrell, James C. Nielsen, Jie Yang, Laurie Panesar

Department: Pediatrics
Division: Cardiology
Institution Affiliation: Stony Brook School of Medicine

Introduction: Limited normative data exists for the size of the LV in premature newborns to aid with staging of hemodynamically significant PDAs.

Hypothesis: To establish the normal range for LVEDV by Bullet method using retrospective ECHO data in subjects with BSA<0.2m2 and explore the optimal method to index LVEDV in premature newborns with PDA.

Methods: LVEDV was calculated retrospectively for 85 normal neonates and analyzed to produce centile nomograms and tabular data for Z-score calculation. The utility of the normal ranges for LVEDV was compared to the current standard for LV size in 19 subjects treated for a PDA. In addition, comparison of LVEDV was made to prior published data of larger newborns and infants.

Results: The indexed LVEDV for the normal cohort was 63.0±11.2 ml/m^2.76 which is significantly different than the published normal value of 70.4±9.1 ml/m^2.76, p<0.001 in larger newborns and infants. For the comparison cohort, the 19 PDA subjects had a mean gestational age of 25±2 weeks and weight of 0.87±0.3kg. The mean LVEDVi of the PDA group (97.7±22.2ml/m^2.76) was significantly larger than the normal group (63.0±11.2 ml/m^2.76), p=0.002. The sensitivity of LVEDV was better (68% versus 11%) compared to LV dimension in detecting a large LV in PDA subjects.

Conclusions: LVEDV calculated by the Bullet can be utilized to determine LV size in neonates with a BSA<0.2m2. An elevated LVEDV > +2 Z-scores can serve as objective data to aid in management decisions in patients with PDA.
Rate of Food Introduction after a Negative Oral Food Challenge in the Pediatric Population

Author Names: Jessica Gau, Jaime Ross, Sally Noone, Beth Strong, Zara Atal, Carly Ehritz, Jennifer Fishman, Julie Wang

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Oral food challenges (OFCs) are performed to diagnose food allergy persistence or outgrowth. This study evaluated the rate of food introduction following a negative OFC and reasons for failure to incorporate the foods into the diet.

Hypothesis: We hypothesize that some children continue to avoid eating the food that they have outgrown even after passing a food challenge to that food.

Methods: An anonymous survey was distributed to parents of children with food allergy during follow-up visits. Fifty respondents reported having had 109 OFCs. Data collected included challenged food, result of the OFC, the frequency of food introduction, and reasons for failure to incorporate the challenged food into the diet on a regular basis.

Results: Of the 109 OFCs, 82 were negative OFCs. Of the 82 negative OFCs, 73 (89%) added food into the diet, 6 (7.3%) did not incorporate the food into their diet. For those who added the food into their diet (n=73), 45 (62%) consumed it daily or weekly, while 28 (38%) consumed the food less often. Reasons for infrequent consumption included dislike (11), fear (9), and the food not being part of the family’s diet (5). The foods most avoided were tree nuts, peanut, and chickpea.

Conclusion: After a negative OFC, about 7% of the foods were still being avoided completely while 34% are seldom introduced (ie: monthly or less) for various reasons. Post-challenge counseling and dietary guidance to address potential barriers may be beneficial to increase rates of consistent food introduction following negative OFCs.
High Dose Steroid Therapy for Liver Abscess in Patients with Chronic Granulomatous Disease (CGD)

Author Names: Yael Gernez¹, Tukisa Smith¹, Angela Tsuang¹, Paul J. Maglione¹, Steven M. Holland,² Charlotte Cunningham-Rundles¹

Departments: ¹Pediatrics and Internal Medicine

Institution Affiliations: ¹Icahn School of Medicine at Mount Sinai Hospital; ²National Institute of Health, Bethesda

Institute Affiliation: Jaffe Food Allergy Institute

Introduction: Liver abscesses in CGD are difficult to treat. IR guided cultures are important, but liver abscesses in CGD are thick and drainage is impractical. Surgery has been most often done, but abscesses may be large, locations difficult and these sites may not heal well.

Hypothesis: High dose steroids and intensive antibiotic coverage may result in better outcomes.

Method: We report two cases of liver abscess in patients with X-CGD. One is a 17-year-old male, with a history of periodontitis and neck abscesses, who had fever for 2 weeks. The second is a 27-year-old male, with previous liver abscess at age 5, and more recently neck and leg abscesses with Phellinus tropicalis [Ramesh et al. J Clin Immunol. 2014 Feb;34(2)]. He had abdominal pain and S. aureus sepsis. Both patients were given broad-spectrum antibiotics with Vancomycin along with antifungal coverage. IR guided aspiration of abscesses was done for microbiology. Surgery was precluded by location, size and risk of the liver abscesses. Both patients were started on steroids (1 mg/kg/day initially for 1 month then slow taper) with monthly CT scans to follow progress.

Results: Under this treatment for one month, both patients have clinically and biologically significantly improved; their liver abscesses have remained stable.

Conclusions: High dose steroids associated with antibiotics may offer an alternative therapy when surgery is precluded.
Hemoglobin A1C in Patients on Growth Hormone During the Initial Year of Treatment

Author Names: Marina Goldis, Lindsey Waldman, Kristen Williams, Christopher Romero, Molly Regelmann, Elizabeth Burtman, Vivienne Cabreza, Elizabeth Wallach, Robert Rapaport

Department: Pediatrics

Division: Endocrinology and Diabetes

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Background: Growth hormone treatment has been suggested to impair glucose metabolism (1-3). While GH therapy did not affect the incidence of type I DM in children (4), diabetes incidence appears to be increased in GH-deficient adults with risk factors (5). Recent reports suggest that GH-deficient pediatric patients have been showing an increase in fasting glucose and A1C level, without evident changes in glucose tolerance or HOMA after 1 year of therapy (6).

Aim: Report the correlation of A1C to GH therapy in GH deficient and non-deficient pediatric patients.

Methods: Patients included were seen at Mount Sinai pediatric endocrinology. Retrospective chart review was done for patients who had a GH stimulation test between September 2012 and October 2013. The Mann-Whitney U test was used for statistical analysis.

Results: Of the 211 patients reviewed, 137 were excluded because of lack of data, other endocrinopathies or other significant past medical history. 74 patients with initial consultation, growth hormone stimulation test and 12 month follow up were reviewed. 19 patients were female and 3 were born small for gestational age (SGA). 26 patients were pre-pubertal at stimulation and at 12 months, while 48 patients were pubertal at 12 months. All patients had normal electrolytes, calcium levels, liver function and thyroid function tests. The average age at stimulation test was 10.9 (± 2.8) years.

Of the 74 patients reviewed, 44 were found to have growth hormone deficiency and 38 were started on growth hormone treatment within the 12 month period after stimulation.

Conclusions: In the population presented, the non-deficient patients who were treated and not treated with GHT had no significant difference in their A1C in the first year of treatment. Z-Score is -1.0185. The p-value is 0.30772. The result is not significant at p≤ 0.05. The U-value is 87. The critical value of U at p≤ 0.05 is 64. Therefore, the result is not significant at p≤ 0.05. In the patients who were deficient, there was a statistical significance in the A1C elevation in the first year of treatment. P value was 0.04, delta A1C was 0.10. While this is statistically significant, the raise in A1C in the deficient patients on treatment was probably not clinically relevant. Long term trend of delta A1C needs to be collected to determine the clinical relevance over the years for treated patients.
Thyroid Imaging in Congenital Hypothyroidism: Focus on Ultrasound

Author Names: Marina Goldis¹, Lindsey Waldman¹, Henrietta Kotlus Rosenberg², Robert Rapaport¹

Departments: ¹Pediatrics, ²Radiology

Division: ¹Endocrinology and Diabetes

Institute Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Sonography (US) is the modality of choice for evaluation of the thyroid gland in both pediatric and adult patients, including neonates. With this non-ionizing, non-invasive, cost-effective readily available imaging modality, both diagnosis and follow up of a wide gamut of abnormalities can be readily accomplished without the use of contrast material, sedation, or anesthesia ¹, ². US plays an important role in determining if the thyroid gland is present and when present, its size, configuration, and echotexture.

Aim: To evaluate the benefit of thyroid ultrasound in infants with suspected congenital hypothyroidism.

Methods: Images of the thyroid gland were reviewed. Literature reviewed on congenital hypothyroidism and value of thyroid ultrasonography.

Results: A normal thyroid gland (Figure 1) has a homogeneous echotexture, which is slightly hyperechoic relative to adjacent neck muscles. Thyroid size and weight varies with patient age. Data for newborns vary from 0.84±0.38 ml to 1.62±0.41 ml ³, ⁴, ⁵. Literature regarding the size of thyroid gland describes no significant difference in thyroid volume observed among newborns examined during the first three weeks of the neonatal period ⁶, ⁷. Variations in the size of the thyroid gland in newborns have been reported from different countries and there are data for Scottish, German, Belgian, and Brazilian infants ⁴, ⁵, ⁶. Data from multiple studies in various populations suggest no significant difference in thyroid volumes in male versus female infants ⁵, ⁶, ⁸.

<table>
<thead>
<tr>
<th>Size of the thyroid gland</th>
<th>Association</th>
<th>Cause</th>
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<tbody>
<tr>
<td>Hypoplastic</td>
<td>1. Small gland</td>
<td>1. Replacement therapy (L-T4)</td>
</tr>
<tr>
<td></td>
<td>2. Hemiagenesis (Figure 4)</td>
<td>2. Failure of development in both lobes, left lobe being absent in 60% of cases ⁹</td>
</tr>
<tr>
<td>Hyperplastic</td>
<td>1. Severe permanent hypothyroidism</td>
<td>1. Iodide organification defects</td>
</tr>
<tr>
<td></td>
<td>2. Dyshormonogenesis</td>
<td>2. Increased level of TSH</td>
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Conclusions: US is helpful in determining whether the gland is enlarged, normal size, small, or absent (Figure 2, 3). While ultrasound is superior to scintigraphy in the assessment of thyroid morphology and reflecting the anatomic status of the gland, it does not provide conclusive data on the functional status of the gland ¹⁰, ¹¹.
Resilient Mice Have Resilient Functional Connectomes

Author Names: Yael Grossman, Dani Dumitriu

Departments: Pediatrics, Neuroscience

Division: General Pediatrics

Institutional Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Like humans, mice subjected to severe social stress display individual variability in stress-response. Susceptible animals succumb to maladaptive symptoms such as social avoidance, while resilient animals continue to behave indistinguishably from controls. The mechanisms that drive this variability in stress-response remain elusive. The goal of this study was to determine if differences in global neural function mediate individual variability to stress-response in mouse.

Hypothesis: We hypothesized that the functional connectome of resilient mice has network properties associated with robust function, such as increased smallworldness and high global efficiency.

Methods: We used a mouse model of acute social defeat in which an intruder mouse is subjected to physical attacks by three different aggressive mice. One hour later, mice were placed in a social interaction test and divided into resilient (social preferment) and susceptible (social avoidance) phenotypes. Mice were immediately sacrificed and brains were stained for a marker of cellular activation. Functional connectivity maps were built for resilient, susceptible, and control mice by quantifying the co-activation of 49 brain regions within each group.

Results: The functional connectome of resilient mice displays features that resemble normal human brain function, such as smallworldness. In contrast, the functional connectome of susceptible mice displays weak connectivity, allowing the amygdala – the brain’s emotional center – to highjack the network during social defeat.

Conclusions: We provide the first evidence that large-scale differences in neural networks mediate selective vulnerability and resilience in mouse. Dissecting the contribution of neurocircuits to stress-outcome could lead to early identification of at-risk individuals and new avenues for prevention.
Phosphoenolpyruvate (PEP) is a Novel Regulatory Mechanism of the Carbohydrate Response Element Binding Protein (ChREBP) in Pancreatic Beta Cells

Author Names: Lee B. Honig, Rebecca L. Cardone, Anila K. Madiraju Richard G. Kibbey, Donald K. Scott

Department: Endocrinology, Diabetes and Bone Disease

Division: Endocrinology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliations: Obesity, Diabetes and Metabolism Institute; The Mindich Child Health and Development Institute

Introduction: Carbohydrate Response Element Binding Protein (ChREBP) is a glucose-sensing transcription factor necessary for glucose-stimulated proliferation in pancreatic beta cells, a process that drives adaptive beta cell mass expansion, a therapeutic goal for Type 1 and Type 2 diabetes. The mechanisms by which metabolism controls ChREBP activity are poorly understood.

Hypothesis: We hypothesized that elevated production of cytosolic phosphoenolpyruvate (PEP) in pancreatic beta cells regulates ChREBP activity.

Methods: To manipulate mtGTP levels (increasing cytosolic PEP through mtPEPCK), we overexpressed either the ADP-binding isoform of Succinyl CoA Synthetase (SCS-ATP, reduced mtGTP) or the GDP-binding isoform (SCS-GTP, increased mtGTP) in INS-1 832/13 cells. To elevate PEP levels, we overexpressed either cytosolic Phosphoenolpyruvate Carboxykinase (PCK1) or Glycerol Kinase (GLYK) along with their respective substrates. The mRNA levels of glucose-responsive genes and glucose-stimulated proliferation were assessed by RT-PCR and BrdU immunostaining, respectively. Promoter activity of glucose-responsive genes was measured using luciferase assays.

Results: We found that SCS-ATP cells exhibited attenuated glucose-stimulated gene expression and proliferation compared to controls (p <0.05). ChREBP Beta promoter activity was significantly decreased in response to glucose in SCS-ATP (low PEP) cells compared to controls (p<0.01). Overexpression of PCK1 with substrate DMM resulted in an additive increase in glucose-responsive steady-state mRNA levels (p<0.05). Overexpression of GLYK and glycerol treatment resulted in activation of ChREBP and proliferation, even in low glucose conditions.

Conclusions: PEP is a novel regulator of ChREBP activity in INS-1 832/13 cells and may serve as a therapeutic target for diabetes treatment by promoting pancreatic beta cell proliferation.
Prenatal Lead Exposure and White Matter Microstructure in Children

Author Names: Megan Horton, Paul Curtin, Victoria Wang, Erika Proal, Téllez-Rojo Martha María, Ernesto Roldan-Valadez, Francisco Xavier Castellanos, Cheuk Tang, Roberta White, Robert Wright

Department: Preventive Medicine

Division: Environmental Health Sciences

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Perinatal lead (Pb) exposure is associated with adverse cognitive and behavioral outcomes that may be mediated by altered brain structure and function. Childhood Pb exposure has been associated with persistent impacts on adult white matter microstructure.

Hypothesis: We hypothesize that prenatal Pb exposure alters white matter microstructure in children.

Methods: We randomly selected 20 subjects (6 years of age) from the ELEMENT cohort in Mexico City for a magnetic resonance imaging (MRI) pilot study. Diffusion tensor images (DTI) were acquired using a 3T scanner. A voxel-wise statistical analysis for diffusivity measures, including fractional anisotropy (FA), was performed along major white matter tracts. All 20 subjects had blood biomarkers of Pb collected during 2nd and 3rd trimesters and at delivery (umbilical cord blood). We examined correlations between Pb biomarkers and FA values. We focused on mean FA and standard deviation (SD) of FA across 48 template regions of interest (ROIs) to capture bidirectional effects that may increase or decrease FA values. Linear regression models examined the association of 2nd, 3rd-trimester and cord blood Pb levels, FA and SD of FA from the ROIs.

Results: Pb levels in 2nd trimester blood were positively correlated with increased global FA after controlling for multiple comparisons (p < 0.05). Higher cord blood Pb was associated with increased FA (β = 0.010, p = 0.05) and increased variability of FA (β = 0.0014, p = 0.07).

Conclusion: These pilot data suggest changes in white matter microstructure associated with perinatal Pb exposure.
Sensitive Windows of Prenatal Air Pollution and Cognitive Function in Pre-School Age Mexican Children

Author Names: Hsiao-Hsien Leon Hsu², Yueh-Hsiu Mathilda Chiu¹², Ander Wilson⁴, Brent A Coull⁴, Allan C. Just², Itai Kloog⁸, Katherine Svensson², Martha M. Téllez-Rojo⁵, David Bellinger⁷, Joel Schwartz⁷, Rosalind Wright¹³, Lourdes Schnaas⁶, Robert O. Wright²³

Departments: ¹Pediatrics, ²Preventive Medicine, ⁴Biostatistics, ⁵Environmental Health, ⁸Geography and Environmental

Institution Affiliations: ¹Icahn School of Medicine at Mount Sinai; ⁴Harvard School of Public Health; ⁸Ben-Gurion University of the Negev

Institute Affiliations: ³The Mindich Child Health and Development Institute; ⁷National Institute of Public Health, Mexico; ⁶National Institute of Perinatology, Mexico

Introduction: The neurotoxicity of air pollution is receiving increasing attention. As for any developmental toxicant, exposure timing may be as important as the dose. We used an innovative statistical method to discover and define the time boundaries of sensitive windows of prenatal exposure to fine particulate matter (PM₂.₅) on children's cognition in a Mexico birth cohort.

Hypothesis: Higher prenatal PM₂.₅ exposure will be associated with lower cognitive function at age 4 years, and the association is particularly strong in a specific time window during pregnancy.

Methods: This analysis included 553 full-term (≥37 weeks) children. Prenatal daily PM₂.₅ exposure was estimated using a validated satellite-based spatio-temporally model. McCarthy Scales of Children's Abilities (MSCA) and its subscales were used to assess children's cognitive function at 4-5 years old (lower scores indicate poorer performance). To define susceptibility windows, we employed Bayesian Distributed Lag Interaction Models (BDLIMs) to examine associations between weekly averaged PM₂.₅ levels and MSCA.

Results: Adjusting maternal age, SES, child age, and gender, BDLIMs showed significant associations of increased PM₂.₅ levels with decreased GCI scores at 34-36 gestation weeks, decreased Quantitative scores at 31-33 weeks, decreased Memory scores at 35-37 weeks, decreased Motor scores at 29-36 weeks and decreased Verbal scores at 29-36 weeks. The estimated cumulative effect of PM₂.₅ across pregnancy showed significant associations with GCI (β=-0.34, 95%CI:-0.63, -0.08), Memory (β=-0.19, CI:-0.37, -0.03), Motor (β=-0.27, CI:-0.42, -0.13), and Perceptual performance (β=-0.18, CI:-0.31, -0.04). We did not find significant gender interactions.

Conclusions: Although multiple windows were found, all occurred in the 3rd trimester. Prenatal exposure to PM₂.₅ was adversely associated with MSCA.
Liver Transplantation for Children with Autoimmune Hepatitis and Primary Sclerosing Cholangitis: An Analysis of the UNOS Database

**Author Names:** Jacqueline Jossen¹, Rachel Annunziato², Jaime Chu¹, Hee-Sung Kim², Shari Sheflin¹, Joanne Lai¹, Ronen Arnon¹

**Department:** Pediatrics

**Division:** Gastroenterology and Hepatology

**Departments:** ¹Department of Pediatrics, Mount Sinai Hospital, New York, New York; ²Fordham University, New York, New York

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Autoimmune hepatitis and primary sclerosing cholangitis are progressive immune-mediated inflammatory diseases that may require liver transplant. Outcomes in children undergoing LT for these diseases are poorly studied in the PELD era.

**Aim:** To characterize the outcome of LT in children with AIH and to compare the outcomes to those of another immune mediated disease, PSC.

**Methods:** Children ≤18 years with PSC or AIH who had a first, isolated LT from 2002-2012 were identified from the UNOS database. Comparisons were made between patients transplanted for AIH vs. PSC as well as chronic AIH versus fulminant AIH.

**Results:** 174 children with AIH and 113 with PSC were transplanted in the study period. Characteristics of our sample are shown in Table 1. Forty-four (25.2%) children with AIH had fulminant hepatic failure (FHF). One and five year patient survival rates after LT of children with chronic compared to FHF were 97.7, 88.6 (p=0.013), and 93.8, 84.1 (p =0.46) respectively. One and five year graft survival rates of chronic versus FHF were 92.3, 86.4 and 84.6, 81.1 (p>0.05 for both). The one and five year patient and graft survival were not significantly different between AIH and PSC patients; patient survival: 95.4, 91.4 vs 97.3, 92.9% respectively, graft survival: 90.8, 83.9 vs 91.2, 83.2%. 19% of children with AIH and 17% of children with PSC lost a graft to recurrent disease.

**Conclusion:** Children with AIH and FHF had significantly lower patient survival rates and similar graft survival rates to children with chronic AIH. Children transplanted for AIH compared to PSC showed no significant differences in 1 and 5 year patient or graft survival.
Phthalate Exposure in Pregnancy is Associated with Preterm Birth in the Mexico City PROGRESS Cohort

Author Names: Allan C. Just, Andrea A. Baccarelli, Dana B. Barr, Joseph M. Braun, Chris Gennings, Adriana Mercado-Garcia, Maria M Tellez-Rojo, Robert O. Wright, Heather H. Burris

Department: Preventive Medicine

Division: Environmental Health

Institution Affiliation: Icahn School of Medicine at Mount Sinai, Harvard T.H. Chan School of Public Health, Rollins School of Public Health at Emory University, Brown University School of Public Health, National Institute of Public Health Mexico, Beth Israel Deaconess Medical Center

Introduction: Preterm birth remains a major public health issue affecting 15 million infants worldwide annually and may be impacted by environmental factors, including the ubiquitous phthalate plasticizers.

Hypothesis: We hypothesized phthalates, either individually or as a mixture, are associated with preterm birth risk.

Methods: We measured 8 urinary phthalate metabolites in 478 pregnant women from the Mexico City PROGRESS cohort in the 3rd trimester. We used multivariable linear regression to analyze associations between each phthalate metabolite with gestational age based on last menstrual period and logistic regression to estimate the odds of preterm birth (< 37 weeks’ gestation), adjusting for maternal age, BMI, socioeconomic status, parity and environmental tobacco smoke. We also modeled the cumulative effect of the mixture of phthalates on odds of preterm birth using weighted quantile sum (WQS) regression.

Results: A 1-interquartile range (IQR) increment of urinary mono-benzyl phthalate (MBzP) was associated with 1.7 days’ shorter gestation (95%CI 0.6, 2.8). Per IQR increment of MBzP, women had higher odds of preterm birth (adjusted OR 1.53, 95%CI 1.05, 2.28). The mixture of phthalates was also associated with preterm birth—rescaled to an adjusted OR of 2.34 (95%CI 1.18, 4.65) per IQR increment in the mixture. The WQS index was dominated by MBzP and MEOHP, with 58% and 29% of the total weight, respectively.

Conclusions: Higher 3rd trimester urinary MBzP concentrations were associated with shorter gestations and higher odds of preterm birth in this cohort. WQS regression revealed a potential mixture effect of phthalate metabolites on preterm birth risk.
Peanut Agglutinin is a Novel Ligand for Dendritic Cell-Specific Intercellular Adhesion Molecule-3-Grabbing Non-Integrin (DC-SIGN)

Author Names: Mohanapriya Kamalakannan, Lisa M. Chang, Galina Grishina, Madhan Masilamani, Hugh A. Sampson

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: The initial binding of allergens to DC-SIGN is thought to be crucial for development of allergic sensitization. However, the precise role of DC-SIGN in food allergy pathogenesis is unknown.

Hypothesis: We sought to characterize DC-SIGN-binding glycoproteins in peanut, soybean and a panel of allergenic and non-allergenic foods.

Methods: DC-SIGN-binding and -blocking assays were performed on human monocyte derived dendritic cells (DCs) using fluorescent-labeled extracts and anti-DC-SIGN antibody respectively. Using a recombinant DC-SIGN-Fc chimera, food protein extracts were tested for DC-SIGN-binding by ELISA and autoradiography. IgE immunoblotting was performed using pooled sera from food allergic subjects.

Results: Peanut agglutinin (PN-A) as identified in DC-SIGN-autoradiographs by mass spectrometry. PN-A specifically bound to DC-SIGN on DCs in a calcium dependent manner and induced the expression of activation markers CD80, CD83, CD86 and HLA-DR in vitro. A number of DC-SIGN-binding proteins in allergenic foods such as peanut, soy, tree nuts, egg and milk were discovered. Foods that generally do not induce allergic responses in humans such as pine nuts, chickpea and corn showed no binding to DC-SIGN. Several DC-SIGN-binding proteins show reactivity in serum IgE immunoblots. We have also identified novel non-IgE-binding proteins that interact with DC-SIGN.

Conclusions: We present a comprehensive report on characterization of DC-SIGN-binding proteins in common allergenic foods. We demonstrate that PN-A, a minor peanut allergen, is a novel ligand for DC-SIGN. Further functional characterization of DC-SIGN-interacting molecules in food allergens may provide new openings in our understanding of the pathology of food allergy and even novel therapeutic targets.
Placental Epi/Genomics as Sensors of the *in utero* Environment and Predictors of Fetal Development

**Author Names:** Maya A. Kappil, Ke Hao, Benjamin B. Green, Luca Lambertini, Carmen J. Marsit, Jia Chen

**Departments:** ¹Preventive Medicine, ²Genetics and Genomic Sciences, ³Epidemiology

**Institution Affiliations:** ¹Icahn School of Medicine at Mount Sinai; ³Geisel School of Medicine at Dartmouth College

**Institute Affiliation:** The Mindich Child Health and Development Institute

**Introduction:** The placenta is the principal organ regulating fetal development with implications for postnatal health outcomes. Appropriate transitioning through gestation requires the tightly coordinated orchestration of the placental epigenome, which sits as the interface between genes and the environment by enabling heritable and persistent changes in gene expression without altering the DNA sequence. Placental epigenetic elements include DNA methylation, histone modifications and non-coding RNAs, which undergo re-programming following fertilization as the zygote differentiates into cell-specific lineages. This is particularly pertinent at imprinted loci, key regulators of fetal development that are mono-allelically expressed based on parent-of-origin. The dynamic state of placental epigenetic marks highlights their sensitivity to perturbations during pregnancy and their potential utility as sensors of the *in utero* environment.

**Methods:** We comprehensively profiled the placental miRNome, imprintome and methylome across multiple birth cohort studies. We explored their associations to *in utero* trace metal exposures as well as to birth outcomes and postnatal neurodevelopment. We also constructed placental gene and eQTL networks by leveraging multi-scale epi/genomic and transcriptomic data.

**Results:** Our findings demonstrate that the placental epi/genome is highly sensitive to environmental stressors, and signature gene patterns are correlated with birth and neurodevelopmental outcomes. Lastly, placental eQTL networks reveal functional enrichment of pathways related to late-onset diseases including asthma and metabolic dysregulation.

**Conclusion:** Emerging technologies are increasingly facilitating the comprehensive profiling of the placental epi/genome. Novel methodologies to derive meaningful signals from the generated data highlight the relevance of the placenta as a sensor of the intrauterine environment and indicator of future health outcomes.
Infant Feeding Disturbance During the First 6 Months of Life and Interventions Performed

Author Names: Matthew R. Kowalik, Stanley Cho, Keith J. Benkov

Department: Pediatrics

Division: Gastroenterology and Hepatology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Infant nutrition during the first six months of life is a major determinant of future health. Multiple organizations recommend breastfeeding for the first six months. The incidence of exclusively breastfeeding, however, remains low and formula feeding remains high. Parent-directed formula changes complicate infant nutrition and frequently occur due to a perceived feeding disturbance. The goal of this study is to assess the incidence of reported infant feeding disturbance and the interventions performed.

Hypothesis: Our primary hypothesis is feeding disturbance in infants is a common complaint and results in an increased number of interventions. Interventions include: anticipatory guidance, formula changes, anti-reflux medication, and GI referral.

Methods: A retrospective chart review of patients, age 6 to 24 months, who had routine pediatric care since birth at Mount Sinai’s Pediatric Associates, was performed. Information from well-child visits was gathered. Infants with feeding disturbance were identified and compared to those without feeding disturbance.

Results: 109 patients were analyzed. 31 patients were exclusively breastfeeding at their initial visit, which decreased to 16 patients by 6 months. Feeding disturbance was noted in 47 patients. There were significantly more interventions in the feeding disturbance group (p <0.05), the majority of which were anticipatory guidance. There was no significant difference in anthropomorphic measurements between groups.

Conclusions: Exclusive breastfeeding remains below goal. Infant feeding disturbance is a common complaint during the first 6 months of life and results in significantly more interventions. Despite the presence of feeding disturbance, there was no significant effect on growth parameters.
Increasing “MyChart” Enrollment in a Teen Parenting Program: A Quality Improvement Project

Author Names: Janet B. Lee, Sari Bentsianov, Caroline Barangan, Anne Nucci, Christine Soghomonian, Linda Olszewski

Department: Pediatrics
Division: Adolescent Medicine
Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: The Teen Parenting Program at the Mount Sinai Adolescent Health Center is a multidisciplinary team of providers who provide comprehensive medical, mental health, nutritional, and psychological care to adolescent parents and their children. Some of the challenges we face in caring for teen parents include being able to have ongoing communication with families outside of clinic hours and improving literacy around health maintenance issues. “MyChart,” is the patient portal utilized by Mount Sinai Hospital to improve patients’ access to their physicians and their medical records.

Hypothesis: We hypothesized that patient access to the “MyChart” health portal would increase if all members of the Teen Parenting Program worked together as a team to improve sign-ups.

Methods: We implemented a quality improvement project to target teen parents and their children to increase “MyChart,” enrollment. The Teen Parenting Program daily flow sheet was modified to remind providers and patients about the importance of signing up for “MyChart.” Additionally, team members were trained to perform patient portal sign-ups within the clinical encounters. The goal of this project was to improve overall enrollment by 20%.

Results: We were able to increase “MyChart,” participation in parents by 19.2% with our intervention and increased children’s access by >1000%.

Conclusions: Multidisciplinary team providers stressing the importance of patient portal usage and assisting families in sign-up is an effective way to encourage patients to participate in “MyChart.” Future projects will focus on the utilization of “MyChart” as a tool to improve appointment adherence and on-time vaccinations with reminder messaging.
Obesity Associates with Heart Failure in Young Adults with Congenital Heart Disease

Author Names: Joseph B. Lerman, Ira A. Parness, Rajesh U. Shenoy

Department: Pediatrics

Division: Pediatric Cardiology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Adults with Congenital Heart Disease (ACHD) are a growing segment of the population, at elevated risk of cardiovascular morbidity and mortality. This risk increases with age, and is primarily attributable to congestive heart failure (CHF). To date, however, the acquired risk factors for CHF in young ACHD are poorly described.

Hypothesis: We hypothesize that acquired cardiovascular risk factors, including obesity, hypertension, hyperlipidemia, and diabetes mellitus, will significantly associate with CHF in young ACHD.

Methods: Retrospective analysis was performed on all ACHD patients (identified via ICD-9 coding), ages 19 – 45 years old, seen at a tertiary care center in 2013.

Results: 493 ACHD patients met inclusion criteria, with 24 identified cases of CHF. CHF patients had a higher prevalence of obesity (p =0.003), diabetes (p < 0.001), and hypertension (p = 0.05), than patients without CHF. CHF was not significantly associated with complex or univentricular CHD. Logistic regression analysis revealed that obese people had greater odds of having CHF (OR 3.29, p <0.01), which retained significance beyond adjustment for traditional cardiovascular risk factors (OR 2.66, p = 0.03). Likelihood ratio testing revealed that only obesity independently predicted the odds of having concomitant CHF ($X^2$ 4.49, p = 0.03).

Conclusions: Obesity was most associated with CHF in this cohort of 493 young ACHD. In the midst of the modern obesity epidemic, these results may indicate a need for improved lifestyle counseling in both children, and young adults, with congenital heart disease. Prospective studies are needed to confirm these findings.
Obesity in Adults with Congenital Heart Disease

Author Names: Joseph B. Lerman, John T. Doucette, Ira A. Parness, Rajesh U. Shenoy

Department: Pediatrics

Division: Cardiology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Obesity may be associated with greater morbidity in Adults with Congenital Heart Disease (ACHD), than in the general population. As ACHD often have exercise limitations, they may be more likely to be obese. To date, however, the prevalence of obesity in ACHD, as compared to the general population, has not been quantified in a large American cohort. Our study therefore seeks to determine the prevalence of obesity (BMI ≥ 30), and morbid obesity (BMI ≥ 40), in a large cohort of ACHD, as compared to matched controls.

Hypothesis: That ACHD have a greater prevalence of obesity and morbid obesity than matched controls.

Methods: Retrospective analysis was performed on all ACHD (identified via ICD-9 coding) seen at a tertiary center in 2013. A control group without CHD was randomly generated matching for age, sex, and ethnicity.

Results: 1,451 ACHD and 1,451 matched controls met inclusion criteria. ACHD had a lower mean BMI than controls (ACHD: 26.9 ±5.7 kg/m², Controls: 27.6±6.3 kg/m², P<0.001). ACHD had a similar prevalence of obesity (OR 1.04, P=0.69), and a lower prevalence of morbid obesity (OR 0.24, P<0.001) than their matched peers. These relationships were not modified by stratification for simple versus complex congenital heart disease.

Conclusion: This is the largest known retrospective cohort study of obesity in American ACHD. ACHD had an equal prevalence of obesity, but a lower prevalence of morbid obesity, than matched controls. Longitudinal studies are warranted to determine the cardiovascular morbidity associated with obesity in this vulnerable population.
Food Insecurity in Families enrolled in the Wholesome Wave Fruit & Vegetable Prescription Program® (FVRx®) in New York City Public Hospitals

Author Names: Pablo Leung¹, Tina Cheng¹, Manaswitha Kare¹, David Rhee¹, Ahmed Torky¹, Martha Ballestas¹, Richa Verma¹, Lwbba Chait-Llamas¹, Anika Clarke¹, Ashley Fitch², Amy Woolever¹, Efnniky Kyvelos, MD¹

Department: ¹Pediatrics

Institute Affiliations: Icahn School of Medicine at Mount Sinai; Elmhurst Hospital Center, Elmhurst, NY; ²Wholesome Wave, Bridgeport, CT

Background: FI has been associated with negative health outcomes. WW is a nonprofit organization that seeks to increase access to healthy, locally grown foods. FVRx® is one of WW's programs that promotes partnerships between healthcare providers, farmers markets and families with diet related disease. In the program, families are provided with vouchers that can be redeemed for fresh fruits and vegetables (F/V) and local participating farmer's markets.

Objective: To examine the prevalence of FI in families of overweight/obese children enrolled in FVRx®.

Design/Methods: Consecutive sample of patients 2-17y; BMI ≥ 85; able to commit to four monthly appointments were enrolled. Measures: Demographics, Household FI assessed at start and end of program (Core Food Security Module questions). Difference in the prevalence of FI was assessed using Wilcoxon signed rank test.

Results: N 263; Mean age 8.8 yrs; F 51%; Hispanic 71%, African or Caribbean American 24%, Asian Pacific Islander 4%; Mom HS grad 51%; FI during first visit 60%; 59% receiving food stamps, 27% receiving WIC benefits. FI indicators showed a significant increase from pre- to post-survey (Z = -3.481, p = .001).

Conclusion: About 60% of children enrolled live in food insecure households. Participation in FVRx® resulted in improved food security. Future development, study and implementation of programs such as FVRx® may be instrumental in the promotion food security and overall health in children.
An Interesting Case of Pervasive Developmental Disorder Secondary to 15q13.3 Microdeletion Presenting with Recurrent Infections

Author Names: Grace V. Liao¹, Parvesh M. Garg², Iya Chikvashvili²

Department: Pediatrics

Division: General Pediatrics

Institution Affiliations: ¹Kravis Children's Hospital at Mount Sinai, NY, ²Elmhurst Hospital Center, NY

Introduction: 15q13.3 microdeletion is associated with a higher risk of neurodevelopmental and psychiatric disorders. However, an increased susceptibility of infection has not been reported in this abnormality.

Objective: We report a case of recurrent infections in a developmentally delayed child with 15q13.3 microdeletion.

Case: A five-year-old male with 15q13.3 microdeletion, autism spectrum disorder, pervasive developmental delay, and vitiligo was admitted for submandibular cellulitis requiring IV antibiotics. The patient had balanoposthitis, multiple acute otitis media, frequent gastroenteritis, viral upper respiratory infections, and chronic impetigo, requiring frequent emergency visits and antibiotic courses. Developmental evaluation was significant for delayed fine motor, social, and language development equivalent to a four-year-old, four-year-old, and three-year-old, respectively. A genetic cause of his developmental disorder was suspected when his mother reported speech delay in patient’s maternal first cousin. Microarray showed 15q13.3 microdeletion. Karyotype was 46XY. Fragile X screen was negative. Results of his audiology and ophthalmologic assessments were normal. Immunologic work up is pending. Immunizations were up to date. He had no allergy. Weight and height were at 75th percentile for age. Head circumference was at 95th percentile. He had no dysmorphic features on exam.

Conclusion: Frequent infections of multiple organ systems in a child with 15q13.3 microdeletion suggests an increased susceptibility of infection in this chromosomal abnormality.
Exploring the Associations Between the Expression Profile of Placenta-Related Genes and Birth Weight in the Human Placenta from the Rhode Island Child Health Study (RICHS)

**Author Names:** Qian Li ¹, Maya Kappil ¹, Men-jean Lee ², Benjamin B Green ³, Carmen J Marsit ³, Jia Chen ¹

**Departments:** ¹Preventive Medicine, ¹Obstetrics, Gynecology and Reproductive Science, Epidemiology, ³Pharmacology and Toxicology

**Institution Affiliations:** Icahn School of Medicine at Mt. Sinai, New York City, NY ¹; ³Geisel School of Medicine at Dartmouth College; Hanover, NH

**Institute Affiliation:** The Mindich Child Health and Development Institute

**Introduction:** Appropriate placental function is crucial to fetal development. The importance of genes involved in placental processes in regulating fetal growth has been long established. However, a comprehensive assessment of the role of these genes on fetal growth has yet to be conducted.

**Hypothesis:** We hypothesize that the expression of placenta-related genes are associated with birth weight.

**Method:** Placental RNA was profiled using a custom-designed code-set containing 22 known placenta-related genes from 672 term placenta. One-way analysis of covariance and multinomial regression analysis, adjusted for batch, maternal insurance and infant ethnicity, were used to identify genes differentially expressed by birth weight categories, including infants born appropriate for gestational age (AGA), small for gestational age (SGA, lowest 10th percentile), and large for gestational age (LGA, highest 10th percentile).

**Results:** Compared to AGA status, a 2-fold increase in the expression of 7 placental genes (ABCG2, PLAC1, CEBPB, CRH, GCM1, INSL4, and PGF) was associated with decreased odds of LGA status ($p < 0.05$), while a 2-fold increase in NR3C1 expression was associated with increased odds of LGA ($p < 0.05$). A 2-fold increase in both ABCG2 and PLAC1 expressions were associated the decreased odds of SGA status ($p < 0.05$). Finally, a trend was observed between a 2-fold increase in NR3C1 expression and increased odds of SGA status ($p = 0.05$).

**Conclusion:** These results suggest that the expression pattern of placental genes has the potential to be developed as a novel biomarker for neonatal outcome.
Is Resident Self-Assessment of Teaching Skills an Accurate Measure of Skills Used?

Author Names: Suzanne Friedman, Scott Moerdler, Alefiyah Malbari, Benjamin Laitman, Kathleen Gibbs

Department: Pediatrics

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Resident as Teacher (RAT) training curricula are growing across residency programs. While multiple studies of these curricula have looked at the impact of resident teaching programs on resident or student perceptions of teaching, few have evaluated resident self-assessment of their teaching skills when compared to their students’ assessments.

Hypothesis: Resident self-assessment of teaching skills will be on par with assessments completed by their students.

Methods: The Mount Sinai pediatric residency has a rigorous required RAT curriculum for all residents as well as an optional longitudinal teaching program (Resident Teaching Group RTG). At the completion of residency, outgoing Mount Sinai pediatric residents and those in the RTG were asked to assess their frequency of using 5 selected teaching skills. The survey was based on a 5 point Likert scale ranging from never (1) to always (5). In addition, medical students were asked to assess the frequency with which their RTG mentors used these 5 specific teaching skills. Differences between student and resident responses were analyzed by t-test.

Results: 23 students and 21 residents (9 RTG and 12 graduating non-RTG) completed surveys. RTG residents had higher self-assessment scores than the students’ assessment for “Settings Goals” (4.1 vs 3.5, P < .05) and “Using Questioning as a Teaching Tool (4.5 vs 3.9, P<.05). Non-RTG residents had lower self-assessment scores than the students’ assessment for “Bedside Teaching” (3.1 vs 4.3, P<.05).

Conclusions: Resident self-assessments of teaching skills often do not correlate with students’ perception of their use and are under or over estimated.
Adolescent Perspectives on Acceptability of Sensitive Health Questions
Posed by Dentists and Physicians

Author Names: John Mansour, Laurence Hyacinthe, John Pfail

Department: Dental Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Discussing sensitive health topics (alcohol use, tobacco use, drug use, sexual activity, pregnancy) with adolescents can be challenging for certain healthcare providers and adolescent patients. The disclosure of such information to a health care provider can be critical for treatment. The attitudes of adolescents on their level of comfort in disclosing this information to a dental provider has never been examined in past literature.

Hypothesis: We hypothesized that adolescents are more comfortable disclosing sensitive health information to physicians than dentists.

Methods: A survey of 100 adolescent patients (ages 13-19) was conducted by random sampling at The Mount Sinai Adolescent Health Center. All patients completed the survey on their level of comfort in disclosing sensitive health information to a physician vs. a dentist.

Results: 100 surveys were collected. The results of comfort levels disclosing the sensitive health information to the provider indicated are in the following respective order: (extremely comfortable, comfortable, unsure/indifferent, uncomfortable, extremely uncomfortable). Alcohol use disclosure: physicians (36%, 47%, 16%, 1%, 0%), dentists (29%, 36%, 24%, 8%, 3%). Tobacco use disclosure: physicians (35%, 49%, 13%, 3%, 0%), dentists (26%, 43%, 23%, 7%, 1%). Drug use disclosure: physicians (31%, 43%, 19%, 6%, 1%), dentists (21%, 34%, 30%, 8%, 6%). Sexual activity disclosure: physicians (24%, 44%, 14%, 16%, 2%), dentists (11%, 24%, 17%, 28%, 20%). Pregnancy disclosure among females: physicians (20%, 55.7%, 15.7%, 5.7%, 2.9%), dentists (11.4%, 27.1%, 31.4%, 10%, 20%).

Conclusions: Adolescents are more comfortable disclosing sensitive health information to physicians than to dentists.
MPI Overexpression Can Protect Against Genotoxic Stress in Zebrafish

Author Names: Shikha Nayar, Nataly Shtraizent, Jaime Chu

Department: Pediatrics

Division: Gastroenterology and Hepatology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: Mindich Child Health and Development Institute

Introduction: Radiotherapy is a mainstay of cancer treatment, but it is not well understood how cancer cells develop mechanisms to bypass cytotoxic effects of radiation. To improve efficiency of radiotherapy, radiosensitizing agents have been proposed as a therapeutic approach. Cazet et al. revealed a role for Mannose Phosphate Isomerase (MPI) as a radiosensitizing target. Here, we utilize zebrafish as an in vivo system to study the role of MPI as a factor promoting cell survival in response to genotoxic stress.

Hypothesis: UV radiation induces p53 stabilization, which decreases Mpi levels and activity. Overexpressing Mpi can radioprotect zebrafish embryos through downregulation of p53.

Methods: mpi mRNA is injected into zebrafish embryos at the one cell stage. Embryos are UV-irradiated 24 hours post fertilization (hpf). Acridine Orange staining is used as a marker for cell death, and p53 protein levels are assessed by Western Blotting. Mpi enzymatic measurements are done using fluorometric assays.

Results: UV radiation to zebrafish embryos at 24 hpf downregulates Mpi transcript and enzymatic activity. When embryos overexpress Mpi, and are exposed to UV radiation, they demonstrate 1) fewer apoptotic cells, 2) reduced p53 protein levels and 3) increased Mpi activity compared to control-injected zebrafish treated with UV.

Conclusions: Here, we show that Mpi is downregulated in response to genotoxic stress, and that overexpressing Mpi prior to UV radiation radioprotects zebrafish embryos, through the dampening of p53. This broadens our understanding of metabolic responses to genotoxic stress, and offers new targets to increase response to radiation therapy.

**Author Names:** Gabriela Ortiz, Todd O’Connor, Jessa Carey, Audrey Paul, Adam Vella, Diane Rode

**Department:** Child Life and Creative Arts Therapy Department

**Division:** Emergency Medicine

**Institution Affiliation:** The Mount Sinai Hospital

**Introduction:** Procedures involving a needle are often a necessary component of the medical treatment that children undergo as part of their emergency care visit. Literature suggests that hospitalized school-aged children often perceive needle procedures as the most stressful part of being in the hospital. Child life specialists and creative art therapists have a unique and integral role in providing psychosocial care to pediatric patients and their families within the emergency department (ED) setting. As such, the aim of the present study was to measure the efficacy of a multi-modal approach involving child life and music therapy interventions that addressed procedure-related distress in pediatric patients ages 4-11.

**Hypothesis:** We hypothesized that the child life and music therapy intervention would significantly decrease a child’s distress, as perceived by their parent, during IV placement.

**Methods:** We measured each child’s distress using a four-question pre-test and post-test questionnaire completed by the parent.

**Results:** We found a decrease in post-test scores for questions 1 & 2, and an increase in post-test scores for questions 3 & 4, with mean differences between pre/post being significant for all questions (p<.05). Crude means analysis of responses from Spanish-speaking and English-speaking parents were compared and showed near parallel trending between pre/post test responses for three of the four questions.

**Conclusions:** A high likelihood of improvement was demonstrated across the four questions, suggesting that the child life and music therapy intervention supported healthy, adaptive coping, and helped to minimize distress experienced during IV placements.
Heart Rate Variability Interactions between Mother-Infant Dyads during Kangaroo Care

Author Names: Sarah Pearce, Donald Pfaff, Lisa Eiland, Robert Green, Ian Holzman

Department: Pediatrics

Division: Newborn Medicine

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Kangaroo Care (KC) is used to promote mother-infant bonding. Long term studies of KC have shown behavioral and developmental benefits such as acceleration of brain maturity and vagal tone. In order to better understand the potential neurodevelopmental effects of maternal-fetal interactions during KC, researchers have studied fetal heart rate variability (HRV) as well as fetal responses to maternal stresses. The goal of this study is to examine the maternal and infant frequency domain (FD) of HRV, which has been shown to correlate with the autonomic nervous system (ANS) status.

Hypothesis: To determine a relationship during KC within the dyad in the FD of the HRV.

Methods: Twenty NICU inpatients, 30-34 weeks gestation, were recruited for KC sessions monitoring vital signs of both mother and infant before and during KC. Interim analysis of 20 KC sessions were performed. The ECG signal was processed and RR intervals calculated employing Kubios HRV analysis software; correlation of maternal and infant signals were analyzed using univariate analysis in the general linear model in SPSS.

Results: Analysis in the FD, calculated using the fast Fourier transform, showed a significant relationship between mother and infant for the low frequency (LF) to high frequency (HF) ratio (p = 0.008).

Conclusions: Interim analysis of the dyads suggests a statistically significant relationship between mother and infant within the FD. This LF:HF ratio can represent the balance between the parasympathetic.
**Post Discharge Follow-Up Calls**

**Author Names:** Adriana Porto, Daniel Rauch

**Department:** Pediatrics

**Division:** General Pediatrics

**Institute Affiliations:** Icahn School of Medicine at Mount Sinai; Elmhurst Hospital Center, Elmhurst, NY

**Introduction:** Post discharge (d/c) follow-up calls are utilized for many reasons: monitoring patient (pt) status, understanding of d/c instructions including medications and appointments, and satisfaction with hospital care. Adult data suggests such calls improve satisfaction and may impact readmission rates. Little data about the impact of such calls in pediatrics exists. Our objective was to examine the utility of d/c calls.

**Design/Methods:** Calls were made to all discharged pts from 7-8/2015.

**Results:** Of 71 patients we were able to contact 51 (72%). 37 respondents were the mother, 8 the father, 1 a brother, and 5 the pt. 50 (98%) reported the pt was in good to excellent condition and 49 (96%) would return to our hospital for a similar condition. 98% felt the treatment was helpful and 96% were satisfied with their care. 88% said they understood the diagnosis and 86% said they understood the treatment. Those who didn't were all parents. 61% were sent home with a prescription. All 31 filled the prescription and claimed to be taking the medicine as prescribed. We did not have any bounce back admissions in that time frame.

**Conclusion/Discussion:** Our d/c follow-up calls were largely successful in reaching our pts, demonstrating that our population can be contacted and possibly followed this way. Based on this we continued our current d/c counseling and the d/c calls while looking for other means of assessing medication adherence.
Analysis of Neonatal Urinary Exosomes: A Window into the Ontogeny of Renal Na Transport Protein

Author Names: Erin M. Qualter, Daniel Flores, Scarlett McKinsey, Lisa M. Satlin

Department: Pediatrics

Divisions: Neonatology, Nephrology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Furosemide promotes a diuresis/natriuresis in premature newborns (NBs). Yet, studies in neonatal animals predict that the target of furosemide, the Na-K-2Cl cotransporter (NKCC2) in the thick ascending limb of kidney, is absent until ~ 36 wks GA in the human. These observations suggest that furosemide mediates its diuretic/natriuretic effects in premature NBs via a NKCC2-independent mechanism of action.

Hypothesis: mRNA and protein expression of Na transport proteins in urinary exosomes increase with advancing GA and correlate with the ability of the NB to reabsorb Na.

Methods: > 6 ml urine was collected weekly after birth from premature NBs via an ostomy bag applied to the perineum. Exosomes were isolated using a differential ultracentrifugation technique (Pisitkun, 2004). Exosomal RNA was quantified by qRT-PCR using the Taqman assay with primers and probes specific for NKCC2 and the α subunit of the epithelial Na channel (αENaC) of the distal nephron.

Results: Total RNA for NKCC2, αENaC, and uromodulin (and the housekeeping gene, GAPDH) was detected in as little as 6 ml of urine collected from premature NBs. In 2 initial subjects for which >3 urinary samples yielded exosomal RNA, relative expression (normalized to GAPDH) of uromodulin, but not NKCC2 or αENaC, increased with GA.

Conclusions: Total RNA is detectable in urinary exosomes isolated from premature NBs. In 2 patients, uromodulin RNA expression increased with advancing GA, as has been described at the protein level by DeFreitas (2016). Expansion of this initial data set will allow us to explore concordance between results of studies performed in animal models with the physiology of the developing human subject.
**Right Atrial Area a Reliable Surrogate for Assessment of Right Atrial Size: A Comparative Prospective Study of Echocardiography Versus Cardiac Magnetic Resonance Imaging in Children**

**Author Names:** Hari Rajagopal, Erika Pedri, Rosalie Castaldo, Simon Lee, Santosh Uppu, Shubhika Srivastava

**Department:** Pediatrics

**Division:** Cardiology

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Increased right atrial (RA) volume correlates with supraventricular arrhythmias and right ventricular diastolic dysfunction. Data on RA size by trans-thoracic echocardiography (TTE) in the pediatric population is sparse. Cardiac magnetic resonance (CMR) imaging is considered the reference standard.

**Hypothesis:** Indexed right atrial area by TTE and not volume would be a more valid measure of RA size.

**Methods:** We prospectively examined 30 consecutive patients (Age median 17 years, range 3-22 years) referred for a clinically indicated CMR imaging. Each patient underwent 2D and 3D TTE and CMR on the same day. RA Volume was assessed by two methods: 1) derived using area length method for both modalities and 2) real-time 3D TTE volumes were compared to CMR volumes derived from steady-state free-precession sequence (SSFP) cine.

**Results:** The agreement between TTE and CMR values using Bland-Altman method showed good agreement between 2D-TTE and CMR indexed RA area (ICC 0.87, CI 0.76-0.93) and derived volumes (ICC 0.81, CI 0.65-0.90, and mean bias of 5 +/- 11.2 ml/m²). There was poor agreement of the 2D TTE derived volume as well as 3D TTE volume when compared to CMR SSFP analysis. The RA area correlated strongly with the RA volume within the MRI group (r= 0.90, p <0.0001). RA area by 2D TTE has excellent reproducibility and repeatability.

**Conclusions:** TTE consistently underestimates the RA volumes by both 2D and 3D methods when compared to CMR imaging. RA area by 2D TTE can be used as a quick, reliable and reproducible surrogate measure of RA size.
Right Atrial Size by Trans-Thoracic Echocardiography: Can We Do Better Than “Eyeballing”? 

**Author Names:** Hari Rajagopal, Erika Pedri, Rosalie Castaldo, Simon Lee, Santosh Uppu, Shubhika Srivastava

**Department:** Pediatrics

**Division:** Cardiology

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Atrial size has been recognized as a prognostic indicator for heart failure and cardiovascular death in adults. Data on right atrial (RA) size by trans-thoracic echocardiography (TTE) and its clinical implication is sparse in Pediatric population. Our objectives were to derive normative data for RA area.

**Methods:** RA area was measured from apical four chamber view in 300 consecutive normal subjects by TTE.

**Results:** BSA range 0.19-2m². The mean indexed RAA ± SD for BSA<1 m² and > 1 m², is 5.79 ± 2.19 cm² and 12.4 ± 2.82 cm² SD respectively. Indexed RA area/m² decreases as BSA increases thereby demonstrating a residual relationship to BSA (r=-0.28, p<0.001). The entire cohort was divided into two groups with a BSA cut off of 1m² to provide the best fit Allometric model(r=0). The AE (Allometric exponent) by least square regression analysis for each group was 0.94 and 0.88 for BSA <1m² and >1 m² respectively, and was validated against an independent sample of 50 subjects. Intraclass correlation coefficient for inter-observer as well as intra-observer variability was 0.90 and 0.96 respectively.

**Conclusion:** RA area from apical four chamber view by 2D TTE unlike 3D TTE is a quick and reliable noninvasive objective measure of right atrial size. RAA/BSA⁰.⁹⁴ for children with BSA <1 m² and RAA/BSA⁰.⁸⁸ for those with BSA >1 m² is accurate and can be used to derive z-scores. The normative data may aid in serial follow-up examinations, and allow for further research on atrioventricular interaction and right ventricular diastolic dysfunction in the pediatric population.
Right Atrial Dilatation is an Early Occurrence in Childhood and Correlates With Right Ventricular Dilatation in Repaired Tetralogy of Fallot

**Author Names:** Hari Rajagopal, Erika Pedri, Rosalie Castaldo, Simon Lee, Santosh Uppu, Shubhika Srivastava

**Department:** Pediatrics

**Division:** Cardiology

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Right atrial (RA) dilation has been shown to be associated with bi-atrial dysfunction and arrhythmia in adults with repaired tetralogy of Fallot (rToF). We have shown that RA area (RAA) can be used as a surrogate for right atrial size.

**Hypothesis:** RAA from apical four chamber view measured by TTE is abnormal in rToF and can be used as an additional surrogate for risk stratification in rToF.

**Methods:** We analyzed 30 consecutive pediatric patients with rToF and compared them to 30 normal subjects. From TTE images, we measured right atrial area (RAA) and calculated z scores from indexed RAA/m^{0.88} for BSA <1 and RRA/BSA^{0.94} for BSA ≥1. We compared RAA z scores to RV diastolic parameters (E/E’ ≥1 vs <1 and antegrade systolic PA flow) and additionally to degree of RV dilatation, tricuspid regurgitation (TR) and pulmonary regurgitation (PR) by TTE.

**Results:** 30 TOF patients: male 52 %. Mean age (Mean 5 years, SD 2.3 years). RAA z scores was significantly higher in rToF compared to normal (Z score Mean of 3.4 vs 1.2, p<0.01). Within the rToF group, RAA z scores correlated significantly with RV dilatation (r=0.38, p<0.05) but not with echocardiographic estimates of diastolic dysfunction.

**Conclusion:** The presence of RA dilation is an early occurrence in childhood. RAA z scores correlate with RV dilatation in rToF. Inclusion of RA z scores for longitudinal follow up of patients with rToF in the routine echocardiography protocols may aid in identifying those at risk for atrial arrhythmias.
A Synonymous Splice Site Variant in MTHFR Gene Causing Hyperhomocysteinemia with Severe Neurological Dysfunction That was Successfully Reversed with Early Initiation of Medical Therapy

Author Names: Dallas A. Reed¹, Hongzheng Dai³, Amy Williamson¹, Jinglan Zhang³, Lee-Jun Wong³, Kimihiko Oishi¹²

Departments: ²Pediatrics, ¹Genetics and Genomic Sciences, ³Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA

Division: Medical Genetics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Mutations in the MTHFR gene cause methylenetetrahydrofolate reductase (MTHFR) deficiency, which is a rare autosomal recessive disorder characterized by sudden onset of respiratory failure, coma, neonatal seizures, and death. Due to its non-specific clinical presentation, diagnosis of MTHFR deficiency is challenging.

Case: A 3 week old female born to consanguineous Pakistani parents presented at 17 DOL with lethargy, poor feeding, and cyanosis, resulting in intubation. Seizures began at 20 DOL. She was non-dysmorphic, edematous, and comatose with a distended bladder. Labs demonstrated elevated plasma homocysteine, decreased methionine, and absence of methylmalonic acid, consistent with hyperhomocysteinemia. Treatment for presumptive MTHFR deficiency was started with leucovorin, vitamin B6, and hydroxycobalamin. Within days, her neurological function was fully recovered.

Methods: Next generation and Sanger sequencing were performed on genomic DNA and total RNA. RT and Real-time PCR were used to examine transcription patterns of MTHFR.

Results: We identified a homozygous synonymous splicing variant c.1530G>A (p.Lys510Lys). RT-PCR for the proband demonstrated abnormal transcripts: an exon 9 skipping form, a 5bp GTGTG insertion at the 3’ end of exon 9, and a novel transcript skipping exons 9 and 10.

Conclusions: A homozygous c.1530G>A synonymous variant in MTHFR resulted in expression of aberrant non-functional transcripts, including the novel transcript we identified. Since neurological outcome is extremely poor in the majority of MTHFR deficient patients, rapid initiation of treatment in patients with acute onset of neurologic depression is essential to prevent severe chronic neurological compromise.
Uncovering the Genetic Architecture of Congenital Heart Disease Using Whole Genome Sequencing

Author Names: Kathryn B. Manheimer, Felix Richter, Nihir Patel, Michael D. Linderman, Ali Bashir, Bruce D. Gelb

Department: Pediatrics

Division: Cardiology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Congenital heart disease (CHD), a complex genetic trait, affects 2-3% of neonates. Aneuploidy, de novo coding SNVs and copy number variants account for 30% of cases, the remainder being unexplained.

Hypothesis: Whole genome sequencing (WGS) data will implicate additional de novo genetic mutations in CHD.

Methods: 30x WGS was performed for 50 parent/affected child trios. SNVs and indels were called with GATK and Haplotype Caller; CNVnator and Lumpy were used to identify structural variants (SVs).

Results: An average of 34 de novo variants/trio were discovered in non-repeat regions. Among 33 trios with prior whole exome sequencing (WES), WGS found 24 de novo coding variants compared to 15 from WES. Expected de novo mutation rates were calculated based on trinucleotide sequence context, recombination rate and other predictors. Burden analysis suggested enrichment of de novo mutations in promoters of genes highly expressed in the heart and regions with H3K27 trimethylation specific to cardiomyocyte development. Preliminary SV analysis identified homozygous recessive and hemizygous deletions with high precision, as well as 1 de novo deletion and 1 de novo duplication. Of note, one male infant had an inherited DMD deletion, diagnosing Duchenne muscular dystrophy pre-clinically.

Conclusions: We designed analysis pipelines to identify de novo genetic mutations implicated in CHD and found WGS to be more robust than WES for exomic defects. Calling of SV, while more challenging, is possible and can be diagnostic. WGS data from 300 CHD trios in progress and 500 control trios will increase our power to detect causal mutations.
Group Cognitive-Behavioral Therapy for Pediatric Obsessive-Compulsive Disorder: A Preliminary Investigation

**Author Names:** Ariz Rojas, Timothy R. Rice, Shannon Gair, Natasha Kostek

**Department:** Psychiatry

**Division:** Child and Adolescent Psychiatry

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Cognitive-behavioral therapy with exposure and response prevention (ERP) is the first-line treatment for pediatric obsessive-compulsive disorder (OCD), yet availability for this treatment is limited. Applying ERP in a group-format can increase accessibility, but there is limited research investigating group ERP treatment. This study evaluated the effectiveness and treatment acceptability of group treatment for pediatric OCD.

**Hypothesis:** We hypothesized that group therapy would be an effective treatment.

**Methods:** 15 children aged 8-12 years with OCD were enrolled in 12-session, 90-minute CBT/ERP group program. The group protocol followed the CBT framework consistent with the goal-standard, manualized individual treatment for OCD. Pre-and post-measures included: CY-BOCS, ROARS, OCI-P and OCI-C, SCARED, FAS, and a post-evaluation form assessing treatment acceptability.

**Results:** Group treatment was effective ($p < .01$), as pre CY-BOCS scores were statistically lower at post-test. All parent-reported indicators revealed clinically significant reductions, including parental accommodation ($p < .05$), interference ($p < .01$), behavior ($p < .05$), and anxiety ($p < .05$). Children and parents on average found the group treatment to be “very helpful.” Open-ended responses indicated that parents thought the group provided insight and confidence to fight OCD, normalized, and fostered acceptance in their child.

**Conclusions:** This study suggests that ERP in group-format elicits clinically significant reductions in symptomology. Over half of the parents found the group-format to be as effective as or more effective than individual treatment. The largest impact of the group-format lies in its ability to foster motivation for treatment, peer support for parent and child, and increase child self-esteem, self-efficacy, and acceptance.
Identifying Sensitive Windows for Prenatal Particulate Air Pollution Exposure and Mitochondrial DNA in Cord Blood

**Author Names:** Maria José Rosa, Allan C. Just, Marco Sánchez Guerra, Adriana Mercado Garcia, Itai Kloog, Deepjyoti Deb, Kasey J. Brennan, Rosalind J. Wright, Martha María Téllez Rojo, Andrea Baccarelli, Robert O. Wright

**Department:** Preventive Medicine

**Division:** Environmental Health

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Institute Affiliation:** The Mindich Child Health and Development Institute

**Introduction:** In utero particulate matter ≤2.5μm (PM$_{2.5}$) exposure produces oxidative stress. Changes in mitochondrial DNA (mtDNA) function can quantify oxidative stress making it a useful tool to assess exposure mechanisms. Our group developed a daily PM$_{2.5}$ exposure model which combined with distribute lag models (DLM) can discern periods of vulnerability to oxidative stress.

**Hypothesis:** Identify sensitive windows to predict cumulative oxidative stress experienced in the prenatal period using mtDNA copy number (mtDNAcn) measured in cord blood.

**Methods:** Women affiliated with the Mexican social security system were recruited during pregnancy. Mothers with cord blood collected at delivery and complete covariate data were included (n=432). Prenatal daily exposure to PM$_{2.5}$ was estimated using a satellite-based spatio-temporally resolved prediction model and place of residence during pregnancy. DNA was extracted from umbilical cord leukocytes. Real-time polymerase chain reaction (PCR) was used to determine mtDNAcn. A DLM incorporating weekly averages of daily PM$_{2.5}$ predictions was used to identify sensitive windows when pointwise 95% confidence bands that did not contain zero.

**Results:** In models including child’s sex, mother’s age at delivery, prenatal environmental tobacco smoke exposure, birth year and assay batch, we found significant associations between higher PM$_{2.5}$ exposure during late pregnancy (33-37 weeks) and decreased mtDNAcn in cord blood.

**Conclusions:** Increased PM$_{2.5}$ during a specific prenatal window in the third trimester was associated with decreased mtDNAcn suggesting heightened sensitivity to PM during this life stage. Refined determination of time windows in which PM has the greatest magnitude of effect can enhance insight into underlying mechanisms.
Parent’s Perception of Food Allergy Management in Schools

Author Names: Jaime Ross, Jennifer Fishman, Sally Noone, Beth Strong, Zara Atal, Carly Ehritz, Jessica Gau, Julie Wang

Department: Pediatrics
Division: Allergy and Immunology
Institution: Icahn School of Medicine at Mount Sinai
Institute Affiliations: The Mindich Child Health and Development Institute; Jaffe Food Allergy Institute

Introduction: Children with food allergies spend much of their day in school. This study examined parental reports of food allergy management in schools.

Hypothesis: We hypothesized that parents of food allergic children are not confident in their school’s ability to protect children from allergic reactions.

Methods: An anonymous written survey was distributed to families in our food allergy practice.

Results: Eighty five surveys were returned. Twenty two (26%) report that their child has had an allergic reaction at school. The majority of the reactions happened in the classroom (60%) and the cafeteria (41%). Of these reactions, 32% were from food provided by the school. Two (9%) of the children received epinephrine for the allergic reaction. The reactions were treated by the nurse (55%), student (9%), and school staff (9%). An emergency allergy action plan was provided by 83% of parents to the school. 45% of parents reported feeling very confident, 38% fairly confident and 17% somewhat confident in the precautions taken by the school.

Conclusion: Allergic reactions happen in schools. While one third of the children have had allergic reactions at school, the majority of families are confident that schools are taking adequate precautions to protect their children from reactions. Continued education for families and school staff will assure safety of food allergic children in the school setting; as well as improved parental confidence in the school’s ability to manage food allergies and reactions.
Measuring Appropriate Use of the Emergency Department (ED) for Pediatric Asthma

Author Names: Sandeep Sharma¹, Barbara Rabin Fastman¹, Adam Vella², Lynne Richardson¹,², Lawrence C. Kleinman¹,³

Departments: Population Health Science and Policy, Emergency Medicine

Institute Affiliations: Icahn School of Medicine at Mount Sinai, New York, NY¹,²
Rainbow Babies Children’s Hospital, University Hospitals³
Case Western Reserve School of Medicine³

Objective: To assess appropriateness of ED use for children with asthma

Study Design: Medical chart review included 1190 emergency department visits from October 2009 – November 2013 for asthma at an academic teaching hospital in East Harlem. Outcomes were age-stratified as: 2 – 5, 6 – 11, and 12 – 18 years. Analysis included descriptive statistics (frequency and cross-tabulations), chi-square, and ANOVA using generalized linear models. We assessed eight appropriate use criteria that would be found in the medical record developed by a Center of Excellence in the federal Pediatric Quality Measures Program.

Population Studied: Random sample of children and adolescents with asthma ED visits

Principal Findings: Criteria could be applied reliably (kappa=0.87) indicating excellent agreement between reviewers. Study population was 40% Black, 53% Hispanic, and 7% other including White. 73% of visits were paid for by Medicaid/CHIP, 16% private, and 7% were uninsured.

The appropriateness proportion of pediatric asthma ED visits was 48%. By age: 54.3% for 2—5 years, 44.3% for 6-11, and 48.3% for 12-18 years were appropriate (p=0.02). By race/ethnicity: 44.1% of visits by Hispanics, 51.3% by Blacks, and 56.5% by whites were appropriate. By insurance status for Medicaid/CHIP was 46.3%, private 59%, and uninsured 38.6%. Gender (P=.017), Hispanic ethnicity (p=.002), private insurance (p=.005), and age group (p=.009) were significantly associated with level of appropriateness.

Conclusion: Less than half of ED visits for pediatric asthma met appropriate use criteria. Reasons may include unavailability of timely primary care, or patient preference for the ED over primary care. As a clinical construct, appropriateness requires data from the medical record.
Spatio-Temporal Ozone Variation in a Case-Crossover Analysis of Childhood Asthma Hospital Visits in New York City, USA

Author Names: Jessie L. Carr Shmool¹, Ellen J. Kinnee¹, Jane E. Clougherty¹, Jiang Zhou¹, Perry E. Sheffield²

Department: Preventive Medicine

Division: Environmental Health

Institution Affiliations: ¹University of Pittsburgh Graduate School of Public Health, Pittsburgh, PA, USA; ²Icahn School of Medicine at Mount Sinai

Introduction: Childhood asthma morbidity has been consistently linked with short-term air pollution exposure. To date, the majority of investigations have used time-series analyses, and it is not well understood how potential exposure misclassification arising from fine-scale spatial air pollution variation may impact upon epidemiological effect estimates. We evaluated effects of short-term fine-scale ozone exposure on childhood asthma exacerbation, adjusting for temperature and co-pollutant exposures.

Hypothesis: We hypothesized that there would be differences in magnitude and precision of effect estimates between a-spatial vs. spatio-temporal exposure estimates.

Methods: Case-crossover analysis included air pollution data from a) highly spatially-resolved intra-urban concentration surfaces, and b) daily regulatory monitoring data. Case data included citywide hospital records for years 2005-2011 warm-season (June - August) asthma hospitalizations (n = 2,353) and Emergency Department (ED) visits (n = 11,719) among children aged 5 to 17 years. Case residential locations were geocoded using a multi-step process to maximize positional accuracy. We evaluated differences in excess risks estimates by comparing results using spatio-temporal ozone exposure and covariate estimates to those using a-spatial daily estimates.

Results: Spatio-temporal models showed percent excess risk per IQR ozone exposure on lag days 1 through 3 ranging from 10.0 (95% CI: 1.2-19.6) to 13.3 (3.6-23.9) for hospitalizations, and on lag days 1 through 4 ranging from 5.0 (95% CI: 1.5-8.6) to 11.7 (7.2-16.4) percent for ED visits.

Conclusions: Excess risk estimates were up to 23% percent higher in spatio-temporal vs. a-spatial models, with stronger effects for shorter lag periods.
MPI Marks a Novel Axis in Glucose Metabolism Regulation in Embryos and Cancer Cells

Author Names: Nataly Shtraizent, Shikha Nayar, Jaime Chu

Department: Pediatrics

Division: Gastroenterology and Hepatology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Rapid cellular proliferation during embryonic development depends on glucose metabolism to fuel biosynthesis of new macromolecules. Metabolic enzymes are known regulators of this glycolysis-driven metabolic program, termed the Warburg Effect, however few have been identified.

Hypothesis: Mannose Phosphate Isomerase (MPI) promotes glycolysis and counteracts p53 to promote biogenesis for cell division.

Methods: We used a morpholino-mediated mpi knockdown model in zebrafish embryos to follow the dynamics of global and candidate gene expression (RNAseq and qPCR, respectively), metabolomics, biochemical assays (Mpi activity and lactate production), in vivo cell death labeling (Acridine Orange) and morphology analyses.

Results: Depletion of Mpi decreased glucose metabolism and DNA biosynthesis in a p53-dependent manner. Specifically, mpi knockdown reduced glycolytic flux, as shown by decreased lactate production, and expression of pfk and ldha genes. Moreover, Mpi was required to maintain normal nucleotide synthesis, based on the decrease in nucleotide precursors (L-Glutamine) and rrm1 gene expression. Consistently, supplementation of L-Glutamine into the embryo water partially rescued the mpi morphant phenotype, with reduction in morphological defects, cell death, and partially restored Mpi enzymatic activity.

Conclusion: We have previously reported the discovery of a negative feedback loop between MPI and p53. Here, we suggest that loss of Mpi induces a p53-mediated response, which further suppresses Mpi, glycolysis, and biosynthetic pathways. Our data expands our understanding of the glycolytic regulation during embryonic development and emphasizes the importance of MPI-p53 negative feedback axis in promoting survival in embryos and cancer cells.
Inflammation Suppresses Developmental Neuroplasticity

**Author Names:** Milo R. Smith¹,² Poromendro Burman¹,³,⁵, Masato Sadahiro¹,³,⁶, Masato Sadahiro¹,³,⁶*, Brian A. Kidd²,⁷, Joel T. Dudley²,⁷*, Hirofumi Morishita¹,³,⁵,⁶*,

**Department:** Neuroscience¹, Genetics and Genomic Sciences², Psychiatry³, Ophthalmology⁴

**Institution Affiliations:** Icahn School of Medicine at Mount Sinai; Graduate School of Biomedical Sciences⁶

**Institute Affiliations:** Mindich Child Health and Development Institute⁵; Icahn Institute for Genomics and Multiscale Biology⁷; Friedman Brain Institute⁸

**Background:** Developmental neuroplasticity is essential for shaping proper brain function and the high prevalence of developmental disorders demand deeper study to determine how diseases may disrupt neuroplasticity.

**Methods:** We developed and applied an integrative bioinformatics approach to systematically match plasticity to 436 disease signatures, to yield a ranked list of diseases most likely to dysregulate plasticity signature genes. We applied a novel Disease Leverage Analysis across the ranked disease list to identify shared pathophysiology that may disrupt developmental plasticity.

**Results/Conclusion:** By inferring shared pathophysiology of diseases signatures that match to plasticity signatures, we predicted inflammation would inhibit developmental plasticity and experimentally validated its suppression of juvenile cortical plasticity. Our findings suggest systemic inflammation in children may have unexpected negative consequences on the neurodevelopment trajectory by disrupting neuroplasticity.

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Detection of Anti-Glutamic Acid Debarboxylase (GAD) Antibodies in Immunoglobulin Products

Author Names: Tukisa D. Smith, Charlotte Cunningham-Rundles

Departments: Pediatrics, Medicine-Clinical Immunology

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Immunology Institute

Introduction: Serum Anti-glutamic acid decarboxylase (GAD) antibodies are highly specific for Stiff-person syndrome (SPS). Two patients with immunodeficiency receiving intravenous immunoglobulin therapy (Gammagard and Gammunex, respectively) had detectable anti-GAD antibodies (160.0 IU/ml and 103.8 IU/ml, respectively) and SPS was suspected. This diagnosis was supported by Electromyography (EMG) findings.

Hypothesis: We hypothesize that the detection of GAD antibodies were secondary to passive transfer from immunoglobulin replacement therapy

Methods: To examine potential passive transfer, we tested anti-GAD antibodies in 4 patients with X-linked Agammaglobulinemia (XLA) receiving immunoglobulin therapy, who, by definition, cannot manufacture antibodies.

Results: Serum from all XLA patients receiving immunoglobulin therapy were positive (3.1-89.0 IU/ml).

Conclusion: Our data suggests that passive transfer of anti-GAD antibody in immunoglobulin products may confound the use of this diagnostic test.
Profiling the Immune Response to Peanut Using Mass Cytometry

Author Names: Leticia Tordesillas¹, Adeeb H. Rahman²,³, Hugh Sampson¹, Cecilia Berin¹

Departments: Pediatrics¹, Human Immune Monitoring Core², Department of Genetics and Genomics³

Division: Allergy and Immunology¹

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: CyTOF (mass cytometry) improves multidimensional single cell analysis, with greater multiplexing capability, lower background, and higher sensitivity for detection of phosphorylation compared to flow cytometry. We developed a CyTOF panel to study acute responses to food allergens in whole blood.

Hypothesis: We hypothesized that apart from basophils and mast cells, other cell types could be involved in acute response to peanut.

Methods: Fresh blood samples from 6 peanut-allergic and 3 healthy subjects were activated with peanut extract, anti-IgE or media for 15 and 30 minutes, all in presence of IL3. Samples were combinatorially barcoded with palladium isotopes to reduce sample-specific batch effects and data collection variation. Pooled samples were stained with a panel of 29 metal-conjugated antibodies including markers for cell identification and activation. Cells were acquired on a CyTOF2, and SPADE analysis performed using Cytobank.

Results: Basophils from patients but not healthy controls upregulated known activation markers (CD63) after peanut stimulation. When activated, basophils increased phospho-ERK, -p38, and –S6, and downregulated CD38 and the FcεR receptor CD32. Aggregation of basophils and platelets was observed upon activation with peanut, measured by co-expression of CD61. In addition to basophils, we confirmed that other cell types showed evidence of activation after exposure to peanut in allergic but not healthy control subjects. cDCs, pDCs and monocytes increased phosphorylation of S6 upon peanut exposure.

Conclusions: We have established methods for profiling antigen-responsive cells in whole blood using mass cytometry. This provides insight into novel cell types activated by allergen, and elucidation of signaling pathways activated in allergic effector cells.
Epinephrine Use in Schools for Food-Induced Anaphylaxis

Author Names: Angela Tsuang, Haidi Demain, Kathleen Patrick, Michael Pistiner, Julie Wang

Department: Pediatrics

Division: Allergy and Immunology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: This study aims to assess epinephrine use in schools to treat food-induced anaphylaxis.

Hypothesis: We hypothesized that some unlicensed staff are giving epinephrine.

Methods: An anonymous questionnaire was distributed to all attendees at the Colorado school nurse training sessions in spring of 2015. Chi-squared test was used to compare proportions (STATA11).

Results: 243 of 341 surveys were completed (71% response). Most of the nurses had 1-5 years of experience (26%) or 6-10 years (24%). 43% of nurses covered 4 or more school buildings, and 10% reported epinephrine being given at least once by unlicensed staff.

In the 2014-2015 school year, 6% reported epinephrine was given by unlicensed staff at least once in schools with one nurse covering ≤ 3 buildings. In contrast, significantly more unlicensed staff members administered epinephrine at least once (16%) in schools with one nurse covering ≥ 4 buildings (p=0.013). There was no significant difference in the proportion of schools where epinephrine was given by unlicensed staff at least once among the 3 settings (rural, urban, and suburban) (p=0.242).

The majority of nurses reported that school staff received anaphylaxis training (80%); however 68% reported that the staff received training for only ≤ 30 minutes. Nurses felt that in-person training (78%) and hands-on demonstration (55%) are the most effective.

Conclusion: Schools with a high building to nurse ratio had significantly more unlicensed staff members administering epinephrine. Training should be extended to non-nursing staff in the form of hands-on training in order to increase student safety.
Alkaline Phosphatase During Growth Hormone Treatment: A Reflection of Growth?

**Author Names:** Lindsey Waldman, Marina E. Goldis, Kristen M. Williams, Christopher J. Romero, Elizabeth Burtman, Molly O. Regelmann, Elizabeth Wallach, Robert Rapaport

**Department:** Pediatrics

**Division:** Endocrinology and Diabetes

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Bone alkaline phosphatase (BALP) is a reflection of growth hormone (GH) treatment effectiveness. Total alkaline phosphatase (TALP) in serum reflects BALP.

**Hypothesis:** TALP levels will reflect changes in growth velocity

**Methods:** A retrospective chart review of children who had a growth hormone stimulation test from 2012-2013 was performed. Auxologic and laboratory data were obtained. Growth hormone deficiency (GHD) was defined as GH peak <10ng/ml. T Test and Mann-Whitney U tests were used for analyses.

**Results:** 74 subjects were included. GHD was diagnosed in 44 and 39 were treated with GH. 30 patients did not have GHD (NGHD), 11 were treated. There was no difference in TALP between GHD or NGHD at baseline (p=0.77) or 12 months (p=0.09).

There was a significant change in TALP in all treated (65.5 ±58.9 U/L) vs untreated (21.5 ±56.6 U/L) patients at p=0.002. In GHD, the change in TALP for those treated (68.8 ±54.2 U/L) vs untreated (46.4 ±89 U/L) showed no significance (p=0.74). In the NGHD group, the change in TALP was greater for treated (53.7 ± 75.2 U/L) vs untreated (14.9 ±46 U/L) at p=0.003.

For all patients, there was a correlation between the change in TALP and change in GV at 12 months (p<0.001). Correlation between GV and TALP in GHD treated was p=0.004 and in the NGHD untreated was p=0.003.

**Conclusions:** Our data suggest that in GHD patients treated with GH, the change in TALP reflect the change in growth velocity. Studies in more patients may elucidate the relationship between TALP and GV in NGHD treated patients.
Prescription of Epinephrine Autoinjectors to Children with Food Allergies in a General Pediatric Clinic

Author Names: Tamar Weinberger¹, Ari Zelig², Allison Gault¹, Julie Wang¹

Department: Pediatrics

Divisions: General Pediatrics, Allergy and Immunology

Institution Affiliations: ¹Icahn School of Medicine at Mount Sinai, New York, NY; ²Montefiore Medical Center, Bronx, NY

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Guidelines recommend that an epinephrine auto-injector should be prescribed to all patients with food allergy and an IgE mediated reaction. However, studies show that epinephrine is under prescribed.

Hypothesis: Pediatricians are more likely to prescribe an epinephrine auto-injector to patients with a history of peanut or tree nut allergy or to those with a history of anaphylaxis.

Methods: A retrospective chart review was performed of children with a diagnosis of food allergy over a one-year period from a general pediatrics practice. Charts were reviewed for prescription of epinephrine auto-injector, type of food allergy, age, sex, prior systemic reaction or anaphylaxis, coexisting asthma, atopic dermatitis, environmental allergies, and emergency department visits for allergic reactions or asthma exacerbations.

Results: Of the 317 charts analyzed, 139 had a documented peanut and/or tree nut allergy. 76.3% of those with peanut and/or tree nut allergy received epinephrine auto-injector prescriptions compared to 53.9% of those without (p<0.001). 78.6% of patient’s with a history of anaphylaxis received a prescription compared to 58.4% (p=0.001) without. After adjusting for confounding variables, children with peanut and/or tree nut allergy were 2.2 times more likely to receive prescriptions for epinephrine (OR=2.20 (95% CI 1.29-3.75, p=0.004)) and children with prior anaphylaxis were twice as likely to receive prescriptions (p=0.014).

Conclusion: Peanut and/or tree nut allergy and a history of anaphylaxis are associated with a significantly higher likelihood of receiving epinephrine auto-injector prescriptions from pediatricians. Further education must be provided to ensure that all children with food allergy are adequately prepared to treat any allergic reactions.
Complications, Length of Stay, and Economic Burden among Children Undergoing Pectus Excavatum Repair

Author Names: E. Hope Weissler¹, Paymon Sanati-Mehrizy¹, Benjamin Massenburg¹, Hillary Jenny¹, Peter J. Taub¹, Peter S. Midulla²

Department: Surgery

Divisions: ¹Plastic and Reconstructive Surgery, ²Surgery

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Pectus excavatum (PEx) is the most common chest wall deformity. The authors aimed to explore the prevalence and financial implications of complications of repair.

Hypothesis: We hypothesized that complications following PEx repair would be infrequent but expensive.

Methods: The HCUP-KID was queried patients from 2000-2012 with a primary diagnosis of PEx admitted primarily for its repair. Independent t-tests, Mann Whitney U tests, and regressions with variables found to be significant on univariate analysis (p<0.05) were used to relate hospital charges and complications.

Results: 9,032 patients were admitted for repair, of whom 85.0% were white and 16.5% were female. The average age was 14.17 years and the average length of stay was 4.64 days. 1,543 patients (17.1%) had at least one complication, most commonly iatrogenic pneumothorax (N=964, 10.7%), post-operative pain (N=436, 4.8%), and pleural effusion (N=239, 2.6%). There were no injuries of the heart or lungs. When controlling for race, hospital size, hospital teaching status, age, and household income quartile, complications were more likely in large hospitals (OR 1.32, 1.15-1.53) and with increasing age (OR 1.06, 1.03-1.08). The average hospital charge was $41,015.58. Using linear regression, hospital charges were associated with western location ($13,070.78, p<0.0001), age ($429.33, p<0.0001), large hospital size ($1,121.92, p=0.18), length of stay ($8,254.58, p<0.0001), and number of procedures ($3,296.04, p<0.0001) and diagnoses ($1,750.76, p<0.0001).

Conclusions: Complications following PEx repair occur in nearly one-fifth of cases; however, complications do not contribute to hospital charges if they’re not associated with additional necessary procedures.
Age, Socioeconomic Status, Race, and Congenital Nevus Excision

**Author Names:** E. Hope Weissler¹, Paymon Sanati-Mehrizy¹, Benjamin Massenburg¹,
Hillary Jenny¹, Peter J. Taub¹, Peter S. Midulla²

**Department:** Surgery

**Divisions:** ¹Plastic and Reconstructive Surgery, ²Pediatric Surgery

**Institution Affiliation:** Icahn School of Medicine at Mount Sinai

**Introduction:** Excision of congenital nevi is recommended for cosmetic benefit, as well as for malignancy prophylaxis. As the lesions grow with the children, excisions at a younger age may be technically easier. The authors examined patterns in congenital nevus excision.

**Hypothesis:** We hypothesized that older age at time of nevus removal would be associated with more complications.

**Methods:** HCUP-KID was queried for patients with diagnoses of congenital nevi and procedure codes indicating excisions between 2000-2012.

**Results:** 1,306 discharges were found, of average age 5.19±5.18 years. In a model controlling for gender, race, private insurance, high income quartile, and hospital region, white race (-1.53 years, p<0.0001) and high household income (-0.96 years, p=0.044) were associated with older age. In regressions controlling for gender, race, private insurance, high-income quartile, and region, increasing age was associated with fewer local excisions (OR 0.97, 0.94-0.99) and less tissue expander use (OR 0.96, 0.92-0.99) and more radical excisions (OR 1.05, 1.02-1.08) and graft use (OR 1.07, 1.04-1.10). 44 patients had complications. In a regression controlling for gender, race, private insurance, region, excision type, age, and income quartile, local and radical excision were less likely to be associated with complications versus tissue expanders, flaps, and grafts (OR 0.11, 0.03-0.45 and OR 0.19, 0.04-0.85, respectively).

**Conclusions:** Patient age drives selection of excision type for congenital nevi. Certain excision types are higher risk. Non-white and poorer patients may be at increased risk for complications following excision of congenital nevi.
Use of Focus Groups to Inform a New Community-Based Youth Diabetes Prevention Program

Author Names: Nita Vangeepuram, Farrah Khan, Sage Lopez, Crispin Goytia, Carol R. Horowitz

Department: Pediatrics, Population Health Science and Policy

Division: General Pediatrics

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Introduction: Rates of diabetes among youth have increased in the last decade and are disproportionately high in minority populations, necessitating new strategies for disease prevention. The study objectives were to explore: 1) peer influences on adolescent lifestyle behaviors, 2) strategies for implementing a peer educator model for diabetes prevention, and 3) use of mobile technologies as part of a youth diabetes prevention program.

Methods: We conducted 6 focus groups with East Harlem youth (ages 13-22). Trained moderators facilitated the groups, which were audio-taped and transcribed verbatim. Two researchers used Grounded Theory to develop a list of inductive and deductive codes, independently coded the transcripts, identified major themes, compared findings, and resolved differences through discussion and consensus.

Results: Participants (32 males and 20 females) self-reported as Hispanic (64%) and Non-Hispanic Black (36%). We identified 44 codes (>90% code agreement) and 3 dominant themes: 1) Adolescents generally encounter unhealthy peer influences related to dietary choices and healthy peer influences related to physical activity; 2) Adolescents endorse a youth peer educator model for diabetes prevention, and describe ideal qualities for peer leaders and methods to motivate, support and evaluate leaders; 3) Adolescents prefer social media for group discussions, support, and sharing pictures/videos, and text messaging to monitor behaviors, track weekly goals, and receive personalized advice and feedback.

Conclusion: A qualitative research approach yielded insight into why youth from urban, ethnically diverse communities are at increased risk for diabetes and how to decrease this risk through a combination of peer education and mobile health technologies.
Identification of the Nature of Cardiac Resident c-kit+ cells

Author Names: Lu Zhang, Nishat Sultana, Jianyun Yan, Jiqiu Chen, Weibin Cai, Shegufta Razzaque, Dongtak Jeong, Wei Sheng, Lei Bu, Mingjiang Xu, Guo-Ying Huang, Roger J. Hajjar, Bin Zhou, Anne Moon, Chen-Leng Cai

Department: Developmental and Regenerative Biology

Institution Affiliation: Icahn School of Medicine at Mount Sinai

Institute Affiliation: The Mindich Child Health and Development Institute

Introduction: Identifying a bona fide population of cardiac stem cells (CSCs) is a critical step for developing cell-based therapies for heart failure patients. For more than a decade, c-kit, a receptor tyrosine kinase expressed in certain types of hematopoietic stem cells, has been recognized as a marker of resident CSCs in mammals. It was shown that c-kit+ cells are multipotent, with differentiation potential to become cardiomyocytes, endothelial, and smooth muscle cells in vitro and after cardiac injury. Here, we provide new insights into the nature of cardiac resident c-kit+ cells.

Hypothesis: We hypothesized that c-kit+ cells in the heart may not represent true CSC population.

Methods: We generated a series of new mouse models by gene targeting to label c-kit+ cells in the heart and trace their progeny during development after injury.

Results and Conclusions: By targeting the c-kit locus with several reporter genes in mice, we unexpectedly found that c-kit+ cells rarely co-localizes with cardiac progenitor marker Nkx2.5 or myocardial marker cTnT. Instead, c-kit labels an endocardial population from embryonic stage to adulthood. After acute cardiac injury, the c-kit+ cells still retain their endothelial identity and do not become cardiomyocytes. Our study supports the notion that cardiac c-kit+ cells are in fact endothelial cells and not CSCs. This finding suggests an urgent need to re-evaluate the mechanisms by which c-kit+ cells contribute to heart repair or regeneration given their endothelial identity.
Maternal Violence, Maternal Lead Exposure, and Child Neurodevelopment in a Cohort of Mothers and Children in Mexico City

Author Names: Laura Zheng, Lourdes Schnaas, Marcela Tamayo y Ortiz, Erika Osorio-Valencia, Brent Coull, David Bellinger, Rosalind Wright, Mara Tellez Rojo, Robert Wright

Department: Preventive Medicine
Division: Environmental Health
Institute Affiliation: Icahn School of Medicine at Mount Sinai
Institute Affiliation: The Mindich Child and Health Development Institute

Introduction: Social stressors and lead exposure are associated with poor neurodevelopment, but research on their interactions is scant. No prior studies have addressed community violence as a stressor.

Hypothesis: Maternal violence exposure in pregnancy, maternal lead exposure, and their interactions, are predictors of infant neurodevelopment.

Methods: Longitudinal analysis of 504 mother-child pairs participating in the PROGRESS Study. Maternal violence exposure was assessed by questionnaire and converted to a continuous measure using Rasch modeling. Infant neurodevelopment was determined by Bayley-III composite scores at 24 months of age. We adjusted for maternal pregnancy blood lead, maternal IQ and age.

Results: The adjusted mean difference (95% confidence interval(CI)) in Bayley Cognitive, Motor, and Language scores per unit increase in violence exposure were -1.31 (-2.62, 0.00), -1.34 (-2.8, 0.12), and -1.48 (-2.87, -0.08). Among children with mean maternal pregnancy lead levels below 5 µg/dL (n=405), the adjusted mean difference (95% CI) in Bayley Cognitive, Motor, and Language scores per unit increase in violence exposure were -1.64 (-3.12, -0.15), -0.79 (-2.41, 0.83), and -1.09 (-2.65, 0.48). For children with maternal pregnancy blood lead levels above 5 µg/dL (n=99), the adjusted mean difference (95% confidence interval) in Bayley Cognitive, Motor, and Language scores per unit increase in violence exposure were -0.32 (-3.09, 2.44), -3.7 (-7.24, -0.17), and -2.84 (-5.89, 0.21). In a model with interaction terms, the p-interaction for violence exposure X maternal blood lead was 0.07.

Conclusions: Increased maternal violence exposure predicted decreased Bayley scores for cognitive and language scales. Evidence of a possible interaction with lead exposure was seen for motor and language scores were seen in the stratified models. This is important as violence and lead often co-exist in disadvantaged populations.