Pediatric Reseach Day is presented by Manning Family Children's, LSU Health New Orleans School of Medicine and Tulane University Medical School.

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Pediatric Research Day

May 2, 2025

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LSU Health NEW ORLEANS School of Medicine Department of Pediatrics

Manning Family Children's LCMC Health

Pediatric Research Day

Friday, May 2, 2025
Manning Family Children's
New Orleans
Worley Hall

Graduating Fellows' Presentations 8:00 am–9:15 am

Moderator: Amanda Messer, MD
Associate Professor of Clinical Pediatrics, LSU Pediatric Hospital Medicine

8:00 am Enhancing Bedside Education: a Pilot Study in the NICU
Mary-Elizabeth Lago, MD, Fellow, LSU Pediatric Neonatology

8:15 am Caregivers' Values during Neonatal Intensive Care Unit Course
Kathryn Cyrus, MD, Fellow, LSU Pediatric Neonatology

8:30 am Significance of Acute Neutrophilic Tubulitis in Kidney Biopsies
Jessica Rosario-Falero, MD, Fellow, LSU Pediatric Nephrology

8:45 am Social Determinants of Health in Pediatric Patients Hospitalized with Growth Faltering
Aamna Hafeez, DO, Fellow, LSU Pediatric Hospital Medicine

9:00 am Development of a Tracheostomy workshop for Pediatric Hospital Medicine Wards
Kayla Griese, DO, Fellow, LSU Pediatric Hospital Medicine

Features of Science 9:30 am–10:15 am

Moderator: Shubho Sarkar, MD
Assistant Professor of Clinical Pediatrics, LSU Pediatric Hospital Medicine

9:30 am A Preliminary Study of Glycemic Outcome During Real World Use of Automated Insulin Delivery Systems (AID) in a High-Risk Population of Youth with Type 1 Diabetes (T1D): Implications for Changes in Outpatient Management
Stuart Chalew, MD, Professor, LSU Endocrinology

9:35 am Comparison of Human-and ChatGPT-Generated Feedback
Bonnie Desselle, MD, Professor, LSU Pediatrics

9:40 am Standardizing and Improving Neurodevelopmental Follow Up of Infants with Hypoxic Ischemic Encephalopathy in the NICU
Michael Evers, MD, Fellow, LSU Neonatology

9:45 am IL-21 and IL-21R Interaction Plays a Key Role in Mounting Humoral and Cell-Mediated Immune Response to Adenoviral and Adeno-Associated Viral Vectors in Lung
Mina Hanna, MD, Resident, Tulane Pediatrics

9:50 am Step 2, AOA, and GHHS, oh my! Which ERAS Filters Predict Applicant Success in Pediatric Residency?
Oluwatosin Igenoza, MD, Resident, Tulane Pediatrics

9:55 am Industrial Toxic Air Pollution and Congenital Heart Defects in Louisiana: A Case-Control Study
Marla Johnston, RN, RN Clinical Trials Coordinator, LSU Pediatric Cardiology

10:00 am Three-Dimensional Sagittal Alignment in Adolescent Idiopathic Scoliosis: Reliability and Clinical Implications
Erik Piedy, Medical Student, LSUHSC School of Medicine

10:05 am Caregivers' Values during Neonatal Intensive Care Unit Course
Kathryn Cyrus, MD, Fellow, LSU Pediatric Neonatology

Faculty Research Forum 10:30 am–11:30 am

Moderator: Bonnie Desselle, MD
Professor of Clinical Pediatrics, LSU Pediatrics

10:30 am Ethnic Disparities in HbA1c and Hypoglycemia Among Youth with Type 1 Diabetes: Beyond Access to Technology, Social Deprivation and Mean Blood Glucose
Stuart Chalew, MD, Professor, LSU Endocrinology

10:38 am Behavioral Outcomes After Inpatient Rehabilitation in Pediatric and Adolescent Trauma Patients
Jessica Zagory, MD, Assistant Professor of Clinical Surgery, LSU Pediatric Sugery

10:46 am Building an Online Orientation, Evaluation and Reference Tool in a Pediatric Academic Center with Two Different Institutions Represented
Shubho Sarkar, MD, Assistant Professor of Clinical Pediatrics, LSU Pediatric Hospital Medicine

10:55 am FERTILE Project
Pinki Prasad, MD, MPH, Associate Professor of Clinical Pediatrics, Hematology/Oncology

11:04 am CRISPR-Mediated Detection of Pneumocystis Transcripts in Bronchoalveolar, Oropharyngeal, and Serum Specimens for Pneumocystis Pneumonia Diagnosis
Brady Younghquist, Doctoral Student, Tulane University School of Medicine

11:12 am Understanding Community Perspectives of Pediatrics Tissue-Based 'Omics Research Across Different Geographies
Sarah Glover, DO, Professor of Medicine, Tulane Gastroenterology and Hepatology

11:20 am Social Determinants of Health Curriculum for the Pediatric Clerkship
Caroline Roth, MD, Assistant Professor of Clinical Pediatrics, LSU Pediatric Hospital Medicine

Lunch/Networking 11:30am–noon

Opening Remarks Noon–12:15 pm

Stuart Chalew, MD, Professor of Clinical Pediatrics, LSU Pediatric Endocrinology

Keynote Speaker 12:15–1:00 pm

Aashim Bhatia, MD, Pediatric Neuroradiologist, Children's Hospital of Philadelphia

Oral Presentations 1:00–2:00 pm

Moderator: Jessica Zagory, MD
Assistant Professor of Clinical Surgery, LSU Pediatric Surgery

1:00 pm Social Determinants of Health in Pediatric Metabolic Dysfunction-Associated Steatotic Liver Disease (MASLD): Utilizing the Childhood Opportunity Index (COI)
Amber Gafur, MD, Fellow, LSU Pediatric Gastroenterology

1:15 pm Differential Gene Expression and Pathway Analysis in Eosinophilic and Reflux Esophagitis
Nicholas Habibian, Medical Student, Tulane University School of Medicine

1:30 pm New Predictive Equation for Oxygen Consumption During Pediatric Cardiac Catheterization Derived from Real Time Measure Data
Jesus Jaile, IV, MD, Fellow, LSU Pediatric Cardiology

1:45 pm Significant Telomere Attrition in Pediatric Ebola Virus Disease Survivors
Charlotte Osterman, Medical Student, Tulane University School of Medicine

Break 2:00–2:15 pm

Poster Session 2:15–3:15 pm

Poster Session 3:15–4:15 pm

Award Ceremony 4:30 pm

Judges

Andrew Abreo, MD
Assistant Professor of Clinical Pediatrics, Allergy/Immunology
LSUHSC School of Medicine

Rebecca Buckley, PhD
Associate Professor of Research, Department of Genetics
LSUHSC School of Medicine

Carter Clement, MD, MBA
Associate Professor of Clinical Orthopedics
LSUHSC School of Medicine

Bonnie Desselle, MD
Professor of Clinical Pediatrics
Vice Chair of Medical Education and Clinical Operations
LSUHSC School of Medicine

Anita Dhanrajani, MD
Associate Professor of Clinical Pediatrics and Section Chief, Rheumatology
Tulane University School of Medicine

Judges

Jessica Gautreaux, MD. MSPH
Associate Professor of Clinical Neurology and Section Chief,
Child Neurology
LSUHSC School of Medicine

Ricardo Gomez, MD
Professor of Clinical Pediatrics and Division Head, Endocrinology
LSUHSC University School of Medicine

Marcella Houser, MD
Associate Professor of Pediatrics, Ambulatory Medicine
LSUHSC School of Medicine

Scott Macicek, MD
Chief Experience Officer
Manning Family Children's

Christy Mumphrey, MD
Associate Professor of Clinical Pediatrics, Neonatology
LSUHSC School of Medicine

Shannon Palombo, MD
Assistant Professor of Pediatrics, Hospital Medicine
LSUHSC School of Medicine

Judges

Kimberly Terrell, PhD
Director of Community Engagement and Research Scientist
Tulane Environmental Law Clinic

Alfonso Vargas, MD
Professor Emeritus
Pediatric Endocrinology
LSUHSC School of Medicine

Stephanie Waldrop, MD, MPH
Assistant Professor, Clinical Sciences
Pennington Biomedical Research Center

Benjamin Watkins, MD
Associate Professor of Pediatrics and Section Chief, Hematology/Oncology
Tulane University School of Medicine

Lolie Yu, MD, MPH
Professor of Pediatrics, Hematology/Oncology
Vice Chair of Clinical Research
LSUHSC School of Medicine

Oral Presentations

Oral Presentation #1
1:00–1:15 pm

Social Determinants of Health in Pediatric Metabolic Dysfunction-Associated Steatotic Liver Disease (MASLD): Utilizing the Childhood Opportunity Index (COI)

Authors: Amber Gafur, MD; Zil Shah, MD; Aamna Hafeez, DO; Patricio Arias, MD; Michael Carver, MD

Presenting Author: Amber Gafur, MD
Fellow
LSU Pediatric Gastroenterology

Abstract:

Background: Research studies from a wide range of disciplines indicate the importance and influence of place-based economic, physical, and social conditions for child development. These factors also have a major impact on obesity in childhood and adolescence. A composite measure designed to evaluate neighborhood conditions that influence children's health and long-term outcomes has been developed called The Childhood Opportunity Index (COI). COI is a 29-item indicator of "the context of neighborhood-based conditions and resources that influence children's healthy development and long-term outcomes" which provides a comprehensive assessment. The COI includes aspects of the built environment including access to healthy food, access to green space, and walkability and the social environment including both economic and educational aspects, such as employment rate, homeownership rate, and high school graduation rate. The study aim is to investigate whether the presence and severity of metabolic dysfunction associated liver disease, or MASLD, has a place-based relationship by utilizing the Childhood Opportunity Index.

Methods: This is a single-center retrospective cohort study completed at a large, freestanding children's hospital (Children's Hospital of New Orleans). Patients will be sorted into COI Index categories based on their home address at time of visit. The study will include patients seen with diagnosis of fatty liver or MASLD in Children's hospital clinics between March 1, 2018 and July 31st, 2024. The study population will include all children aged 5 through 17 years old with ICD diagnosis of fatty liver or MASLD. Patients will be excluded if they have any other suspected etiology of elevated liver enzymes such as viral hepatitis, Wilson's disease, alpha-1-anti trypsin disease, Crohn's or Ulcerative Colitis, autoimmune hepatitis. This list is not exhaustive and will be up to discretion of chart reviewer. A retrospective chart review was conducted using electronic health records.

Results: Overall, 870 charts were reviewed and 441 patients met inclusion criteria. There was a slight male predominance 258 (59%) male patients vs. 183 (41%) female patients. The average age of diagnosis of MASLD was 12.6 years old and did not vary greatly amongst COI categories. There was a large predominance of patients in very low and low COI categories and distribution was as follows: Very Low 121 (27%) Low 168 (38%) Moderate 105 (24%) High 30 (7%) and Very High 17 (4%). Average BMI amongst all patients was 96.68 and demonstrated lower average BMI in those in the Very High COI category. Other metabolic markers were also measured with lipid panel being the only significant lab available in a large portion of the population study and demonstrated results as follows: Normal 135 (31%) Abnormal 215 (49%) No Sample 91 (21%).

Conclusions: This retrospective review highlights a significant association between lower COI categories and an increased prevalence of MASLD in children. The findings emphasize the need for public health policies that address the underlying social and environmental factors contributing to pediatric metabolic diseases. By focusing on improving childhood opportunities in disadvantaged communities, we can reduce the burden of MASLD and other related conditions in vulnerable pediatric populations.

Oral Presentation #2
1:15–1:30 pm

Differential Gene Expression and Pathway Analysis in Eosinophilic and Reflux Esophagitis

Authors: Nicholas Habibian; Hani Nakhoul, MD; Melody Badoo; Nazih Nakhoul, PhD; Solange Abdulnour-Nakhoul, PhD

Presenting Author: Nicholas Habibian
Medical Student
Tulane School of Medicine

Abstract:

Eosinophilic esophagitis (EoE) and reflux esophagitis (RE) are inflammatory diseases of the esophagus that can share similar clinical symptoms but differ in their pathophysiology and primary demographic. EoE is an immune mediated inflammation characterized by eosinophilic infiltration while RE results from reflux of acidic contents into the esophagus. Clinical symptoms of EoE include dysphagia and food impaction, whereas RE is characterized by heartburn and regurgitation, although these symptoms can be interchangeable. Despite similarities in the symptoms, EoE and RE have distinct clinical and histological features. The aim of this study is to use the gene expression profiles in EoE and RE to better understand molecular mechanisms of these diseases.

To study genes and pathways involved in esophageal tissue damage we analyzed gene expression data in pediatric pinch biopsies obtained from three patient groups, EoE, RE, and normal (NL). Three RT^2 Profiler™ PCR Arrays (Qiagen), 84 genes each, "*Human Extracellular Matrix & Adhesion Molecules*", "*Inflammatory Cytokines*" & "*Human Nitric Oxide Signaling Pathway*", were used in biopsies from EoE patients and gene expression was compared to NL patients. "*Human Extracellular Matrix & Adhesion Molecules*", & "*Inflammatory Cytokines*" arrays were used to study gene expression profile in biopsies from RE patients compared to NL. We used Ingenuity Pathways Analysis (IPA) to map differentially expressed genes in EoE and RE to biological pathways, causal networks, upstream and downstream effects.

In EoE samples, differential expression of genes (DEG) indicates the activation of canonical pathways including, pathogen induced cytokine storm signaling, CGAS-STING signaling, IL10 & IL 8. Upstream activating molecules include E. coli B5 lipopolysaccharide, CXCL12, NFκB & IL1A. In EoE samples, DEG indicates activation of biological pathways like movement of granulocytes and leukocytes. **In RE samples**, DEG indicates activation of canonical pathways PTEN & RhoGDI and inhibition of extracellular matrix organization & wound healing pathways. Upstream regulators include aryl hydrocarbon receptor, multiple cytokines and molecules including TNF-α, fibronectin-1, β catenin & CD40 ligand. Cell movement & cell-to-cell signaling, among other biological functions, are expected to be decreased in RE.

Based on gene expression analysis, the drivers of EoE and RE diseases differ significantly. In EoE, the analysis is consistent with activation of multiple cytokines pathways strongly confirming the inflammatory allergic origin of the disease. On the other hand, gene expression in RE indicates alterations in pathways and molecules mostly involved in tissue remodeling like integrins and collagen, indicating a distinct pattern of tissue injury and repair. This analysis also identified multiple factors; cytokines, chemical molecules and pharmacological agents; that could potentially act as candidates to modify gene expression in EoE and RE.

Oral Presentation #3
1:30–1:45 pm

New Predictive Equation for Oxygen Consumption During Pediatric Cardiac Catheterization Derived from Real Time Measured Data

Authors: Jesus Jaile, IV, MD; Thomas Kimball, MD; Marla Johnston, RN, MSN; Ernesto Mejia, MD; Zhide Fang, PhD

Presenting Author: Jesus Jaile, IV, MD
Fellow
LSU Pediatrics

Abstract:

Background: Determination of oxygen consumption (VO2) in the pediatric catheterization lab has been traditionally extrapolated through predictive equations which have become outdated. As technology has improved, the opportunity to measure VO2 in real time (M-VO2) now exists. However, some pediatric catheterization laboratories may not have access to such equipment. The purpose of this study was to develop a new predictive equation for VO2 in pediatric patients based on real time measurement of VO2 . Such an equation could be used by laboratories without real time monitoring abilities.

Methods: M-VO2 data were collected prospectively in 152 patients undergoing cardiac catheterization using a GE CARESCAPE monitor. Inclusion criteria were weight more than 2.5 kilograms and need for intubation during cardiac catheterization procedures The M-VO2 data were statistically compared against the most popular current predictive equation, the LaFarge equation, as well as the reference standard thermodilution method. A predictive equation (The Jaile Equation) was created and underwent post-testing of its predictive value in a new subset of test patients not used to build the formula.

Results: Correlation between LaFarge VO2 (L-VO2) and measured VO2 (M-VO2) using a Pearson Correlation Coefficient revealed the variables to be highly correlated with r =0.81 (p <0.0001). The correlation of M-VO2 to thermodilution was even more significant and closer to unity than the correlation of L-VO2 to thermodilution. (r 0.9765 p<0.0001, vs r 0.789 p<0.0008). Univariate and multivariate analyses identified gender, age, weight, and heart rate as variables significantly contributing to M-VO2 (p 0.0065, 0.015, 0.005, and <0.0001 respectively). These variables were used to build a predictive equation for M-VO2:

M-VO2 = 84.51 + 5.60 Gender– 0.51 Age – 0.14 Weight (kg) + 0.37 Heart Rate BPM

The predictive equation was then prospectively tested against M-VO2 and L-VO2 in the test group of patients. The equation proved to be a more accurate predictor of M-VO2 compared to LaFarge based on a lower Root Mean Squared Error (4.82 vs 29.05), and higher correlation coefficient (0.93 vs 0.86).

Conclusion: LaFarge estimates of VO2 provides a consistent overestimate of VO2 which can have impact on clinical decision making. Real-time measurement of VO2 provides a more accurate measurement as validated by simultaneous measurement with thermodilution. For heart centers who do not have the ability to measure VO2, the equation: the Jaile equation can be used to obtain a more accurate estimation of VO2 when compared to the LaFarge table.

Oral Presentation #4

1:45–2:00 pm

Significant Telomere Attrition in Pediatric Ebola Virus Disease Survivors

Authors: Charlotte Osterman; Trevor Roy, RN; Stacy Drury, MD, PhD; John Schieffelin, MD; Nell Bond, PhD

Presenting Author: Charlotte Osterman
Medical Student
Tulane School of Medicine

Abstract:

Background: Viral infections in childhood have a profound impact on the immune system and cause long-term clinical sequelae in several contexts, including Ebolavirus disease (EVD). Published data show that EVD survivors—including those infected as children—experience a significantly higher burden of adverse health complications compared to their peers, a phenomenon termed post-Ebola syndrome (PES). However, the mechanism behind PES remains unclear. Additionally, the long-term health and developmental consequences of Ebolavirus infection in childhood are unknown. One proposed mechanism for developing PES is accelerated physiologic aging as measured by prematurely shortened telomeres. Shorter telomere length (TL) is associated with poor health outcomes across several chronic and infectious disease states (i.e.: cardiovascular disease, HIV/AIDS). Underlying drivers of the disparate clinical burden experienced by young EVD survivors must be understood to mitigate negative health outcomes over the life-course. In this study we set out to determine 1) whether telomere length is shorter in EVD survivors compared to uninfected controls, and 2) whether telomere length is associated with PES in young EVD survivors.

Methods: Intravenous blood samples were collected during an ongoing study in Eastern Sierra Leone. Buffy coat was processed and stored according to standard methods. EVD survivors between 8-25 years of age were enrolled along with age/sex-matched controls at a 1:2 ratio. DNA was extracted and relative TL was quantified using monochrome multiple (MM) qPCR. Samples were plated in duplicate-triplicates and the intraclass correlation coefficient (ICC) was calculated for quality control. Relative mean TL was then calculated compared to a standard curve. Statistical analysis was performed in GraphPad/PRISM. Chi-square was used to compare categorical variables and non-parametric T-tests were used for continuous variables.

Results: Between 2016 and 2019, 117 EVD survivors and 234 age- and sex- matched uninfected controls were enrolled into the study. Cohort demographics were well matched with median age of 18 years (IQR 12-21) and 55% female participants in both groups. EVD survivors had significantly shorter telomeres compared to controls (TL=1.118 and 1.236, respectively, p=0.0016). However, there was not a significant difference in TL between survivors with and without PES (TL=1.101 and 1.082, respectively, p=0.9784).

Conclusions: We identified telomere attrition in pediatric survivors compared to their peers, which likely relates to the increased burden of health complications they experience long after recovery. We did not see an association between TL and PES within the survivor cohort, however, the small sample size may preclude detection of significant differences between sub-groups of survivors. PES may be associated with other biological processes involved in immune dysregulation and senescence which are currently under investigation. These findings are significant and have the potential to impact the long-term health of young EVD survivors who are likely more vulnerable to the early onset of aging related disorders. Studies are ongoing to determine the impact of TL and immunosenescence on the long-term health outcomes of EVD survivors infected as children.

Funding Acknowledgement: This study was supported by the NIH (1R01AI123535-01A1, 5U19AI135995-04, and 5U01AI151812-02 [PI: JSS]; and 5U24AG066528 [PI SSD]).

Poster Presentations

Students

A Case Report and Literature Review of Gabriele-de Vries Syndrome

Authors: Keisha Patel; Sloane Clay;
Regina Zambrano, MD

Presenting Author: Keisha Patel
Student
LSUHSC School of Medicine

Abstract:

Introduction: Gabriele-de Vries syndrome (GADEVS) (OMIM 617557) is rare autosomal dominant disorder caused by Pathogenic variants in Yin and Yang 1 (*YY1*) (OMIM 600013), a zinc-finger transcription factor that both represses and activates genes. GADEVS is characterized primarily by developmental delay/intellectual disability, facial dysmorphisms, intrauterine growth restriction, and feeding difficulties. Other reported features include hypotonia, abnormal movements, behavioral, skeletal, eye, cardiac, and renal abnormalities. In 2017, Gabriele et. al. reported the first 10 cases, and since then, a total of 32 cases have been published. Here, we present an additional case along with a literature review to further characterize the current phenotypic spectrum of this rare disorder.

Case: A 5-year-old male was referred to the Genetics Clinic at Children's Hospital of New Orleans for re-evaluation. First seen at 5 weeks old, initial testing included a chromosome micro array (normal). He continued to have developmental delay, dysmorphic features, micrognathia, feeding difficulties with gastrostomy tube in place, hypotonia, ADHD (attention deficit hyperactivity disorder), strabismus, patent foramen ovale, hypercalciuria, bilaterial nephrolithiasis, seizure like activity, vomiting, recurrent pneumonia, and obstructive sleep apnea. Upon re-evaluation and after a neurodevelopmental disorder panel was non-diagnostic, whole exome and mitochondrial sequencing (GeneDx XomeDxPlus) were recommended and identified a Likely Pathogenic variant in *YY1*, c.1106 A>G p. (Asn369Ser).

Literature Review: A case from Dos Santos et. al. was identified whose variant is identical to our case. They reported novel and infrequent findings such as non-febrile seizures, severe scoliosis, hearing impairments, and chorioretinitis. Our case shares major features with Dos Santos et. al.'s case such as developmental delay (33/33), facial dysmorphisms (33/33), feeding difficulties (25/33), behavior issues, eye abnormalities, hypotonia, vomiting, and pneumonia. They both also had seizure like activities, an uncommon feature of GADEVS (5/33). Our case also demonstrates features not present in the published case such as cardiac (6/33), renal (4/33), and sleep disturbances (4/33) which are also infrequent findings reported in GADEVS.

Discussion: A complete loss of *YY1* in mice resulted in peri-implantation lethality, but heterozygous mice showed neurulation defects and developmental restrictions, which suggest haploinsufficiency of *YY1* to be the cause for GADEVS. Additionally, *YY1* haploinsufficiency leads to loss of acetylation, allowing methylation of lysine 27 on histone 3 (H3K27) via polycomb repressive complex 2 (PRC2) which inhibits gene expression. In all, this case study and literature review contributes to the characterization and expansion of the phenotypic and clinical spectrum of GADEVS while demonstrating that patients with identical variants may display a variable phenotype. Future directions include clarifying the pathophysiology of this rare syndrome via functional studies, especially for infrequent, multisystemic features such as sleep, renal, and cardiac abnormalities.

Metastatic Primary Renal Ewing Sarcoma in a Pediatric Patient

Authors: Ngan Tran; Anna Simon, MD; Zachary LeBlanc, MD

Presenting Author: Ngan Tran
Student
LSUHSC School of Medicine

Abstract:

Background: Primary renal Ewing sarcoma (ES) is an extremely rare and aggressive tumor. ES is most commonly a primary bone malignancy, often occurring in adolescents. Very few cases with renal involvement have been reported in the literature. There are limited cases describing the management and treatment for this subtype of ES, especially in the pediatric population.

Case presentation: We report a 16-year-old male patient with metastatic primary renal ES of the left kidney who presented with non-specific lower abdominal pain and dysuria. A CT scan revealed a left kidney mass measuring 12 x 9 x 16 cm with encasement of renal vasculature and displacement of the aorta. MRI demonstrated bony metastases to the femur and pelvic griddle, and PET CT confirmed extensive skeletal metastasis. Additionally, cytogenetic review of the tumor biopsy was positive for ESWR1-FLI1 fusion protein characteristically seen in ES. The patient completed induction chemotherapy per COG protocol AEWS 1221 with excellent response. He underwent local control with left nephrectomy, with a successful complete resection. He continues to receive consolidation chemotherapy per this protocol.

Conclusion: This case highlights the rare incidence of primary renal Ewing Sarcoma with skeletal metastases in a pediatric patient

Case Series of Thickened Calvaria in Infants with History of Prematurity

Authors: Mohammed Rais; Gregory Fulton, MD; Matthew Blessing, MD

Presenting Author: Mohammed Rais
Student
LSUHSC School of Medicine

Abstract:

Background & Purpose: Infants with a history of severe prematurity, chronic lung disease, and other comorbidities associated with prematurity presented with distinct head shape and significant calvarial thickening. Recent research (Zapatero et al., 2023) identified a correlation between neonates with thicker calvaria and severe lung disease, along with a predisposition to craniosynostosis. While the etiology of this skull thickening is unknown, we hypothesize that intramedullary hematopoiesis causes proliferation of the red marrow in the diploic space. This phenomenon has been well documented in older children with cyanotic heart disease and hemoglobinopathies, but not in neonates and infants. We aim to contribute to this emerging literature by detailing similar cases across multiple craniofacial centers, as there are currently no publications describing the clinical presentation of these patients; this initial pilot case series seeks to address this gap.

Methods/Description: A pilot case series of patients were evaluated by craniofacial pediatrics at two centers due to concerns of head shape with prominent frontal bone malformation or calvarial thickening found incidentally on imaging. Patients were examined and the following data was recorded: birth history, pulmonary history, relevant medical history, head shape exam findings, imaging findings (if available), clinical photos (if available), clinical course, and outcomes.

Results: A retrospective chart review across two institutions identified the most recent 12 infants with prematurity evaluated by craniofacial pediatrics for prominent frontal bone malformation or calvarial thickening. After compiling information, multiple patterns emerged, with the majority of infants having a history of: extreme prematurity, bronchopulmonary dysplasia with chronic hypoxic and chronic hypercapnic respiratory failure requiring long-term respiratory support with or without tracheostomy, anemia, and osteopenia of prematurity. The most common head shape findings were significant brachycephaly and bilateral prominences of the central portions of the frontal and parietal bones, with relative concavity along the metopic and sagittal sutures. When imaging was available, it showed thickened calvarium, including proliferation of the diploic space, most prominently in the mid-frontal bones. Craniosynostosis was also noted in 27% of patients.

Conclusions: This case series details patterns in medical history and head shape that are common in infants with prematurity, chronic lung disease, and other comorbidities. It supports the findings of Zapatero et al., while highlighting variability in clinical presentations. Further research is needed to determine etiology of the calvarial thickness and to explore whether improved treatment of chronic lung disease, anemia, and osteopenia of prematurity can mitigate calvarial thickening in infants with a history of prematurity.

Novel Co-occurrence of Maternal Uniparental Disomy and a Pathogenic Variant in the SHOX Gene in a patient with Langer Mesomelic Dysplasia: A Case Report

Authors: Jordan Brignac; Greta Geiger; Kennedi Jones; Jariya Upadia, MD

Presenting Author: Kennedi Jones, MS, LCGC
Genetic Counselor
Tulane Pediatrics

Abstract:

Pathogenic Variants in the short stature homeobox-containing (SHOX) gene, located on the short arm of the X and Y chromosomes, are associated with a range of physical malformations. These include Leri-Weill Dyschondrosteosis (LWD), which is characterized by short stature, mesomelia, and Madelung deformity, a more severe form to nonspecific short stature. SHOX deficiency typically results from haploinsufficiency and follows pseudo-autosomal dominant inheritance. In rare cases, homozygous or compound heterozygous pathogenic variants in SHOX can lead to SHOX nullizygosity, resulting in a more severe phenotype known as Langer mesomelic dysplasia (LMD). LMD is marked by severe short stature and shortening of the long tubular bones. Some patients with LMD have inherited SHOX variants from parents with LWD. However, LDM caused by maternal uniparental disomy (UPD) of the X chromosome with a pathogenic SHOX variant has not been previously reported. We present a novel case of maternal UPD of the X chromosome with a pathogenic SHOX variant in a patient diagnosed with mesomelic dysplasia. The patient exhibited severe short stature along with shortening of the forearms and tibia-fibula. Molecular testing identified a homozygous nonsense variant c.582C>A (p.C194*) in SHOX due to maternal UPD. Microarray analysis confirmed the diagnosis, revealing a region of homozygosity indicative of UPD. This is likely the first reported case of LMD caused by maternal UPD of the X chromosome with the SHOX, c.582C>A (p.(C194*) variant, thereby expanding the genotypic spectrum of LMD.

Rare Pediatric Case of Diabetic Ketoacidosis in Cystic Fibrosis

Authors: Sydney Rein; Mary Younger-Rossi, MD; Adrienne Savant, MD

Presenting Author: Sydney Rein
Student
Tulane School of Medicine

Abstract:

Introduction: Cystic fibrosis (CF) is one of the most common autosomal recessive diseases, and it is associated with various multisystem comorbidities, including CF-related diabetes (CFRD). CFRD differs from type-1 diabetes (T1D) as it is caused by both insulin deficiency and insulin resistance. Diabetic ketoacidosis (DKA) is a serious, life-threatening complication of T1D; however, it is rare in patients with CFRD.

Treatment for CF includes CF transmembrane conductance regulator modulators (CFTRm), such as elexacaftor/tezacaftor/ivacaftor (ETI). CFTRm corrects the abnormal protein function causing CF, thus changing the underlying pathophysiology and leading to significant health improvements. However, the effects of CFTRm on CFRD are still under investigation. This case report highlights an episode of DKA in a patient with CF on ETI.

Case: This case is of a 16-year-old girl with a past medical history of CF (diagnosed after presentation with meconium ileus, F508del homozygous, sweat chloride 105/103 mmol/L) and pancreatic insufficiency. She was on routine CF therapies and was started on elexacaftor/tezacaftor/ivacaftor (ETI) at age 13. At age 15, she presented with polyuria, weight loss, fatigue, anorexia, and hyperglycemia, and she was diagnosed with CFRD. At the time of diagnosis, she had normal C-peptide and insulin levels and negative autoantibodies. Since her CFRD diagnosis, she has had suboptimal compliance with her insulin regimen.

The patient presented to the emergency department (ED) with one day of nausea and vomiting. One week prior, the patient presented to her primary care provider with a cough and was prescribed a seven-day course of prednisone. She had stable vital signs and normal examination in the ED; however, based on her lab values, she was diagnosed with DKA.

In the ED, she received standard treatment for DKA – one liter each of normal saline and lactated ringer's, five units of insulin aspart, and ondansetron and was admitted to the floor for further management. In the hospital, the patient's insulin regimen was adjusted. She received counseling on the management of her diabetes and demonstrated understanding of her new regimen and the effects of uncontrolled diabetes on CF. The patient was discharged without complications 38 hours after ED presentation.

Discussion: This case is unique as DKA is extremely rare in people with CF (PwCF) and has not been reported while on CFTRm. The multifactorial risk factors for DKA are highlighted – treatment with a CFTRm, infection, corticosteroids, and insulin omission. The pathophysiology of CFRD with remaining islet cells producing insulin alters the risk of DKA. However, the pathologic features of CFRD may be improved by CFTRm – such as potential reduced islet cell loss, increased residual insulin secretion, decreased hepatic insulin resistance, and improved incretin effects – raising the question of potential alteration in the risk for DKA in CFRD.

Assessment of Anticipated Behavioral Changes of Medical Students Following an Interactive Didactic Session on Adolescent Interviews

Authors: Kathryn Dillman; Brianna Bourgeois; Maria Lugo; Jade Lemoine; Caroline Roth, MD

Presenting Author: Kathryn Dillman
Student
LSUHSC School of Medicine

Abstract:

Purpose: In response to national recommendations and a gap at our institution, an educational activity was developed to target the objectives:
-Utilize the SSHADESS framework for conducting interviews with adolescent patients
-Demonstrate effective communication techniques, including active listening and non-verbal communication skills, when engaging with this population

Methods: The activity was included within a didactic session co-created by fourth-year medical students and a clerkship director. Third-year medical students participated in a session co-facilitated by a clerkship director and a fourth-year student chief where the SSHADESS assessment was introduced followed by a paired student role play activity. Participants voluntarily submitted a post-survey which included the question for thematic analysis: “Based on your experience during the role play, what do you plan to do differently when interviewing an adolescent patient during your clerkship?” We are employing the six steps of a reflexive thematic analysis outlined by Braun & Clarke to qualitatively analyze the responses to this question.

Results: 121 students attended the session between July 2024-January 2025 with a response rate of 92% to the survey question. Reflexive thematic analysis has resulted in four initial themes: working to create a welcoming and/or nonjudgemental environment, using techniques to elicit desired information, optimizing the provider-patient relationship, and using a patient-centered approach.

Conclusions**:** Preliminary analysis revealed a variety of planned changes by students which fall into the four abovementioned themes. Next steps include collection and analysis of responses for the remainder of the academic year with refinement of themes via the reflexive thematic analysis process.

Three-Dimensional Sagittal Alignment in Adolescent Idiopathic Scoliosis: Reliability and Clinical Implications

Authors: Erik Piedy; Claudia Leonardi, PhD; Carter Clement, MD; Amit Bhandutia, MD

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Abstract:

Introduction: The diagnosis, severity, and treatment of adolescent idiopathic scoliosis (AIS) are typically determined based on the major coronal curve as measured by the Cobb technique. However, spine literature examining adult patients suggests that the most important anatomic features for long-term spine health are radiographic parameters such as sagittal vertical axis (SVA), the difference between pelvic incidence (PI) and lumbar lordosis (LL), and other sagittal measurements. This study investigates the sagittal parameters of patients with AIS who reach skeletal maturity with a moderate-to-severe major coronal curve that does not meet the criteria for surgery. We hypothesized that many of these patients will exhibit sagittal alignment that raises concerns for their long-term spine health.

Case: A retrospective chart review was conducted on patients diagnosed with AIS between September 2016 and February 2022. Inclusion criteria included a major coronal curve between 30º and 50º upon reaching skeletal maturity and "graduating" from further monitoring. Clinical, demographic, and radiographic data were collected. Radiographic measurements included the major coronal curve, thoracic kyphosis, lumbar lordosis, cervical lordosis, pelvic incidence, sacral slope, sagittal vertical axis (SVA), pelvic tilt, T1-pelvic angle, and Roussouly sagittal classifications. Concerning sagittal parameters were defined as: SVA greater than 5 cm, pelvic tilt greater than 25º, lumbar lordosis-pelvic incidence (LL-PI) mismatch greater than 10º, cervical lordosis less than 20º, thoracic kyphosis less than 20º, and T1-pelvic angle greater than 14º. Patients were categorized based on Roussouly classifications using sacral slope and pelvic incidence. The percentage of patients with concerning sagittal parameters was reported.

Results: Sixty-seven patients met the inclusion criteria, with an average major coronal curve of 38.2º at the time of skeletal maturity. The average (standard deviation, range) for thoracic kyphosis, lumbar lordosis, cervical lordosis, pelvic tilt, and LL-PI were: 38.7º (13.3º, 9º–89º), 47.6º (11.05º, 18º–70º), 17.8º (19.3º, -20º–61º), 10.2º (7.88º, -10º–31º), and 8.9º (7.83º, 0º–37º), respectively. The average SVA was 4.16 mm (28.7 mm, -70 mm–70 mm), and the average T1-pelvic angle was 7.5º (6.23º, 1º–37º). Overall, 4.48% of patients had an SVA above 5 cm, 32.84% had an LL-PI mismatch above 10º, 10.45% had a T1-pelvic angle greater than 14º, 4.48% had a thoracic kyphosis less than 20º, and 4.48% had a pelvic tilt greater than 25º. Regarding the Roussouly classification, 10.45%, 7.46%, 56.72%, and 25.37% of patients were classified as type 1, type 2, type 3, or type 4, respectively.

Conclusions: This study suggests that AIS patients who barely miss the criteria for posterior spinal fusion (PSF) often demonstrate sagittal parameters that, based on adult spine literature, would be considered unacceptable. From a sagittal perspective, these patients are not significantly different from surgical AIS patients who undergo correction of sagittal alignment during PSF. These findings indicate that some AIS patients may benefit from PSF during adolescence, based on sagittal alignment rather than solely on traditional coronal curve criteria. Future long-term research is needed to determine if sagittal parameters should be included in surgical decision-making for AIS.

Sociodemographic Variables Are Rarely Reported in Randomized Controlled Trials Investigating Posterior Spinal Fusion For Adolescent Idiopathic Scoliosis: A Systematic Review

Authors: Erin Brown, BA; Kenneth Nguyen, BS; Bhavesh Koppala, BS; Alexander Kuzma, MD; Nicholas Baksh, MD

Presenting Author: Erin Brown
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Abstract:

Introduction: Adolescent Idiopathic Scoliosis (AIS) is a three-dimensional structural spinal deformity that affects children from the age of ten until they reach skeletal maturity. Untreated AIS can lead to significant physical deformity, as well as psychosocial and medical morbidity. Surgical intervention, particularly posterior spinal fusion (PSF), is considered when conservative treatment such as bracing fails. Demographic and sociodemographic variables influence AIS care, yet their reporting in randomized controlled trials (RCTs) remains unclear. The purpose of this study is to determine the rate of reporting of demographic and sociodemographic variables in RCTs investigating PSF for AIS.

Methods: Following PRISMA guidelines, PubMed, Embase, and Scopus databases were searched on April 9, 2024, using the terms “posterior spinal fusion,” “randomized controlled trial,” and “adolescent idiopathic scoliosis.” Inclusion criteria were RCTs on PSF in AIS, published in English, with accessible full texts. Exclusions included non-English publications, unavailable full texts, cadaver studies, technique articles, and non-RCT designs. Two authors screened studies for inclusion, resolving disagreements with a third author. Data collection involved recording the presence or absence of demographic variables like age and sex, as well as sociodemographic variables including race, ethnicity, familial insurance status, familial income/socioeconomic status, housing status, familial work status, and familial education level. Descriptive statistics, chi-squared tests, and Fisher's exact tests were used for analysis, with significance set at $P<0.05$.

Results: The initial search identified 148 studies, with 44 meeting inclusion criteria after screening and removal of duplicates. Demographic variables such as age and sex were reported in 97.7% and 95.5% of studies, respectively. Sociodemographic variables, including race and ethnicity, were reported in 6.8% and 9.1% of studies, respectively. Other sociodemographic variables like familial income, insurance, housing, work status, and education were absent in all studies. There was a significant disparity in the reporting of demographic versus sociodemographic variables ($P<.001$). No significant differences were observed in sociodemographic variable reporting by journal ($P=0.999$) or year of publication ($P=0.185$), although a trend towards increased reporting post-2017 was noted.

Conclusion: This systematic review highlights the under-reporting of sociodemographic variables in RCTs on PSF for AIS, with demographic factors like age and sex more frequently reported. Despite the significance of sociodemographic factors in patient care, their inclusion in RCTs remains limited. Improved reporting of these variables is essential for assessing pre-operative and post-operative outcomes, ultimately enhancing AIS patient care. Future research should prioritize comprehensive demographic and sociodemographic data to ensure equitable healthcare outcomes.

Orthopaedic Management of Bladder Exstrophy: Single Institution Experience of Seven Cases

Authors: Michael Starns; Srikanth Mudiganty, MBBS; William Accousti, MD; Brian Le; Rakin Nasar

Presenting Author: Rakin Nasar
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Abstract:

Background: Bladder exstrophy is a rare congenital condition characterized by malformations of the bladder, urethra, pelvic diaphragm, and bony pelvis. Surgical correction requires a multidisciplinary approach, with pediatric urologists and orthopaedic surgeons playing key roles. Orthopaedic treatment typically involves pelvic osteotomies to minimize pubic symphysis diastasis. This study aims to evaluate the immediate and long-term outcomes of seven bladder exstrophy cases from a single institution from an orthopedic perspective, focusing on functional surgical outcomes and patient quality of life.

Methods: This is an IRB-approved retrospective chart review study of seven pediatric patients from Children’s Hospital New Orleans who underwent bladder exstrophy repair surgeries between August 2009 and January 2024 with data sourced from the electronic medical record. Procedure and anesthesia reports were reviewed to gather data regarding the date of surgery, operation time, estimated blood loss, number of osteotomies performed, and the placement and removal of external fixators. The length of the pubic symphysis diastasis was measured during three instances: preoperative, immediately postoperative, and at last follow up visit. The guardian of each patient was contacted by telephone to inquire about the patient’s presence of hip stiffness, hip pain, limp, or a leg-length discrepancy. Overall contentment with surgical results was inquired by asking the patient’s guardian if there is satisfaction with the surgery outcome, if the guardian would elect to have the surgery performed again, and if the guardian would recommend the surgery to someone else.

Results: A total of seven patients (5 male and 2 female) were included in this study, with age of operation ranging from 4 to 44 months (12.57 mean, 7 median), mean operation time of 12 hours and 8 minutes, and mean estimated blood loss of 247.5 mL. Each surgery resulted in bilateral osteotomies that had all unionized by final follow up visit. Six of the seven patients received external fixator devices, with the devices being removed on average 49 days postoperatively. Preoperative, immediate postoperative, and final follow up diastasis length averages measured 46. 52 mm, 16.87 mm, and 39.99 mm, respectively. By final follow up, three patients experienced hip stiffness, five reported hip pain, three reported limp, and two reported a leg length discrepancy. The guardians for all seven patients endorsed satisfaction with surgery outcomes, a willingness to have the surgery performed again, and a willingness to recommend the surgery.

Conclusion: Pelvic osteotomies in bladder exstrophy repair effectively achieve bony union and initial reduction of pubic symphysis diastasis, though some postoperative widening occurs over time. While postoperative complications such as hip stiffness, pain, limp, and leg-length discrepancies were noted in some patients, overall guardian satisfaction with surgical outcomes was high. These findings support the role of orthopaedic intervention in bladder exstrophy management and highlight the need for long-term follow-up to address functional concerns.

The Utilization and Effects of Earmuffs on Neonates

Authors: Andrea Yazbeck; Meghan Howell, MD; Laura Wright, MPH

Presenting Author: Andrea Yazbeck
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Abstract:

Purpose: Preterm infants receiving treatment within the Neonatal Intensive Care Unit are at risk of exposure to hazardous sound environments which can result in long-term negative effects. The use of ear protective devices or earmuffs could help attenuate sound and reduce risk to newborns.

Methods: A systematic literature search was conducted using PubMed, SCOPUS, CINAHL, and Web of Science databases in September of 2023 through October of 2024. The search included all relevant peer reviewed studies conducted with preterm infants within the NICU who utilized ear protective devices. One independent reviewer performed the study selection, data extraction, and assessment of quality. The inclusion criteria for this review included infants born < 37 weeks gestation, infants that were admitted to the NICU and that individual ear protective devices (ex. earmuffs, ear plugs, etc.) were utilized. Additionally, the studies reviewed must have been interventional or observational studies, written in English language, published between 2000 - 2024, and had a minimal sample size of 10 participants. Exclusion criteria included non-individual ear protective devices or devices not designed specifically for auditory protection (i.e. incubators, review articles, systematic reviews, meta-analysis or case reports.

Results: Several studies found that the utilization of earmuffs decreased neonatal heart rate and respiratory rate both significantly and insignificantly. Additional studies demonstrated an increase in oxygen saturation with the intervention as opposed to without. Lastly, this review summarizes studies that have shown infants with sound attenuation interventions have lower scores on the Anderson Behavioral State Scoring System and better sleep outcomes.

Conclusions: This comprehensive review of previous studies suggests positive and statistically significant outcomes relative to infant physiology and behavior during the utilization of ear protective devices on neonates hospitalized in the NICU. However, given the limited information regarding this topic, further studies are recommended. The utilization of ear protective devices is a relatively low-cost adjustment (as compared to restructuring NICU infrastructure) and can help mitigate potential hazardous effect of noxious NICU sounds on premature newborns. Finally, as statistical significance does not correlate with clinical significance, studies examining more comprehensive markers of infant physiology and behavioral states, including longitudinal data, are warranted.

Literature Review: Enhancing Perioperative Management in Neuromuscular Scoliosis (NMS)

Authors: Andrew Suh; Andrew Gross; Solomon Baah; Kenneth Chiep; Eman Toraih, MD, PhD; Timothy Skalark, MD; Michael Kelly, MD; Carter Clement, MD

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Abstract:

Introduction: Neuromuscular scoliosis (NMS) presents unique perioperative challenges due to its association with severe spinal deformities and systemic complications. Effective perioperative care is critical to minimize risks and improve outcomes for these patients. This literature review synthesizes current research focusing on infection mitigation, respiratory complication management, and surgical safety to provide a framework for optimizing perioperative protocols. By synthesizing past research and current best practices, this review seeks to provide a cohesive framework to guide evidence-based perioperative protocols, ultimately improving outcomes in this vulnerable patient population.

Methods: The review analyzed peer-reviewed studies and clinical guidelines addressing perioperative strategies for NMS patients. Key topics included antibiotic prophylaxis, respiratory assessments, surgical techniques, and nutritional optimization. Data was synthesized to identify best practices and areas requiring further research.

Results: Antibiotic prophylaxis with tailored regimens, including cefazolin and amikacin, is likely to show increased efficacy in reducing surgical site infections (SSIs). Preoperative assessments, such as pulmonary function tests and sleep studies, were found valuable for risk stratification, though not universally predictive of outcomes. Surgical techniques like halo traction and posterior-only spinal fusion reduced complications compared to anterior-posterior approaches. Nutritional optimization, particularly vitamin D supplementation, was shown to enhance recovery and reduce complications.

Discussion: Tailored approaches to infection mitigation, including advanced antibiotic regimens and adjunctive therapies like incision negative pressure wound therapy, are critical for minimizing SSIs, though it is questionable on its validity on mitigating microbes. Respiratory assessments, while not mandatory, provide critical insights for perioperative planning. The adoption of less invasive surgical techniques and strategic nutritional interventions improves safety and recovery. However, variability in protocols highlights the need for standardized guidelines.

Conclusion: Optimizing perioperative care in NMS requires a multidisciplinary approach centered on infection control, respiratory management, and surgical safety. Future research should address gaps in evidence, particularly the efficacy of adjunctive therapies and the standardization of preoperative assessments, to improve outcomes for this vulnerable population.

Poster Presentations

Residents

Tacrolimus and Celiac Disease: When Medications and Gluten Don't Mix

Authors: Leah Smith, DO; Ushma Bhandary, MD; Corinna Victor, MD; Amber Gafur, MD; Christy Gallois, RN; Diego Aviles, MD

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Abstract:

Celiac disease is an autoimmune condition in which patients are unable to properly digest gluten due to a combined genetic predisposition and environmental trigger. The body's reaction is an abnormal and overwhelming immune response. This causes the hallmarks of celiac disease – mucosal damage of the small intestine and the production of autoantibodies. Symptoms typically include nausea, vomiting, diarrhea, bloating, and weight loss. While drug induced celiac disease is rare, it has been reported in some patients following kidney transplants. When a patient receives an organ transplant certain medications are necessary to suppress the immune system. Tacrolimus is a calcineurin inhibitor, which is involved in T-cell activation and proliferation, and is commonly used in these patients to decrease the likelihood of rejection. If graft failure occurs, immunosuppression medications are tapered and stopped. In this case report, we will discuss a 16-year-old female who received a deceased donor kidney transplant four months prior to presentation. Since the transplant she has remained on maintenance immunosuppression therapy with tacrolimus and mycophenolate. During this admission, she reported five weeks of nausea, daily vomiting, a twenty-pound weight loss, and recent diarrhea. Common causes of weight loss and vomiting following a transplant include graft rejection, decreased oral intake, infectious diarrhea, and deconditioning secondary to prolonged hospitalizations – all of which were considered in this patient. A stool sample was collected and negative for an infectious cause. An esophagogastroduodenoscopy was performed, and biopsies showed chronic duodenitis with increased epithelial lymphocytes, which is consistent with celiac disease. A tissue transglutaminase IgA was also collected and unremarkable, supporting the likelihood that the patient's celiac disease was drug induced, rather than antibody mediated. As seen in this patient, antibody testing may be negative and should not exclude the diagnosis of celiac disease. Therefore, if clinical suspicion is high it is recommended to move forward with a scope regardless of IgA results. Tacrolimus has many known adverse effects including abdominal pain, vomiting, diarrhea, headaches, insomnia, nephrotoxicity, and electrolyte abnormalities. Diarrhea is a very common symptom in patients taking tacrolimus; this may be due to the macrolide structure of the medication which mimics motilin and ultimately increases gut motility. There is also a risk of developing a variety of infections, which can be secondary to the immunosuppressive properties of the medication. For this reason, it is important to check trough levels frequently to ensure appropriate dosing. While celiac disease is a less common side effect of tacrolimus, it is important to consider the diagnosis in patients who are on immunosuppressive therapy following an organ transplant and are suffering from gastrointestinal symptoms and weight loss.

Treatment of Hereditary Angioedema with Variant in Plasminogen Gene in a Pediatric Patient

Authors: Hope Retif, MD; Devyn Rohlfs, MD; Kenneth Parish, MD; Andrew Abreo, MD

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Abstract:

Hereditary angioedema with variant in plasminogen gene is classified as HAE with normal C1-INH. It is a missense mutation with an amino acid exchange p.Lys330Glu in the kringle 3 domain of plasminogen. In patients with HAE-PLE, tongue swelling is the most common symptom, with two reports of asphyxiation due to upper airway obstruction. Icatibant, a selective, competitive beta-2 receptor antagonist, has been effective in treating HAE with normal C1-INH compared to corticosteroids alone and corticosteroids combined with antihistamines. In one study, Icatibant shortened the duration of swelling episodes on average by 88%.

This is a 5-year-old male with no past medical history who developed five episodes of lip and tongue angioedema associated with eating various foods. These episodes were unresponsive to epinephrine with only two instances aborting with Benadryl and the majority self-resolving in one to two days. The presence of angioedema did not share a common theme amongst food groups, and the patient was subsequently able to tolerate these same foods at a later date. Additionally, the incidents were not associated with pruritus, urticaria, fever, respiratory compromise, gastrointestinal symptoms or cardiovascular events. Interestingly, there was no family history of hereditary angioedema. Over the course of several ED and office visits, patient had a normal C4 (38 mg/dL), a normal C3 (146 mg/dL), a normal C1 esterase inhibitor (34 mg/dL), a normal C1 esterase inhibitor function (102%), and normal tryptase levels (5.3-6.6). Genetic testing showed a variant of uncertain significance in Plasminogen (c.493T>C) which can be associated with autosomal dominant hereditary angioedema. This patient was started on Icatibant 0.4 mg/kg/dose at the onset of an attack. He subsequently had an episode of lip angioedema where he administered Icatibant within one hour of symptom onset; however, lip swelling did not decrease or resolve as expected. Patient was evaluated in the ED and subsequently discharged after several hours of monitoring.

Since there are very few reports of patients affected with HAE-PLE, there are no randomized control trials and limited literature available on effectiveness of treatments, with Icatibant being the most effective treatment option studied. While this patient did not respond as well to Icatibant as others with this condition, further investigation is warranted in the dosage for pediatric patients and monitoring for decrease in duration and frequency in subsequent outbreaks.

Complex Challenges in the Nutritional Management of a Medically Complex Neonate

Authors: Sandya Vikram, MD and Maradith Skalak, MD

Presenting Author: Sandya Vikram, MD
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Abstract:

Congenital gastrointestinal anomalies and inborn errors of metabolism affect many infants. Some studies have shown that inborn errors of metabolism may be associated with an increased risk of congenital gastrointestinal anomalies. Managing nutrition in infants with gastrointestinal and metabolic conditions can be challenging. Each condition can lead to unique nutritional needs which must be balanced. Our case describes a premature infant with closed gastroschisis, short bowel syndrome secondary to jejunal atresia, and phenylketonuria and the management required to adequately support his growth and development.

The patient was born at 33 weeks' gestation, with prenatal ultrasounds concerning for gastroschisis. At birth, the gastroschisis was noted to have closed, but the infant was subsequently diagnosed with jejunal atresia, congenital short gut syndrome, and phenylketonuria (PKU). The patient was initially managed on total parental nutrition (TPN) following surgical repair of the jejunal atresia, while small volumes of enteral feeds were introduced. Due to limitations associated with short gut syndrome, the patient has remained TPN-dependent throughout his months-long hospitalization. He requires careful monitoring of amino acid levels in the setting of PKU, with specialized phenylalanine-free amino acid formulations in his TPN and specialized enteral formula. These specialized nutritional supplements result in the need for frequent changes to his TPN prescription, limiting his options for outpatient care. The complex interplay of his multiple medical conditions has required a multidisciplinary team, including neonatologists, gastroenterologists, surgeons, dieticians, and geneticists, to optimize his nutritional status and subsequent growth and development.

Nutritional management in neonates with multiple conditions affecting absorption and dietary restrictions can be complex. This case demonstrates an infant with unique medical challenges and underscores the importance of a multidisciplinary, personalized approach to managing nutrition for infants with multiple gastrointestinal and metabolic conditions.

Double Trouble: Unraveling the Rare Concurrence of IBD and Sickle Cell Disease

Authors: Shivani Patel, DO; Zil Shah, DO;
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Abstract:

Sickle cell disease (SCD) is a hemoglobinopathy caused by a point mutation in the beta-globin gene, which alters red blood cell shape and leads to vascular occlusion and reduced oxygen delivery. This causes variable manifestations including pain, anemia, splenic sequestration, and gallstones. Accurate diagnosis of co-existing conditions can be challenging in these patients due to overlapping symptoms. Inflammatory bowel disease (IBD) poses a unique challenge as its primary features- abdominal pain, weight loss, and diarrhea – are prevalent in patients with SCD. Concurrence of these diseases is rare but important to highlight given their impact on quality of life and the importance of early intervention.

A 9-year-old female with a history of sickle cell disease and irritable bowel syndrome presented to her hematologist reporting abdominal pain and an unintentional 18-pound weight loss, prompting a referral to gastroenterology. Initial evaluation indicated mildly elevated fecal calprotectin and elastase but was otherwise benign. Three months later, she presented to the emergency department with worsened abdominal pain, emesis, anorexia, and fever. Initial evaluation showed anemia (Hb 6.2 gm/dl), hypokalemia (2.4 mmol/L), and hypoalbuminemia (2.4 g/dl). She was admitted to the hematology service for a suspected vaso-occlusive episode, with gastroenterology consulted. Further evaluation was diagnostic of acute pancreatitis, raising concern for exocrine pancreatic insufficiency given her hypoalbuminemia and low fecal elastase. Repeat testing showed elevated fecal calprotectin, fecal elastase, and stool alpha-1-anti-trypsin (A1AT) and low fat-soluble vitamin levels.

The patient received albumin infusions during admission, without notable improvement. This poor response coupled with her elevated A1AT level was consistent with a protein-losing-enteropathy and potential IBD. A colonoscopy was performed, which showed evidence of Crohn's Disease. The patient was initiated on Infliximab and appropriate nutritional supplementation and discharged.
Since starting treatment, her abdominal pain and diarrhea have improved, and she has returned to her previous growth trajectory. The IBD team will continue to follow her monthly, with plans for repeat endoscopy.

This case highlights the difficulty of identifying comorbidities within the sickle cell population, leading to prolonged discomfort and symptom exacerbation. It reinforces the importance of maintaining a broad differential, and to avoid anchoring on typical SCD complications when a patient reports atypical symptoms or does not respond to treatment as expected. In conclusion, recognizing the potential for inflammatory bowel disease in patients with sickle cell disease is vital; timely diagnosis and intervention not only improve quality of life but also help prevent serious long-term complications, underscoring the need for a collaborative, multidisciplinary approach in managing these complex cases.

Ascending the Odds: A Rare Case of Pediatric Onset Colon Adenocarcinoma

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Abstract:

Background: Colorectal cancer (CRC) is the third most commonly diagnosed cancer and a leading cause of cancer death worldwide. The incidence of CRC is increasing; recent studies show this increase is confined to younger patients. Early onset CRC (EO-CRC) is defined as diagnosis in patients aged <50 years old. In certain regions the rate of EO-CRC has been shown to outpace cancer rates in older adults. However, CRC in the pediatric population (ages 0-19 years old) remains exceedingly rare, accounting for <0.1% of cases. Pediatric – onset CRCs (PO-CRCs) tend to be more aggressive in their biological behavior and are often diagnosed at advanced stages.

Case Presentation: A 16 year-old female originally presented with abdominal pain, nausea, vomiting, and blood in her stools. Colonoscopy revealed a mass in the ascending colon. She underwent surgical resection with left hemicolectomy and lymph node dissection. Pathology was consistent with Stage IIIc (pT4a,pN0,M0) colon adenocarcinoma. Genetic testing revealed a pathogenic mutation in the ATM gene and MEN1 gene variant of unknown significance. She began adjuvant treatment with FOLFOX (fluorouracil, leucovorin, and oxaliplatin) but completed only 3 cycles due to social challenges.

Three years later, she re-presented with abdominal pain and CT imaging showed a mass at the base of the mesentery, concerning for disease recurrence. Laparoscopic biopsy confirmed recurrence of CRC, and CT imaging showed mesenteric lymphadenopathy and liver lesion consistent with metastatic disease. She was initiated on FOLFOX + bevacizumab and has received 4 cycles thus far with clinical improvement.

Conclusion: Due to the rarity of PO-CRC and lack of age specific treatment guidelines, these patients are often treated with adult protocols. Despite advances in treatment strategies improving outcomes in adult CRC patients, PO-CRC has not seen these same advances. Research shows tumors in this age group may be biologically distinct from those seen in adults. Cases such as this highlight the need for additional research to find effective treatment strategies in this unique population.

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A Case of Pediatric Skin and Soft Tissue Infection with Interdisciplinary Management due to Underlying Fibrodysplasia Ossificans Progressiva

Authors: Eleanor Oubre, MD; Stuart DePaulo; Kayla Griese, DO; Caroline Roth, MD

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Abstract:

Purpose: Fibrodysplasia Ossificans Progressiva (FOP) is a rare genetic disease characterized by sporadic heterotopic bone formation. Soft tissue inflammation can lead to devastating ossification and rigidity. Patients are often treated with steroids and immunosuppressants. The immunocompromised status and high risk of ossification adds challenges to caring for patients with FOP presenting with common pediatric illnesses including skin and soft tissue infections. While rare, it is important to recognize patients with FOP and utilize resources for their management.

Case: 9-year-old female with FOP who presented with an acute onset, erythematous, fluctuant patch on the left buttock after a horsefly bite consistent with cellulitis and abscess formation. Prior to admission, patient received doxycycline which has cross reactivity with Palovarotene, a retinoic acid receptor-gamma agonist which decreases aberrant heterotopic ossification, so both were discontinued. She was then transitioned to Bactrim and cefdinir. In the ED, she underwent an incision and drainage (I&D) then was started on vancomycin and admitted. Wound cultures obtained were positive for MRSA, sensitive to Bactrim. The lesion demonstrated clinical improvement, so the patient was transitioned to Bactrim and discharged home after continued improvement. However, after discharge she developed worsening symptoms, and she returned to ER with signs of abscess reformation. She was readmitted and started on IV Linezolid per Infectious Disease recommendations.

The patient's primary endocrinologist and rheumatologist, experts in management of FOP, were contacted given the risks of progression with medical management alone verses complications secondary to FOP for surgical intervention. It was decided that surgery was the best option. Pediatric surgery and anesthesia were consulted and provided copies of FOP surgical procedure and anesthesia guidelines. To prevent further ossification the patient was also started on high dose steroids. The patient underwent a successful I&D with loop drain placement. Cultures obtained remained positive for MRSA. She was transitioned to oral linezolid for a 21-day course and discharged home. During her hospitalization, the patient experienced a FOP flair in her shoulder secondary to delay of her immunosuppressant in the setting of infection. The flair improved following the high dose steroids. We were able to resume her home medications prior to discharge from the hospital.

Conclusion: Our patient highlighted the risk analysis which should occur for missed medication doses, medication interactions, and procedural intervention. The International FOP Association has guidelines for how providers should manage patients with FOP, with examples applicable to our patient listed in table 1. This patient was discharged home with no known ossification emphasizing the impact of inter-specialty collaboration when treating a patient with unusual chronic illnesses and high risks of complications

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A Diagnostic Journey in a Patient with Suspected Pneumonia

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Abstract:

Purpose: This case study highlights the diagnostic challenges of botulism, a rare but life-threatening neuroparalytic condition. Due to the non-specific nature of symptoms, botulism can often go misdiagnosed during first presentation. The progressive development of hypotonia and respiratory in our patient underscores the importance of considering neuromuscular etiologies such as botulism in patients with atypical respiratory decline. By detailing this case, we aim to raise awareness among clinicians regarding the early recognition of botulism, its distinguishing clinical features, and the need for timely intervention to prevent respiratory failure.

Case: 4-week-old ex. 39w6d female presented to OSH with 1 day of feeding difficulty, congestion, and sleepiness. Mother described patient as having poor latch with breast feeding and appeared to be choking with bottle feeds as well as decreased wet diapers than normal, prompting her to present to OSH ED. Upon arrival to ED, patient was found to be grunting and hypoxic, so was suctioned and placed on low flow nasal cannula. Patient was observed in ED with feeds and would have frequent desaturations during feedings to low 80s. Patient was then transferred to our ED for further management. Upon admission, RVP negative and CXR with concern for consolidation, so patient started on IV Rocephin. Overnight, patient continued to have difficulty with feeds, so NGT was placed and patient started on continuous Pedialyte feeds. Throughout the morning, patient developed rapid decompensation. Noted to be lethargic with minimal response to pain, had short apneic spells with underlying bradypnea, and poor perfusion. Patient was started on 10L HFNC and admitted to PICU. Patient continued to show respiratory decline and required intubation. Upon intubation, patient noted to have decreased gag reflex. Urine drug screen and further infectious workup resulted negative. Patient developed worsening hypotonia, and decision was made to collect stool and blood for botulism screening. After collection, patient received botulism immunoglobulin. Patient then showed progressive improvement on exam, improved alertness, tone, strength, and reflexes. Patient remains intubated in PICU at this time, formal diagnostic testing pending.

Conclusion: Infant botulism can initially present with nonspecific symptoms such as feeding difficulties, hypoxia, and respiratory distress, often leading to a misdiagnosis of pneumonia or other infectious etiology. The progressive development of hypotonia, bradypnea, and diminished reflexes necessitate a broader differential diagnosis. Early identification and prompt administration of botulism immunoglobulin are critical in improving outcomes and preventing prolonged respiratory failure.

Delayed Diagnosis of Juvenile Dermatomyositis: A Multidisciplinary Challenge

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Abstract:

Introduction: Juvenile dermatomyositis (JDM) is a rare autoimmune disorder characterized by muscle weakness and cutaneous involvement. Diagnosing JDM in young children is particularly challenging due to its rarity, overlapping differential diagnoses, and limited access to pediatric rheumatology expertise. This report presents a case of delayed JDM diagnosis due to a narrowed diagnostic approach focused on genetic and congenital myopathies and prolonged specialist referral times.

Case Presentation: A 3-year-old female with no family history of autoimmune disease was referred for inpatient evaluation of suspected JDM. The patient initially met all developmental milestones; however, at 18 months, her parents noted difficulty walking, frequent tripping, and an inability to rise from a seated position without assistance. At 2.5 years, pediatric neurology assessed her for muscular dystrophy, with electromyography confirming myopathy, prompting further genetic testing. Whole exome sequencing identified variants of uncertain significance, delaying the recognition of an inflammatory myopathy.
Prior to hospital admission, the patient's primary care provider (PCP) noted firm subcutaneous nodules, prompting laboratory evaluation. Despite mild inflammatory markers, creatine kinase (CK) remained normal. However, calcinosis was observed in the trunk, neck, and extremities. A pediatric dermatology consultation four months later confirmed extensive calcinosis cutis, violaceous rashes, and nailfold capillary dropout. Urgent pediatric rheumatology evaluation identified proximal muscle weakness, diffuse calcinosis, heliotrope rash, and positive NXP2 autoantibodies, leading to a JDM diagnosis. MRI confirmed muscular and fascial edema, and the patient was admitted for treatment with high-dose corticosteroids, IVIG, and tofacitinib.

Discussion: The European League Against Rheumatism/American College of Rheumatology (EULAR/ACR) 2017 criteria for JDM include proximal muscle weakness, characteristic rashes, elevated muscle enzymes, muscle biopsy findings, MRI evidence of inflammation, and myositis-specific antibodies (MSAs). The patient's atypical presentation, absence of early cutaneous signs, and normal CK levels contributed to diagnostic delays. The differential diagnosis of JDM is broad, encompassing genetic, metabolic, and neurological disorders, necessitating a multidisciplinary approach.
Most JDM patients experience diagnostic delays exceeding three months, with younger patients and those residing far from pediatric rheumatology specialists facing longer delays. Calcinosis, a known marker of severe disease, is associated with delayed diagnosis and treatment initiation, especially in patients with anti-NXP2 autoantibodies. Increased awareness and expedited evaluation pathways for pediatric muscle weakness are needed to prevent complications.
Early recognition of JDM by PCPs is essential to improving outcomes. Educational initiatives should target general pediatricians and neurologists to include JDM in the differential diagnosis of unexplained muscle weakness. Early referral to rheumatology and collaborative care models can facilitate timely diagnosis and intervention. Future research should focus on strategies to enhance access to pediatric rheumatology care, develop algorithms for early identification of inflammatory myopathies, and mitigate the burden of delayed diagnosis on families.

Conclusion: This case highlights the need for improved awareness of JDM among non-rheumatology specialists and emphasizes the importance of timely diagnosis. Increased accessibility to pediatric rheumatology services, enhanced diagnostic algorithms can significantly affect disease outcomes and reduce complications associated with delayed treatment.

Starved by the Brain: A Case of ARFID Coinciding with Lupus Cerebritis

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Abstract:

Lupus cerebritis[1], a rare complication of systemic lupus erythematosus (SLE)[2], can cause a variety of neuropsychiatric symptoms, such as altered mental status, psychosis, and cognitive deficits. However, the development of avoidant/restrictive food intake disorder (ARFID)[3] secondary to neurocognitive changes is rarely described[4]. Case series have described Anorexia Nervosa as a prodrome to fully developed lupus. ARFID is an eating disorder marked by the avoidance of food due to sensory sensitivities, fear of consequences, or low appetite, resulting in weight loss or nutritional deficiency. A 17-year-old female with a history of juvenile idiopathic arthritis and a recent diagnosis of SLE, complicated by acute encephalopathy, presented with concerns of significant weight loss, reduced caloric intake, and dysphagia. She had previously been hospitalized for lupus cerebritis, during which an EEG revealed diffuse slowing, and a negative lorazapam challenge test ruled out catatonia. An NG tube was required due to poor oral intake, and earlier attempts to stimulate her appetite with cyproheptadine were unsuccessful. Despite ongoing outpatient medical management, she continued to experience nausea, reported difficulty swallowing, generalized weakness, food disinterest and fatigue and was readmitted. Further symptoms included dysarthria, psychomotor slowing, and mood disturbances, with the patient opting to sleep and avoid daily activities. Neuropsychological testing subsequently revealed a diagnosis of Major Neurocognitive Disorder, and psychiatric evaluation ruled out a primary eating disorder and body dysmorphia, attributing her reduced appetite and food aversions to prolonged hospitalization and stress and possible taste disruption. Her clinical presentation is consistent with ARFID in nature, likely influenced by her neurological insult, anxiety, and depression, although her oropharyngeal mechanism of swallowing remained intact as assessed by speech therapy. During this hospital stay a G-tube was placed for minor uptake of eating with multiple modalities. This case illustrates the intersection between neuropsychiatric manifestations of SLE and ARFID. Although lupus cerebritis often presents with altered mental status and cognitive deficits, its contribution to secondary eating disorders like ARFID is underreported and highlight the importance of monitoring neurocognitive and psychiatric symptoms in autoimmune diseases.

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Unexpected Congenital Tongue Mass in a Neonate: A Case of Glial Heterotopia

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Abstract:

Glial heterotopia is a rare but benign congenital tumor involving the displacement of neuroglial tissue in extracranial sites. This anomaly has an incidence of one in every twenty to forty thousand patients and greater than sixty percent of patients are diagnosed prior to one year of age. Accurate diagnosis requires histopathological examination, therefore, up to ninety-nine percent are misdiagnosed prior to biopsy or excision. Most commonly they occur in the nose, but can also be seen in the palate, tongue, scalp, orbit, and middle ear. Symptoms may differ greatly depending on the location of the lesion.

We report a case of a term female neonate with an unexpected large oral mass at the time of delivery. The infant had no respiratory compromise, however prompt transfer to a higher level NICU was warranted in the event a possibly difficult intubation was required. Transport to a center with pediatric imaging, multidisciplinary surgical services, and interdisciplinary support was critical. Additionally, oral feedings were compromised due to the location of the mass.

A neck MRI revealed a well-defined, heterogeneously enhancing mass arising at the posterior aspect of the tongue abutting the inferior surface of the uvula measuring 3.2 x 2.2 x 1.2 cm. Imaging was concerning for lingual hamartoma versus lingual hemangioma. Pediatric Otolaryngology-Head and Neck Surgery and Craniofacial teams were consulted upon admission. A yellow-tan mass was resected on the sixth day of life which consisted of glioneuronal tissue showing both neurons, positive for NeuN1, and glial cells staining positive for GFAP (glial fibrillary acidic protein) in a fibrotic background. NeuN1 is a protein marker exclusively expressed in nervous tissue. GFAP is uniquely expressed in astrocytes, which make up most of the central nervous system. Pathology also identified multinucleated and gemistocytic neurons, increased mitotic figures, and structures consistent with choroid plexus, all of which support the finding of neural tissue outside of the nervous system. Histopathological evaluation confirmed the diagnosis of benign glioneuronal heterotopia. A chromosomal microarray was obtained and did not identify any pathogenic variants.

The neonate recovered well with no acute complications, however she experienced feeding difficulties related to post operative pain requiring a sixteen-day NICU admission. A follow-up appointment at two months of age demonstrated no regrowth of the mass and no residual feeding difficulties.

This report highlights a unique patient case involving the rare diagnosis of a glial heterotopia of the oropharynx. Early diagnosis and intervention prevented further growth and potential impact of mass on feeding and breathing.

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Development of Lennox-Gastaut Syndrome in Patients with Previously Stable Rett Syndrome

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Abstract:

Rett Syndrome is a neurodevelopmental disorder, commonly associated with epilepsy, many times pharmacoresistant. Progression to Lennox-Gastaut syndrome (LGS) is rarely described. Though epilepsy is seen in Rett Syndrome, there are no specific guidelines about obtaining electroencephalograms or their frequency. These cases highlight how more frequent EEG monitoring in Rett patients with epilepsy could have made an earlier diagnosis of LGS. This can help ameliorate diagnostic and therapeutic challenges encountered during hospitalization. Furthermore, a diagnosis of LGS can ensure access to disease specific pharmacotherapy.

Case 1: A 9-year-old Hispanic female with Rett and prolonged QT syndrome was admitted after an episode of unresponsiveness with perioral cyanosis. Continuous EEG revealed atypical absence, tonic and atonic seizures. Generalized slow spike-and-wave activity in wakefulness and sleep with paroxysmal fast activity in sleep was noted concerning for a new diagnosis LGS. This EEG was much worse than prior studies done 3 years ago. Despite initial stabilization with an increase in perampanel, her seizure frequency worsened. A lorazepam dose improved the EEG background temporarily. Multiple other anti-seizure medications, including valproic acid, clobazam, and phenobarbital, were trialed which caused significant sedation lasting days. Cannabidiol improved seizure control, and MRI and cerebrospinal fluid studies were unremarkable. The patient was discharged home with continued subtle seizures with plans for further medication adjustments outpatient.

Case 2: An 11-year-old female with Rett syndrome, GMFCS V cerebral palsy, and progressive neuromuscular scoliosis developed altered mental status following posterior spinal fusion. An EEG revealed slow spike-and-wave discharges and left predominant paroxysmal fast activity in sleep. This EEG was different from her baseline EEG six years prior. MRI showed no acute abnormalities, although a small corpus callosal defect was reidentified. No changes were made to her home medications of lacosamide and perampanel. The patient returned to baseline days later with only supportive care.

A Case of Juvenile Xanthogranuloma Mimicking Maculopapular Cutaneous Mastocytosis

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Abstract:

Introduction: Maculopapular cutaneous mastocytosis (MPCM), also known as urticaria pigmentosa, typically appears as yellow-brown to red-brown, polymorphic macules or papules with an asymmetric distribution.[1] MPCM often develops in the first year of life, and is the most common form of cutaneous mastocytosis in children. The lesions are abnormal accumulations of mast cells; however, systemic symptoms and elevated tryptase levels are uncommon. The diagnosis of MPCM is typically based on the characteristic appearance, but skin biopsy may be useful in unusual cases that present a diagnostic challenge.

Case: A 10-month-old male with atopic dermatitis presented with diffuse yellow-tan macules on the face and trunk. The lesions first appeared at four months of life and were not associated with systemic symptoms. The lesions became erythematous and slightly raised with hot baths, emotional stress, and infections. He was diagnosed with MPCM based on clinical appearance. The lesions progressed into widespread reddish, yellowish, and brown papules. A skin biopsy performed by Dermatology confirmed the diagnosis of juvenile xanthogranuloma (JXG). He shows no signs of ocular or internal organ involvement, but will be closely monitored by multiple specialists.

Discussion: JXG is a non-Langerhans cell histiocytosis that also occurs in young children with a similar distribution. JXG typically presents as a solitary lesion, although multiple lesions or systemic involvement can occur. Both MPCM and JXG follow a benign course and usually resolve spontaneously without treatment. It is important to consider a skin biopsy and explore less common etiologies when skin lesions have an atypical appearance or the diagnosis of MPCM is uncertain.

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Beyond the Norm: A Male Infant with Turner and Down Syndrome Mosaic Patterns

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Abstract:

Background: Mosaicism refers to the presence of two genetically distinct cell populations within an individual originating from a single fertilized egg. Chromosomal mosaicism occurs when some, but not all, chromosomes are affected by abnormalities. Patients with Turner syndrome can present with mosaicism for X chromosome monosomy (eg. 45,X/46,XX) or (45,X/46,XY). Similarly, Trisomy 21 mosaicism results in some cells having a duplicate copy of chromosome 21. The varying degrees of mosaicism in different tissues can significantly affect phenotypic severity. Our case presents a rare instance of both Trisomy 21 and monosomy X (Turner) mosaicism in a male infant, leading to perplexing prenatal testing and a unique phenotypic presentation.

Case Presentation: The patient is a 2-month-old male infant with a prenatal history of maternal cell-free DNA testing indicating a 45,XO chromosomal abnormality. Ultrasound imaging showed male phenotypic features, raising concerns about a potential error in non-invasive prenatal testing (NIPT), as Turner syndrome typically presents with female phenotypic features. The parents declined confirmatory amniocentesis, opting for cord blood testing after birth. The mother had a NRFHRT, and the ultrasound showed mild pleural effusions and ascites, prompting admission. At birth, the patient required resuscitation with PPV and CPAP and was admitted to the NICU for respiratory distress and management of hydrops. Physical examination revealed male external genitalia with descended testes and no major features of Down's or Turner's syndrome. Chromosomal microarray confirmed mosaicism of two cell lines: one with trisomy 21 and a male XY sex chromosome complement, and the other with two copies of chromosome 21 and a 45,X chromosome complement, with mosaicism percentages of 40% and 60%, respectively. Additional testing revealed a vein of Galen malformation and a fenestrated ASD, requiring specialist evaluation. The patient will follow up with pediatric endocrinology for monitoring comorbidities, especially as puberty approaches, and with genetics for counseling.

Discussion: Turner syndrome karyotypes include 45 X, 45 X/46 XX, 45 X/46 XY, or structural abnormalities of the second X chromosome. Phenotypic manifestations of 45 X/46 XY can range from females with typical/atypical Turner syndrome features to ambiguous genitalia or males with incomplete masculinization. Individuals with Turner syndrome featuring a Y chromosome and normal male phenotypes with undescended testicles are both risk factors for developing gonadoblastoma, which can precede invasive tumors. Although virilization and marker chromosome elements often prompt Y chromosome mosaicism testing in Turner syndrome, phenotypic males with normal genitalia and descended testicles are less likely to be tested for chromosomal abnormalities. As trisomy 21 commonly presents with characteristic dysmorphic features that prompt confirmatory testing, this diagnosis may be missed if the physical exam is benign. However, these infants require thorough testing and vigilant follow-up for associated abnormalities such as cardiovascular, thyroid, renal as well as infertility issues. Furthermore, the increased risk of gonadoblastoma necessitates shared decision-making regarding clinical monitoring or surgical intervention. Thus, recognizing the rare coexistence of Turner and Down's mosaicism in phenotypically normal male infants during prenatal testing is crucial.

Wrist Chondrodesis After Failed Centralization Procedure in a Patient with Radial Longitudinal Deficiency

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Abstract:
Radial longitudinal deficiency (RLD) is a congenital condition that involves a spectrum of dysplasia long the pre-axial border of the upper limb, Presentation can range from mild thumb hypoplasia to complete absence of the radius. Patients with RLD often experience functional impairment due to thumb hypoplasia, wrist instability with radial deviation of the hand on the forearm and a shortened forearm. Treatment depends on a variety of factors such as, the severity of the wrist and forearm deformity, degree of thumb involvement, and the patients age. In this case report, we discuss a patient with radial longitudinal deficiency who previously underwent a centralization procedure at one and a half years old and presented to clinic with continued wrist instability and thumb hypoplasia. Initial radiographs demonstrated failure of the previous centralization procedure with a pin present in the forearm soft tissues. Patient subsequently underwent hardware removal and chondrodesis of the wrist with successful stabilization of his wrist. Literature and consensus on treatment of RLD after failed centralization procedure is scarce. This case report presents an effective surgical procedure for this complex issue.

IL-21 and IL-21R Interaction Plays a Key Role in Mounting Humoral and Cell-Mediated Immune Response to Adenoviral and Adeno-Associated Viral Vectors in Lung

Authors: Mina Hanna, MD; Robert Clark, MD, MPH; Alex Martin; Dora Pungan; Dong Lin, PhD; Jay Kolls, MD

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Abstract:

Purpose of Study: Although cystic fibrosis (CF) was discovered ~ a century ago, it remains without an available cure for all mutations. 8-10% of CF patients remain without therapy options despite advancements in targeting misfolded CFTR. Gene therapy is an active area of investigation in hopes of tackling this unmet need; however, the need for re-dosing remains a challenge for any means of viral vector delivery. Thus, mitigating the immune response may be the key to overcome this challenge. Given the central role of IL-21R in regulating T and B cell immune response, we hypothesized that targeting IL-21R may attenuate B and T cell responses and enhance secondary transgene expression.

Methods: Naïve or IL21R-/- mice received $4x10^{10}$ viral particles and $1x10^{11}$ viral particles intratracheally of an Ad5 vector for first and repeat doses, respectively, at days 0 and ~27. Serum anti-vector antibodies were assayed 2 weeks post-dosing. All mice were euthanized 3 days following 2nd viral dose. Lung tissue was harvested for analysis of B cells and CD8 tissue-resident memory (TRM) cells as well as secondary transgene expression. Similarly, naïve or IL21R-/- mice received $1x10^{11}$ vg of adeno-associated virus 6.2 (AAV6.2) by endotracheal intubation and monitored with *In Vivo* imaging system (IVIS) to assess for transgene expression.

Summary of Results: At 2 weeks, serum anti-AdV IgG ($P < 0.05$) and IgG1 ($P < 0.01$) were significantly decreased in the IL21R-/- mice by 2-way ANOVA. Flow cytometry analysis showed a significant reduction of CD8 TRM cells in IL-21R-/- mice ($P < 0.01$) by nonparametric T-test and a reduction in CD19 B cells ($P < 0.05$) by 2-way ANOVA. Moreover, secondary transgene expression was significantly enhanced in IL-21R-/- mice compared to B6 controls ($P < 0.01$). Similarly, in the context of AAV the generation of anti-AAV IgG requires IL-21R signaling ($P<0.0001$). IVIS also showed significantly enhanced secondary transgene expression of AAV6.2 in IL-21R-/- ($P<0.0001$).

Conclusion: Our results consistently showed how IL-21R and IL-21 play an important role in the development of humoral and CD8 TRM-mediated responses to adenoviral and AAV vectors. This suggests how targeting IL-21R signaling may be the key to overcoming challenges for gene therapy using viral vectors as a means of delivery. Future studies will investigate the role of IL-21R in adult mice using antibody blockade.

Birds, Bees, & Clinical Expertise: A Qualitative Exploration of Healthcare Providers' Perspectives on Sex Education in Louisiana

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Abstract:

Background: Louisiana consistently ranks among the worst states for new diagnoses of chlamydia, gonorrhea, HIV, and syphilis, and currently ranks third among states for teen births. Meanwhile, sex education in Louisiana is not required by law and if taught, it must emphasize abstinence as the expected social standard. Previous research has identified healthcare providers as a trusted source for information on sexual and reproductive health. Little is known, however, about Louisiana healthcare providers' perspectives on sex education and the barriers they face to providing sex-ed in clinical settings.

Methods: Using qualitative methods, we conducted semi-structured interviews with eligible participants via the online Zoom platform. Eligibility criteria include anyone currently providing primary care services to adolescents 10-24 years old in Louisiana. A convenience sample of participants were recruited via email from a previous pool of research subjects, although prior study participation was not a requirement for study inclusion. Participants were also recruited via snowball sampling. Participants were asked a series of questions regarding their knowledge, attitudes, and experiences with sex education as well as barriers to providing sex-ed in healthcare settings. Preliminary data was transcribed using NVivo transcription software and coded for common themes using NVivo analysis software. Data collection is ongoing.

Results: Preliminary results among nine healthcare providers include six resident physicians, two attending physicians, and one nurse practitioner. Many participants cited low levels of health literacy, difficulties accessing sexual health resources, and a lack of sex-ed in Louisiana schools as contributing factors to the state's current adolescent sexual health outcomes. Several participants mentioned the importance of interviewing adolescents alone during healthcare visits to discuss sexual health topics. Most participants expressed confidence educating patients on anatomy/physiology and general sexual health and expressed difficulties talking to patients about gender and sexuality. All participants provided examples of "time" as a barrier to providing sex-ed in their clinical practice. Several participants expressed surprise at the current HIV Pre-Exposure Prophylaxis (PrEP) guidelines and revealed that PrEP is not something they commonly discuss with all their sexually active adolescent patients. Many participants expressed fear and worry over the future of sex-ed in Louisiana. Finally, all participants believed that healthcare providers play an important role in educating Louisiana youth on their sexual and reproductive health.

Conclusions: Louisiana healthcare providers have important knowledge and experiences to share regarding sexual and reproductive health education. Overall, study participants see themselves as trusted sources of information for sexual and reproductive healthcare topics and strive to incorporate this education into their clinical practice. Louisiana healthcare providers are filling in sex-ed gaps for their patients and families, however, they cannot shoulder this burden alone. Future research and resources should aim to strengthen healthcare settings as access points for sex education.

Step 2, AOA, and GHHS, Oh My! Which ERAS Filters Predict Applicant Success in Pediatric Residency?

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Abstract:

Study objectives: While medical education programs nationwide have transitioned to competency-based assessment, United States Medical Licensing Exam (USMLE) scores remain the metric most frequently used by residency programs to rapidly narrow the large applicant pool. The recent shift to Step 1 pass/fail reporting has increased reliance on Step 2 CK scores, despite weak and conflicting evidence of the association between USMLE scores and clinical skills in residency. This study examines whether several filterable medical school metrics reported in the Electronic Residency Application Service (ERAS) predict success in pediatric residency, as measured by milestone achievement on the Accreditation Council for Graduate Medical Education (ACGME) pediatric competencies.

Methods: Pediatric residency programs participating in Association of Pediatric Program Directors (APPD) Longitudinal Educational Assessment Research Network (LEARN) were invited to join this study in 2020; ten of 149 programs provided data on categorical residents entering between 2016-2019. Metrics included type of medical degree, resident graduation from a US medical school, membership in Alpha Omega Alpha (AOA) and Gold Humanism Honor Society (GHHS), Step 1 score, Step 2 Clinical Knowledge (CK) score, and Step 2 Clinical Skills (CS) first attempt pass/fail status. Primary outcome was mean Pediatric Milestone (PM) ratings at mid-point and end of each residency year, analyzed as a continuous variable. Year of training served as a positive control.

Results: Data from 518 residents across 10 programs were analyzed. Ninety percent graduated from US medical schools; 14% and 16% were AOA and GHHS members, respectively. Year of residency training was positively associated with success in all competency domains. GHHS membership and Step 2 CK were positively associated with overall PM performance, while Step 1 was negatively associated. By competency domain, Step 2 CK was positively associated with Medical Knowledge (MK), Patient Care (PC), and Practice-Based Learning and Improvement (PBLI). GHHS membership was positively associated with Interpersonal and Communication Skills, MK, PC, PBLI, and Professionalism. The effect size of GHHS on PM ratings was higher than Step 2 CK across all domains. Step 1 was negatively associated with PBLI and Professionalism.

Conclusion: Among filterable ERAS metrics, GHHS membership showed the strongest association with pediatric residency success, as measured by PM ratings in this multi-site cohort of pediatric residents; Step 2 CK had a weaker positive association.

Metabolic Basis of Post-Infectious Sequelae After Ebola Virus Disease

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Abstract:

Background: Ebola virus disease (EVD) survivors present with various sequelae collectively described as post-Ebola syndrome (PES). The etiology of PES development after Ebola virus (EBOV) infection is not well described, although theories including autoimmune effects, chronic infection or reactivation have been proposed. Metabolomics can demonstrate viral influence on metabolism, broadening our understanding of mechanisms of viral sequelae development. Metabolomics has been applied to post-viral sequelae in long COVID but has not been utilized to study post-viral sequelae of many other viral infections. Certain metabolites are known to influence immune response to pathogens, including Krebs cycle intermediates. Here, we present results from a metabolomic pilot study in EVD survivors with and without PES.

Methods: Eighty participants were included as part of an ongoing cohort study of EVD survivors in Sierra Leone. Survivors were selected for the PES group if they demonstrated musculoskeletal (MSK) and/or gastrointestinal (GI) sequelae or cardiopulmonary (CP) sequelae. The featured analysis compares survivors with PES sequelae (n=37) and EBOV antibody positive asymptomatic survivors (n=20). Plasma samples were sent to a third party for liquid chromatography mass spectrometry (LC-MS). Peak intensities were analyzed in MetaboAnalyst. To minimize the number of false positive results, false discovery rate (FDR) was set at .05. Logistic regression for significance of demographic characteristics was performed in RStudio.

Results: EVD survivors with PES demonstrate alterations in short chain fatty acid (SCFA) levels (acetate, propanoate/propionate) and Krebs cycle metabolites. Downregulated Krebs cycle metabolites include succinate and malate, among others. Partial least squares discriminant analysis (PLS-DA) revealed separation of metabolites between the PES and asymptomatic survivor groups. Demographic characteristics including age, sex, and date of plasma collection did not predict PES when comparing survivors with and without PES using logistic regression.

Conclusions: Survivors of EVD with PES express a unique metabolic footprint, characterized by downregulation of many Krebs cycle intermediates and possible gut microbiome disruption noted by changes in SCFA levels. The alterations in Krebs cycle intermediates is significant given the implications for disrupted energy metabolism and these metabolites' roles in immunometabolism. Similar patterns of downregulated Krebs cycle activity have been noted in exhausted T cells in chronic viral infections, but the significance of these findings in plasma levels is not well described. Further studies exploring connections between metabolism, gut microbiome and immune responses in EVD survivors are required to fully understand the implications of these results.

Understanding Causes of Disparate Outcomes in Pediatric Acute Leukemia: Social Vulnerability Index as a Predictor for Increased Hospitalizations for Infections during Intensive Chemotherapy

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Abstract:

Background: Outcomes for pediatric leukemia are excellent due to modern cooperative group treatment protocols and advances in supportive care. However, disparities persist among patients from minoritized and lower socio-economic backgrounds. Infections remain a significant cause of morbidity and mortality in pediatric leukemia patients. We hypothesized vulnerabilities associated with social determinants of health (SDoH) would correlate with a higher incidence of infections requiring hospitalization during intensive chemotherapy in this patient population.

Methods*:* A retrospective chart review was performed on 94 patients treated during the initial stages of chemotherapy treatment for acute leukemia at a tertiary care pediatric hospital from 2017 to 2024 in Louisiana. We collected data on patients' gender, race/ethnicity, insurance status and preferred language. SDoH was assessed using two indices based on home address: the Childhood Opportunity Index (COI) and the Social Vulnerability Index (SVI). Data on all infections leading to or extending hospitalization was collected as well as whether alive or deceased at last contact. Chi-square tests were used for association analysis and Wilcoxon Rank-Sum test was used to compare continuous variables. Patients were categorized into groups based on the number of infections during the study period: 0, 1-2, 3-4 or more than 5 infections.

Results*:* The study population included patients with B or T cell ALL (74), AML or APML (18), or blastic plasmacytoid dendritic cell leukemia. Six patients had relapsed leukemia. Demographics were as follows: 50% female, 30% Black, 24.5% Hispanic, and 22.3% with non-English language preference; 69% of patients had public insurance, 5 were uninsured. 75% lived more than 70 miles from our hospital. Compared with national standards, 81% of patients had either very low (37, 39.4%) or low (40, 42.6%) COI score. Compared with state standards, 46% of patients had either very low (20, 21.3%) or low (24, 25.5%) COI score. 61.7% (58) of patients lived in areas with high SVI, indicating greater social vulnerability. 67% of patients were diagnosed with at least one infection requiring readmission or extending hospitalization; 28 patients had 1-2 infections, 22 had 3-4 infections, and 12 had 5 or more infections. While no single factor predicted an increased risk for infection, patients coming from areas of high SVI were more likely to be hospitalized for multiple infections (p= 0.0596). The overall survival rate of the group was 82%. No factor was associated with an increased risk for mortality.

Conclusion: Our findings suggest pediatric patients with acute leukemia coming from areas of high SVI based on zip code are hospitalized for more infections compared to patients from areas of low SVI. Our findings may help illuminate the causes of broader disparities in pediatric acute leukemia outcomes. This underscores the need for ongoing efforts to address vulnerabilities related to SDoH to improve oncological outcomes, especially in underserved regions like ours where treating leukemia can be uniquely challenging.

Poster Presentations

Fellows

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That Sinking Feeling: Two Cases of Adolescents with Gastroptosis

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LSU Pediatric Gastroenterology

Abstract:

Gastroptosis is a rarely reported condition where the stomach is displaced inferiorly towards the iliac crest while standing. It is typically diagnosed with upper gastrointestinal tract barium contrast studies in the upright position. It may present with non-specific gastrointestinal symptoms such as abdominal pain, vomiting, dyspepsia, anorexia, nausea, bloating. After meal activity may induce symptoms with positional changes being a common alleviating factor. Gastroptosis is thought to be due to the decrease of the tone of abdominal musculature as well as relaxation of the stomach from its mesenteric attachments and ligaments.

Gastroptosis is associated with delayed gastric emptying. It is unknown if the gastric dysmotility is a result of this condition or if gastroptosis and its symptomatology are due to an alternate etiology.
Treatment may include physical therapy for abdominal musculature strengthening exercises, abdominal binding, prokinetic medicines. Previously surgery was considered a first line approach but now that is reserved for cases with refractory symptoms.

Particularly in pediatrics there is a lack of research on this condition. This highlights the need to raise awareness for this rare finding and to inquire about alleviating factors such as positional changes when investigating symptoms that are common presenting complaints to a pediatric gastroenterology clinic.

We present two cases of children who presented to our tertiary children's hospital with similar symptoms who were diagnosed with gastroptosis on upper gastrointestinal tract studies.

Our first patient was a 13-year-old male who presented to our clinic with chronic intermittent periumbilical abdominal pain. Due to the intermittent nature and that he had dropped weight percentiles an upper gastrointestinal series was performed revealing that the greater curvature of his stomach was extended into the pelvis to the level of mid sacrum on standing. He had relatively normal positioning of his stomach in supine position. He was treated with physical therapy with noted improvement in symptoms.

Our second patient was a 11-year-old female who presented to our clinic also with chronic intermittent periumbilical pain but associated with vasovagal syncope and pallor. An upper gastrointestinal series was obtained showing in the upright position the greater curvature of the stomach projected below the level of the iliac crests. She had delayed gastric emptying in the upright position as well. While supine her stomach ascended to the normal position and displayed normal gastric emptying. She was also referred to physical therapy and was lost to follow-up.

As chronic abdominal pain and dyspeptic symptoms are common presenting complaints to a pediatric gastroenterology clinic these cases highlight the importance of the inquiry of positional alleviating factors and obtaining upper gastrointestinal barium studies when indicated.

Hypereosinophilic Syndrome vs. Eosinophilic Gastrointestinal Disease in a Pediatric Patient: A Diagnostic Conundrum

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Presenting Author: Meaghan Bank, MD
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Abstract:

Introduction: Eosinophilic gastrointestinal disease (EGID) can present with a variety of clinical manifestations. In this case, we describe a pediatric patient who presented with protein-losing enteropathy and hypereosinophilia, ultimately found to have eosinophilic esophagitis and gastritis and NOTCH1 mutation.

Case Presentation: A 14-month-old female presented with 1 week of edema of her eyelids, abdomen and lower extremities. Symptoms were preceded by adenovirus and SARS-CoV-2 infection. After initial suboptimal response to systemic steroid treatment she underwent bone marrow biopsy which was notable for mild hypereosinophilia as well as genetic testing which was notable for NOTCH1 mutation, known to be implicated in about 25-30% of cases of idiopathic hypereosinophilic syndrome. Ultimately, her peripheral eosinophilia did resolve after 6 weeks of swallowed budesonide treatment.

Discussion: While this patient's acute presentation would be unusual for hypereosinophilic syndrome given the acute onset of symptoms and single organ system involvement, there was initial concern for HES based on her noted NOTCH1 mutation. However, in this case the patient's eosinophilia resolved with glucocorticoid treatment. While NOTCH1 mutations have been associated with idiopathic HES, they have not been described in association with eosinophilic gastrointestinal disease.

Conclusion: This case highlights an unusual presentation of eosinophilic gastrointestinal disease presenting as protein losing enteropathy in a patient with severe peripheral eosinophilia and NOTCH1 mutation.

Variable Clinical Phenotype of Takenouchi-Kosaki Syndrome

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Abstract:

CDC42 is a gene that encodes a Rho GTPase that regulates intracellular signaling and cytoskeletal reorganization. Heterozygous mutations in CDC42 lead to autosomal dominant Takenouchi-Kosaki syndrome (TKS), a rare congenital developmental disorder. We describe a patient with TKS with unique clinical manifestations. An 18-year-old female presented with recurrent viral respiratory infections, lymphedema, and hepatosplenomegaly. Past medical history included developmental delay, dysmorphic facial features, bilateral sensorineural hearing loss, bicornuate uterus, and cardiac defects. Initial labs were notable for anemia, macrothrombocytopenia, and lymphocytopenia. Bone marrow biopsy showed normocellular marrow. Chest imaging revealed reticulonodular interstitial markings with innumerable nodules, and subsequent lung biopsy was consistent with obliterative bronchiolitis. Extensive infectious evaluation was unremarkable. Immunologic evaluation was notable for T, B, and NK cell lymphopenia and poor polysaccharide vaccine response. Chromosomal microarray was normal. Primary Immunodeficiency panel revealed a heterozygous pathogenic variant c.203G>A (p.Arg68Gln) in CDC42 consistent with a diagnosis of TKS. Pathogenic variants in CDC42 can lead to TKS. The p.Arg68Gln mutation is classified as a group I mutation that affects CDC42 binding to signaling partners. We describe a case with unique clinical manifestations, including obliterative bronchiolitis and immunologic abnormalities. While there is no specific immunologic phenotype associated with this syndrome, previous cases have identified variable T-cell lymphopenia, B-cell lymphopenia, hypogammaglobulinemia, and poor T-cell independent vaccine response.

Live Birth of an Infant with Triploidy (69, XXY) and Normal Prenatal Genetic Testing

Authors: Amanda Barkemeyer, MD; Victoria Aguilar, MD; Jeffrey Surcouf, MD; Julie Gallois, MD

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Abstract:

Triploidy is an abnormal genetic condition in which an additional haploid set of chromosomes are present resulting in a 69-chromosome karyotype. It is a frequent chromosomal cause of pregnancy loss, and it is rare in live born infants with most dying shortly after birth. Common phenotypic findings include intrauterine growth restriction, hypotonia, craniofacial anomalies, syndactyly, malformation of the extremities, cardiac defects, and brain anomalies.

A 690-gram male was born to a 30-year-old G2P1 female via Cesarean section at 29 3/7 weeks gestation. The prenatal history was concerning for decreased fetal movement, uteroplacental insufficiency, subchorionic hematoma at 8 weeks with recurrent bleeding, maternal hypertension, and intrauterine growth restriction with normal cell free DNA. Apgar scores were 1, 5, and 7 at one, five, and ten minutes. After birth, the infant required intubation, conventional ventilation, and surfactant for respiratory distress syndrome. The infant had multiple congenital anomalies including cleft palate, micrognathia, low hairline, syndactyly of 3rd and 4th digits of left hand, abnormal left thumb, webbed third and fourth digits of right hand, left genu varum, small great toes, and hypotonia. Initial echocardiogram demonstrated significant pulmonary hypertension, tricuspid regurgitation, small atrial septal defect. Due to worsening pulmonary hypertension associated with hypotension and right ventricular enlargement, the infant was treated with inhaled nitric oxide, epinephrine, and milrinone and was transferred to our NICU on day of life 15. Additional problems at the birth hospital included apnea, hypocalcemia, thrombocytopenia and anemia.

The infant was gradually able to wean off epinephrine, milrinone, and nitric oxide, but remained intubated due to respiratory failure. He tolerated slow advancement of enteral feeds. Additional problems uncovered included direct hyperbilirubinemia and bilateral anomalous optic discs. Rapid whole genome sequencing demonstrated triploidy (69, XXY). After conversations with family, limitations in resuscitative care were placed, and the infant died on day of life 51.

While prenatal genetic testing can be valuable, it is important to consider test limitations. If a genetic test analyzes the relative proportions of genetic material across all chromosomes, then certain genetic conditions such as triploidy may go unrecognized because triploidy (69, XXY) can appear similar to a typical chromosomal complement (46, XY). Triploidy is a diagnosis which should be considered in an infant born with multiple congenital anomalies despite normal noninvasive prenatal testing. Although most cases of triploidy do not result in live births, this unique case contributes to the existing literature on the clinical course of a surviving infant.

A Unique Case of Chronic Cough in a Toddler

Author: Emma Domangue; Joseph Sansone, MD; Kathryn Wiggins, MD; Hannah Sibley, MD; Amy Prudhomme, DO; Shubho Sarkar, MD

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Abstract:

Feeding difficulties and cough are common but nonspecific symptoms in pediatric patients. One potential cause is a tracheoesophageal fistula (TEF), a rare congenital anomaly that allows for the abnormal passage of food and liquids into the airway occurring in approximately 1 in 3500 births. The H-type TEF is the most rare type of congenital TEF occurring in 4% of cases of TEF. If not diagnosed or treated, TEF can lead to chronic respiratory infections and nutritional deficits among other complications. Recognizing TEF early is essential in children with recurrent aspiration and feeding issues, as delayed diagnosis can result in ongoing complications despite standard treatments for more common conditions.

A 13 month old girl with a history of premature birth at 34 weeks gestational age and reactive airway disease was admitted for feeding difficulties and persistent cough in the setting of a recent viral upper respiratory infection and concern for aspiration pneumonia. She had recently been admitted to an outside hospital for hypoxia in the setting of confirmed viral URI and concurrent pneumonia. She experienced coughing episodes with post-tussive emesis and poor PO intake requiring nasogastric tube feeds during her previous admission. She completed a course of antibiotics and was discharged, but re-presented for persistent fever, feeding difficulty, and worsening cough. Her symptoms were notable for progressively worsening lung sounds and inability to tolerate oral nutrition. Concern for aspiration with feeds prompted a swallow study which revealed aspiration across all consistencies during and after swallowing. A laryngoscopy and bronchoscopy were then performed which identified the cause of aspiration as a distal H-type tracheoesophageal fistula (TEF). While surgical planning was underway, her NG tube was converted to NJ due to persistent respiratory symptoms concerning for reflux through the TEF. She underwent a thoracoscopy converted to an open repair of the TEF. Her work-up for other VACTERL and CHARGE abnormalities was negative. After repair of her TEF, she was able to tolerate an oral diet without complication, and she was then discharged home with surgical follow up scheduled.

This case illustrates the importance of considering TEF in children with recurrent respiratory infections, chronic cough, and feeding difficulties. These symptoms, while commonly attributed to gastroesophageal reflux or other common pediatric ailments, may indicate an underlying TEF. Early diagnosis and surgical correction can alleviate chronic complications like recurrent infections and growth faltering. Clinicians should remain vigilant for TEF in cases of unresolved respiratory and gastrointestinal problems to ensure timely treatment and improved outcomes.

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Subcutaneous Fat Necrosis with Extensive Hematoma Following Therapeutic Hypothermia

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Abstract:

Subcutaneous fat necrosis (SCFN) is an uncommon but often self-limited panniculitis. In term neonates it can be associated with a perinatal stress event, particularly in neonates who undergo therapeutic hypothermia. There are several known complications of SCFN including, most commonly, hypercalcemia which can be severe as well as hypoglycemia, thrombocytopenia, anemia, and hypertriglyceridemia. We present the case of an infant with a rare complication of SCFN who developed a hematoma requiring surgical debridement and skin grafting.

A term male infant was born via emergent Cesarean Section due to non-reactive fetal heart tones. At delivery, the infant had a nuchal cord, body cord, and thick meconium. He required significant resuscitation with APGARS of 0/0/3/7. The infant met criteria for therapeutic hypothermia due to concern for hypoxic ischemic encephalopathy and was transferred to our Level IV NICU. His initial hospital course was consistent with severe multi-organ dysfunction given the fetal distress, ischemia, and acidosis.

On DOL 2, the infant was found to have erythematous indurated plaques to the back and posterior shoulders most consistent with SCFN. Therapeutic hypothermia was continued. In the following days, the erythematous skin evolved to a violaceous color and became more nodular. On DOL 9, there was an acute change with the development of large, fluctuant, violaceous nodules overlying the back and bilateral posterior shoulders with a large central mass measuring ~15cm x 15cm. This was associated with a concomitant drop in hemoglobin and platelets requiring multiple blood products. An ultrasound of the mass was obtained showing extensive subcutaneous fat edema with scattered fluid. On DOL 10, following correction of coagulopathy and, in collaboration with dermatology, general surgery incised the large central nodule which evacuated 250 mL of serous fluid and blood. Pathology was significant for SCFN with hematoma. The infant was followed closely by plastic surgery to determine the need for soft tissue reconstruction. On DOL 13, the patient underwent debridement of the non-viable tissue with placement of a wound vac measuring 8cm x 9cm. Over the next month, the infant underwent six wound vac exchanges with placement of dermal regeneration template on DOL 16. A split thickness skin graft was performed on DOL 34. The wound vac was ultimately removed prior to discharge on DOL 45. He has since followed up in dermatology and plastic surgery clinics and, by 7 weeks of age, the skin graft was noted to be well healing and closed. Additionally, due to the extensive SCFN he required continued surveillance of calcium levels until three months of age which all remained normal.

This unique case of SCFN complicated by the development of a significant hematoma contributes to a small collection of existing literature on this rare complication of SCFN. Secondary hematomas require multidisciplinary care and may need intensive, repeated surgical intervention. Therapeutic hypothermia is critical in the management of term infants (>36 weeks) status post a significant HIE event to optimize long term neurodevelopmental outcomes. Awareness of rare sequelae is important to the comprehensive care of these patients.

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Acute Onset of Internuclear Ophthalmoplegia in a 12-Year-Old with Acute Lymphoblastic Leukemia

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Abstract:

Background: Internuclear ophthalmoplegia (INO) is a discrete, localizing neuro-ophthalmic phenomenon that rarely occurs in childhood. It is therefore more difficult to identify etiologies of INO in the pediatric population; however, it is not a known common side effect of pediatric acute lymphoblastic leukemia (ALL) therapy.

Objectives: To describe the clinical and radiologic findings of INO in a patient receiving chemotherapy for pre-B high-risk ALL.

Design/Method: Case Report

Results: A previously healthy twelve-year-old female first presented with nausea, vomiting, and massive hepatosplenomegaly. Workup revealed a diagnosis of HR CNS1 pre-B ALL, and she was started on standard 4-drug Induction therapy. Of note, she was found to have hypogammaglobulinemia prior to starting therapy, and she received her first IVIG infusion on Induction Day 6. Her Induction course was complicated by tumor lysis syndrome, pancreatitis, and hyperinsulinemia; but her treatment was not interrupted. Unfortunately, she had an M2 Day 29 marrow and started Consolidation therapy prior to count recovery. Shortly after starting Consolidation, she began having staring spells. A brain MRI displayed T2 and T1 hyperintensity with diffusion restriction at the cortex of her bilateral parietal lobes without enhancement. These findings were consistent with a subacute ischemic infarct with cortical laminar necrosis. An EEG was negative for seizure activity. No evidence of thrombosis was seen on imaging. A repeat MRI a week later for blurry vision showed no change. Approximately three weeks from the initial symptoms, she began to exhibit abnormal eye movements which ophthalmology and neurology diagnosed as internuclear ophthalmoplegia. She had received intrathecal methotrexate three days prior. Clinically, she had limitations in eye adduction and upward gaze bilaterally. REPE notably had superficial enhancement of the meninges and prominent superficial cortical vessels. Workup, including for rheumatologic etiologies, remained negative. She received an IVIG infusion five days later, and her symptoms seemed to resolve in the next 36-48 hours without any recurrence. A definitive etiology for her symptoms was not established.

Conclusion: The scarcity of INO cases described in the pediatric population presents a challenge to diagnose and manage patients consistently. Current treatment guidelines recommend simply addressing the underlying etiology which can include ischemia, infection, or a brainstem tumor. There is only one case report in the literature which attributes INO to intrathecal methotrexate. Among the differential for causes in our patient were ischemia, chemotherapy adverse effect, and vasculitis. The timing of resolution after receiving IVIG also makes it peculiar. Since that time, we have kept the IgG level above 500mg/dL and have also given further doses of intrathecal methotrexate; and it has not recurred. However, this does not prove causation. This case report provides evidence for the variability of patients presenting with INO and raises awareness for its detection as well as expands the pool of potential adverse events associated with ALL therapy.

Changing the View of Pain: Using Virtual Reality as Adjunctive Therapy for Sickle Cell Pain in Pediatric Patients

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Abstract:

Introduction: A vaso-occlusive pain crisis is the most common presentation requiring inpatient admission for patients with sickle cell disease. As seen in many fields of medicine, advancements in technology have provided opportunities to introduce new therapies and procedures as complements to the current standard practices with the intent of improving patient care. For this proposed study, we will assess whether virtual reality (VR) can be an effective complementary therapy to treat vaso-occlusive pain in pediatric patients with sickle cell disease.

Methods: A randomized, controlled trial was designed to elicit the best results for assessing virtual reality's ability to address sickle cell pain. Assenting children (7-17 y/o, n=5) and consenting adults (18-21 y/o,n=7) admitted to MFC for a vaso-occlusive pain crisis were randomly assigned as either control patients to receive standard therapy (IV fluids, NSAIDs, opiates) alone or as case patients to receive standard therapy plus a daily VR experience. No blinding was performed for this study. Initial target sample size is 36 patients. Each patient's participation lasted for a maximum of 3 hospital days. The primary outcome measure was the reported change in location, severity, and quality of pain at three scheduled time points on each hospital day. This measure was assessed by answers recorded on the Adolescent Pediatric Pain Tool (APPT). Case patients were asked to fill out two additional surveys to assess for adverse events and to provide feedback about their experience of using virtual reality.

Results: Thus far, twelve patients (n=7 controls and n=5 cases) have completed the study to some extent. All collected data was used for patients who either were removed early from the study or discharged prior to completion of 3 days. There is no statistical data at this time given the low sample size in each study group. However, there is one notable trend of all 5 case patients having at least one recorded reduction in pain severity following a VR session. To analyze the primary outcome measure, pain scores and qualitative assessments from the APPTs will be first calculated as medians with interquartile ranges. These will be analyzed using paired t-tests to compare differences between the means for each category (pain severity, number of affected body parts, and qualitative descriptions). P-values will then be generated to determine clinical significance. To ensure equality between case and control groups, block stratification based on age and gender will be utilized at the time of data analysis.

Conclusion: The current standard therapy for vaso-occlusive pain is heavily reliant on opioid therapy, which is why efforts have been made to find other complementary therapies with the intention of reducing this need. By conducting this study, the goal is to provide evidence that virtual reality can effectively reduce vaso-occlusive pain and serve a complementary role in the multi-disciplinary approach to treat pediatric patients with sickle cell disease.

Eosinophilia, Biomarkers, and Atopic Dermatitis: A Retrospective Review

Authors: Sarah Campbell, MD; Hope Retif, MD; Nicholas Culotta, MD; McKenzie Burian; Alexandra Streifel, MD; Devyn Rohlfs Rivera, MD; Michelle Korah-Sedgwick, MD; Christopher Haas, MD

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Abstract:

Background/ Aims: Atopic dermatitis is a chronic inflammatory skin disease hypothesized to be due to skin barrier dysfunction and immune dysregulation. The goal of this study is to examine the relationship between factors such as atopic conditions, environmental allergies, and response to treatment and atopic dermatitis. We aim to investigate the utility of detectable surrogate markers for clinical response and disease course.

Methods: This was a retrospective chart review of pediatric and adult allergy & immunology and dermatology clinic patients at large tertiary care centers between 2020 and 2024. Data including allergen sensitization, presence of other atopic conditions, use of topical treatments and/ or immunosuppressive therapies, eosinophil level and percentage, IgE level, and response to treatment was collected.

Results: Of the 120 subjects analyzed, the average age was 16 years old with 93% of subjects diagnosed in childhood or infancy. Only 37% had documented dust mite allergy and 32% with grass allergy. 61% had another documented atopic condition. 58% had a recent eosinophil count (mean= $557x10^3$ uL), and 30% had an IgE level (mean= 2,338 kU/L). Many subjects did not have follow up regarding treatments.

Conclusions: No relationship could be found between the above factors and control of atopic dermatitis. This finding fits with previous studies, although limited by lack of regular laboratory testing. Potential biomarkers including total IgE and specific allergen testing were not found in most subjects. This could be noted as an area that could be improved upon along with overall better characterization of our patient population.

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Development of a Tracheostomy workshop for Pediatric Hospital Medicine Wards

Authors: Kayla Grise, MD; Leslie Reilly, MD; Amanda Messer, MD; Caroline Roth, MD

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Abstract:

Introduction: Tracheostomy is one of the most common pediatric lifesaving procedures. These patients are at increased risk for complications including obstruction and tube displacement. Despite the increasing number of pediatric tracheostomy patients there is no standardized tracheostomy management education for trainees at our hospital. A needs assessment of 16 recently graduated residents found that majority had <3 experiences of bagging a patient through a tracheostomy tube (82%) or changing a tracheostomy tube (93%). 37% of residents reported less than 3 experiences suctioning a tracheostomy tube. All residents disagreed with the statement: "I am confident in my ability to train a family in management of common tracheostomy problems such as sterile suction, bagging a patient through the tracheostomy tube and changing a tracheostomy tube". As a result of the needs assessment, we created a tracheostomy workshop which addresses common complications and is provide to all residents during their pediatric hospital medicine rotation.

Objective(s):
1. Following a hands-on tracheostomy workshop residents will demonstrate improved skills with tracheostomy care including suctioning, bagging and changing a tracheostomy tube.
2. Following a hands-on tracheostomy workshop residents will report improved confidence in tracheostomy care and training families with tracheostomy management.

Methods: Developed a tracheostomy workshop with video instructions demonstrating how to identify parts of a tracheostomy tube, provide sterile suction through a tracheostomy tube, bag a patient who is trach dependent and change a tracheostomy tube. Residents underwent pre and post evaluation of the skills. Skills assessment sheets were made in conjunction with members of the safe trach team and evaluators underwent inter-rater reliability testing. Additionally, residents were surveyed on self confidence in the skills listed above both before and after the workshop.

Results: 45 residents have participated in the workshop, with 30 of those completing pre and post assessment. After completing the workshop all residents indicated an improved confidence in the ability to train a family in common tracheostomy management and address acute tracheostomy issues in a setting with limited healthcare resources. Residents also demonstrated improved skills in identifying parts of a tracheostomy tube, providing sterile suction, bagging and changing a tracheostomy tube.

Conclusion: A hands-on tracheostomy workshop is a valuable addition to pediatric hospital medicine wards. Following this workshop, residents are better equipped to address acute complications in a growing patient population.

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Evaluating the Phoenix Sepsis Score as a Predictor of In-Hospital Mortality in Pediatric Hematopoietic Cell Transplantation

Authors: Anna Simon, MD; Ira Shulka, MD; Kayla Phelps, MD, MPH; Lolie Yu, MD, MPH; Zachary LeBlanc, MD

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Abstract:

Background: Hematopoietic cell transplantation (HCT) is a potentially curative intervention with inherent risks. Pediatric HCT patients often require Pediatric Intensive Care Unit (PICU) admission for organ dysfunction, severe infection, and respiratory failure. Mortality rates reach 33%, especially when intubation and continuous renal replacement therapy (CRRT) are required. Within 100 days post-HCT, the primary cause of death is organ failure. Predicting critical care needs is difficult, and there are no validated tools for predicting pediatric HCT outcomes. The Phoenix Sepsis Score (PSS) is a novel criteria used to predict in-hospital mortality in pediatric patients with suspected infection by grading organ dysfunction within 24 hours of admission. According to Sanchez-Pinto (2024), a PSS ≥ 2 strongly predicts in-hospital mortality.

Objective: This study demonstrates how the PSS can predict in-hospital mortality for pediatric HCT patients requiring PICU admission.

Design/Method: We conducted a retrospective analysis of pediatric patients who received HCT at our tertiary care center from 2018 to 2024. Medical record data were analyzed using descriptive statistics. We evaluated intubation status, pressor support, and CRRT use for each PICU encounter. A PSS (0 -13) was assigned for each PICU admission using lab values, vital signs, and physical exam findings from the first 24 hours. Given redundancy in the PSS criteria, not all variables are needed for composite score calculations. We omitted PICU admissions for routine post-operative monitoring and brief admissions (<24 hours) resulting in floor step-down.

Results: Our center performed 112 HCTs on 86 patients. Our cohort included 54.6% male and 45.4% female patients, with 38.4% autologous and 60.7% allogeneic HCTs. Thirty patients required critical care for a total of 42 PICU admissions. The in-PICU mortality rate was 35.7% (n=15). Rates of PICU admissions were higher for allogeneic compared to autologous HCTs (32.4% vs 18.6%). Escalations of care were common, with nearly half of the PICU admissions requiring intubation (45.2%, n=19), similar rates of PICU admissions requiring pressor support (45.2%, n=19), and one in five PICU admissions requiring CRRT (19.1%, n=8). The average PSS was 3.2 (range 1-11). The mean PSS among PICU survivors was lower compared to non-survivors (2.04 vs 4.80). The odds of PICU death were 7.9 times higher in patients with PSS ≥ 2.

Conclusion: We found an in-PICU mortality rate (35.7%) comparable to the literature (33%). By assessing organ dysfunction and the risk of septic shock, the PSS could be a useful early predictor of in-hospital mortality for HCT patients admitted to the PICU.

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Association of Genetic Variants and Clinical Course in Pediatric Patients with Cardiomyopathy

Authors: Cyndee Jocson, MD; Marla Johnston, MSN, RN; Christine Spaeth, MS, LCGC; Zhide Fang, PhD; Tamara Bradford, MD

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Abstract:

Purpose: Pediatric cardiomyopathy (CM) has different causative genes as well as multiple mutations in each gene. Multi-gene testing has paved the way to better understanding the relationship between cardiomyopathy and genetic results. Recent literature in pediatric CM has demonstrated that greater genetic variant burden, including both pathogenic and variants of unknown significance (VUS), correlate with worse clinical outcomes.
We aim to correlate genetic variants with clinical course in pediatric patients with cardiomyopathy. We will describe the demographic profile, clinical characteristics, and genetic test results of cardiomyopathy patients. We will further explore the relationship between genetic test findings related to combination(s) of pathogenic variants and VUS.

Methods: Retrospective data will be collected from patients receiving treatment for pediatric heart failure at a single-center tertiary care pediatric facility. We will include patients who meet the following criteria: diagnosed with hypertrophic, dilated, or LV non-compaction cardiomyopathy; age less than 21 years at diagnosis; and have undergone multigene testing for cardiomyopathy. Patients will be assigned to one of the following categories of cardiomyopathy: hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), and non-compaction cardiomyopathy (NCM). Basic demographic information, length of illness, presenting symptoms, and family history of cardiomyopathy will be recorded. Diagnostic test results will be used to further define the severity of the CM.

Conclusion: We will describe the characteristics of our patient sample focusing on the need for advanced cardiac therapies (i.e. implantable cardioverter defibrillator (ICD), ventricular assist device (VAD), or surgery) as well as significant events including transplant evaluation or aborted sudden cardiac death. We plan to explore the potential relationship of genetic test results, including negative findings, pathogenic variant only, VUS only, or both pathogenic variants and VUS.

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Relationship of the Index of Severity for Eosinophilic Esophagitis and Esophageal Distensibility in Pediatric Eosinophilic Esophagitis

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Abstract:

Background: Eosinophilic esophagitis (EoE) is a chronic immune mediated disorder of the esophagus diagnosed and monitored via endoscopy with mucosal biopsies to assess for inflammation as well as fibrostenosis. The Index of Severity for Eosinophilic Esophagitis (I-SEE) is a metric used to standardize EoE disease components (symptoms, complications, inflammatory and fibrostenotic features) to grade disease severity.
Early fibrosis may not be detected on mucosal biopsies and Endoscopic Functional Luminal Imaging Probe (EndoFLIP) has been identified as a potential tool to identify inflammatory or fibrostenotic disease phenotype by measurement of a distensibility index (DI). Measurement of distensibility is important because it has been shown to correlate with clinical outcomes such as food impaction, need for esophageal dilation, and abnormal esophageal motility. There is a need to determine disease phenotype early in diagnosis to identify patients who may be at higher risk for EoE complications. It is unknown if EndoFLIP findings correlate with I-SEE scores.
Our rationale is that patients with high I-SEE scores should have reduced DI and EndoFLIP is a tool that could be used as a surrogate for disease activity. But if patients have low I-SEE scores and reduced DI this could be indicative of early fibrosis. Our hypothesis is that pediatric patients with EoE with higher distensibility index (DI) measured with EndoFLIP will have lower I-SEE scores.

Methods: Our primary objective is to assess for correlation between I-SEE scores and DI as well as EoE quality of life screeners. Our secondary objective is observing if patients who are being treated for EoE have an improvement in their DI across multiple assessments. This is a prospective, single center study of pediatric patients ages 5-18 with dysphagia or known EoE undergoing endoscopy and EndoFLIP. Descriptive statistics will be used to characterize the cohort's I-SEE scores, DI score, and QOL screeners.

Results: Six patients have been enrolled and had data collection with the expected enrollment of thirty to appropriately power the study. This is an ongoing research project with the aim of finishing data collection by the end of 2025.
Conclusions: This is a novel study as the comparison between the distensibility index measured with EndoFLIP and the Index of Severity of Eosinophilic Esophagitis has not previously been examined. Additional data will be obtained to fully assess the correlation between the DI and I-SEE as well as EoE quality of life screeners.

Enhancing Bedside Education: a Pilot Study in the NICU

Authors: Mary-Elizabeth Lago, MD; Amy Creel, MD; Jeffrey Surcouf, MD; Christy Mumphrey, MD

Presenting Author: Mary-Elizabeth Lago, MD
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Abstract:

Bedside teaching has declined over the last 40 years due to reasons such as: different learning styles, increased physician responsibilities, increased patient load, time constraints, technology, and multiple levels of learners. Most literature showing improved learner behavior and medical care associated with bedside rounds stems from 2000-2010s Internal Medicine studies, leaving a current Pediatric gap.
This study evaluates the effect of an educational intervention on Neonatal Intensive Care Unit (NICU) bedside teaching, with a secondary focus on eliciting the educational needs of trainees and faculty and addressing their confidence related to educating diverse groups of learners.

The frequency of faculty bedside teaching of students, residents and/or fellows during daily rounds was monitored January 2024-January 2025 in a level IV academic NICU. Bedside teaching criteria were defined based on literature then inter-rater reliability of data collectors was tested. A needs assessment was sent to trainees and faculty to determine preferred teaching styles and faculty confidence and experience educating multiple levels of learners. An assessment informed educational intervention occurred in the middle of the study period. The workshop style intervention focused on: setting up a learning environment, adult learning principles, NICU specific techniques, and the importance of feedback. Pre- and post-intervention results were compared using descriptive statistics.

Faculty survey results indicated a lack of training on educating medical learners (76%), but no lack of confidence in ability to address multiple levels of learners. Data showed learners experience bedside teaching during rounds 0 - 6 times per day (average 1.94), increasing post-intervention to 1-9 times per day (average 4.41), a statistically significant change (p=0.002). Eighty percent of education moments were targeted at medical students, decreased to seventy-two percent post-intervention. Stratified by experience level, faculty 5-10 yrs post-training have the highest pre-intervention (average 2.75), and less experienced faculty (<5 yrs post-training) have the lowest (average 1.63). Faculty >15yrs post-training had the largest change and highest post-intervention median of 5.98. Analysis of patient census during the study showed no correlation of frequency of educational moments to census.

This study demonstrated an education intervention can increase bedside educational moments for trainees. While frequency of educational moments was not influenced by census, it was influenced by experience of faculty. Learners and educators may benefit from further, larger studies of targeted bedside educational interventions and expansion to other Pediatric specialties.

Do Social Determinants of Health Impact a School's Preparedness to Prevent Sudden Cardiac Death?

Authors: Mike Charles, MD; Amelia Haydel, MD; Marla Johnston, RN, MSN; Zhide Fang, PhD; Thomas Kimball, MD; Kelly Gajewski, MD

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LSU Pediatric Cardiology

Abstract:

Introduction: Other than accidents, homicide/suicide and cancer, sudden cardiac death (SCD) is the leading cause of death in young people. Screening is imperfect for the prevention of SCD. However, school cardiac emergency response plans (CERP), prompt cardiopulmonary resuscitation (CPR) and use of automatic external defibrillator (AED) have been shown to increase survival. School preparedness remains variable. The Jump Start Your Heart Act formed through Act 234 of the 2023 Louisiana Regular Legislative Session mandates that all educational facilities possess an AED on their premises and must have a CERP. Social determinants of health are known to impact pediatric public health and may play a role in a school's ability to adhere to this new law.

Purpose: The objective of this study is to determine the association of SDH and high schools' ability to adhere to the Jump Start Your Heart Act in their emergency preparedness.

Method: Survey will be administered to nurses of 245 high schools (public and private) in23 parishes of Southeast Louisiana. Social Determinants of Health (SDoH) will be assessed using the address of the schools. The following indices will be utilized to measure SDoH: Social Vulnerability Index, Index of Relative Rurality, and The Childhood Opportunity Index. Association with social determinants of health will be evaluated

Conclusion: We will explore whether high schools in geographical areas with increased social vulnerability have decreased adherence to the Jump Start Your Heart Act, i.e. less preparation for prevention of sudden cardiac death.

Standardizing and Improving Neurodevelopmental Follow Up of Infants with Hypoxic Ischemic Encephalopathy in the NICU

Authors: Michael Evers, MD; Michelle Knecht, MD; Christy Mumphrey, MD; Raegan Gupta, MD; Julie Gallois, MD

Presenting Author: Michael Evers, MD
Fellow
LSU Neonatology

Abstract:

Hypoxic Ischemic Encephalopathy (HIE) is a potentially devastating type of brain injury that affects newborns due to brain oxygen deprivation in the peripartum period. Infants diagnosed with HIE are at increased risk for neurodevelopmental delays. Access to early screening and interventions, including outpatient therapies (physical, occupational, speech), Early Steps (a state-run early intervention service), audiology, ophthalmology, and NICU follow-up, are linked to improved neurodevelopmental outcomes.

To improve the care and outcomes for patients with HIE at a tertiary-care NICU, a multidisciplinary group including neonatologists, nurse practitioners, nurses, and family partners was formed. We aimed to improve referral rates and clinic attendance by 20% over a year period. Referral rates and appointment attendances were tracked for these infants including outpatient therapies, Early Steps, audiology, ophthalmology, and NICU follow-up clinic. Bi-weekly meetings reviewed progress and provided staff and families opportunities to discuss barriers and interventions. In December of 2023, an electronic medical record smart phrase was created to clearly communicate outpatient neurodevelopmental needs for infants with HIE. These recommendations were disseminated to providers and dedicated NICU case managers, who discussed and arranged outpatient appointments with families. Smart phrase utilization rates were tracked. Clinic names were also clarified on family patient portals.

54 infants with HIE were identified over a period of three years from January 2021- December 2024. Applying the above interventions, electronic smart phrase utilization rates improved from 0% to 71% resulting in improvement in outpatient referrals and clinic attendance exceeding our initial goal of 20%. Outpatient Therapy and Early Steps referral rates improved from 43% to 94% and 51% to 88% respectively. The percentage of children receiving outpatient therapy increased from 49% to 76%. Ophthalmology referral rates improved from 3% to 100% and clinic attendance improved from 5% to 56%. NICU follow-up clinic referral rates improved from 81% to 100% and clinic attendance improved from 57% to 78%. Audiology referral rates were maintained from 92% to 94% and there was an increase in audiology clinic attendance from 62% to 89%.

HIE is a complicated diagnosis with a wide range of outcomes. This QI initiative improved the rates of both referral and follow-up visits, which are critical interventions to improve the neurodevelopmental outcomes of infants with HIE.

Poster Presentations
Faculty/
Research Professionals

Velopharyngeal insufficiency: A novel feature in PPP1R12A-related disorder?

Authors: Gregory Fulton, MD; Maryam Ijaz, MS, LCGC; Regina Zambrano, MD

Presenting Author: Gregory Fulton, MD
Associate Professor
LSU Pediatrics

Abstract:

Background/Purpose: *PPP1R12A*-related urogenital and/or brain malformation syndrome (UBMS) usually presents with multiple congenital anomalies, most commonly involving the brain and/or urogenital systems. Of the 12 cases reported, none describe palatal anomalies or velopharyngeal insufficiency (VPI). The objective of this report is to add another clinical feature to this rare syndrome.

Methods/Description: Our patient is now a 12-year-old male who presented to the craniofacial team at 9-years-old for evaluation of VPI. On exam and nasal endoscopic evaluation of speech he had poor palatal movement, causing nasal escape of air on phonation. He did not have evidence of submucous or occult cleft palate. His history was significant for retractile testicles and mild hypospadias status post orchiopexy and circumcision as an infant, delayed initiation of speech until 4yo, ADHD, learning disability, dysarthria, VPI, and myopic astigmatism. Chromosomal microarray was normal. Whole exome sequencing reported a *de novo* pathogenic variant in *PP1R12A*: c.1409 C>G p.(S470*).
PPP1R12A-related disorder is characterized by variable genitourinary anomalies and brain anomalies, with developmental delay and/or intellectual disability. However, no brain involvement and normal intelligence have been reported in some cases.

Results: Our patient has a learning disability and ADHD, and brain MRI was unremarkable besides hypoplasia of the left transverse and sigmoid sinus. He has a normal external genitourinary exam for age, although he does have a history of retractile testicles and hypospadias. He underwent a Furlow palatoplasty for his VPI, with dramatic improvement of his hyper nasal speech, but continues to have significant changes in intelligibility due to dysarthria. He has a full neuropsychological evaluation pending to help determine his level of learning or intellectual disability. The pathogenic variant identified in the proband is novel and arose *de novo*; it is predicted to result in protein truncation or nonsense mediated decay. There are no clear genotype-phenotype correlations established in the reported cases, but relative to the reported cohort, his presentation is mild.

Conclusion: *PPP1R12A*-related disorders present with a variable phenotype most commonly affecting the brain and GU tract. We present a novel pathogenic variant in a patient with no major structural brain anomalies, minor GU anomalies (hypospadias) and VPI and palatal dysfunction. We aim to expand the clinical characterization of the rare disorder linked to pathogenic variants in *PPP1R12A,* and recommend speech pathology evaluation in affected patients.

A Case Report of ECMO for Persistent Air Leak Syndrome in a Neonate

Authors: Lisa Barbiero, MD; Amanda Barkemeyer, MD; Leah Nuss, MD; Timothy Pettitt, MD; Staci Olister, MD

Presenting Author: Lisa Barbiero, MD
Assistant Professor
Tulane Neonatology

Abstract:

Neonatal air leak is a problem encountered by neonates with a variety of underlying etiologies. Most cases of pneumothorax resolve with placement of pleural drains. In rare cases, the air leak can last longer than 5 days, signifying the development of persistent air leak (PAL) syndrome. This is a rare indication for neonatal ECMO. However, the ability to oxygenate and ventilate using ECMO support while avoiding high-pressure respiratory support makes air leak syndrome reversible.

We present the case of a term neonate who recovered from bronchopleural fistula and persistent air leak syndrome using ECLS. He was initially transferred to our Level IV NICU for ECMO evaluation due to hypoxic respiratory failure from meconium aspiration syndrome. He received medical management and did not initially require ECMO. On DOL 10 he developed a spontaneous right-sided pneumothorax treated with chest tube placement. On DOL 22, pleural air was noted on the left as well. Despite maximal medical management of the bilateral pneumothoraces including HFOV, sedation, prone positioning, neuromuscular blockade, and numerous well-placed chest tubes, he had persistent large air leaks. The infant subsequently developed acidosis and persistent hypoxia despite medical management of the air leak and associated pulmonary hypertension. To rest the lungs and to seal the ongoing air leak, the patient was placed on VA ECMO support on DOL 35. Pediatric pulmonology and pediatric cardiothoracic surgery agreed with this approach. An ultra protective lung rest strategy was utilized on ECMO with no bagging, minimal rest settings (CPAP +5 cm H20) and no suctioning. By ECMO day 10 the pneumothorax was radiographically resolved and did not recur with increasing lung recruitment strategies. He remained on ECMO for a total of 13 days. There was no recurrence of air leak post ECMO or post chest tube removal. The infant was extubated on DOL 97 and transitioned to room air one month later on DOL 129.

This report adds to the sparse literature on management of neonatal persistent air leak syndrome with ECMO. Minimizing ongoing ventilator induced lung injury is crucial to allow healing of the damaged underlying lung tissue and to mitigate ongoing damage. In neonates with persistent air leak syndrome, the use of ECMO to allow for adequate gas exchange with much lower ventilatory settings is feasible to facilitate self-healing. In this unusual case, we were able to demonstrate a permanent recovery and to seal the refractory air leak using very protective lung rest with the aid of extra corporeal life support.

Supporting Fellows' Scholarly Project Success: An intensive, focused, scholarship curriculum

Authors: Amanda Messer, MD; Amy Creel, MD; Colleen LeBlanc, MD

Presenting Author: Amanda Messer, MD
Associate Professor
LSU Pediatric Hospital Medicine

Abstract:

Introduction: Fellowship programs are charged with supporting fellows in meeting their scholarly activity requirement, yet specific guidance on practicably and effectively doing this is lacking. Fellow scholarship influences the likelihood of faculty scholarly pursuits, which is important given decreasing numbers of pediatric subspecialty investigators nationally. Exploration of methods to enhance fellows' experiences with scholarship is needed.

Objective: Our objective was to create a departmental based scholarship curriculum for all first-year pediatric fellows, aiming to increase fellows' confidence and knowledge of specific scholarly topics.

Methods: We developed a one-week curriculum with principles of the zone of proximal development theory. First-year pediatric fellows participated in morning group learning sessions, then afternoons of individualized activities. The week culminated with fellow presentations. Fellows' project mentors were included in select sessions. Pre and post intervention survey data were collected for knowledge and confidence. Evaluations (5-point Likert scale) and free text comments were also collected.

Results: All first-year pediatric fellows (n = 14) participated. There was 100% completion of pre and post intervention surveys and 97% completion of evaluation items. Median scores for confidence items significantly increased (15 items, all p values < 0.05). There was no statistically significant change in pre and post intervention knowledge scores (19 items, all p values > 0.05), though more participants answered correctly post intervention. Overall program rating was high (Mean 4.71, p < 0.0001). Participants rated 20 of 25 curriculum components as valuable or highly valuable (Mean > 4, p value < 0.05). Means for sessions on refining the question, biostatistics, hypothesis, methods, and survey design were lower (Mean 3.14 to 4). Qualitative themes were overwhelmingly positive.

Discussion/Conclusion: A dedicated scholarly inquiry curriculum can enhance fellows' confidence in completing their required scholarly project and be well received. Though there was not a statistically significant increase in knowledge, participants reported feeling an increase in knowledge. Notably high scores on the pre-intervention knowledge survey may have contributed to the inability to measure significant change. Study limitations include being a single center study and only having data to support short term changes. Future directions include tracking outcomes longitudinally including rates of fellow scholarly project dissemination at national conferences and through peer reviewed publications.

Industrial Toxis Air Pollution and Congenital Heart Defects in Louisiana: A Case-Control Study

Authors: Kimberly Terrell, PhD; Marla Johnston, RN; Ketaki Mukhopadhyay, MD; Taylor Katt, MD; Gianna St. Julien; Diego Lara, MD, MPH; Kelly Gajewski, MD

Presenting Author: Marla Johnston, RN
RN Clinical Trials Coordinator
LSU Pediatric Cardiology

Abstract:

Purpose: investigate the relationship between industrial air pollution exposure and congenital heart disease incidence among Louisiana births.

Methods: Case-control study of 5198 Louisiana births from 2018-2021 (916 cases and 4282 matched controls, excluding maternal smoking/alcohol/drug use), using Environmental Protection Agency RSEI-Microdata. We evaluated associations using logistic regressions conditioned on matched variables (race, birth year, and region) with relevant covariates (e.g. diabetes status) and sensitivity analysis to address potential confounding.

Results: Risk of CHD overall increased with ethylene oxide thresholds in a dose-response manner. The highest odds ratio (OR) was observed in the highest ethylene oxide quartile (≥6.0 ng/m^3; adjusted OR = 3.65; 95% CI: 1.75, 7.57). when divided into CHD subtypes, ethylene oxide concentrations ≥2.0ng/m^3 were associated with an increased risk of septal defects (adjusted OR = 3.13, 95% CI: 1/41, 6.51). Associations with other defect subtypes were non-significant, though statistical power was limited.

Conclusions: The results indicate that prenatal exposures to ethylene oxide at current environmental concentrations are associated with increased congenital heart disease risk.

Comparison of Human-and ChatGPT-Generated Feedback

Authors: Bonnie Desselle, MD; Emma Simon, MBA; Amy Prudhomme, DO; Shubho Sarkar, MD; Amy Creel, MD

Presenting Author: Bonnie Desselle, MD
Professor
LSU Pediatrics

Abstract:

Faculty feedback is vital for the professional growth of students, residents, and fellows. Resources, including published expert-authored scenarios with feedback scripts, can be referenced but may not be easily accessible to faculty. Generative large language models have the potential to offer just-in-time support to faculty, tailored to specific trainee scenarios. The purpose of this study was to compare the quality of ChatGPT-generated feedback scripts with expert-authored scripts based on standardized scenarios of learner deficiencies. Six faculty were asked to blindly judge two narrative scripts for each of the eighteen learner deficiency scenarios. The learner deficiency scenarios were entered into ChatGPT-4 with an associated prompt asking to create a verbal feedback script. Published feedback scripts served as the expert-authored script. The faculty rated each script in four domains using a 3-point Likert scale (0-2), yielding a total minimum score of 0 and a maximum score of 8. The mean scores of the expert-authored and ChatGPT-generated scripts were compared. The faculty were also asked to select which response script they preferred for each of the case scenarios. Faculty rated ChatGPT scripts higher than expert-authored scripts (mean = 7.55, 6.23; $p<0.05$). ChatGPT scripts were preferred in 84 of the 108 ratings (78%). ChatGPT-generated feedback scripts can be of high quality. Large language models may be a useful, just-in-time tool for faculty preparing to give verbal feedback or seeking to improve their feedback skills.

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A Preliminary Study of Glycemic Outcome During Real World Use of Automated Insulin Delivery Systems (AID) in a High Risk Population of Youth with Type 1 Diabetes (T1D): Implications for Changes in Outpatient Management

Authors: Nicholas Christakis; Brantlee McConaughey, MD; Patrick Quebedeaux, MD; Ricardo Gomez, MD; Dania Felipe, MD; Arlette Soros, MD; Alan Delamater, PhD; Stuart Chalew, MD

Presenting Author: Stuart Chalew, MD
Professor
LSU Pediatric Endocrinology

Abstract:

Data from our clinic at MFCH and other investigators around the US have shown that Non-Hispanic Black youth (NHB) with T1D have suboptimal glycemic control compared to Non-Hispanic White youth which eventually would lead to higher occurrence of diabetes complications. Potentially this disparity in glycemic control is due to less access by NHB to advanced technologies for insulin delivery. Pivotal trials of AID systems have shown overall improvement in glycemic control for users. Potentially use of AID would lead to improved glycemic outcome for NHB patients.

To test this hypothesis we studied the impact of real world AID usage on glycemic control in NHB patients. Patients from MFCH diabetes clinic were included if they self-identified as NHB, had chosen to use AID, had had T1D longer than 6 months, had no other chronic medical or psychologic conditions besides T1D, had used A1D for longer than 90 days, had A1c data available before and during A1D use. Patients were using AID devices manufactured by Tandem (Control IQ), Omnipod 5, or Medtronic 670G AHCL. Instruction in use of the AID system was provided to the patient/family by a specially trained health care professional or in the case of Omnipod5 could also be done through an internet program. Medtronic users initially had available frequent contact with a designated CDCES nurse educator for the first month of use but otherwise patients had standard outpatient follow up.

The table below summarizes results for each of the AID by manufacturer and overall.

	n	M/F	Age (Y)	Use (d)	A1c Pre	A1c Last	diff A1c	p
Tand	8	5/3	6.1±2.8	312 ±147	9.0±1.2	8.9±1.2	-0.09±1.6	0.88
Omni	10	8/2	12.0±3.6	325±153	9.6±1.7	9.6±2.3	0.2±1.1	0.95
Medt	6	2/4	15.1±3.3	177±6.8	8.8±0.82	8.9±1.3	0.16±1.5	0.82
All	24	15/8	14.6±3.3	288±141	9.2±1.4	9.2±1.9	0.01±1.3	0.96

Patients with a decrease in A1c ≥0.5 during AID use were considered Responders, n=9 (38%), and had an average drop in A1c of 1.2±0,7 and last updated A1c of 8.0±0.8 compared to A1c of 9.2±1.1 before use. There were no differences between Responders and non-Responders with regard to pre AID A1c, age, sex or duration of AID use.

This preliminary study suggests that use of AID devices with routine clinic follow up did not lead to an overall improvement in A1c in a historically high risk population. However 38% of patients did have a clinically meaningful improvement in glycemic control. These results suggest that changes in the AID on-boarding process and follow up will be needed to improve glycemic outcomes needed to reduce and prevent diabetes complications in a majority of high risk patients. Further investigation into the differences between Responders and non-Responders may help guide innovations to improve glycemic outcomes in patients using AID.

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Does a Comprehensive Home Monitoring Program Mitigate the Negative Effects Related to Social Determinants of Health on the Outcomes of Babies with Complex Congenital Heart Disease?

Authors: Marla Johnston, RN; Stephanie Bush, NP; Lynn Bardales, RN; Zhide Fang, PhD; Thomas Kimball, MD

Presenting Author: Marla Johnston, RN
RN Clinical Trials Coordinator
LSU Pediatric Cardiology

Abstract:

Purpose: Complex congenital heart disease requires multiple stages of surgical palliation. The interstage period in the first 4 months of life and between the first and second surgical palliation, has the highest morbidity and mortality. Interstage weight gain is associated with improved clinical outcomes. We analyzed the relationship between social determinants of health [SDoH] and interstage weight gain in infants before and after the initiation of a comprehensive home monitoring program [HMP1 and HMP2].

Methods: Retrospective data collection on 32 patients [16 HMP1 and 16 HMP2] who were discharged home between the first (Time 1) and second (Time 2) surgical palliation. Interstage growth was measured by raw data and calculated data, Weight-for-age Z scores (WAZ). Social determinants of health were calculated based upon the address available in the electronic medical record. Social Vulnerability Index, Index of Relative Rurality, and United States Department of Agriculture Food Access Research Atlas were used to determine SDoH. Hospital readmission was collected.

Results: HMP2 infants were younger than HMP1 infants at both Time 1 and Time 2 (54.5 (IQR;30.5-67.5) vs 60.5 (IQR;47-88.5 at Time 1;) 151 (IQR;134-194) vs 170 (IQR;152-239 at Time 2). Weight for age Z [WAZ] scores at Time 1 for HMP1 patients was -2.23 and for HMP2 patients was -1.34. WAZ scores at Time 2 were significantly better in the HMP2 vs. HMP1 infants (-0.93 vs -1.73, $p<0.05$), indicating that HMP2 patients had significantly higher weight than HMP1 patients at the time of their second surgery. Readmission rates were significantly higher for HMP 2 patients 94% vs 56%, p=0.04. The change in WAZ from Time 1 to Time 2 was significantly and positively related to SVI in HMP2 patients [r = 0.4949, p=0.0513] but not in HMP1 patients indicating that in the HMP2 group, the more socially vulnerable patients tended to have better interstage weight gain.

Conclusions: Despite undergoing earlier stage 2 surgery, the HMP 2 infants had better age-adjusted weight at the time of their second stage. Because the HMP2 group had higher interstage admission rates, and their weight gain was positively associated with being more socially vulnerable, we speculate that the home monitoring program identified the more socially vulnerable infants and successfully mitigated the potentially adverse effects of their social vulnerability.

Interactive E-Learning Modules for Pediatric Board Prep

Author: Christy Mumphrey, MD; Brian Barkemeyer, MD; Julie Gallois, MD; Michelle Knecht, MD; Mary Johnson, MD; Chelsey Sandlin, MD

Presenting Author: Christy Mumphrey
Associate Professor
LSU Neonatology

Abstract:

Background: Finding time and being motivated to study during residency can be challenging. In our program, residents are provided with the MedStudy® Pediatrics Core books, but regular use is likely scarce during the first and second years. The objective of this study was to create a curriculum that allowed residents to review board-type material while on their neonatal intensive care unit (NICU) rotation and to evaluate if that translated into increased Pediatric In-Training Exam (ITE) scores for Neonatology subject content. Residents' feedback on the curriculum was also reviewed.

Methods: Interactive self-study modules were developed using the e-learning platform Articulate®. The curriculum was based on Neonatology board study content in MedStudy® for PGY2 residents rotating at a lower acuity NICU. Module completion was strongly encouraged. On-service faculty and/or fellows held weekly in-person reviews via Jeopardy®-style games. Individual PGY2 and PGY3 resident ITE scores were compared for the percentage of correct NICU content questions. Changes in scores for residents who completed the entire curriculum (Class of 2024) versus those who completed half of the curriculum (Class of 2023) or no curriculum (Class of 2021 & 2022) were compared. Residents were also surveyed on their end-of-the-year program evaluation for feedback on the curriculum.

Results: Variability was present in the percent change of correct NICU ITE questions from the years 2021-2024 with no definite trends identified. Median percent change per graduating class ranged from –7% to 14.5%. Program surveys revealed that 83% of participating residents felt the interactive curriculum had a positive impact on their NICU learning experience (n=24).

Discussion: Using an e-learning platform to teach multiple, short interactive lessons provided a different approach to resident board preparation. Weekly content review games reinforced important concepts via a flipped classroom model. Although changes in NICU specific ITE scores were not observed, the curriculum was overall viewed positively by the residents. Several limitations existed for this study. Inherent variability exists in ITE scores for each individual resident and between residency classes, which makes score comparisons difficult to interpret. Additionally, the low number of NICU content questions on the ITE may not accurately reflect the knowledge gained by the curriculum. While module development required a faculty time investment, these enduring teaching materials provide a sustainable and feasible resource for resident board preparation.

Impact of a Dedicated NICU Educational Attending on Resident Education and Faculty Satisfaction

Authors: Mary Johnson, MD; Karleigh Barkemeyer, MD; Christy Mumphrey, MD

Presenting Author: Karleigh Barkemeyer, MD
Chief Resident
LSU Pediatrics

Abstract:

Background and Study Objectives: Resident education in the NICU is crucial for developing clinical skills and confidence, but the high acuity and demanding clinical duties often limit the time available for structured teaching. This can result in dissatisfaction among trainees and frustration for providers who are eager to teach but struggle to find time amidst their busy schedules. Furthermore, the reduction in ICU requirements by the ACGME highlights the importance of clear and effective teaching during a trainee's limited time in the ICU setting. Two years ago, feedback from residents revealed a need for more structured teaching during their NICU rotation. In response, a dedicated NICU educational attending rotation and curriculum was created in July 2022, facilitated by off-service NICU faculty leading didactic sessions and in-person simulations. This study aims to evaluate the impact of this rotation on resident education as well as faculty satisfaction.

Methods: This study used a survey-based method with a 5-point Likert scale to assess current perspectives on NICU education following the initiation of the NICU educational attending rotation. Surveys were anonymous and administered to current and former residents who experienced dedicated educational sessions during their NICU rotations as well as current faculty members involved in the rotation. Surveys were sent to 55 residents and 13 participating faculty members.

Results: Resident survey results (n=25, 45.5% response rate) revealed that the implementation of the educational attending had a positive impact on their overall rotation experience (96% of respondents) and current clinical practice and/or knowledge (96% of respondents). The residents viewed the implementation of the educational attending as a valuable learning experience (96% of respondents). Faculty survey results (n=13, 100% response rate) revealed a positive impact on their stress level (92% of respondents), ability to focus on bedside teaching (92% of respondents), overall view of the rotation (85% of respondents) and workflow (92% of respondents) during service time. Only 46% of faculty respondents perceived a positive impact on resident knowledge during rounds.

Conclusions: The implementation of a dedicated educational attending in the NICU positively impacted faculty and residents. Although this requires a time investment from faculty, the benefits for on-service faculty and residents provide strong justification to sustain the rotation. These positive outcomes may also support future expansion of the curriculum and rotation.