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6th Edition - World Congress on
PEDIATRICS & NEONATOLOGY
Sep 25 - 26, 2025 at Berlin, Germany

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6th Edition - World Congress on **PEDIATRICS & NEONATOLOGY** Sep 25 - 26, 2025 at Berlin, Germany

Scientific Program

Day 1- September 25, 2025

Meeting Hall-Conference 1

Registrations Open (08:00-08:45)

Welcome Ceremony (08:45- 09:00)

Moderator:

Jennifer Massa, *Harvard School of Public Health, USA*

Keynote Talks

Preben Christensen,
SPZ Lab A/S, Denmark

09:00-09:35

Is there a link between sperm DNA fragmentation and malformations in newborns as well as the development of diseases in infants, children, and adolescents?

Cody Arnold,
Stanford University, USA

09:35-10:05

Unbound Bilirubin and Risk of Severe Neurodevelopmental Impairment in Extremely Low Birthweight Newborns

**Chandra Sekhar
Devulapalli**,
Helgeland Hospital, Norway

10:05-10:35

Hidden Deficiency: Immune-Mediated Vitamin D Loss in Children with Chronic Inflammatory and Allergic Conditions

Group Photo & Refreshment Break (10:35-10:55)



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Time: 10:55-11:25

Title: The New Norm: A Safe Oral Feeding NICU Culture

Sandra Aubuchon, *President, Dr. Brown's Medical* and
Lisa Kleinz, *Director of Education, Dr. Brown's Medical*

Technical Session - I

Session Themes: Neonatology & Child Health
Pediatric Nutrition
Pediatric Metabolism Disorders

Session Chairs:

Cody Arnold, *Stanford University, USA*
Chandra Sekhar Devulapalli, *Helgeland Hospital, Norway*

Jorge Sales Marques,
*Department Head of
Center of Child and
Adolescent, Hospital Cuf
Porto, Portugal*

11:25-11:45

Screening for Pompe, Fabry,
Gaucher disorders using dried blood
spot: 3 years experience

Beatrice Stefanescu,
*Indiana University School
of Medicine, United States*

11:45-12:05

Assessing Early Screening for
Cortical Visual Impairment in the
NICU: Preliminary Findings from a
Pilot Study

Jennifer Massa,
*Harvard School of Public
Health, USA*

12:05-12:25

Improving Cooking Skills, Lifestyle
Behaviors, and Clinical Outcomes for
Adults at Risk for Cardiometabolic
Disease: Protocol for a Randomized
Teaching Kitchen Multisite Trial (TK-
MT)

Yara Zehnder,
*Swisstransplant,
Switzerland*

12:25-12:45

Initiative International organ
exchange: Life-saving for children on
the organ waiting list

Kiumars Pirkalani,
*Mehr medical group,
Iran*

12:45-13:05

Enzymatic approach to neonatal
hyperbilirubinemia

Lunch Break @ Restaurant 13:05-14:00

Technical Session - II

Session Themes: General Pediatrics
Neonatology & Child Health
Allergy, Infections & Immunology
Advanced in Neonatal intensive care

Session Chairs:

Jorge Sales Marques, *Department Head of Center of Child and Adolescent, Hospital Cuf
Porto, Portugal*
Keith Zarahi Amaral, *Summit Hospital, United States*

Melissa Beck Wells,
*SUNY Empire State
University, USA*

14:00-14:20

Navigating the Intersection of
Giftedness and ADHD: Addressing
Misdiagnosis and Supporting Twice-
Exceptional Learners

Keith Zarahi Amaral,
*Summit Hospital, United
States*

14:20-14:40

Acute Liver Failure: Is
Acetaminophen the Only Culprit?

Bernadett Kertész
*Semmelweis University,
Hungary*

14:40-15:00

Approaching feeding difficulties in
infants and preterm newborns from
the perspective of physiotherapy

Erika Isolauri,
University of Turku, Finland

15:00-15:20

The Disappearing Bifidobacterium
Microbiota in Infants Over the Past 25
Years

Andleeb Badar,
RCPI, Ireland

15:20-15:40

Case Report: PHP Type 1b in a
14-Year-Old Boy with Hypocalcemia
and Spasms After Flucloxacillin Intake

Refreshment Break (15:40 - 15:55)

Fei-Yan Deng, <i>Suzhou Medical College of Soochow University, China</i>	15:55-16:15	Birth weight is positively associated with adult osteoporosis risk: observational and Mendelian randomization studies
Zahra Talaeeerad, <i>Mehr medical group, Iran</i>	16:15-16:35	Pharmacological approach to neonatal hyperbilirubinemia
Rajai Rofail Raja Al-Bedaywi, <i>Hamad Medical Corporation, Qatar</i>	16:35-16:55	Glucose-6-phosphate dehydrogenase deficiency and neonatal indirect hyperbilirubinemia: a retrospective cohort study among 40,305 consecutively born babies
Sanna Hellström Schmidt, <i>Lund University, Sweden</i>	16:55-17:15	Overuse of EEG and ECG in children with breath-holding spells and its implication for the management of the spells
Aftab Yusuf Raj, <i>Square Hospital, Dhaka, Bangladesh</i>	17:15-17:35	Role of human milk oligosaccharides in immuno regulation and epigenetic modulation of pathogenetic bacteria on infant and toddlers health

Pannel Discussion & Day 1 Concludes

Day 2- September 26, 2025

Meeting Hall-Conference 1

Moderator:

Jennifer Massa, *Harvard School of Public Health, USA*

KEYNOTE FORUM

Per Wierup,
*Cleveland Clinic
Foundation and Skane
University Hospital, USA*

10:00-10:30

Mitral valve repair using leaflet expansion and subpartial annuloplasty in children

Scientific Session-III

Session Themes: General Pediatrics
Pediatric Pulmonology
Pediatric Oncology/ Hematology
Advances in Neonatal Intensive Care

Session Chair:

Per Wierup, *Cleveland Clinic Foundation and Skane University Hospital, USA*
Sean Gamble, *Stanford University School of Medicine, USA*

Sean Gamble,
*Stanford University School
of Medicine, USA*

10:30-10:40

Building coalitions for strategic interventions to improve adolescent health in the technology age

**Shanthosh Raj Srinivasan
& Geethanjali**,
*ESIC medical college and
hospital, India*

10:40-11:00

Comparative Evaluation Of Erosion Level Of Commonly Prescribed Pediatric Liquid Medicines On Primary Teeth : An Invitro Study

Beatrice Stefanescu,
*Indiana University School
of Medicine, United States*

11:00-11:20

Contemporary Genetic Testing Patterns in Neonatal Seizures

Refreshment Break @ (11:20-11:40)

Elise Margaretha Adriana Slob,
University Medical Center Groningen, The Netherlands

11:40-12:00

ADRB2 genotype-guided treatment for childhood asthma: an individual patient data meta-analysis of the PACT and PUFFIN trial

Kiumars Pirkalani,
Mehr medical group, Iran

12:00-12:20

Treatment of 1182 early type I DM with a combination of 3 immune modulatory drugs Eight years of experience

Pravana Naicker,
Whittington Health Trust, UK

12:20-12:40

Hereditary Pyropoikilocytosis: A rare cause of neonatal jaundice

Hannia Bertha Matt Hernandez,
Christus Muguerza Reynosa Hospital, Mexico

12:40-13:00

Immunological tolerance acquisition after dietary elimination in mexican infants with cow's milk protein allergy mediated by ige

Richard Fry.
Dr Fry & Associates, UK

13:00-13:20

"ADHD / ASD and Neurodiversity". Is it time to move on?

Lunch Break @ Restaurant 13:20-14:20

Panel Discussions & End of the Day



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VIRTUAL SESSION

6th Edition - World Congress on
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Sep 25 - 26, 2025 at Berlin, Germany
(Hybrid Event)

Time Zone: Central European Time (Germany Time)



[Join Zoom Meeting](#)

Meeting ID: 864 9110 9463

Pass code: 530520



Day 1- September 25, 2025

Virtual Program (Central European Time)

Kavita Ratna,
India

14:30-14:50

Centering Children in Child
Safeguarding

Sandra S Stone,
*University of South Florida,
USA*

14:50-15:10

The Intersection of Child Sexual
Abuse and Sex Trafficking
Victimization

Ping-Hui Liu,
*Cincinnati Children's
Hospital, USA*

15:10-15:30

Secretion Movement During
Mechanical Ventilation Superimposed
on Intrapulmonary Percussive
Ventilation (IPV) Versus Continuous
High-Frequency Oscillation (CHFO): A
Bench Study

**Maria Belen Ocampo
Ordóñez,**
*Universidad San Francisco
de Quito, Ecuador*

15:30-15:50

Clinical and surgical physician's
perception of nutrition knowledge

Sandra Fucile,
*Queen's University,
Canada*

15:50-16:10

Current Oral Feeding Management
Approaches in NICUs: Results from a
National Survey

Katie Carlton,
*Medical College of
Wisconsin, USA*

16:10-16:30

Machine learning risk stratification for
high-risk infant follow-up clinic

Ana Fresán Orellana,
*Instituto Nacional de
Psiquiatría Ramón de la
Fuente Muñiz, Mexico*

16:30-16:50

Mistakes are not an option: correlates
of anxiety and depression in
pediatricians in training

Sanjay Vepa,
Kaiser Permanente, USA

16:50-17:10

Prenatal detection rates for
congenital heart disease using
abnormal obstetrical screening
ultrasound alone as indication for fetal
echocardiography

Stanley P. Galant,
*University of California
Irvine, USA*

17:10-17:30

Adding oscillometry and fractional exhaled nitric oxide to guidelines potentially enhances identification of exacerbation risk factors in asthmatic children

Yuxuan Liu,
Stanford University, USA

17:30-17:50

Uridine Metabolism as a Targetable Metabolic Achilles' Heel for chemo-resistant B-ALL

Day-1 Virtual Concludes

Day 2- September 26, 2025

Virtual Program (Central European Time)

Guanghui Qian,
*Children's Hospital of
Soochow University, China*

10:30-10:50

Involvement of USP7 in aggravating Kawasaki disease by promoting TGF β 2 signaling mediated endothelial-mesenchymal transition and coronary artery remodeling

M.G.T.W. Premadasa,
National Hospital, Srilanka

10:50-11:10

Molybdenum Cofactor Deficiency with A Novel Pathogenic Variant in the Molybdenum Cofactor Synthesis 1 (MOCS1): A Case Report

**Amira Elserwy Aly
Raslan,**
*Dallah Hospitals, Saudi
Arabia*

11:10-11:30

Magnetic Resonance Imaging (MRI) Findings in Pediatric Autoimmune Encephalitis: A Case Report

Dubravka Bačaj Ivanić,
*Clinical Hospital
"Sveti Duh", Zagreb,
Croatia*

11:30-11:50

Unusual case of Plesiomonas shigelloides sepsis and meningitis in a neonate with targetoid vasculitic rash

Bala Munipalli,
Mayo Clinic, USA

11:50-12:10

Long COVID and Hypermobility have shared pathophysiology

Inger Kjær, <i>University of Copenhagen, Denmark</i>	12:10-12:30	Location in early childhood of the permanent mandibular canine explain the "mysterious mandibular permanent, canine" observed in puberty
Mustafa Alhamed, <i>University of Gezira, Sudan</i>	12:30-12:50	The Role of Faculty of Medicine University of Gezira Students in improving maternal and child health among Families through primary health care centers practice and family medicine course at Wad Medani City, Gezira State, Sudan, 2023
Sleh Eddine Saâdi, <i>Mohamed Taher Maamouri Hospital, Tunisia</i>	12:50-13:10	Executive functions and their influence on academic performance in school-aged children in Tunisia
John Patrick T Co, <i>Mass General Brigham, USA</i>	13:10-13:30	Promotion in Place: A Model for Competency-Based Time-Variable Graduate Medical Education
Berrahal Mounir, <i>Algeria</i>	13:30-13:50	Metagenomic approach to renal failure in children
Shuai Tan, <i>Xuanwu Hospital, Capital Medical University (XWH- CMU), China</i>	13:50-14:20	New immune targets derived from platelets: PF4 & TGF- β ?
Anne Kathinka Aslaksen, <i>Haukeland University Hospital, Norway</i>	14:20-14:40	Children had increased risks of impaired motor and visual-motor skills after prenatal exposure to opioid maintenance therapy
Sylvain Chemtob, <i>Université de Montréal, Canada</i>	14:40-15:00	Innovative modulators of IL-1 and IL-6 receptors to prevent inflammation-induced preterm birth and improve newborn outcomes

Amanda Winkler, <i>Massachusetts General Hospital, USA</i>	15:00-15:20	Prevention of NICU Admission Hypothermia in Moderate- and Late-Preterm Infants
Rajindra Prasad Aryal, <i>BIDMC, Harvard Medical School, USA</i>	15:20-15:40	Impact of Pathogenic C1GALT1C1 Variants on Extended O-Glycan Functions
Christina Murphy, <i>Children's Hospital of Philadelphia, USA</i>	15:40-16:00	The relationship between headache, mood, and functional disability
Christie Del Castillo-Hegy, <i>Fed Is Best Foundation, USA</i>	16:00-16:40	A Hidden Epidemic: Complications of Insufficient Feeding in Term Breastfed Neonates
Bernard A Cohen, <i>Johns Hopkins School of Medicine, USA</i>	16:40-17:00	Keynote: Tinea in Tots
Michael H. Malloy, <i>University of Texas Medical Branch at Galveston, USA</i>	17:00-17:20	Keynote: Ethical Dilemmas on the Limits of Viability of Perivable Fetuses
Miles Weinberger, <i>Volunteer Clinical Professor University of California San Diego, USA</i>	17:20-17:40	Keynote: Nonpharmacologic Cessation of Refractory Chronic Cough in Adults
Howard Saft, <i>National Jewish Health, USA</i>	17:40-18:00	The pulse oximeter as a source for racial disparities, how we got here, updates from the ISO and path towards never again
Eugene Arnold L, <i>Nisonger Center Clinical Trials Program, USA</i>	18:00-18:30	Keynote: Problems of most concern to parents of children with ADHD and emotional dysregulation in a randomized controlled trial of multinutrients: MADDY secondary outcome

End of the Day

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BOOK OF ABSTRACTS

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KEYNOTE PRESENTATIONS DAY-1

6th Edition - World Congress on
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Is there a link between sperm DNA fragmentation and malformations in newborns as well as the development of diseases in infants, children, and adolescents?

Preben Christensen*

SPZ Lab A/S, Fruebjergvej 3, DK-2100 Copenhagen OE, Denmark



After 1 year of unprotected intercourse, 15% of couples are unable to conceive a child. Reduced fertility in the men is behind at least half of these cases and sperm DNA damage (fragmentation) is a frequent cause. Male health at the time of conception or sperm DNA damage has been directly linked to miscarriage, stillbirth, preterm birth, and low birth weight.

It has been known for some time that de novo point mutations and structural rearrangements have a preferential paternal origin. When sperm DNA damage is single stranded, it may be repaired by the oocyte in the brief window that exists between fertilization and the S phase of the first mitotic division. However, this repair may be incorrect and result in a de-novo mutation. It is unlikely that double-stranded breaks (fragmentation) in the sperm DNA will be repaired correctly by the oocyte, and this type of damage may therefore result in chromosomal aberrations.

Male IVF patients frequently carry high levels of sperm DNA damage, which increases with male age. In addition, the oocyte's ability to repair DNA is compromised with increasing female age. When the level of sperm DNA damage is moderate (defined as a DFI value between 15 and 25), the fertility of the couple is reduced and more time is required to achieve pregnancy naturally.

Moderate or high levels of sperm DNA damage may result in miscarriage but may also result in de-novo genetic complications in the offspring such as rare diseases, e.g., Costello or Smith-Magenis Syndrome. However, since sperm DNA damage is very common, it may also explain the growing problem with myopia and mental disorders in children and adolescents.

The way forward appears to be to pay closer attention to the causative factors involved in sperm DNA integrity. A more detailed evaluation of the couple prior to conception may improve their long-term health and may also improve the health of the offspring.

Biography:

Preben Christensen has been researching sperm quality and reduced fertility since 1992 and received his PhD in 1995. His research focus has been on developing and validating various flow cytometric methods for assessing sperm quality. In 2006, his research led to the founding of SPZ Lab, which today offers tests of sperm DNA integrity (SDI®-test) to many European fertility clinics and childless couples. He has a keen research interest in exploring male fertility as a marker for a man's general long term health as well as the health of any future children.

Unbound Bilirubin and Risk of Severe Neurodevelopmental Impairment in Extremely Low Birthweight Newborns

Cody Arnold*

Stanford University School of Medicine, USA



Background: Unbound bilirubin (UB) was measured on day 5 ± 1 in 1101 ELBW newborns in the Aggressive vs Conservative Phototherapy randomized controlled trial. We accessed this dataset to quantify the UB-mediated risk of severe neurodevelopmental impairment (sNDI) in extremely low birthweight (ELBW) newborns.

Methods: UB levels were standardized within laboratories as z-score percentiles. A dichotomous UB exposure variable was based on sharply increased risk of sNDI at the 85th percentile ($UBz > 85th$). Targeted maximum likelihood estimation with ensemble machine learning (TMLE+ML) and logistic regression (LR) were used to estimate sNDI risk attributable to $UBz > 85th$.

Results: UB - total bilirubin correlation was poor: R^2 0.24, 95% CI: 0.19, 0.30. The UB level corresponding to the $UBz > 85th$ cut-point in this cohort was estimated to be between 0.56 and 0.79 $\mu g/dL$. In 825 survivors with known sNDI status the crude risk difference (RD) associated with $UBz > 85th$ was 19.5%. With TMLE+ML, the RD was 14.4% (95% CI: 5.7%, 23.1%) corresponding to a number need to harm (NNH) of 7 (95% CI: 4, 18). LR results were similar: RD 12.4% (95% CI: 4.7%, 20.1%), NNH 8 (95% CI: 5, 21).

Conclusions: Exposure to $UBz > 85th$ was associated with a large increase in risk of sNDI in ELBW newborns. The current incidence of exposure to harmful UB levels in very premature newborns is unlikely to be rare. TB levels provide a false sense of security to clinicians caring for very premature newborns.

Biography:

Cody Arnold MD, MSc is an associate professor in the Division of Neonatal and Developmental Medicine, Department of Pediatrics at the Stanford University School of Medicine. He is a neonatologist and epidemiologist, the principal investigator in the randomized controlled trial Cycled Phototherapy Dose Finding Study for Extremely Low Birth Weight Infants (JAMA Pediatrics 2020, PMID: 32338720). Currently his work is focused on the application of modern methods for causal inference to the study of risks secondary to exposure to unbound (free) bilirubin in premature newborns.

Hidden Deficiency: immune-mediated vitamin d loss in children with chronic inflammatory and allergic conditions

Chandra Sekhar Devulapalli*

Senior Paediatrician, Department of Medicine, Helgeland Hospital, Sandnessjoen, Norway



Vitamin D deficiency is a common challenge in pediatric practice, often attributed to inadequate diet, limited sunlight, or low physical activity. Yet, mounting evidence reveals a crucial, yet underappreciated, role of immune-driven mechanisms—especially in children with chronic inflammatory, allergic, or infectious conditions.

In disorders such as asthma, atopic dermatitis, allergic rhinitis, and recurrent respiratory infections, pro-inflammatory cytokines disrupt vitamin D metabolism by suppressing activating enzymes like 1 α -hydroxylase and enhancing catabolic pathways, while activated immune cells consume vitamin D locally, further reducing circulating levels. Parallel mechanisms are observed in autoimmune diseases including juvenile idiopathic arthritis and type 1 diabetes, where persistent inflammation and organ involvement impair vitamin D activation and absorption.

Compounding these effects, lifestyle factors such as decreased outdoor exposure and suboptimal nutrition often accompany chronic illness, increasing vulnerability. Genetic variations in the vitamin D receptor and binding proteins add another layer of complexity, influencing individual susceptibility.

Vitamin D deficiency in this context should be seen not simply as a nutritional deficit but as a dynamic biomarker of immune dysregulation. Pediatricians are encouraged to adopt a holistic, immunologically informed perspective when assessing vitamin D status, particularly in children with chronic or recurrent diseases. Understanding these interwoven pathways is essential to optimizing supplementation strategies and improving overall pediatric care.

Biography:

Dr. Chandra Sekhar Devulapalli, MD, PhD, is a senior pediatric consultant at Helgeland Hospital, Norway, with expertise in pediatric pulmonology, gastroenterology, nutrition, and allergies. He earned his PhD from the University of Oslo, focusing on asthma and lung function in children. He has authored over 40 peer-reviewed articles, including in NEJM, Thorax, and BMJ, with 1,700+ citations. His research includes vitamin D, COVID-19, and chronic pediatric diseases. Recognized with multiple national awards, he also serves as a journal reviewer and has published two academic books. Dr. Devulapalli is known for uniting clinical excellence with impactful pediatric research.

SPEAKER PRESENTATIONS | DAY 1

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Screening for Pompe, Fabry, Gaucher disorders using dried blood spot: 3 years' experience

Jorge Sales Marques*

Center of Diagnosis and Evaluation of Rare Disorders Hospital Cuf Porto – Portugal

Background: Lysosomal storage diseases (LSD) are a group of 50 metabolic diseases, with the origin changes cause by bad function of the lysosome. Pompe, Fabry and Gaucher are one of these diseases. The diagnosis can be realized in filter paper with only some drops of blood. Enzyme replace therapy (ERT) is the treatment of choice.

Purpose: The purpose of this study is find cases of Pompe, Fabry or Gaucher disorders, using dried blood spot (DBS) during a period of 3 years.

Material: The screening of these 3 disorders was during a period of 3 years, in two hospitals, one public in Macau, China and another private in Portugal, between 1-1- 2020 to 31-12-2020 and 1-1-2023 to 31-12-2024, respectively.

Methods: We used DBS samples for enzyme study of these 3 disorders. The criteria for determination was based on the author enzyme flowchart of Pompe, Fabry and Gaucher diseases. All positive test need to be confirmed by whole blood and DNA study.

Results: A total of 27 cases (Macau:16, Portugal:11) were screened using flowchart criteria: Pompe – 22, Fabry – 2, Gaucher – 3. The age of the patients was between 1 month to 31 years old. The gender ratio was 1:1. We found 3 cases: Pompe -1, Fabry – 1 and Gaucher – 1.

Discussion: ERT in diseases like Pompe, Fabry, Gaucher has change the final prognosis of these patients, giving them a better quality of life and decreasing the risk of death. These 3 disorders are underdiagnosed because they affected multiple organs and systems. The patient can be lost in different consultations, like pediatrics, neurology, orthopedics, hematology or cardiology. Whenever we have a patient with clinical symptoms, the first step is to do the enzymatic screening in the filter paper . The confirmation test is DNA study . We can offer prenatal diagnosis and genetic counselling for the future pregnancy in these 3 diseases. Pompe and Gaucher are inherited as autosomal recessive disorder and Fabry is a x-linked disease.

Conclusion: If we suspect of Pompe, Fabry or Gaucher disorders, we can decide to do the DBS based on the flowchart of these diseases. In our study, we found 3 cases, one of each disorder, in 27 patients that were screened. ERT is the treatment of choice and the earlier we diagnose the disease, the better will be the prognosis and quality of life of the patient.

Assessing Early Screening for Cortical Visual Impairment in the NICU: Preliminary Findings from a Pilot Study

Beatrice M. Stefanescu* and Amanda B. Deford

Department of Pediatrics, Indiana University School of Medicine, Indianapolis, IN, USA

Background: Cortical visual impairment (CVI), which results from damage to neurologic pathways, is the leading cause of childhood blindness and low vision. Identification of CVI is often delayed by years, prolonging time to therapies. The Neonatal Assessment Visual European Grid (NAVEG) screen has been suggested as a tool to help identify premature infants at the highest risk for CVI. The feasibility and validity of NAVEG screening has not been evaluated among other high-risk populations prior to discharge from the NICU.

Objectives: Evaluate the feasibility of implementing NAVEG screening for at-risk neonates prior to NICU discharge; validate NAVEG tool for prediction of CVI in specific high-risk populations.

Design/methods: This is an ongoing cohort study of infants admitted to our level IV NICU, with enrollment expected to be complete in winter of 2025-2026. Neonates are screened for eligibility by identification of risk factors for CVI including prematurity of <32 weeks gestation at birth, or neurologic conditions including seizure, perinatal asphyxia, brain anomaly, intracranial hemorrhage, or stroke. Enrolled neonates undergo NAVEG screening by trained examiners, consisting of ocular, motor, and perceptual visual components. Infants with abnormal results are referred for additional evaluation with specialized early intervention services and results collected. A one-year parental survey captures perceptions of the infants' visual function.

Results: Of 292 eligible infants, 85 were enrolled. 39% of the screened infants had abnormal scores. The perceptual visual component was most often affected, and 34% of screened infants had all three visual domains affected (Figure 2). There were no significant associations between baseline and demographic infant characteristics and screening risk classification (Table 2). In binary logistic regression, congenital hydrocephalus and intracranial hemorrhage were positively associated with increased odds of medium or high risk for CVI on the screening test ($p=0.0020$ and $p=0.0297$, respectively; Table 2). Overall, the implementation of NAVEG screening within the level IV NICU has been well received by providers, nurses, and families.

Conclusion: The implementation of the NAVEG screening protocol has been shown to be feasible, well- received, and smoothly executed. The perceptual visual component was the most likely portion of the NAVEG screen to elicit an abnormal score. Congenital hydrocephalus and intracranial hemorrhage have a statistically significant association with abnormal NAVEG score. The outcomes of this study have potential to change the way CVI is identified and improve outcomes for at-risk neonates.

Improving Cooking Skills, Lifestyle Behaviors, and Clinical Outcomes for Adults at Risk for Cardiometabolic Disease: Protocol for a Randomized Teaching Kitchen Multisite Trial (TK-MT)

Jennifer Massa*

Harvard T.H. Chan School of Public Health, United States

This protocol describes a study to investigate the feasibility and preliminary efficacy of a novel Teaching Kitchen Multisite Trial (TK-MT) for adults with cardiometabolic abnormalities. The TK-MT protocol describes a hybrid lifestyle intervention combining in-person and virtual instruction in culinary skills, nutrition education, movement, and mindfulness with community support and behavior change strategies. This 18-month-long randomized controlled trial aims to evaluate the feasibility of implementing a 12-month, 24 class program, assess preliminary study efficacy, and identify barriers and facilitators to implementation. Methods: The intervention program includes 16 weeks of intensive hands-on culinary and lifestyle education classes followed by eight monthly virtual classes. Psychometric assessments and biometric data will be collected at baseline, 4, 12, and 18 months. Semi-structured interviews and open-ended surveys will be conducted during the 12-month follow-up assessment. Results: Feasibility will be assessed through recruitment, attendance, and fidelity data. Secondary outcomes will analyze changes in health behaviors, biometric data, and anthropometric measures using mixed-effects regression models. Qualitative data will undergo thematic analysis. Conclusions: As envisioned and described in detail in this manuscript, this study will inform the development and implementation of reproducible, scalable teaching kitchen interventions. The protocol described here is intended to set the stage for future investigations to evaluate evidence for the impact of teaching kitchen interventions on dietary habits, physical activity, and overall health and well-being.

Biography:

Jennifer Massa is a Lead Research Scientist at Harvard. Dr. Massa helped design and lead the seminal study on the efficacy of teaching kitchens in 2014. As a Doctor of Science in epidemiology and nutrition science, she has the expertise to develop and implement rigorous scientific studies on nutrition and lifestyle interventions. Dr. Massa also serves as director of research for the Teaching Kitchen Collaborative where she provides strategic guidance on program development and mentors researchers both nationally and internationally. Dr. Massa sits on the National Institutes of Health steering committee for Whole Person Health where she advocates for Food Is Medicine policies and initiatives.

International organ exchange: Life-saving for children on the organ waiting list

Zehnder Yara* and Immer F

Swisstransplant, National Foundation organ donation and transplantation, Berne, Switzerland

Background: Finding the right organ for children with terminal organ failure on the waiting list is a major challenge. On the one hand, the number of paediatric organ donors is very small and, on the other hand, it is important to bear in mind that it is much more complex to find the right organ in the country of the organ donor due to size and weight. Against this backdrop, international organ exchange via the FOEDUS (Facilitating Exchange of Organs donated in EU member states) platform is particularly important for children. This paper aims to provide an insight into the paediatric organs that were offered via FOEDUS and ultimately transplanted.

Methods: All organ offers since FOEDUS went live on 1 June 2015 until 31 December 2024 were evaluated. Organ offers from paediatric organ donors (< 18 years) were recorded in detail with regard to organ type, age and origin of the organ donor and quality of the organ. The children were divided into age categories and evaluated.

Results: Since the launch of FOEDUS, 902 paediatric organ donations have been offered via the platform. This corresponds to 32.0% of total activity. Of these, 256 were hearts (28.4%), 222 were lungs (24.6%), 182 were livers (20.2%), 106 were kidneys (11.8%) and the rest were pancreases and small intestines. Most of these organs came from the 1 to 5 age category. Only 14 of the 25 Member States actively participated in paediatric organ exchange. Of the 902 organs offered, 263 (29.2%) were transplanted. Of these, 110 transplants were for the heart (46.6%), 83 for the liver (11.4%) and 36 for the lungs (13.7%). Fifty percent of the transplanted hearts came from children who had been resuscitated beforehand.

Conclusions: International organ exchange is particularly important for paediatric organs in order to find a suitable recipient internationally. International cooperation is very demanding for teams, especially in the paediatric field, so that organs are often not uploaded to the platform in order to avoid delays. The number of offers for children could be increased in the long term if all Member States took action and, despite the challenging conditions, took this important step to improve the supply of organs for paediatric recipients in the Member States.

Enzymatic approach to neonatal hyperbilirubinemia

Kiumars Pirkalani*

Mehr Medical Group, Tehran, Iran

Background: The simple ultra -old problem of neonatal hyperbilirubinemia has been solved by very complex settings of exchange transfusion and phototherapy and beside time/cost and ward consuming issues; treatment is not optimum. Till now it has not been clarified whether low grade hyperbilirubinemia is harmful, harmless or beneficial regarding the antioxidant nature of unconjugated bilirubin. Besides Albumin binding, transportation to the liver, uptake, conjugation and secretion into bile has not been fully investigated for therapeutic intervention. It is clear that conjugation within the circulation, if possible, omits these steps and will be faster than natural absorption, conjugation and release. We proposed that peripheral conjugation with a canine glucose transferase short cut all these steps without the need for the energy rich uridyl- glucuronic-transferase which is relatively deficient in neonates and much so in premature babies.

Aim: Therapeutic and prophylactic reduction of unconjugated bilirubin and complete replacement of phototherapy and exchange transfusion with a pharmacological approach.

Methods: After successful preclinical study, two enzymes i.e. human glucuronic transferase and canine glucose transferase were used at picogram concentrations in 12 patients in three clinical phases. Type I Crigler Najjar models were not available so moribund neonates with life expectancy of less than three months were used for toxicity studies.

Results: The human enzyme was unable to conjugate bilirubin in human serum because of the lack of uridyl-diphosphate. Addition of uridyl-diphosphat completed the task within minutes. Conjugation with glucose under canine glucose transferase was easy, without the need for uridyl-diphosphate and resulted in water soluble monoconjugates. The reaction was completed within seconds without complex needed agents or by products in vitro.

In vitro studies of hundreds of blood samples were extremely successful with our best glucose transferase. The details are out of the scope of this presentation.

A single injection caused a reduction of harmless Bilirubin of 8mg/dl down to zero in a timely curve in 12 human neonates.

Conclusion: Treatment of neonatal hyperbilirubinemia with canine glucose transferase is safe without side effects done at picogram concentrations and can be accomplished as treatment and prophylaxis without hesitancy about immunologic reactions or need for retreatment. A large study is underway.

Biography:

Chairman of Mehr Medical group since 2005

Navigating the Intersection of Giftedness and ADHD: Addressing Misdiagnosis and Supporting Twice-Exceptional Learners

Melissa Beck Wells*

Department of Educational Studies, State University of New York – Empire State College, USA

The misdiagnosis of gifted students as having Attention Deficit Hyperactivity Disorder (ADHD) presents a significant challenge due to the overlapping behavioral traits of both profiles. Characteristics such as high energy, impulsivity, and intense focus on specific interests may be misinterpreted as symptoms of ADHD, while difficulties with sustained attention in unchallenging tasks may obscure underlying giftedness. This paper explores the complexities of twice-exceptionality, the historical evolution of ADHD and giftedness diagnoses, and the consequences of misclassification on academic performance, social belonging, and emotional well-being.

Drawing from the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5), the study examines diagnostic criteria for ADHD and compares them with the cognitive and behavioral characteristics of gifted individuals. Theoretical perspectives, including Gardner's theory of multiple intelligences and developmental systems theories, provide insight into the cognitive and social development of twice-exceptional students. Additionally, the impact of the COVID-19 pandemic on diagnostic trends and educational outcomes is analyzed. Findings indicate that reliance on conventional ADHD screening tools, such as the Conners 3 Rating Scale, without consideration of gifted traits can lead to misdiagnosis, inappropriate interventions, and hindered educational support.

This presentation will discuss best practices for identifying and supporting twice-exceptional learners, emphasizing Universal Design for Learning (UDL) as a framework for addressing their diverse needs. Policy recommendations include refining assessment protocols, implementing teacher training programs focused on differential diagnosis, and fostering stronger collaboration between educators, clinicians, and families. A strength-based approach that prioritizes individual learning profiles over rigid diagnostic labels is critical for creating inclusive and equitable learning environments.

References:

1. American Psychiatric Association. Diagnostic and Statistical Manual of Mental Disorders (5th ed.). APA, 2013.
2. François-Sévigny, J., Pilon, M., & Gauthier, L.-A. Differences in Perceptions of Behaviors Manifested by Gifted Children with ADHD. *Brain Sciences*, 2022, 12, 1571.
3. Rinn, A. N., & Reynolds, M. J. Overexcitability and Giftedness: A Guide for Educators. *Gifted Child Today*, 2012, 35, 34-39.
4. Webb, J. T., Gore, J. L., Amend, E. R., & DeVries, A. R. Misdiagnosis and Dual Diagnoses of Gifted Children and Adults: ADHD, Bipolar, OCD, Asperger's, Depression, and Other Disorders. Great Potential Press, 2005.
5. Mulholland, S., Cumming, T. M., & Lee, J. Accurately Assessing Teacher ADHD-Specific Attitudes Using the Scale for ADHD-Specific Attitudes. *Journal of Attention Disorders*, 2023, 27, 554-568.
6. Frolii, A., Ricci, M. C., Iovane, M., Piscitelli, C., & Iandolo, G. Universal Design for Learning: A New Framework for Special Education. *International Journal of Disability, Development and Education*, 2023.
7. Wood, S. C. Examining Parent and Teacher Perceptions of Behaviors Exhibited by Gifted Students Referred for ADHD Diagnosis. *Roeper Review*, 2012, 34, 194-204.
8. Terman, L. M. Genetic Studies of Genius: Vol. 1. Mental and Physical Traits of a Thousand Gifted Children. Stanford University Press, 1925.

Acute Liver Failure: Is Acetaminophen the Only Culprit?

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⁴ Saint Louis University, Saint Louis, MO, USA

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Epstein-Barr virus (EBV) is a common herpesvirus associated with infectious mononucleosis and rare complications such as hepatitis. EBV-associated hepatitis during acute infection may alter liver metabolism, compounding the risk of drug-induced toxicity. We report the case of a 16-year-old female with acute EBV infection who developed severe acetaminophen toxicity after reported use for migraine relief. Her condition was refractory to N-acetylcysteine but improved with the addition of fomepizole, which may have mitigated liver injury by reducing N-acetyl-p-benzoquinone imine (NAPQI) production via CYP2E1 inhibition and suppressing inflammation. This case underscores the complexity of managing pediatric acute liver failure with coexisting hepatic insults, particularly in the presence of viral infections that disrupt liver metabolism, and highlights fomepizole as a potential adjunct in pediatric N-acetylcysteine-refractory acetaminophen toxicity. Further research is warranted to explore the interactions between viral hepatitis and drug-induced liver injury.

Biography:

Keith Zarahi Amaral, MD is a General Pediatrician at Summit Hospital in Show Low, Arizona. He had first trained at Saint Louis University where he was heavily involved in various research projects and case reports articulating some of the uncommon presentations of pathology observed during his training in Saint Louis. As a General Pediatrician today, he strives to increase public knowledge in attempt to empower parents with the ultimate goal of building strong children – the future for our civilization.

Approaching feeding difficulties in infants and preterm newborns from the perspective of physiotherapy

Fruzsina Erdélyi¹ and Bernadett Kertész^{1,2*}

¹Bethesda Children's Hospital,

²Semmelweis University Faculty of Health Science, Physiotherapy Department, Hungary

Breastfeeding difficulties are occur often in case of any muscle abnormality, causing physical and psychological difficulties for both the child and the mother. Counsellors and specialists of the health care system try to support mothers in breastfeeding difficulties and questions around the world such as in Hungary. In many cases, help is provided directly by the counsellor, but very often parents are referred to physiotherapists, because feeding difficulties in infancy can develop in a complex way, acting from many directions. A wide-ranging study is needed to describe this extensively. [1,2,3]

Muscle tone disbalance may involve a small area of the body or a muscle group, maybe only one main muscle, or the whole body and it cause a feeding problem. This is particularly the case for premature babies and for babies diagnosed with an abnormality (torticollis, stroke, lack of oxygen, genetic abnormality).^[5] Early physiotherapy intervention is essential to prevent the development of persistent changed muscle function and help to correct the potential for breastfeeding. [1,4,5,6]

In the event of any abnormality, it is worth thinking about physiotherapeutic care. Physiotherapy assessment and treatment should be applied to the whole body, as all muscles interact with each other, often resulting in functional abnormalities further away from the affected area.^[3]

In our presentation, our aim to present a comprehensive picture of the relationship between infant and premature babies feeding challenges physiotherapy in the aspect of a special hungarian method call Dévény therapy.

Biography:

Dr. Bernadett Kertész is a member of the Physiotherapy Department at Semmelweis University, Budapest, Hungary. She specialized in infant and premature physiotherapy. She works at university as an assistant professor, teach and research contributions in peadiatric and infant physiotherapy. This has led her to be the Physical Therapy Infant Workgroup Leader.

References:

- Guler, S., Cigdem, Z., Knoll, B., Ortabag, T., & Yakut, Y. (2022). Effect Of The Premature Infant Oral Motor Intervention On Sucking Capacity In Preterm Infants In Turkey: A Randomized Controlled Trial. [Article]. *Advances In Neonatal Care*, 22(6), E196-E206.
- Kolb, B., Harker, A., & Gibb, R. (2017). Principles Of Plasticity In The Developing Brain. [Review]. *Developmental Medicine And Child Neurology*, 59, 1218-1223.
- ZS Mézám et al. The Dévény Special Manual Technique – Gymnastics Method, *Developments in Health Sciences*, 2018, Pages: 18–20
- Parau, D., Todoran, A., & Balasa, R. (2024). Physiotherapy Intervention On Premature Infants-A Pilot Study. [Article]. *Medicina-Lithuania*, 60(1).
- Thakkar, P., Rohit, H., Das, R., Thakkar, U., & Singh, A. (2018). Effect Of Oral Stimulation On Feeding Performance And Weight Gain In Preterm Neonates: A Randomised Controlled Trial. [Article]. *Paediatrics And International Child Health*, 38(3), 181-186.
- Vonderlin, E., Nöcker-Ribaupierre, M., & Wilken, M. (2010). Supportive Interventions In Newborn. *German Newborn Intensive Care Units*. [Article]. *Monatsschrift Kinderheilkunde*, 158(1), 42-47.

THE DISAPPEARING BIFIDOBACTERIUM MICROBIOTA IN INFANTS OVER THE PAST 25 YEARS

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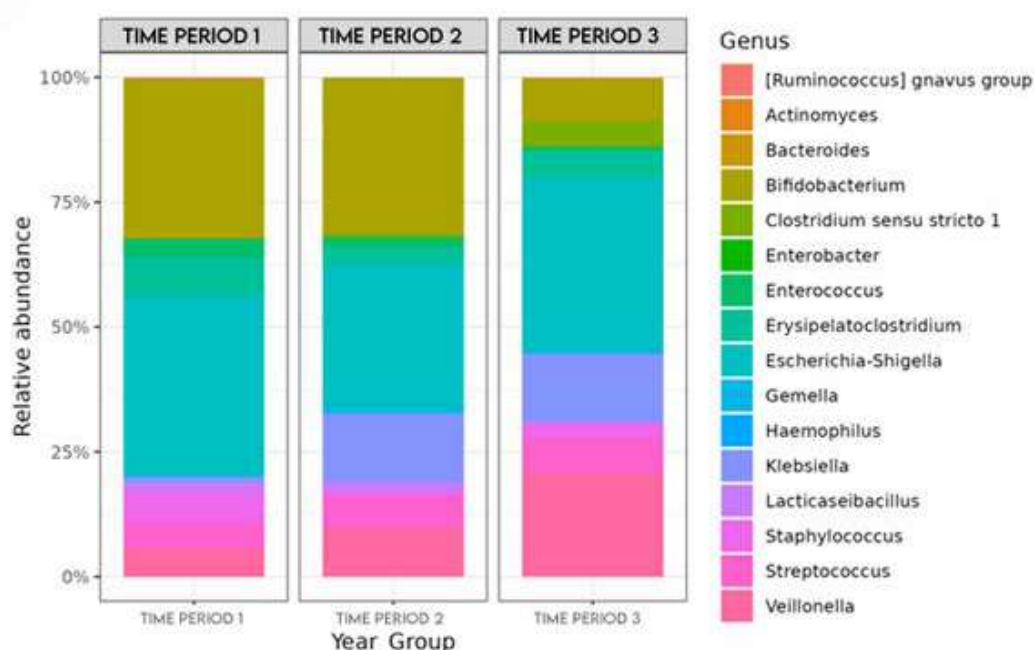
⁴ Department of Pediatrics and Adolescent Medicine, Turku University Hospital, Finland

Bifidobacteria typify the gut microbiota composition of healthy, breast-fed infants. Low bifidobacterial abundance in infancy has been linked with a heightened risk of non-communicable diseases in later childhood. Our objective was to assess factors associated with the gut microbiota composition in infants throughout the allergy and obesity epidemics of the past decades.

We studied 72 full-term, healthy breastfed infants participating in prospective clinical studies, grouped by the year of birth into three time periods (1997-2001, 2005-2009, 2015-2022). Faecal samples were collected and stored for microbiota profiling by 16S rRNA gene sequencing at the age of 1-3 months. Perinatal factors including mode of birth, antibiotic exposure during pregnancy and at birth were taken into account in the analysis.

The microbial richness and diversity of the infant gut microbiota decreased significantly over the three time periods, mainly as a result of reduced abundance of the phylum Actinobacteria, and its genus Bifidobacterium in children born in 2015-2022 as compared to those born during the time periods 1997-2001 and 2005-2009 (**FIGURE 1**). The time period of birth was the strongest determinant of the gut microbiota composition, followed by mothers' pre-pregnancy body mass index, intrapartum antibiotic contact and mode of birth. The relative abundance of members of the genus Bifidobacterium was negatively associated with elapsed time (1997-2022 $p=0.0005$) and intrapartum antibiotic exposure ($p=0.0026$).

The abundance of the genus Bifidobacterium has gradually decreased over the past 25 years. The depletion of gut microbiota richness and diversity, and the selective disappearance of the genus Bifidobacterium has occurred parallel to the increase in the prevalence of non-communicable diseases.



Biography:

Dr Isolauri is Professor of Pediatrics at the University of Turku, and Turku University Hospital, Finland. She has received numerous Academic Honours such as The John Harris price (European Society for Paediatric Gastroenterology and Nutrition); Niilo Hallman price (Foundation for Paediatric Research); Institut Candia (France); Marvin L. Dixon Memorial Lecture (Harvard Combined Program in Pediatric GI&Nutrition Unit, Children's Hospital, Massachusetts General Hospital, USA); Nanna Svartz Lecture (Karolinska Hospital, Stockholm, Sweden); Tissier Price and Lecture (The Japan Bifidus Foundation); The IDF Elie Metchnikoff Prize in the category Nutrition and Health and the Matti Äyräpää Award from The Finnish Medical Society Duodecim, and The European Award in Medicine: the Gastroenterology category.

She has supervised 26 doctoral theses (PhD) and acted as Opponent in Finland, France, Sweden, The Netherlands and Denmark. Her Major Research Interests are: Gastroenterology, gut microbiota; Nutrition, probiotics, and Immunology, food allergy in the Neonatal exposures, Adverse outcomes, Mucosal immunology and Intestinal microbiota (NAMI) programme. Author of 466 scientific publications, she has written 287 original publications, 112 Review articles/ Chapters in books and 67 Scientific articles in Finnish. In addition she has 191 Invited Lectures in International Academic Meetings. H-index 101 (Jan, 2025).

PHP Type 1b in a 14-Year-Old Boy with Hypocalcemia and Spasms After Flucloxacillin Intake

Andleeb Badar*

Paediatrics BST trainee Year 1

Trainer: Dr Sinead Glackin

Consultant Paediatrician, Sligo University Hospital, Ireland

Pseudohypoparathyroidism (PHP) Type 1b is a rare disorder caused by resistance to parathyroid hormone (PTH), leading to hypocalcemia and hyperphosphatemia. This case describes a 14-year-old boy who presented with hypocalcemic spasms following flucloxacillin treatment. Notably, his family had a history of chronic body aches and migraines, which had been previously overlooked. Initial genetic testing was misinterpreted as negative, delaying diagnosis. This case highlights the importance of accurate genetic testing interpretation and the need to consider familial symptoms in cases of unexplained hypocalcemia.

Case Presentation:

Patient Information:

- Age/Sex: 14-year-old male
- Presenting Symptoms:
 - Muscle spasms, tetany, perioral numbness, carpopedal spasms
 - Generalized body aches and fatigue (which had been ongoing for months)
 - Chronic migraines, similar to other family members
- Medical History: No prior diagnosis of metabolic disorders; occasional muscle cramps and headaches were present but not investigated
- Family History: Parents and siblings reported chronic body aches and migraines, but no formal diagnosis of metabolic disease
- Recent History: Treated with flucloxacillin for a bacterial skin infection one day before symptom onset

Physical Examination:

- Positive Chvostek's and Trousseau's signs
- No features of Albright's hereditary osteodystrophy (AHO) (e.g., short stature, brachydactyly)
- Generalized muscle tenderness, consistent with reported body aches

Laboratory Investigations:

Test	Result	Reference Range
Serum Calcium	1.48 mg/dL	(low) 2.10-2.55 mg/dL
Serum Phosphorus	2.12 mg/dL	(high) 0.95-1.65 mg/dL
Parathyroid Hormone (PTH)	Elevated	15-65 pg/mL
Vitamin D	26(low)	50-125 ng/mL

Diagnostic Challenges:

Given the biochemical profile of hypocalcemia, low vitamin D, and elevated PTH, Vitamin D deficiency was suspected initially. But persistent high phosphate wasn't in support of this diagnosis. Congenital hypoparathyroidism e.g. DiGeorge Syndrome and CHARGE syndrome can cause low calcium, low PTH, High phosphate and normal vitamin D. But it wasn't in keeping with all the labs and there was absence of any physical features of these syndromes.

Chronic kidney disease can cause hyperphosphatemia due to impaired secretion, low calcium due to low vit D activation, high PTH due to secondary hyperparathyroidism. But the renal function tests of the pt. were completely normal. McCune-Albright Syndrome can present with PTH resistance, but the typical clinical features of this syndrome were absent.

The only probable diagnosis that was in keeping with all the symptoms and the lab investigations was Pseudohypoparathyroidism type 1b (as PHP Type 1a has classic AHO phenotype e.g., short stature, brachydactyly which was not seen in this case). To confirm this diagnosis, genetic testing was requested.

However, initial genetic testing was done for the panel of FER05 i.e. for Congenital Hypothyroidism and Thyroid Dysmorphogenesis as there was no specific gene panel for hypocalcemia and pseudohypoparathyroidism. So, the initial genetic test came back negative.

Later on, the analysis was reviewed to identify any additional genes that might need to be included. There are also not many genes associated with pseudohypoparathyroidism, the most important one, GNAS, had already been analyzed as part of FER05. Furthermore, the diagnostics were extended to STX16. The paternal UPD of 20q (where both GNAS and STX16 are located) which required additional tests (MLPA) was done as well.

Upon re-evaluation, there was an aberrant Methylation pattern in GNAS Complex locus which was detected via Methylation-sensitive MLPA, which showed methylation pattern on both alleles, this aberration confirmed the clinical suspicion of pseudohypoparathyroidism type 1b (PTP1B)

Discussion:

PHP Type 1b is caused by epigenetic alterations at the GNAS gene, leading to selective PTH resistance in the renal tubules. Unlike PHP Type 1a, it lacks the classic AHO phenotype (e.g., short stature, brachydactyly).

Familial Symptoms and PHP:

This case is notable because multiple family members had chronic body aches and migraines, raising suspicion that they may also have undiagnosed PHP Type 1b. Hypocalcemia can cause neuromuscular irritability, leading to muscle pain and headaches, which might explain their symptoms. This suggests PHP Type 1b may have been running undiagnosed in the family for years.

Flucloxacillin as a Trigger for Symptomatic Hypocalcemia:

The patient's hypocalcemic crisis was precipitated by flucloxacillin, potentially due to:

1. Renal Calcium Wasting: Flucloxacillin may cause mild renal tubular dysfunction, exacerbating hypocalcemia.
2. Increased Metabolic Demand: Infection and medication use may unmask latent calcium regulation disorders.
3. Gut Microbiome Disruption: Antibiotics can alter intestinal flora, affecting calcium absorption.

Diagnostic Delay Due to Nonspecific Gene Panel for PHP type 1b:

The absence of a specific gene panel for genetic testing of PHP Type 1b led to a delayed diagnosis, emphasizing the importance of:

- Methylation-specific testing for PHP Type 1b if standard sequencing is inconclusive
- Considering familial symptoms when evaluating suspected metabolic disorders

Management & Outcome:

- Acute treatment: Intravenous calcium gluconate to relieve tetany
- Long-term therapy: Oral calcium carbonate and calcitriol for calcium homeostasis, vitamin D supplements.
- Family screening: Given the chronic symptoms in family members, they were referred for calcium, PTH, and genetic testing
- Patient education: Emphasis on lifelong monitoring to prevent recurrence

Conclusion:

This case underscores several key points:

1. PHP Type 1b can present with chronic body aches and migraines, mimicking other conditions and leading to underdiagnosis.
2. Genetic testing errors can delay diagnosis, emphasizing the need for expert interpretation.
3. Medication use (flucloxacillin) or infections can unmask underlying PHP Type 1b, triggering acute hypocalcemia.
4. Familial patterns of symptoms should prompt further metabolic investigations.

By recognizing these factors, clinicians can avoid diagnostic delays and ensure early intervention for affected individuals and their families.

References:

<https://rarediseases.info.nih.gov/diseases/10680/pseudohypoparathyroidism-type-1b>

<https://www.orpha.net/en/disease/detail/94089>

Mantovani, G. (2011). Pseudohypoparathyroidism: Diagnosis and Treatment. *Journal of Clinical Endocrinology & Metabolism*, 96(10), 3020-3030.

Levine, M. A. (2012). Genetics of Pseudohypoparathyroidism and Related Disorders. *Hormone Research in Paediatrics*, 78(5-6), 334-341.

Thiele, S., & Mantovani, G. (2015). Clinical and Molecular Aspects of Pseudohypoparathyroidism. *Endocrine Reviews*, 36(1), 67-116.

Bastepe, M. (2013). GNAS Mutations and the Role of Imprinting in Pseudohypoparathyroidism. *Journal of Molecular Endocrinology*, 50(2), R53-R64.

Birth weight is positively associated with adult osteoporosis risk: observational and Mendelian randomization studies

Fei-Yan Deng*

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The relationship between birth weight and osteoporosis was inconsistent in previous observational studies. Therefore, we performed a systematic evaluation to determine the inconsistent relationship and further make causal inference based on the uk biobank datasets (~500,000 individuals) and individual/summary-level genetic datasets. Observational analyses found consistent negative associations either between birth weight and estimated bone mineral density (ebmd) or between genetic risk score (grs) of birth weight and ebmd in total subjects, and sex-stratified subgroups. Mediation analyses detected significant mediation effects of adult weight and height on associations between birth weight and ebmd. Birth weight was causally associated not only with three bmd phenotypes (ebmd, total body [tb]-bmd, and femoral neck [fn]-bmd) under two effect models (total and fetal effect), but also with the risk of fracture using different mendelian randomization (mr) methods. Multivariable mr analyses detected the pleiotropic effects of some environmental factors (e.g., gestational duration, head circumference, hip circumference) on the associations between birth weight and bmd/fracture. Three bmd phenotypes (ebmd, tb-bmd, and fn-bmd) have significant mediation effects on the associations between birth weight and fracture by using a novel mediation mr analysis under the multivariable mr framework. This multistage systematic study found consistent causal associations between birth weight and osteoporosis risk, fetal origin of genetic effects underlying the associations, and several mediation factors on the detected associations. The results enhanced our understanding of the effects of fetal original phenotypes on outcomes in late adulthood and provided helpful clues for early prevention research on osteoporosis.

Keywords: Birth Weight; Fracture; Mendelian Randomization (Mr); Osteoporosis

Biography:

Dr. Fei-yan deng is a professor in public health and preventive medicine at suzhou medical college of soochow university. Her research interest is focused on molecular and genetic epidemiology for the chronic musculoskeletal disease osteoporosis, with the purpose to develop novel biomarkers and models for early diagnosis and precision prevention of osteoporosis.

Pharmacological approach to neonatal hyperbilirubinemia

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Mehr Medical Group, Tehran, Iran

A very conceptual and uncomprehensive point in the strategy is that chelation of Bilirubin is not as easy as conjugation. It is bound to albumin within the circulation and soluble in fat. Besides, in contrast to general notion the lipophilic site of the molecule is not where conjugation occurs. (Fig. 1) Conjugation does not cover the fat-soluble side of the molecule but makes the molecule larger and more water soluble from the water-soluble pole. As the figure shows the molecule retains its lipophilic site but this loses importance because of the larger size of the molecule.

So, it is difficult to approach the molecule with solubilizing agents from the fat-soluble site because chelating agents are themselves water-soluble. The only hope would be extraction of Bilirubin from albumin in the circulation and increasing the binding potential of albumin to extract bilirubin from fat-rich tissues like the brain. As figure 2 shows chelation of bilirubin via the lipophilic site is extremely difficult as this site is per se covered by cellular fat. To attack this site, you need to approach from the inner site with a water-soluble molecule from a lipid-laden nano-environment which is impossible from the standpoint of physical chemistry.

Aim: Increasing the "Albumin pool" to extract more unconjugated Bilirubin from fat-rich tissues and hence therapeutic and prophylactic reduction of unconjugated bilirubin and complete replacement of phototherapy and exchange transfusion with a pharmacological approach.

Methods: Twelve neonates with harmless hyperbilirubinemia in the range of 8mg/dl were treated with a single dose 500mg of a triguanide derivative called TR-225. Bilirubin was measured every half hour for 24 hours. Patients with stable levels under 2 were discharged from the hospital.

Results: TR-225 is 100% capable of reducing hyperbilirubinemia in all patients studied. The conjugate could be traced within the urine in a proportional manner. Speed of bilirubin reduction is 18 times that of exchange transfusion. An increase of hemolysis was suspected but not shown. The time/concentration curve denotes extreme effectiveness without rebound due to re-distribution of bilirubin from peripheral fat to circulation. The treatment is not without side effects and can hardly be used as a prophylaxis.

Conclusion: Treatment of neonatal hyperbilirubinemia with a chelator is chemically extremely difficult and needs sophisticated drug development techniques such as artificial intelligence. But it is a very interesting issue which can possibly close thousands of neonatal wards worldwide. More widespread studies at all three phases and preclinical studies are underway.

Biography:

Chairman of Mehr Medical group since 2005

Glucose-6-phosphate dehydrogenase deficiency and neonatal indirect hyperbilirubinemia: a retrospective cohort study among 40,305 consecutively born babies

Rajai albedaywi*

Hamad medical corporation, Qatar

Glucose-6-phosphate dehydrogenase deficiency (G6PDD) being highly prevalent in the Middle East, the primary objective was to estimate the incidence of neonatal jaundice among G6PD- deficient neonates and to explore its association with various risk factors.

This retrospective cohort study includes 7 years data of neonates diagnosed with G6PDD between 1st January 2015, and 30 September 2022, from Al Wakra Hospital, HMC Qatar.

Among the 40,305 total births, 1013 had G6PDD with an incidence of 2.51%. Of all the G6PDD babies, 24.6% (249/1013) received phototherapy and three babies required exchange transfusion. Statistically significant associations were noted between the need for phototherapy and gestational age, gestational age groups, birth weight, and birth weight groups, but logistic regression analysis showed significant association for phototherapy only with the gestational age group.

Conclusion: Universal screening and proper follow-up is essential for G6PDD as it plays crucial role in neonatal jaundice.

Biography:

Dr.Rajai albedaywi (jordanian nationality) join hamad medical corporation, Qatar since 2013 and work at neonatal intensive unit as consultant since 2019 and have great research interest. Currently, he is co-investigator for many research projects. He had seven publications during the last 6 years. Dr Rajai is Instructor of Clinical Pediatrics at Weill Cornell Medicine-Qatar and involved in teaching of students and resident at HMC.

Overuse of EEG and ECG in children with breath-holding spells and its implication for the management of the spells

Sanna Hellström Schmidt*

Lund University, Sweden

Aim: Breath-holding spells (BHS) are common in children; however, evidence-based clinical guidelines are currently lacking. This study investigated a large population-based cohort of patients with BHS in order to propose a refined description of typical BHS presentations and to develop management guidelines.

Methods: A cross-sectional retrospective study was conducted on patients diagnosed with BHS in Southern Sweden between 2004 and 2018. Data on disease characteristics and diagnostic procedures were extracted from patient medical records.

Results: The study included 519 patients, with a mean age at diagnosis of 19.8 ± 13.8 months and an equal gender distribution. In 48.3% of cases, a BHS diagnosis was made following a single episode. During spells, 78.0% of patients were unresponsive, and atonic, tonic, tonic-clonic, or myoclonic seizure-like activity were reported for 71.5% of cases. Additionally, 78.0% of spells lasted less than one minute. Electroencephalography (EEG) was performed in 30.4% of patients, and electrocardiography (ECG) in 45.1%. Pathological EEG findings were observed in six children (3.8%), four of whom had coexisting epilepsy. Only 0.9% of patients showed ECG findings suggestive of pathology, with no cases of long QT syndrome identified.

Conclusion: Children with BHS were frequently exposed to unnecessary diagnostic investigations. The study characterises the typical presentation of BHS and introduces a management algorithm aimed at reducing the overuse of EEG and ECG in this patient group.

Role of human milk oligosaccharides in immuno regulation and epigenetic modulation of pathogenetic bacteria on infant and toddlers health

Aftab Yusuf Raj*

Senior Consultant, Pediatrics & Neonatology, Square Hospital, MBBS, MD (Pediatrics), MD (Neonatology), Fellow Newborn Medicine (Canada), MPH(NSU)

Human milk oligosaccharides (HMO's) are the third most abundant solid component of breast milk, after lactose and lipids. HMO's play an important role in Infant and toddlers' growth and development, cellular gene expression, immunogenesis. Health benefits of HMO's include modulation of gut commensal bacteria, anti-inflammatory effect in our systemic circulation, and preventing entry of toxic bacteria by strengthening the gut mucosal barrier. HMO's Play a crucial role in epigenetic modification, influences the expression of our genes. HMO's silence the bad genes, helping good genes to express more. HMO's optimize proper functions of Gut-Brain Axis, reduce neuroinflammation, potentiates cortical neuronal activities, Maintain good cognitive skill and memory functions. This review article also discusses the role of HMOs on our immune system that is immuno modulation effects, focuses on concept of Berker's hypothesis (the fetal origin of adult disease)

Biography:

Prof. Dr. Aftab Yusuf Raj graduated from Sher-E-Bangla Medical College. He then trained in the field of Pediatrics and Neonatology and served in different Medical Colleges & Hospitals across the country and held some vital positions including Associate Professor of Pediatrics and Neonatology for several years. He received training in his respective field from all India Institutions of Medical Sciences and Calgary Medical University (Neonatology) and Alberta Children Hospital, Canada. He also worked as a Consultant at United Hospital, Bangladesh, Dhaka.

KEYNOTE PRESENTATIONS DAY- 2

6th Edition - World Congress on
PEDIATRICS & NEONATOLOGY
Sep 25 - 26, 2025 at Berlin, Germany

Mitral valve repair using leaflet expansion and sub-partial annuloplasty in children

Per Wierup*, Jens Johansson Ramgren, Johan Sjögren, Kiet Tran Phan, Igor Zindovic, Shabab Nozohoor, Lea Christiersen³⁴ and Nina Hakacova.

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Objective: Mitral valve reconstruction in the pediatric population is a challenge due to the frequent combination of annular dilatation and leaflet restriction and the need for growth. We present a novel strategy using leaflet expansion and sub-partial annuloplasty with polytetrafluoroethylene (PTFE) reinforcement.

Methods: From January 2014 through May 2021, 11 children aged 5 months-14 years (median 24 months) underwent elective mitral valve repair due to severe mitral valve regurgitation (MR). The mitral valve abnormalities included congenital malformations (n=7), postoperative leakage following commissurotomy (n 1), and functional MR due to dilated cardiomyopathy (n = 3). Surgery consisted of leaflet expansions with autologous, untreated pericardium and sub-partial annuloplasty with PTFE reinforcement.

Results: All children survived their surgeries with uneventful postoperative courses, except for one patient who needed an early reoperation to resolve a functional stenosis due to a spinnaker phenomenon. At discharge, mean gradient was 3.5 ± 3.9 mmHg, with trivial MR in 9 patients (82%). All patients were alive and asymptomatic during the median follow-up of 3 years (range 1-7 years). Their echocardiographic data showed a mean trans-mitral gradient of 4.4 ± 1.7 mmHg and remained unchanged. Residual mitral valve regurgitation was trivial or mild in 9 patients (82%) and moderate in 2 patients (18%).

Conclusions: Leaflet expansion with autologous pericardium and sub-partial annuloplasty with polytetrafluoroethylene reinforcement for mitral regurgitation in the pediatric population gives stable and satisfactory results both early and at intermediate follow-up, permitting growth of the mitral valve.

SPEAKER PRESENTATIONS | DAY 2

6th Edition - World Congress on
PEDIATRICS & NEONATOLOGY
Sep 25 - 26, 2025 at Berlin, Germany

Building coalitions for strategic interventions to improve adolescent health in the technology age

Sean J. Gamble*

Stanford University School of Medicine, United States of America

Social media and other digital technologies are important parts of young people's lives and they bring both potential to benefit development and expose them to harms. Pediatricians are utilizing new resources to center youth experience and promote strategies like family media plans, but it is also important to recognize that provider efforts are pitched against powerful forces. Like the fight against Big Tobacco, which intentionally engineered its products to be highly addictive and appealing to youth, the health of young people in the technology age is also in the crosshairs of corporate profit. Recent research has demonstrated a connection between addictive behavior with digital technology and youth suicidality. Technology companies have made intentional design choices to foster compulsive use and make it difficult for young people to put down their devices, and the result is a crisis in youth mental health.

Policymakers have crafted legislation to try to address these issues but have typically failed to achieve real success. New legislation has often failed to address the disparate lived experience and needs of diverse young people, including LGBTQ and immigrant youth, or failed to adequately confront the underlying surveillance business model that often drives overuse by young people. Many new laws have also run into legal challenges.

I will discuss the research, policy, and legal landscape surrounding legislative efforts to improve youth wellbeing in the technology age. I will bring together resources and best practices from research, clinical work, law and policy, illustrate the importance of cross-disciplinary collaboration to achieve real change, and propose paths forward.

Biography:

Dr. Sean Gamble is a Clinical Assistant Professor of Pediatrics at the Stanford University School of Medicine. Dr. Gamble supports training for Stanford residents on advocacy efforts and is the lead author of a March 2025 Journal of Adolescent Health commentary on young wellbeing and technology.

Comparative evaluation of erosion level of commonly prescribed pediatric liquid medicines on primary teeth: an invitro study

Shanthosh raj Srinivasan*

ESIC medical college and hospital Chennai, India

Maintaining good oral health is crucial for the overall health, growth, and development of children. Dental caries is the most widespread oral disease, Epidemiological studies have shown a frequent occurrence of enamel erosion in young people. Paediatric liquid analgesics (PLAs) are frequently prescribed and well-accepted by young patients. However, prolonged use of these medications can increase the risk of dental caries and dental erosion. This study aims to evaluate the erosive potential of commonly prescribed paediatric liquid medicinal syrups on primary teeth enamel. The objectives include assessing changes in the enamel prismatic pattern of deciduous teeth using scanning electron microscopic (SEM) analysis and comparing the erosive effects of these medications through surface profilometric analysis.

Biography:

Dr shanthosh Raj Srinivasan has completed his MDS(Pedodontics and preventive dentistry)from Dr MGR Educational Research Institute Chennai, India. He is currently working as Associate Professor in Department of Dentistry ESIC Medical college and hospital Chennai India (Govt of India). He has been serving as an editorial board member of several reputed journals.

Contemporary Genetic Testing Patterns in Neonatal Seizures

Beatrice M. Stefanescu*, Anna C. Cole, Kristen R. Suhrie, and Asmatahasin Mohammed

Department of Pediatrics, Indiana University School of Medicine, Indianapolis, IN, USA

Introduction: Emerging research has shown that genetic disease may be the underlying etiology for many neonatal seizures. Prior studies focused specifically on neonatal epilepsies have shown the superiority of exome (ES) and genome (GS) sequencing to confirm a diagnosis as well as evidence of cost-effectiveness and shorter hospital length of stay.¹⁻³ However, most existing studies evaluating genetic testing in infants with seizures limited their scope to neonatal epilepsies, excluding infants with seizures attributed to other underlying etiologies, such as hypoxic ischemic encephalopathy (HIE), hemorrhage, stroke, etc. Recent research in the HIE population suggests that genetic evaluation in this group may be important and necessary, even in infants with a perinatal sentinel event.⁴⁻⁵ Additionally, small studies and case reports have identified other neurologic conditions initially presenting with apparent isolated neonatal seizures. These findings raise the question of whether broader genetic testing in all infants with seizures may improve the diagnostic yield and inform long-term management and strategies.

Objectives: To provide a contemporary account of the patterns of genetic testing and associated diagnostic yield in a cohort of NICU patients with confirmed electrographic seizures.

Methods: A retrospective observational cohort study was conducted in NICU patients with confirmed electrographic seizures admitted to a level IV NICU in Indianapolis, IN between January 2021 and December 2023. Patient demographics, perinatal history, seizure history, neuromonitoring reports, genetic testing performed with associated results, and NICU outcome were collected. Infants were categorized based on primary neurological abnormality(s): HIE, intracranial hemorrhage, congenital hydrocephalus, brain malformation, stroke, brain infection, metabolic, other, and none.

Results: There were 222 infants included in this study. Demographic data are outlined in Table 1. Most of the infants were term, and 125 (56%) were admitted due to a neurologic concern or seizures. Over half of patients (127, 57%) underwent genetic testing, with 172 tests performed. A total of 73 tested infants (57% of tested infants, 33% of entire cohort) had some reported finding on genetic testing and 40 of these infants (31% of those tested, 18% of entire cohort) had a reported diagnostic result. Patterns of genetic testing and diagnostic yield for underlying seizure etiology are described in Table 2. GS was the most commonly used test, with 77 patients (61%) receiving this test.

Discussion: An underlying genetic diagnosis was confirmed in 18% of the cohort when only 57% of patients underwent some form of genetic testing, most commonly, GS testing. HIE was the most common underlying neurologic diagnosis in this NICU cohort, but only 13% underwent some form of genetic testing. However, out of those who underwent testing 27% received a genetic diagnosis, which is consistent with two recent studies reporting yields of 25% and 36% in the HIE entire population (not just focusing on infants with seizures).⁴⁻⁵ The remaining underlying diagnoses had high diagnostic yields ranging from 42-100%.

Conclusions: These findings suggest that genetic testing in all neonates with seizures may have greater clinical utility than previously recognized. Genetic testing in this population may provide better prognostic assessments and support more targeted therapeutic interventions. A completed statistical analysis will be presented at the time of presentation.

References:

1. Berg AT, Coryell J, Saneto RP, Grinspan ZM, Alexander JJ, Kekis M, Sullivan JE, Wirrell EC, Shellhaas RA, Mytinger JR, Gaillard WD, Kossoff EH, Valencia I, Knupp KG, Wusthoff C, Keator C, Dobyns WB, Ryan N, Loddenkemper T, Chu CJ, Novotny EJ, Koh S. Early-life epilepsies and the emerging role of genetic testing. *JAMA Pediatr.* 2017;171:863-871.
2. Sanchez Fernandez I, Loddenkemper T, Gainza-Lein M, Sheidley BR, Poduri A. Diagnostic yield of genetic tests in epilepsy: A meta-analysis and cost-effectiveness study. *Neurology.* 2019;92:e418-428.

3. Akbari H, Sunderraj A, Sanchez-Pinto N, Berg AT, George AL, Pardo AC. Genetic testing and hospital length of stay in neonates with epilepsy. *Pediatr Neurol.* 2022; 133:30-33.

4. Woodward KE, Murthy P, Mineyko A, Mohammad K, Esser MJ. Identifying genetic susceptibility in neonates with hypoxic-ischemic encephalopathy: A retrospective case series. *J Child Neurol.* 2023;38(1-2):16-24.

5. Parobek CM, Zemet R, Shanahan MA, Burnett BA, Mizerik E, Rosenfeld JA, Vossaert L, Clark SL, Hunter JV, Lalani SR. Clinical exome sequencing uncovers genetic disorders in neonates with suspected hypoxic-ischemic encephalopathy: A retrospective analysis. *Clin Genet.* 2024;106(1)95-101.

Table 1. Infant birth, admission, and discharge demographics

Birth & Admission Demographics	
Gestational age, n (%)	
≤24 weeks	7 (3)
25-28 weeks	10 (5)
29-32 weeks	9 (4)
33-36 weeks	21 (9)
≥37 weeks	175 (79)
Male sex, n (%)	
116 (52)	
Birth weight, g (median (IQR))	
3070 (2517-3519)	
Birth location, n (%)	
Inborn	63 (29)
Other hospital	148 (67)
Home	3 (1)
Birthing center/not hospital	4 (2)
Other site	1 (0.4)
Unknown	3 (1)
APGAR scores (median (IQR))	
1 minute	4 (1-7)
5 minutes	7 (4-9) 6
10 minutes	(4-7)
Age at admission, days (median (IQR))	
1 (0-3)	
Reason for admission to the NICU, n (%)	
Neurologic or concern for seizures	125 (56)
Other	97 (44)
Discharge Demographics	
Disposition, n (%)	
To home or medical foster care	126 (57)
Transferred out of NICU	51 (23)
Died	45 (20)

Cause of death, n (%)	
Anomaly or syndrome	CNS
injury	13 (29) 12 (27)
Multi-organ system failure	9 (20)
Respiratory failure	4 (9)
Infection	3 (7)
GI/Intra-abdominal catastrophe	2 (4)
Inborn error	2 (4) 2 (4)
Other	
Age at discharge, days (median (IQR))	
22 (11-48)	
Discharged on AEDs, n (%)	
79 (45)	

Table 2. Genetic testing rates and outcomes in neonates with seizures, stratified by primary neurological diagnosis.

Underlying Neurologic Diagnosis	Infants, n (%)	Received genetic testing, n (%)	Diagnostic result, n (%)
Hypoxic ischemic encephalopathy	82 (37)	11 (13)	3 (27)
Intracranial hemorrhage	71 (32)	17 (24)	8 (47)
Stroke	33 (15)	12 (36)	5 (42)
Brain malformations	21 (9)	15 (71)	10 (67)
Brain infection	17 (8)	2 (12)	2 (100)
Congenital hydrocephalus	6 (3)	3 (50)	2 (67)
Metabolic	6 (3)	6 (100)	6 (100)
Other neurologic condition*	16 (7)	9 (56)	7 (78)
No known neurologic condition	28 (13)	14 (50)	8 (57)

*Other neurologic conditions: PVL, cystic encephalomalacia (isolated), brain tumor, kernicterus, white matter volume loss (isolated), dural venous sinus thrombosis

ADRB2 genotype-guided treatment for childhood asthma: an individual patient data meta-analysis of the PACT and PUFFIN trial

E.M.A. Slob^{1,2,3*}, S.J.H. Vijverberg¹, L. Noij¹, S. Mukhopadhyay^{4,5}, T. Ruffles^{4,5}, R.H.N. van Schaik⁶, E.T.G. Kersten⁷, J.W.R. Twisk⁸, M.W. Pijnenburg⁹, G.H. Koppelman⁷, A.H. Maitland-van der Zee^{1,2}, on behalf of the PUFFIN and PACT investigators.

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Background: Long-acting β_2 -agonists (LABA) are commonly used to treat asthma, however, some children do not respond to LABA. This might be due to variation in the A allele of rs1042713 (Arg16 amino acid) in the ADRB2 gene encoding the β_2 receptor. We investigated whether Arg16Gly genotype-guided treatment improved asthma-related outcomes.

Methods: We recruited Dutch and Swiss 6-18-year-old (PUFFIN) and 12-18-year-old children from England and Scotland (PACT) with uncontrolled asthma despite inhaled corticosteroids (ICS) and performed an individual patient data meta-analysis of the two randomised controlled trials. Patients requiring step-up were randomised to genotype-guided treatment (adding LABA (Gly16Gly) or montelukast (PACT)/a double dose of ICS (PUFFIN) (Arg16Arg/Arg16Gly)) or standard care with a follow-up of at least 6 months. The primary outcome was change in asthma control. Secondary outcomes were change in exacerbation rate and time to exacerbation. Repeated measures mixed models (RMMM) and cox regression were used. Due to missing data, PACT baseline exacerbations were coded as 0, no exacerbations.

Results: 59 out of 102 (PUFFIN) and 59 out of 91 (PACT) children had at least one A allele. These 193 children were included. There was no difference in asthma control between both arms during follow-up. Genotype-guided treatment resulted in lower asthma exacerbation rates (-0.08 (-0.16 - -0.00, $p=0.038$)) and a trend towards longer time to exacerbation (hazard ratio 0.62 (95%CI 0.38-1.01, $p=0.054$)) compared to standard care.

Conclusion: Genotype-guided step-up treatment for children with uncontrolled asthma on ICS may lower asthma exacerbation rates.

The PUFFIN trial was funded by the Lung Foundation Netherlands (projectnumber: 5.1.16.094); ClinicalTrials.gov number: NCT03654508. The PACT trial was funded by The Henry Smith Charity and Action Medical Research (grant number GN2203); ClinicalTrials.gov number: NCT02758873.

Treatment of 1182 early type I DM with a combination of 3 immune modulatory drugs Eight years of experience

Kiumars Pirkalani*

Mehr Medical Group Tehran Iran

Background and Aims: Immune modulation of type I diabetes has failed in the last 90 years ago. Increase of C-Peptide has never been seen. Based on experience with harmless immune modulatory drugs in other disease such as alopecia areata, multiple sclerosis, and we tried to treat type I DM in the last 7 years ago. The treatments were changed gradually and the most effective protocol is presented.

Materials and Methods: Eleven hundred eighty-two patients were treated with Azithromycin 250mg, Vit A 25000IU twice weekly, and Atorvastatin 10-40mg every night as the immune modulatory block. Dapagliflozin (or Empagliflozin), Glucophage and Pioglutazone were used to reduce the need for insulin. In refining and defining response rate, serum fasting and 2hpp Insulin and C-Peptide, HbA1C, anti-Gad and anti-Islet cell Ab were measured at beginning and every 3 months.

Results: Hundred percent of the patients responded to at least one of the seven parameters with a reduction of HbA1c at 1.4-9.4 percent. Patients could omit their short acting insulin at evening, morning and noon in 100, 87 and 69% respectively.

Reduction of anti-GAD and Anti Islet cell AB were encountered in 96 and 92 percent from a high of up to 2000 and 150 down to below 5 and 1.2. Fasting and 2hpp Insulin, fasting and 2hpp C-Peptide were increased in 86%, 88%, 68% and 82% from a low of 0.2, 0.4, 0.001, 0.001 to a high of 4.2, 16.2, 0.3 and 7.6 respectively.

Three hundred eighty-eight (32.8%) patients discontinued their insulin completely and 198 (16.7%) awaits this.

Discussion: The astonishing results of increased C-Peptide with declaration of 2hpp C-Peptide as a novel response marker denotes that either increased secretion of insulin or increased beta cell proliferation (like DYRK-1 pathway) is possible in vivo. More sophisticated techniques are needed.

Biography:

Chairman of Mehr Medical group since 2005

Hereditary Pyropoikilocytosis: A rare cause of neonatal jaundice

Pravania Naicker*, Nischal Rao, and Dhivya Raajan

Whittington Health Trust, London, United Kingdom

Background: Hereditary pyropoikilocytosis (HPP) is a rare red blood cell (RBC) membrane disorder within the spectrum of hereditary elliptocytosis, caused by defects in cytoskeletal proteins such as alpha-spectrin, beta-spectrin, and protein 4.1R.^{1,2} These abnormalities result in RBC membrane instability, leading to haemolytic anaemia and neonatal jaundice.^{2,3} We report a case highlighting the diagnosis of HPP in a neonate.

Case Report: A one-month-old female infant presented with febrile illness secondary to COVID-19 infection. Initial investigations revealed severe haemolytic anaemia: haemoglobin 65 g/L, mean corpuscular volume 85.5 fL, and red cell distribution width 27.5%. Peripheral blood film showed hypochromia, microcytosis, polychromatic macrocytes, poikilocytosis, pencil cells, and red cell fragments. She was admitted for further evaluation and received a red cell transfusion. She was born at term following an uncomplicated pregnancy. Shortly after birth, she was admitted to the neonatal intensive care unit for pathological jaundice, with serum bilirubin levels exceeding the exchange transfusion threshold. No underlying cause was identified at that time; she responded to phototherapy. Family history included maternal G6PD deficiency and beta-thalassaemia trait, with no ABO incompatibility noted. Given the abnormal blood film, a RBC membrane gene panel was performed, identifying a pathogenic variant in the spectrin alpha erythrocytic 1 (SPTA1) gene, confirming hereditary pyropoikilocytosis. The infant was also found to be a heterozygous carrier for G6PD deficiency. She was managed supportively with blood transfusions, folic acid supplementation and was referred for haematology follow-up and genetic counselling.

Conclusion: RBC membrane disorders such as hereditary pyropoikilocytosis should be considered in neonates with early-onset jaundice and haemolytic anaemia, particularly when common causes such as ABO incompatibility and G6PD deficiency are excluded. Careful blood film examination is critical for diagnosis and guiding further investigation.

References:

1. More, T. A.; Kedar, P. Unravelling the Genetic and Phenotypic Heterogeneity of SPTA1 Gene Variants in Hereditary Elliptocytosis and Hereditary Pyropoikilocytosis Patients Using Next-Generation Sequencing. *Gene* 2022, 843, 146796. <https://doi.org/10.1016/j.gene.2022.146796>
2. Brancamp, R.; Hughes, C. E.; Dar, A.; Polic, A.; Zuckerwise, L. C.; Booth, G. S. Homozygous SPTA1-Associated Hereditary Pyropoikilocytosis Presenting as Hydrops Fetalis. *Transfusion* 2024, 64 (1), 189–193. <https://doi.org/10.1111/trf.17617>
3. Niss, O.; Chonat, S.; Dagaonkar, N.; et al. Genotype–Phenotype Correlations in Hereditary Elliptocytosis and Hereditary Pyropoikilocytosis. *Blood Cells Mol. Dis.* 2016, 61, 4–9. <https://doi.org/10.1016/j.bcmd.2016.07.00>

Biography:

Pravania Naicker is Junior Clinical Fellow in Paediatrics and Neonatology at Whittington Health Trust in London, United Kingdom. She graduated from the University of Cape Town in South Africa in 2020 and completed her internship at Groote Schuur Hospital and Red Cross War Memorial Children's Hospital in Cape Town, South Africa. Pravania has a special interest in quality improvement projects and research is actively involved with various projects at Whittington Health Trust.

Immunological tolerance acquisition after dietary elimination in mexican infants with cow's milk protein allergy mediated by ige

Hannia B. Matt Hernández*, Virginia María González Perez, and Víctor Soto Torres Septiem

Pediatrics Allergy and Clinical Immunology Dept

Christus Muguerza Reynosa Hospital, Mexico

Cow's milk protein allergy (CMPA) is one of the most common allergic diseases in infancy, with a global prevalence of 2%–7.5% in infants under one year^{1 2}. In Latin America, data remain limited, though comparable prevalence is observed⁵. Between 40%–60% of CMPA cases are IgE-mediated, with rapid-onset symptoms and positive skin prick test (SPT)³. Casein, α -lactalbumin, and β -lactoglobulin are the primary allergenic proteins, with casein being the most immunogenic and thermoresistant^{6 7}. CMPA, particularly its IgE-mediated form, is associated with the atopic march and later allergic diseases^{10–12}.

This retrospective study analyzed 285 Mexican infants diagnosed with IgE-mediated CMPA between 2 and 3 months of age. Infants were grouped by diet: exclusive breastfeeding with maternal dairy elimination, amino acid-based formula, or hydrolyzed rice formula. SPTs were performed at diagnosis and after 12 months. Oral food challenge (OFC) at 12 months determined tolerance. Dietary adherence and allergic comorbidities were recorded.

Clinical tolerance was achieved in 62.1% of infants. The highest tolerance rate was observed in breastfed infants (78.9%), followed by amino acid formula (68.4%) and rice formula (65.4%). SPT wheal sizes for all three proteins significantly decreased after 12 months. Adherence to diet was identified as the strongest predictor of tolerance (OR >100, $p < 0.001$). Although rice formula showed the highest adherence (76.8%), it yielded the lowest tolerance rate, suggesting a weaker immunomodulatory effect. Allergic comorbidities were significantly associated with reduced tolerance ($p < 0.001$), while family history of atopy was not statistically significant.

These results support the immunological benefit of breastfeeding and amino acid formulas in promoting tolerance. They highlight the importance of adherence and continued immunological evaluation in managing CMPA during early infancy.

References:

1. World Allergy Organ J 2023, 16, 296.
2. Arch Dis Child 2014, 99, 296.
3. BMJ 2013, 347, f5424.
4. WAO J 2010, 3, 57.
5. Arch Argent Pediatr 2012, 110, 202.
6. Rev Alerg Mex 2019, 66, 394.
7. Rev Cubana Pediatr 2016, 88, 123.
8. Acta Pediatr Mex 2024, 45, 38.
9. Rev Gastroenterol Mex 2022, 87, 46.
10. Pediatrics 2020, 145, e20192735.
11. J Allergy Clin Immunol 2015, 135, 349.
12. Ann Allergy Asthma Immunol 2008, 100, 278.

“ADHD / ASD and Neurodiversity”. Is it time to move on?

Richard Fry*

Dr Fry & Associates, Winchester, UK.

In this address he will discuss his journey to questioning the continued utility of the diagnoses “ADHD, ASD and Neurodiversity” and look at other ways of thinking about those presentations, which appear to be mushrooming to an almost endemic level.

We will look at the drivers, use and meaning of these diagnoses. We will also consider the inter-relationship between Paediatrics and Psychiatry in these conditions.

In the final section we will consider new ways of thinking about the problems these children have and how to address them.

Biography:

Dr Richard Fry is a Consultant Integrative Child Psychiatrist practicing in the private sector in the UK after many years leading services in the NHS. He also spent some years in General Practice. In the last fifteen years he has moved from purely mainstream and conventional approaches to a more holistic view of brain and emotional health. He now practices in an Integrative fashion trying to consider the drivers of these presentations and address them. The aim is to minimise the exclusive use of medications and to optimise their effects where they are used. Metabolic and genetic health and constitution are key to his thinking. He places great emphasis on working in collaboration with colleagues skilled in Physical Medicine as well as those with Psychological expertise.

WCPN-2025



VIRTUAL ABSTRACTS DAY-1

6th Edition - World Congress on
PEDIATRICS & NEONATOLOGY
Sep 25 - 26, 2025 at Berlin, Germany

Centering Children in Child Safeguarding

Kavita Ratna*

The Concerned for Working Children, India

Children and young people, especially the most vulnerable, bear the brunt of socio-political and economic gaps. They also lack of access to effective complaint mechanisms. We see these as a violation of the **Principle of the Best Interest of the Child**, children's right to participation, and all the other rights outlined in the **UN Convention on the Rights of the Child (UNCRC)**. Children are also denied accountability of the state and other duty bearers in their protection and nurturing - a fundamental premise of the rights discourse.

In this framework, this presentation will highlight the importance of reviewing and re-assessing our perceptions and views about multi-dimensional poverty which compels many young people to resort to labour – sometimes even choosing work over abysmal schooling systems. We will take a close look at the grassroots experiences of such children and adolescents, juxtaposed with the commitments of the state and global communities. It will also highlight the implications of these vis a vis their physical, emotional and mental wellbeing.

Working children and adolescents are, at best, placed at the receiving end of state and public attention. At worst, they are abused, neglected, and erased from meaningful policy and programs. From our work experience of over four decades, we are aware that while many of these young people face physical, emotional, and existential challenges, what they need first and foremost is to be **'seen' and 'heard'**. The presentation will cover why young people require respect and understanding; how their concerns must be prioritised; why responses from the state and civil society-including the medical community - must align with their individual narratives and how to ensure our responses are geared to uphold their rights and personhood.

Biography:

Kavita Ratna is the Executive Director of the Concerned for Working Children (CWC), which has been thrice nominated for the Nobel Peace Prize. Engaged with child rights since 1990, she holds numerous national and international portfolios, including membership of the UN Expert Group Committee on Children's Participation, the International Working Group on Child Labour (Asia), the International Interagency Steering Group on Monitoring and Evaluating Children's Participation, and a Board Membership of the Child Friendly Governance Project, Geneva. She has provided technical expertise to the Commonwealth, UNICEF, and other international and national agencies on child centered policy and programming.

New immune targets derived from platelets: PF4 & TGF- β

Shuai Tan^{1†*}, Huizhen He^{1†}, Yuxin Li^{1†}, Mingyue Shang¹, Yaofang Cao¹, Dongmei Zou¹, Ronghua Hu¹, Wuhan Hui¹, Xiaoli Chang¹, Jing Ni¹, Qiang Ma¹, Li Su¹, Jing Sun¹, Wanxue He², Yumeng Li¹, Yaochi Chen¹, Xingmin Feng^{3*} and Wanling Sun^{1*}

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Platelets are key factors in hemostasis and thrombosis, and are fully involved in immune response processes such as atherosclerosis. Immune cells such as T cells and macrophages are drivers of immune responses. An increasing number of studies have shown that crosstalk between platelets and immune cells is likely one of the critical bridges in inflammatory responses and immune responses. Among these, platelets release mediators such as PF4, TGF- β , and RANTES, which bind to T cell membrane receptors, activate downstream mitochondrial energy metabolism signaling pathways, affect nuclear transcription, and thereby regulate the differentiation and proliferation of immune cells—this is currently one of the strong hypotheses for platelet regulation of immune cells. AA is a bone marrow failure syndrome characterized by pancytopenia and decreased HSPCs in the bone marrow, having a strong immune background color, it can be either congenital or acquired, predominantly affecting adolescents and the elderly, with higher incidence in Asia compared to Europe and America. Current treatment options include allogeneic hematopoietic stem cell transplantation or immunosuppressive agents, yet proximately a third of patients fail to reach long-term survival. AA is primarily driven by immune-mediated destruction of HSPCs, initiated by self-activated T cells. Early stages feature a Th1 response, which later shifts to Th17 and effector memory CD8⁺ T cells. Key cytokines including IFN- γ and TNF- α play crucial roles in this immune dysregulation, influencing HSPCs and contributing to bone marrow failure. Furthermore, BMF, particularly M1 subtype, are implicated in AA via the TNF- α /TNF- α receptor pathway, leading to T cell activating and subsequent HSPC damage. Some studies suggest that platelet may modulate T cell responses through mechanisms such as Akt-PGC1 α -TFAM pathway or PF4-mediated activity, which could play a role in AA. However, direct evidence connecting platelet regulation to T cell-mediated HSPC damage is limited, and current research has largely focuses on CD8⁺ T cells. Moving forward, it is essential to investigate the interactions between platelets, CD4⁺ T cells, and mitochondrial energy metabolism. We propose that platelet-derived factors such as PF4 and TGF β may activate mitochondrial pathways, influencing T cell activation and leading to HSPC destruction in AA. For another immune background color disease, atherosclerosis, platelets are the main source of circulating TGF β and significantly regulate CD4⁺ T effector cell responses. However, the effect of platelet-derived TGF β on atherosclerosis is still unclear. Using CRISPR-Cas9/Cre LoxP gene editing technology, we constructed "PF4 Cre⁺- TGF - β 1^{f/f}" mice, and found that Platelet-specific TGF β deficiency aggravates atherosclerosis, vascular inflammation and hypercholesterolemia in mice. In summary, this hypothesis "Platelet-PF4/TGF β -Immune cells" could provide new insights into the molecular mechanisms and pave the way for novel therapeutic strategies.

Biography:

Shuai Tan MD PhD | Physician-Scientist Beijing High-Level Returned Overseas Talent; Beijing High Tech Innovation Program Spring Bud Project Scholar; Xuanwu Hospital Wisdom Convergence Project Scholar

Xuanwu Hospital, Capital Medical University (XWH-CMU), Beijing, China

Problems of most concern to parents of children with ADHD and emotional dysregulation in a randomized controlled trial of multinutrients: MADDY secondary outcome

Eugene Arnold L*

Nisonger Center Clinical Trials Program, USA

Objective: To objectively quantify parent observations of child's behavior and identify problems most amenable to change. In a randomized clinical trial (RCT) of children with attention-deficit/ hyperactivity disorder (ADHD), one primary outcome, blinded Clinical Global Impression-Improvement, showed a significant ($p=0.001$) advantage of multinutrients (54% responders) over placebo (18% responders) and the other, parent symptom ratings, showed significant improvement in both treatment groups, without significant difference between them. We examine a secondary outcome, blinded clinician ratings of parent-selected target problems.

Method: The multisite 8-week Micronutrients for ADHD in Youth (MADDY) RCT randomized 126 children ages 6-12 with ADHD and emotional dysregulation to broad-spectrum micronutrients ("multinutrients") vs. placebo. Parents were asked to describe their child's most concerning problem(s) at baseline; they quantified them by frequency, duration, impairment, and consequences. At subsequent visits, parents re-quantified the problem(s). Blinded child psychiatrists independently reviewed the PTPs and rated change at mid- and end-treatment, compared to baseline on a 9-point scale with 5=no change and 1=problem gone. PTPs were grouped into 9 categories. Mean ratings were compared between active and placebo groups and explored by category.

Results: By week 8, a significant separation favored multinutrients: 38% of the multinutrient group vs. 25% of the placebo group were "definitely improved" or better ($p=0.04$). Inattention (72.2%) and emotional dysregulation (69.1%) were the most frequent PTPs. Inattention and internalizing symptoms improved more with multinutrients than placebo ($p=0.01$, $d=0.55$; $p=0.03$, $d=0.80$, respectively). The multinutrient advantage was not significant for 7 other problems.

Conclusions: This secondary outcome found that multinutrients improved parental concerns significantly more than placebo overall, and especially in two: inattention and internalizing symptoms (anxiety/depression), but not significantly in hyperactivity/impulsivity, aggression, autistic symptoms, physiological symptoms, peer relationships, or emotional dysregulation/irritable oppositionality.

Keywords: ADHD, inattention, internalizing symptoms, multinutrients, micronutrients, parent reports, RCT

Location in early childhood of the permanent mandibular canine explain the “mysterious mandibular permanent canine” observed in puberty

Inger Kjaer*

University of Copenhagen, Denmark

Two studies in odontology document the importance of insight in embryology and fetal pathology for understanding clinical and radiological questions in the dentition.

The first study (Kjær et al. Eur Arch Paediatr Dent 2023;24(5):613-619) demonstrates how radiographs in early childhood can reveal abnormal location of initial crowns of permanent canines. In 31 orthopantomograms taken from children younger than 5 years of age, the early permanent mandibular crown was diagnosed. The indication for taking these radiographs was failure in primary molar eruption. This study demonstrates different malpositions of the initial permanent canine crowns, misplaced distally or mesially compared to the preceding primary canine.

The second study (Kjær et al. Eur Arch Paediatr Dent, accepted for publication 2025) is a radiographic study consisting of more than two radiographs taken at different times from 15 children at puberty. All individuals were diagnosed with permanent mandibular canines, initially located distally, compared to the site of the primary canine. One child appeared in both studies. Some of the canines erupted from the initial position in an upward direction and others in a horizontally and downward direction ending far from the primary canine, and even in the contralateral side of the mandible. These last mentioned canines are the so-called mysterious mandibular permanent canines with a formerly unknown etiology.

The study demonstrates that a mysterious mandibular canine originates from a permanent tooth bud, rotated and misplaced distally compared to the developing primary canine. In the embryologic period, the permanent tooth bud has developed from the mucosa of the primary canine. This fetal pathological event caused the abnormal eruption path of the mysterious canine observed in the clinic.

Biography:

Inger Kjær. Professor emerita in orthodontics, Specialist in orthodontics, Dr Odont et Dr Med. Research profile: Embryology and fetal pathology as a biological base for understanding postnatal normal and pathological development. Consultant at the University Hospital in rare diseases in the craniofacial area.

Prevention of NICU Admission Hypothermia in Moderate-and Late-Preterm Infants

Amanda E. Winkler^{a*}, Joseph H. Chou^a, Elizabeth West^b, Carolyn Bleilera, Janet Bell^a, Jeanne Gilbert^a, Christina M. Duzyj^b, Sergei Roumiantsev^c, Paul H. Lerou^a, Megan Aurora^a.

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Background: Neonatal hypothermia is a common and preventable cause of neonatal morbidity and mortality. While hypothermia prevention has been extensively studied in infants < 32 weeks' gestation, few studies have targeted moderate- and late-preterm infants (MLPI) in the delivery room (DR).

Methods: This quality improvement initiative was conducted from June 2019 to June 2023 at the Massachusetts General Hospital Neonatal Intensive Care Unit (NICU) and Labor and Delivery Unit. All inborn MLPIs, 32 + 0/7 to 36 + 6/7 weeks' gestation admitted to the NICU were included. We expanded thermoregulatory measures typically used in protocols for infants < 32 weeks' gestation including increased DR ambient temperature to 74°F and thermal mattress use. The primary outcome was hypothermia (< 36.5°C upon NICU admission). The balancing measure was hyperthermia (≥38°C).

Results: During the study period, there were 566 inborn MLPIs with a mean gestational age of 34 + 3/7 weeks and a mean birth weight of 2269 g. Special cause variation in neonatal hypothermia incidence was observed with a decrease from a mean baseline of 27% to 7.8% post-intervention. Special cause variation was observed in hyperthermia incidence with an increase from 1.4% to 6.2% post-intervention largely initially associated with noncompliance with protocol for thermal mattress removal.

Conclusions: Expansion of several thermoregulation techniques commonly used in infants < 32 weeks' gestation, particularly thermal mattress use, was associated with decreased incidence of NICU admission hypothermia in MLPIs with an increase in mild hyperthermia predominantly associated with concomitant polyethylene wrap use.

Biography:

Dr. Amanda E. Winkler is a neonatologist and physician-scientist at Massachusetts General Hospital in Boston, Massachusetts, USA. She specializes in clinical translation of medical devices to optimize patient outcomes in neonates. Her work in quality improvement in the use of exothermic mattresses in the delivery room management of moderate- and late-preterm infants has led to significant improvements in thermoregulatory management in this population.

Magnetic Resonance Imaging (MRI) Findings in Pediatric Autoimmune Encephalitis: A Case Report

Amira E Raslan*

Dallah Hospitals, Saudi Arabia

Autoimmune encephalitis (AE) is a rare condition that involves an immune-mediated attack on the brain, often presenting with seizures, altered consciousness, and other neuropsychiatric symptoms. This case report describes a seven-year-old girl who presented with acute brain dysfunction, including seizures and disturbed consciousness, following a gastrointestinal illness. Magnetic resonance imaging (MRI) findings revealed early signs of AE, including hyperintensities in the external capsules and medial temporal lobe. Following negative infectious workups, the diagnosis of AE was confirmed, and the patient was treated with immunomodulatory therapy, leading to gradual improvement. This case emphasizes the importance of early recognition, the role of MRI in diagnosis, and the need for prompt treatment to improve outcomes in pediatric AE.

Biography:

Amira Raslan is a radiologist completed 13 years of experience in interpreting medical images, I has residency program in Tanta cancer center in Egypt, and Master degree in transcranial US in neonates and passed first two parts of fellowship of royal collage of radiologists, I am working now in Dallah hospitals in Saudi Arabia as radiology registrar and have great interest in pediatric and women imaging.

Mistakes are not an option: correlates of anxiety and depression in pediatricians in training

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Introduction: Pediatricians in training are a population at risk for anxiety and depression: they face long working hours; they are confronted with the suffering and death of a vulnerable population and may have personal characteristics that put them in danger¹⁻². Nonetheless, few studies have looked at their prevalence and associated factors. We aimed to identify demographic, professional activities, adversities and perfectionism features related to the presence of anxiety and depression in pediatricians in training. **Methods:** As part of a cross-sectional study, pediatricians in training took part in an online survey from October 2019 to April 2021 where demographic information, professional activities and adversities, perfectionism, depression, and anxiety were evaluated³. **Results:** 936 pediatric residents answered the survey. 11.6% reported having depression and 20% anxiety. Being single, a history of anxious or depressive episodes, being the resident in charge of a patient who died, aggressions from colleagues and a high concern for errors were associated with current depression or anxiety. **Discussion:** Screening for depressive and anxious symptoms should be routinely performed from entry to the residency to favor early intervention⁴. A shift from promoting perfectionism to a more compassionate training could serve a population facing so many adversities⁵.

Biography:

Dr. Ana Fresán is Doctor in Psychology and Health from the National Autonomous University of Mexico (UNAM), Head of the Clinical Epidemiology Laboratory and Medical Sciences Researcher in the Clinical Research Directorate at the National Institute of Psychiatry in Mexico City. She has received several awards for her research contributions. She is also a tutor for the specialty in Psychiatry, Master's and Doctorate in Medical Sciences and Psychology at the National Autonomous University of Mexico. Her main areas of research are aggressive behavior, stigma in mental disorders, and the physical and mental health of physicians. She has more than 280 presentations at national and international conferences and meetings, more than 200 scientific articles and 40 book chapters. Her academic work currently has more than 4,000 bibliographical citations. She also is an Honorary Member of the Mexican Psychiatric Association.

Children had increased risks of impaired motor and visual-motor skills after prenatal exposure to opioid maintenance therapy

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Aim: Preschool children prenatally exposed to opioid maintenance therapy (OMT) have an increased risk of neurodevelopmental impairments. We aimed to investigate long-term motor and visual-motor integration outcome in children aged 5–13 Years, born to mothers in OMT.

Methods: From January 2018 to June 2021, 63 children prenatally exposed to OMT and 63 comparison children matched for age and gender, were examined at two Norwegian hospitals. Motor skills were assessed by the Movement-ABC test and visual-motor integration by the Beery VMI test. A motor function neurological assessment test was used to examine neuromotor soft signs.

Results: In the OMT-exposed group, 16% had motor impairment, 35% had motor problems and 19% had visual-motor integration problems. Forty-three percent of the exposed children had neuromotor soft signs. Strabismus had some influence on motor and visual-motor outcomes but could not explain the group differences.

Conclusion: Children prenatally exposed to opioid maintenance therapy have an increased risk of long-term motor impairment and visual-motor problems. In addition, they exhibit significantly more neuromotor soft signs, which may affect general well-being, leisure activities and school performance.

Long COVID and Hypermobility have shared pathophysiology

Bala Munipalli*

Mayo Clinic, USA

Hypermobility spectrum disorders (HSD) and hypermobile Ehlers–Danlos syndrome (hEDS) are the most common joint hypermobility conditions encountered by physicians, with hypermobile and classical EDS accounting for >90% of all cases. Hypermobility has been detected in up to 30–57% of patients with myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS), fibromyalgia, postural orthostatic tachycardia syndrome (POTS), and long COVID (LC) compared to the general population. Extrapulmonary symptoms, including musculoskeletal pain, dysautonomia disorders, cognitive disorders, and fatigue, are seen in both LC and HSD. Additionally, ME/CFS has overlapping symptoms with those seen in HSD. Mast cell activation and degranulation occurring in both LC and ME/CFS may result in hyperinflammation and damage to connective tissue in these patients, thereby inducing hypermobility. Persistent inflammation may result in the development or worsening of HSD. Hence, screening for hypermobility and other related conditions including fibromyalgia, POTS, ME/ CFS, chronic pain conditions, joint pain, and myalgia is essential for individuals experiencing LC. Pharmacological treatments should be symptom-focused and geared to a patient's presentation. Paced exercise, massage, yoga, and meditation may also provide benefits.

Tinea in Tots

Bernard A Cohen

Johns Hopkins School of Medicine

We will discuss the management of tinea in tots with a review of dermatophyte infections in children and a focus on tinea capitis. Until recently there were no specific guidelines for the management of tinea capitis in children which prompted our survey study of pediatric dermatologists on the management of children with this infection. The study was supported by the Pediatric Dermatology Research Alliance, and the results and recommendations for treatment were published in the Journal of Pediatrics in 2021. We will also include results from other studies we completed on dermatophyte infections in children.

Biography:

Dr. Cohen is Professor of Pediatrics and Dermatology at the Johns Hopkins University School of Medicine. He is Pediatric Dermatology Editor for Contemporary Pediatrics and a member of the Transitional Retirement Program focusing on teaching, clinical studies, and publications with students and residents.

Metagenomic approach to renal failure in children

Berrahal Mounir*

Microbiology medical; University of Algiers 01.Algeria

Purpose: The study of the urinary microbiota makes it possible to establish the epidemiological of urolithiasis infected or colonized sometimes by multiresistant and cunning microorganisms necessary in the prevention of infectious or functional complications and control his clinical recurrence.

Methods and materials: Culture and antibiotic sensitivity tests from microorganisms in the stone kidney after fragmentation have been realized. A direct metagenomic ribosome 16 S V3-V4 PCR in EP 2x300 LP (one MiSeq illumine) of the intraoperative pyelic pyuria urine has been analyzed.

Results: Culture of the stone returns positive at bacterial urealytic positive: *Morganella morganii* is sensitive to the calforan, imipenem, and resistant to Cefazolin, Augmentin, and ampicillin. After pejorative evolution of the patient towards the multiple abscess of the left kidney and acute renal insufficiency with 180 creatinin micromoles and contracting and calcifying urétérale and fibrin to be objectively measured by the surgeon urologist. A direct metagenomic analysis of intraoperative pyelic pyuria and bioinformatics software includes four algorithms for preprocessing: CleanPrimer, Slice Genome, PCR Clean, and Sample Size. It included a single metagenomic; data simulation algorithm, simulateR for error and correction, and the processing to correct errors in Next Generation. Sequencing data included eMER-kmer process coverage. Analysis gave the results after parental consent according to mainly Entérobactéries (60.68%), and Anaerobic bacterium of the Bacteroides and others.

Conclusion: From this genomic experience, we will have to review the threshold microorganisms and especially the place of Anaerobes or emerging agents by a genomic multicenter study of the urinary microbiota of patients with infection urolithiasis to have better manage this public health pathology.

Key words: urinary microbiota, urolithiasis infected; children; renal failure; metagenomic analysis; Anaerobic

A Hidden Epidemic: Complications of Insufficient Feeding in Term Breastfed Neonates

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Fed Is Best Foundation, CHI St. Vincent Little Rock, United States

Medical complications of insufficient feeding in breastfed newborns, namely jaundice, dehydration, and hypoglycemia, have steadily increased in incidence—an unintended consequence of efforts to promote exclusive breastfeeding before hospital discharge, a key metric of the Baby-Friendly Hospital Initiative. This lecture reviews the case of a term newborn infant who had evidence of poor intake while exclusively breastfeeding during his hospital stay that were not recognized by his health providers. The infant was discharged home and was subsequently found to be in cardiac arrest 12 hours after hospital discharge due to hypernatremic dehydration. Although he was resuscitated and placed on life support, he ultimately died at 19 days of age due to extensive brain injury from hypernatremic dehydration leading to hypoxic-ischemic encephalopathy and cardiac arrest. This lecture highlights multiple pitfalls of current perceptions of normal vs. abnormal newborn feeding behavior, weight loss percentages, elimination patterns, and clinical thresholds widely believed to be safe for neonates. It discusses newer data showing how common insufficient feeding complications occur in term breastfed newborns even at weight loss percentages previously deemed normal by most health professionals and hospital protocols. It also reviews newer data on the clinical and laboratory thresholds at which neonatal brain injury, developmental delays, and permanent disabilities occur. Lastly, this lecture reviews in-hospital strategies to monitor and prevent excessive weight loss and other comorbid conditions in response to signs of inadequate newborn feeding that have the potential to prevent extended and repeat admissions, neonatal brain injury, long-term disability, and rare deaths.

Biography:

Dr. Christie del Castillo-Hegyi is an American board-certified emergency physician who obtained her medical degree from University of California, San Francisco and completed her residency training in emergency medicine at the University of New Mexico, U.S.A. She is a cofounder of the Fed Is Best Foundation, a non-profit organization dedicated to advocating for hospital and global health policy reforms on infant feeding management and providing parent and health professional education to prevent complications of insufficient infant feeding that result in brain injury, neurodevelopmental delays, disabilities, and rare deaths. She is a member of the Global Development Disability Research Collaborators and has co-authored papers on hypernatremic dehydration and the global rise in childhood disabilities.

The relationship between headache, mood, and functional disability

Christina Murphy*

Children's Hospital of Philadelphia, USA

Objective: To explore the relationship between anxiety, depression, and headache-related disability in a pediatric clinic-based retrospective cross-sectional study.

Background: Anxiety and depression are commonly considered comorbidities of headache disorders and frequently seen in children and adolescents. All these conditions can contribute to disability and may have a cumulative impact. In this study we tested whether self-reported anxiety and/or depression in youth were associated with headache-related disability.

Methods: This was a retrospective cross-sectional study of children ages 6-17 years old who completed a headache intake questionnaire at the time of new outpatient neurology visit. Those who reported on behavioral health symptoms, involvement of a behavioral health provider (i.e., yes/no), and the PedMIDAS (a validated metric of headache-related disability) were included. The relationship between anxiety and/or depression and headache-related disability was examined.

Results: Of the 12,660 questionnaires queried, 9,118 met criteria for inclusion. Respondents were 64.0% female and had a median age of 13.5 [IQR 10.3, 15.7]. Compared to patients without self-reported anxiety/depression, patients with anxiety and depression reported higher headache-related disability ($M = 17.0$, [IQR 6.0, 41.0]) even after accounting for covariates (estimated difference = 6.0, 95% CI [4.4 - 7.5]). For participants with anxiety and/or depression, having a behavioral health provider was associated with greater headache-related disability (estimated difference = 7.0; 95% CI 4.6 - 9.3).

Conclusions: Patients with self-reported anxiety and/or depression reported higher headache-related disability. Having a behavioral health provider was associated with greater headache-related disability, indicating the complexity and high level of need for this population. Further research is needed to understand the directionality of these results; however, patients with headache as well as depression and or anxiety are a vulnerable group who may benefit from an integrated care model.

Biography:

Christina Murphy, PhD is a pediatric psychologist in the Headache Program, within the Division of Neurology at the Children's Hospital of Philadelphia and an Assistant Professor of Clinical Psychiatry at the Perelman School of Medicine, University of Pennsylvania. Dr. Murphy's clinical and research interests include assessment and treatment of pediatric headache, cognitive behavior therapy for headache management, risk and resilience among pediatric patients with headache, and quality improvement in the division of neurology.

Unusual case of *Plesiomonas shigelloides* sepsis and meningitis in a neonate with targetoid vasculitic rash

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A female infant weighing 2840-g was born vaginally after 36 weeks of uncomplicated pregnancy. In 36th hour of life newborn appeared septic and physical examination revealed vasculitic rash. Well-defined annular erythematous patches of different size (5-15mm) with targetoid shape and central hyperpigmented purple duskiness and red to pink border spread over feet, knees, abdomen, neck and chin (Figure 1) were noticed. Upon admission to neonatal intensive care unit (NICU), the infant's temperature was 37.1°C, heart rate 146 beats/min, respiratory rate 46/min and mean arterial blood pressure was 38 mmHg. Laboratory findings showed leukopenia with leukocyte count of $2.2 \times 10^9/L$ and mild neutropenia with an absolute neutrophil count (ANC) of 1386 cells/ μL , C-reactive protein was 35.5 mg/L, prothrombin time 0.42 seconds, and activated partial thromboplastin time 36.2 seconds. Empirical antibiotic treatment with intravenous ampicillin (100 mg/kg, every 8 hours) and cefotaxime (50 mg/kg/day) was instituted. Due to altered coagulation parameters and skin lesions resembling purpura fulminans fresh frozen plasma was given. Because of development of respiratory insufficiency newborn was supported by nasal continuous positive airway pressure ventilation. After initial stabilization, lumbar puncture was performed. Cerebrospinal fluid leukocyte count was $584 \times 10^9/L$ (492 neutrophils). Glucose and protein concentrations were 2.1 mmol/L and 0.95 g/L. Gram-negative bacterium with coliform morphology was detected in blood culture, while CSF remained sterile. Bacterium was identified as *Plesiomonas shigelloides* by MALDI-TOF MS, Bruker analysis. Susceptibility to meropenem, cefotaxime, ceftazidime, ceftriaxone, piperacillin/tazobactam and cefepime was established, with resistance to ampicillin and amoxicillin/clavulanic acid. Fecal culture taken from mother three days postpartum was also positive for *P.shigelloides*. Interestingly, mother had no evident symptoms of acute infection. Due to deterioration in clinical appearance and unpredictable clinical course, dual antibiotic therapy was changed to rescue therapy with intravenous meropenem (40 mg/kg, every 8 hours). Newborn was treated with meropenem for 21 days. Brain magnetic resonance image carried out after therapy showed normal finding. Patient was discharged from the hospital with no evident sequelae in neurological and clinical appearance.

Involvement of USP7 in aggravating Kawasaki disease by promoting TGFβ2 signaling mediated endothelial-mesenchymal transition and coronary artery remodeling

Guanghui Qian*

Children's Hospital of Soochow University, China

Kawasaki disease (KD), characterized by systematic vasculitis, is a leading cause of pediatric heart disease. Although recent studies have highlighted the critical role of deubiquitinases in vascular pathophysiology, their specific contribution to KD remains largely unknown. Herein, we investigated the function of the deubiquitinase USP7 in both KD patients and a CAWS-induced KD murine model. USP7 expression level is increased both in HCAECs induced by KD sera and cardiac CD31+ endothelial cells of KD mice. Whereas knockout of USP7 in-creses the cellular proportion of endothelial cells and potentially attenuates the elevated EndoMT, fibrosis, and inflammation in cardiac tissue of KD mice, consistently with the in vitro experiment observed in HCAECs induced by TGF-β2. Mechanistically, USP7 interacts with SMAD2/3, enhancing their protein stability by removing the K48 ubiquitin chain from both proteins and preventing their proteasome degradation, thus increasing the p-SMAD2 levels and nuclear entry. Importantly, intraperitoneal injection of USP7 inhibitor, P22077 elicited a robust anti-EndoMT and anti-vascular inflammation effect in KD model mice. Therefore, our study uncovered a previously unrecognized function of increased USP7 in KD by augmenting TGFβ2/SMAD2/SMAD3 signaling, thus facilitating the transcription of genes implicated in the EndoMT, cardiac fibrosis, and vascular remodeling. Our finding suggests that USP7 could serve as a potential therapeutic target for the prevention and treatment of coronary artery lesions in KD and related vascular diseases.

Biography:

Guanghui Qian has a Personal Chair in Genetics at Soochow University of China. He is a Principal Investigator at Institute of Pediatric Research at Children's Hospital of Soochow University. He has dedicated research career to the study of the molecular mechanisms that regulate IFN signaling- mediated antiviral function and Kawasaki disease (KD), which is mainly characterized by systematic immune vasculitis.

The pulse oximeter as a source for racial disparities, how we go here, updates from the ISO and path towards never again

Howard Saft*

National Jewish Health, University of Colorado School of Medicine, United States

The pulse oximeter is an important device that provides, what many consider to be a vital sign. Since its development, evidence suggested that its readings include a racial/skin color bias. This bias only became more widely known since the COVID pandemic. This biased reading resulted in skin color biased medical-decision making leading to racial disparities. Currently, the International Organization for Standardization (ISO) and the United States Food and Drug Administration (FDA) are in the process of updating their regulatory guidance. Similar to understanding disease mechanisms to develop treatments, we should do the same for such a source of systemic skin color biased care. The speaker will (1) provide an overview of how the pulse oximeter works, (2) summarize key literature demonstrating skin color bias including pediatrics and neonatology, (3) describe the gaps in regulatory policy and practice (4) provide an update (expected Summer of 2025) of regulatory body agencies by the ISO and U.S. FDA with implications and (5) present a pathway to solving this issue and framework for preventing similar sources of disparities.

Biography:

Howard Saft MD, MS is an Associate Professor at National Jewish Health in the United States. He currently serves as a U.S. Expert to the International Organization for Standardization for the pulse oximeter panel. Dr. Saft is boarded in Clinical Informatics, Internal Medicine, Pulmonary and Critical Care Medicine. He completed postdoctoral training in health services research which included policy research, medical anthropology related to socio-technical systems and patient safety. He currently services on the Society of Critical Care Medicine's Diversity, Equity and Inclusion committee.

Promotion in Place: A Model for Competency-Based Time-Variable Graduate Medical Education

John Patrick T. Co*

Mass General Brigham and Harvard Medical School

Boston, Massachusetts, United States of America

Graduate medical education has historically been time based, leading to variable and sub-optimal learner outcomes. Models that allow for competency-based advancement are needed to better ensure that learners are prepared before entering independent practice. In the United States, there are several barriers for implementing competency-based time-variable (CBTV) graduate medical education (GME), including reliance on residents for providing patient care, hospital credentialing, medical licensing, malpractice insurance, as well as regulatory standards. Promotion in Place (PIP) is a model of CBTV in GME that addresses these challenges.¹

We implemented the PIP model in the Massachusetts General Hospital Pathology Residency Program starting in academic year 2020-21.² Since then, each year there have been residents who were deemed ready for independent practice before the standard time of residency training as defined by the Accreditation Council for Graduate Medical Education (ACGME) and the American Board of Pathology. To support the program implementation of PIP, we developed several tools that can be utilized by other programs and specialties, including a template for decision making, Clinical Competency Committee (CCC) simulation guide, Common Program Commitments, guide for semi-structured discussion with residents interested in PIP, a Frequently Asked Questions document, and Process Map for gaining clarity and shared mental model between the trainee, involved faculty, and program as to how the resident would spend their time in sheltered independence, including length of time, clinical service on which the resident would be working, and faculty that would be available for help. We are implementing the PIP model in two other specialties in the United States.

Biography:

John Patrick T. Co is Vice President of Education at Mass General Brigham and Associate Professor of Pediatrics at Harvard Medical School. He is a Principal Investigator of an American Medical Association Reimagining Residency Grant, Promotion in Place, to develop, implement, and evaluate a model for competency-based time-variable advancement in graduate medical education.

References:

1. Goldhamer, MEJ, Pusic MV, Nadel ES, Co JPT, Weinstein DF. Promotion in Place: A Model for Competency-Based, Time-Variable Graduate Medical Education. *Academic Medicine*. January 29, 2024.
2. Martinez-Lage M, Goldhamer MEJ, Pusic M, Branda JA, Weinstein DF, Black-Schaeffer WS, Co JPT. Implementing the Promotion in Place Model of Competency-Based Time-Variable Graduate Medical Education in Pathology. *J Grad Med Educ*. 1 May 2025; 17 (2s): 64–71.

Machine learning risk stratification for high-risk infant follow-up clinic

Katie Carlton*

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Term and late preterm infants cared for in the neonatal intensive care unit (NICU) are not consistently referred to high-risk infant follow-up programs at discharge. Infants ≥ 34 weeks gestation without congenital anomalies are presumed to be at low risk for developmental impairment, and therefore, they do not qualify for high-risk infant follow-up based on prematurity alone. However, NICU hospitalization and the accompanying life-sustaining interventions increase the risk for developmental delays. Therefore, we aimed to identify clinical NICU factors associated with abnormal developmental screening and develop a risk-stratification model using machine learning for high-risk infant follow-up enrollment. To accomplish this goal, we performed a retrospective cohort study identifying abnormal developmental screening prior to 6 years of age in infants born ≥ 34 weeks gestation admitted to a United States level IV NICU. Five machine learning models were developed by classification and regression tree (CART), random forest, gradient boosting TreeNet, multivariate adaptive regression splines (MARS), and regularized logistic regression analysis using NICU predictors. Performance metrics included sensitivity, specificity, accuracy, precision, and area under the receiver operating curve (AUC). Within our 1355 patient cohort, 87% received developmental screening, and 47% had abnormal results. NICU predictors across all models were oral feeding, follow-up appointments, and medications prescribed at NICU discharge. Each model resulted in an AUC >0.7 , specificity $>70\%$, and sensitivity $>60\%$. Therefore, stratification of developmental risk in infants ≥ 34 week is possible utilizing machine learning methodology. These machine learning models, specifically the CART decision tree, will be used to revise high-risk infant follow-up enrollment criteria used at time of NICU discharge.

Biography:

Dr. Katie Carlton is an Assistant Professor of Pediatrics in the Division of Neonatology at the Medical College of Wisconsin, Milwaukee, WI, USA. She earned her medical degree at the Medical College of Wisconsin and completed both her pediatric residency and neonatal and perinatal medicine fellowship at the Medical College of Wisconsin and Children's Wisconsin. Her research focuses on developmental outcomes of infants cared for in the neonatal intensive care unit (NICU), with a focus on utilizing machine learning and artificial intelligence to optimize high-risk infant follow-up after NICU discharge. This project was supported by the NIH (UL1TR001436, TL1TR001437).

Molybdenum Cofactor Deficiency with A Novel Pathogenic Variant in the Molybdenum Cofactor Synthesis 1 (MOCS1): A Case Report

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Molybdenum Cofactor Deficiency (MOCOD) is a rare often fatal autosomal recessive disorder. We report a case of genetically confirmed MOCOD in an infant presenting with refractory neonatal seizures, with a family history of Dandy-Walker malformation (DWM). A term baby girl was the second born to consanguineous parents. Her elder sister had spasticity, developmental delay, seizures, and DWM, passed away at 8-years-old. On day two of life, the baby developed respiratory distress, recurrent apneic attacks, poor sucking, feeding difficulties, and refractory generalized tonic-clonic convulsions.

CT brain revealed global cerebral ischemia, and EEG was normal. Biochemical tests showed hypouricaemic hypouricosuria with a serum uric acid level of 17 $\mu\text{mol/L}$ (119-327), and uric acid to creatinine ratio (spot urine sample) of 0.1 $\mu\text{mol}:\mu\text{mol}$ (0.7-1.5). In the presence of neurological manifestations, hypouricaemic hypouricosuria, potentially indicative of MOCOD. Further analysis revealed increased plasma xanthine 70 $\mu\text{mol/L}$ and hypoxanthine 542 $\mu\text{mol/L}$ with elevated urine xanthine 0.202 mmol/L (0.01-0.09), though urine S-sulphocysteine levels were normal 2 $\mu\text{m/mM}$ (0-10).

Unfortunately, the baby's condition deteriorated further, and passed away on day 15 of life.

All exons and flanking intronic regions of the MOCS1 and MOCS2 were amplified and analyzed by PCR-Sanger sequencing from genomic DNA. The patient was homozygous for a novel variant MOCS1 c.1807G>C, p.(Ala598Pro) in exon 10 which was consistent with the MOCOD phenotype which has not been reported in the literature.

This case underscores the importance of early biochemical and genetic testing for MOCOD. Uric acid screening is a cost-effective tool for identifying neonates at risk for MOCOD.

MOCOD type A is now treatable, starting Cyclic pyranopterin monophosphate (cPMP) substitution early yields rapid and sustained biochemical improvement, enhancing neurodevelopmental outcomes.

Clinical and surgical physician's perception of nutrition knowledge

María Belén Ocampo Ordóñez*

Universidad San Francisco de Quito, Ecuador

Background: Due to the significant increase in the prevalence of food-related diseases, the value that physicians place on nutritional advice may have implications for patient treatment. The objective of this study was to evaluate the perception of the importance of nutritional intervention among physicians in the Universidad San Francisco de Quito's (USFQ) healthcare system.

Methods: This cross-sectional study employed a telephone survey administered to a subset of all medical doctors (MDs) working in the healthcare system clinics of USFQ between 2021 and 2022. Study participants were recruited through voluntary response sample from a complete list of 253 MD. The single time questionnaire consisted of a 22-item validated survey in which attitudes, self-perceived capacity, and knowledge about nutrition of medical doctors were evaluated. Data was analyzed using descriptive statistics, two-sided t test, bivariate associations and linear and logistic regressions.

Results: 136 MDs completed the survey yielding a response rate of 54%. Our analysis grouped participants into clinical (CE) and non-clinical specialties, hereafter referred to as surgical MDs. While a higher percentage of physicians in CE are confident in their ability to provide examples of recommended food portions based on national or international guidelines, 1 in 10 do not know how to use and interpret BMI or waist circumference, and around 1 in 3 do not know how many calories there are in one gram of fat, protein, or carbohydrates, and their basic metabolic functions. Almost all survey participants believe MDs can have an impact on the eating behavior of a patient if time is used to discuss the problem, however, almost half of survey participants believe nutrition counseling is not an effective use of time.

Conclusion: It is important to explore the perceptions and self-confidence of physicians around nutrition related issues. Our results demonstrated that nearly 1 in 4 surgical MDs do not feel capable of recognizing nutritional risk in patients, which highlights the essentiality of physicians having an updated understanding of basic nutrition principles.

Future research should examine how commonly MDs refer patients to

nutritionists/dietitians, as well as strategies for improving physician knowledge on basic nutrition concepts.

Biography:

María Belén Ocampo holds a bachelor's degree in Human Nutrition from the Universidad San Francisco de Quito (USFQ) and a master's degree in Public Health from the University of North Carolina at Chapel Hill. She is also a registered Dietitian nutritionist with the American Academy of Nutrition and Dietetics. Her research focuses on dietitian nutritionist training, as well as infant feeding and nutrition education.

Ethical Dilemmas on the Limits of Viability of Perivable Fetuses

Michael H. Malloy*

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Shaeffer and Avery's textbook, *Diseases of the Newborn* (1971) estimated the limit of viability to be around 28 weeks gestation and/or 1,000 grams. In the United States in 2022 there were 382 infants who survived out of 1576 born at 22 weeks gestation (24% survival for 22 weeks). Clearly the "limit of viability" is a moving target and the acceptable risk of intervening to attempt to "save" these perivable fetuses is a value judgement and not one that can be answered by science. With the possibility of the implementation of human trials using an artificial placenta or womb on the imminent horizon one must pause to ask, is there any moral quandary to using these advanced technologies or does the research/technologic imperative always outweigh any moral imperatives that might question the appropriateness of attempting to push the limits of viability to ever lower stages of gestation?

The ethics of using these technologies in perivable fetuses are challenging at best and dystopian at worst. Aldous Huxley in his dystopian novel, *Brave New World* (1932) foresaw the problems that arise when society controls reproduction. Should this new technology be promoted as a life-saving opportunity or conveyed as an experimental technology the results of which are uncertain? Vulnerable parents confronted with the loss of a perivable fetus present a challenging problem to obtain truly informed consent. Classical ethical concepts reduce ultimately to truth-telling and value judgements.

Reconciling the dilemma of pursuing the research/technologic imperative vs the moral imperative of questioning the virtue of pushing the limits of viability ever downward will not be easily resolved. Nevertheless, in considering this dilemma one must take into consideration not only the sanctity of life, but also the cultural, social justice, financial justice, and religious implications of the ever-moving target of the limits of viability.

Biography:

Michael H. Malloy is a Professor in the Department of Pediatrics/Division Neonatology and is the Emeritus Chair of Oslerian Education at the University of Texas Medical Branch in Galveston, Texas. He has over 40 years of clinical experience caring for infants in the neonatal intensive care unit. He has a Master of Science in Epidemiology and a Certificate in the History of Medicine and has spent the last several years studying the issues surrounding the limits of viability of extremely preterm infants and the factors influencing the cultural designation of when a conceptus acquires the status of an independent being.

The Role of Faculty of Medicine University of Gezira Students in improving maternal and child health among Families through primary health care centers practice and family medicine course at Wad Medani City, Gezira State, Sudan, 2023.

Mustafa Abdullah Alhamed*

University of Gezira, Yemen

Introduction: University of Gezira through its community based and socially accountable program plays an active role in improving maternal and child health in families at Wad Medani city. Through their PHC Health Centre Practice and family Medicine course, the students link the families with primary health care centers where they were trained.

Objective: To highlight the role of students in improving maternal and child health in families at Wad Medani city.

Methods: Cross sectional survey using a pretested questionnaire interviewing the mothers in the catchment area of the health centers.

Results: 120 families were studied. Different maternal and child health status were addressed. Under five children: were in 80 families. The students detected incomplete vaccination in 25%, all of them were fully vaccinated and weighed them using the road to health chart. The students advised them how to feed their children according to age. Malaria, pneumonia and diarrhea were detected among under five children. The students send them to the health centers, followed them at home, educated them regarding prevention and control of these diseases. The complicated cases were referred to hospital.

Pregnant women: Were 30%. Students provided health education about ANC and told them about the frequency of ANC visits. Vaccinated them against tetanus. Educated them to prepare and described the appropriate attachment and techniques of breast feeding. Educated them about Personal hygiene.

Conclusion: The students provided services regarding maternal and child health.

Recommendations: continuation of this program and further large-scale studies should be done.

Biography:

Mustafa Abdullah Alhamed, MBBS, University of Gezira in Sudan.

Secretion Movement During Mechanical Ventilation Superimposed on Intrapulmonary Percussive Ventilation (IPV) Versus Continuous High-Frequency Oscillation (CHFO): A Bench Study

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Cincinnati Children's Hospital, United States

This bench study evaluates secretion movement, pressure and volume delivery, and patient-ventilator interaction during pediatric mechanical ventilation superimposed with high-frequency assisted airway clearance devices: intrapulmonary percussive ventilation (IPV) and continuous high-frequency oscillation (CHFO). Using a simulated pediatric patient model, the study found that both IPV and CHFO led to less distal secretion movement compared to conventional ventilation (CV) only. The CV-IPV group showed higher peak inspiratory pressure (PIP) and mean airway pressure (MAP) compared to the CV-CHFO group, indicating better alveolar recruitment. However, the CV-CHFO group had a higher asynchrony index, suggesting more patient-ventilator asynchronies. Both IPV and CHFO effectively loosened secretions, with IPV showing a wider range of secretion dispersion. These findings suggest that superimposing high-frequency airway clearance devices on mechanical ventilation can enhance secretion clearance and alveolar recruitment in pediatric patients. This study provides valuable insights into the application of high-frequency assisted airway clearance devices in pediatric mechanical ventilation, highlighting the benefits and potential drawbacks of IPV and CHFO therapies. Further research is needed to optimize these techniques for clinical use and to address the challenges associated with patient-ventilator asynchronies.

Biography:

Ping-Hui Liu holds multiple degrees, including a Master of Science in Sports Medicine and a Master of Science in Respiratory care, and she is currently a Research Respiratory Therapist at Cincinnati Children's Hospital, USA and a Doctoral Resesarcher in Dr. Michael Riley, Professor in College of Allied Health Science at University of Cincinnati, USA. She has extensive professional experience as an Respiratory Therapist at Kaohsiung Chang Gung Memorial Hospital, Cleveland Clinic and Cincinnati Children's Hospital, and has received awards for her contributions to respiratory care. Liu is actively involved in bench studies and clinical researches in respiratory care.

Impact of Pathogenic C1GALT1C1 Variants on Extended O-Glycan Functions

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Cosmc (encoded by C1GALT1C1) is an ER-localized specific molecular chaperone for T-synthase, and serves as a master regulator of mucin type O-glycosylation. Global deletion of Cosmc (an X-linked gene) in mice is embryonically lethal. However, in human development, the function of Cosmc is largely unknown. Recently, we found two pathogenic C1GALT1C1 variants, which are associated with a congenital disorder of glycosylation (CDG). One is a de novo mosaic variant (c.202C>T, p.Arg68*; Cosmc-R68) discovered in a 5-year-old female, and the second one is a germline hypomorphic, hemizygous variant c.59C>A (p.Ala20Asp; A20D-Cosmc) resulting in C1GALT1C1-CDG (COSMC-CDG) found in two boys (7 and 21 years), mother, and grandmother. The female 5-year-old patient carries a C1GALT1C1 variant causing complete loss-of-function as a mosaic (with a 27% allelic fraction in buccal cells), and she was prematurely born with non-immune hydrops fetalis at 33 weeks of gestation, which later resolved. She presented with global developmental delays and also screened positive for autism (at 18 months). She also showed persistent proteinuria (at 10 months) which persisted throughout, but she had no known renal dysfunction. Her complete blood count remains normal, but the neonatal period was complicated by thrombocytopenia. Patient leukocytes showed reduced Cosmc and T-synthase expression and loss of T-synthase activity which is consistent with cells expressing the Tn antigen. Our second study on the germline C1GALT1C1, who carry a mutation causing a A20D mutation within the TMD of Cosmc; this results in a remarkable loss of both Cosmc and T-synthase proteins, and activity. Patients presented with developmental delay and hematological disorders. Interestingly, the patients have acute kidney failure (AKI), which is fully responsive to treatment with Eculizumab, a complement inhibitor. Collectively, our study provides insight into the role of O-glycans in human health and disorders and provides a treatment option for AKI related to COSMC-CDG.

Biography:

Raj is a faculty member at the Center for Glycoscience at Harvard Medical School, where he serves as a Principal Investigator on various research projects. His research group is dedicated to integrating the fields of glycobiology, biochemistry, genetics, and cell biology to investigate the functions of membrane proteins, with a particular emphasis on glycosyltransferases and the implications of glycosylation in human diseases. Currently, a key focus of his work is to elucidate the functional roles of O-glycans within the context of human diseases, contributing to a deeper understanding of their biological significance and potential therapeutic avenues.

Current Oral Feeding Management Approaches in NICUs: Results from a National Survey

Sandra Fucile*, Alyssa Alguire and Janine Kowalczyk

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Objective: Up to 40% of infants who require neonatal intensive care experience pediatric feeding disorders, delaying the transition from full tube feeding to independent oral feeding and extending hospital stays¹⁻⁴. Effective management of oral feeding, including its initiation and advancement, is essential in the neonatal intensive care unit (NICU)⁵⁻⁷. Current practice approaches for oral feeding management are unknown. This study aims to evaluate oral feeding management practices across Canadian NICUs.

Methods: A national on-line survey study was conducted among level II and III NICUs. Sixty-five NICUs were invited to participate in the survey. The survey consisted of three sections: Hospital demographics, clinical approaches for introducing and advancing oral feeds, and strategies for management of pediatric feeding disorders.

Results: Forty-eight (74%) respondents completed the survey. The most reported practice for initiating and advancing oral feeds was the use of custom-developed NICU guidelines (46%). Few respondents utilized evidence-based oral feeding protocols (33%) or validated standardized assessments (2%). The key clinical factors considered when introducing as well as advancing oral feeds was infant behavioral cues, such as alertness, physiological stability (heart rate, respiratory rate and oxygen saturations) and respiratory support. Most surveyed units (79%) employed a multidisciplinary team-based approach that included family engagement in managing oral feeds. Furthermore, 65% of surveyed NICUs noted they addressed feeding matters only until hospital discharge, and only 35% provided continued follow-up service after discharge from the NICU.

Conclusion: This survey highlights three key findings regarding oral feeding practices in Canadian NICUs: (1) There is a shift from using gestational age alone toward more infant-driven behavioral cues to guide oral feeding initiation and advancement; (2) Most NICUs manage oral feeding only until hospital discharge, underscoring the need for improved post-discharge feeding follow-up; and (3) The widespread use of custom-developed unit guidelines points to the need for standardized, evidence-based protocols to improve consistency and optimize feeding outcomes across NICUs.

References:

1. Viswanathan S, Jadcherla S. Feeding and Swallowing Difficulties in Neonates: Developmental Physiology and Pathophysiology. *Clin Perinatol*. Jun 2020;47(2):223-241.
2. Robinson L HL, Fucile S. . Investigating the developmental trajectory of long-term oral feeding problems in preterm infants. *Developmental neurorehabilitation*. 2022;11:1-5.
3. Kwon J, Kellner P, Wallendorf M, Smith J, Pineda R. Neonatal feeding performance is related to feeding outcomes in childhood. *Early Hum Dev*. Dec 2020;151:105-202.
4. Goday PS, Huh SY, Silverman A, et al. Pediatric Feeding Disorder: Consensus Definition and Conceptual Framework. *J Pediatr Gastroenterol Nutr*. Jan 2019;68(1):124-129.
5. Fucile S, Caulfield A, Geleynse S. An oral feeding model for infants in the NICU: A checklist for practitioners. . *NEONATAL NETWORK THE JOURNAL OF NEONATAL NURSING*. 2020;38(6):348-356.
6. Rhooms L, Dow K, Brandon C, Zhao G, Fucile S. Effect of Unimodal and Multimodal Sensorimotor Interventions on Oral Feeding Outcomes in Preterm Infants: An Evidence-Based Systematic Review. *Adv Neonatal Care*. Feb 2019;19(1):E3-E20.
7. Borges Nery P, Snider L, Camelo JSJ, et al. The Role of Rehabilitation Specialists in Canadian NICUs: A 21st Century Perspective. *Phys Occup Ther Pediatr*. 2019;39(1):33-47.

Biography:

Dr. Sandra Fucile is a registered Occupational Therapist and Assistant Professor at the School of Rehabilitation Therapy with a cross-appointment in the Department of Pediatrics at Queen's University. She is passionate about giving every infant a bright start and has dedicated her research towards enhancing the health and development of infants born with complex medical conditions with a particular focus on infants born preterm. Her research program encompasses three areas: 1) Identification of neurodevelopmental biomarkers; 2) Creating and testing innovative assessments tools; and 3) Design and evaluation of novel family-delivered intervention approaches. She leads the Infant Neurodevelopment, Feeding Advances And New technologies (I.N.F.A.N.T.) Research lab located at Kingston Health Sciences Center which consists of an interdisciplinary team of students, clinicians, researchers and families conducting clinical investigations aimed at promoting the well-being of infants and their families.

The Intersection of Child Abuse and Sex Trafficking Victimization

Sandra S. Stone*

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This paper explores the critical link between child sexual abuse (CSA) and commercial sexual exploitation (CSE), highlighting how childhood trauma increases vulnerability to sex trafficking. Survivors of CSA often experience long-term psychological harm, including low self-esteem, impaired relationships, and susceptibility to manipulation by traffickers. Evidence shows that CSA is a significant risk factor for later trafficking, with systemic failures—such as inadequate child protection, lack of professional training, and misidentification of victims—compounding the issue. The consequences are severe, leading to complex PTSD, depression, and substance abuse. To address this intersection, the paper calls for comprehensive prevention strategies, early intervention for at-risk youth, professional training, cross-sector collaboration, and survivor-centered, trauma-informed care. A holistic approach is essential to break the cycle of abuse and exploitation and to ensure lasting support and healing for survivors.

Prenatal detection rates for congenital heart disease using abnormal obstetrical screening ultrasound alone as indication for fetal echocardiography

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Objective: To determine the live born prenatal detection rate of significant congenital heart disease (CHD) in a large, integrated, multi-center community-based health system using a strategy of referral only of patients with significant cardiac abnormalities on obstetrical screening ultrasound for fetal echocardiography. Detection rates were assessed for screening in both radiology and maternal fetal medicine (MFM). The impact on fetal echocardiography utilization was also assessed.

Methods: This was a retrospective cohort study using an electronic health record, outside claims databases and chart review to determine all live births between 2016 and 2020 with postnatally confirmed sCHD that were prenatally detectable and resulted in cardiac surgery, intervention, or death within 1 year.

Results: There were 214,486 pregnancies resulting in live births. Prenatally detectable significant CHD was confirmed in 294 infants. Of those 183 were detected for an overall live-born detection rate of 62%. Detection rates in MFM were 75% and in radiology were 52%. The number of fetal echocardiograms needed to detect (NND) sCHD was 7.

Conclusions: A focus on quality and standardization of obstetrical screening ultrasound with referral to fetal echocardiography for cardiac abnormalities alone achieves benchmark targets for live-born detection of significant CHD requiring fewer fetal echocardiograms.

Executive functions and their influence on academic performance in school-aged children in Tunisia

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Executive functions are crucial mental processes that enable children to navigate the challenges of the school environment, ultimately supporting their academic success. This research employed a correlational approach to explore the connection between executive functions and academic performance in school-aged children in Tunisia. The study specifically focused on examining how key components of executive functioning, such as working memory, mental flexibility, and inhibition, relate to performance in core academic subjects, including language, mathematics, and science. A total of 180 children, ages 7 to 12, participated in the study. Their executive functioning was assessed using the Behavioral Rating Inventory of Executive Function (BRIEF), a reliable tool completed by both parents and teachers. The BRIEF offers a comprehensive evaluation of children's behavior in various contexts, providing insight into their cognitive abilities in different environments. Results showed strong correlations between working memory and performance in language and mathematics, especially in teacher ratings. Mental flexibility and inhibition were weakly associated with language but showed moderate correlations with mathematics. Parent ratings showed generally weaker correlations for language and moderate ones for mathematics. No significant association was found with science in either parent or teacher evaluations. Planning and organization also demonstrated strong correlations with language and mathematics in teacher assessments, and moderate ones in parent reports. Inhibitory control followed a similar pattern. These findings highlight discrepancies between teacher and parent perspectives, with teachers consistently reporting stronger links between executive functions and academic outcomes. The absence of correlation with science contrasts with previous research and may reflect contextual or methodological differences. This study underscores the importance of executive functions in academic success and the need for early identification of children at risk. Future research should explore predictive models and contextual variables affecting both assessment and intervention in educational

Biography:

Sleh Eddine Saddi is a psychologist and psychotherapist specializing in various therapeutic approaches, including brief therapy, hypnosis, psychodrama, and schema therapy. He is the author of a recent study titled Executive Functions and Their Influence on Academic Performance in School-Aged Children in Tunisia (2025).

Adding oscillometry and fractional exhaled nitric oxide to guidelines potentially enhances identification of exacerbation risk factors in asthmatic children

Stanley P. Galant, MD* and Tricia Morphew

Children's Hospital of Orange County and University of California, Irvine, United States.

Children's Hospital of Orange County and Morphew Consulting LLC, Bothell, Washington, USA.

A possible explanation for the continued risk of asthma exacerbations while utilizing current asthma guideline-based management is the failure to consider small airway dysfunction (SAD) and eosinophilic airway inflammation in assuming asthma-controlled status. To construct a practical algorithm that potentially identifies additional risks of not well-controlled (NWC) asthma and exacerbations beyond guideline criteria with oscillometry and fractional exhaled nitric oxide (FENO) determination. This was a retrospective, posthoc analysis of 183 children, ages 4-18 years, with moderate-to-severe asthma, characterized by demographic factors, National Health Lung and Blood Institute (NHLBI) determined asthma-controlled status, therapy step, and lung function status. Impulse oscillometry (IOS) and FENO were performed before spirometry. Abnormal lung function values were determined for spirometry, SAD, and FENO based on established thresholds. Statistical analysis included a decision tree and Venn diagram. For 53 patients with guideline-based NWC, 15 (28.3%) had an $FEV_1 \leq 80\%$ predicted. An additional 30 patients (56.6%) had abnormal SAD and/or FENO with a preserved FEV_1 . Of 130 patients, well-controlled (WC) by guidelines 76 (58.5%) exhibited SAD and/or abnormal FENO. These data demonstrate that over 50% of WC patients with moderate-to-severe asthma, with few symptoms and preserved FEV_1 , had SAD and/or abnormal FENO levels, suggesting the potential of early phenotype-specific therapy to prevent risk of future exacerbations.

Biography:

Dr. Galant is a Clinical Professor of Pediatrics at the University of California, Irvine. He is board certified in pediatrics and allergy-immunology. His major focus currently is the peripheral airways in relationship to asthma control and asthma exacerbations.

Innovative modulators of IL-1 and IL-6 receptors to prevent inflammation-induced preterm birth and improve newborn outcomes

Habelrih T,¹ Côté F,¹ Quiniou C,¹ Ferri B,¹ Nadeau-Vallée M,¹ Hou X,¹ Loiselle SE,¹ Sawaya K,¹ Leimert K,² Lubell WD,¹ Olson DM,² **Sylvain Chemtob^{1*}**

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Preterm birth (PTB) is a major cause of neonatal mortality and morbidity. To date there are no approved drugs that effectively tackle PTB; currently utilized tocolytics are ineffective in prolonging gestation and in improving fetal/neonatal outcome. Evidence supports a determinant role for interleukins-1 and -6 (IL-1 and IL-6) in the pathophysiology of PTB. We have developed small stable all-d peptides, specifically 'rytvela' and HSJ633 (amino acid sequence: vrkfqnsps; each peptide: 0.9-1 kDa), that respectively antagonize the interleukin-1 and -6 receptors (IL-1R and IL-6R). Binding and signal transduction assays confirm specificity of rytvela to IL-1R and that of HSJ633 to IL-6R, albeit biasing receptor-coupled signaling, indicative of desirable (and safer) pharmacologic selectivity through allosteric modulation. In an in vivo murine model of lipopolysaccharide (LPS)-induced PTB, both rytvela and HSJ633 administered prophylactically as well as in treatment mode (2-4 h post-LPS), diminished inflammation in gestational and fetal tissues, reduced the rate of PTB, improved survival of neonatal progeny and preserved integrity of fetal organs. Combined treatment of rytvela and HSJ633 was superior to either drug candidate, as it abrogated PTB and annulled fetal/neonatal mortality. FITC-labelled modulators confirms transplacental crossing. Finally, we discriminately uncovered that soluble IL-6R (rather than membrane-bound IL-6R) is the major participant in eliciting inflammation-induced PTB antagonized by HSJ633. Findings reveal first-in-class efficacy of small peptide antagonists of IL-1R and IL-6R, respectively rytvela and HSJ633, as potential drug candidates in restricting the inflammatory cascade associated with PTB and mitigating adverse neonatal outcomes. Administration of rytvela and/or HSJ633 to safely prolong gestation and prevent ensuing complications related to PTB substantiate a prominent preclinical development and offer unprecedented perspectives to address the unmet medical need in PTB.

Biography:

Sylvain Chemtob is a reputed neonatal pharmacologist and physiologist, with expertise in ischemic retinopathies and design of therapeutics to counter premature birth. He is professor of Pediatrics and Ophthalmology at University of Montreal, Canada, and has been a Canada Research Chair (2000-24) and Leopoldine Wolfe Chair (2009-25). He is a Fellow of the Canadian Academy of Health Sciences and Fellow of the Royal Society of Canada. He is author of >350 publications (many in major journals) and inventor of 30 patents. This work has been supported by CIHR.

Uridine Metabolism as a Targetable Metabolic Achilles' Heel for chemo-resistant B-ALL

Yuxuan Liu*

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Relapse continues to limit survival for patients with B-cell acute lymphoblastic leukemia (B-ALL). Previous studies have independently implicated activation of B-cell developmental signaling pathways and increased glucose consumption with chemo-resistance and relapse risk. Here, we connect these observations, demonstrating that B-ALL cells with active signaling, defined by high expression of phosphorylated ribosomal protein S6 ("pS6+ cells"), are metabolically unique and glucose dependent. Isotope tracing and metabolic flux analysis confirm that pS6+ cells are highly glycolytic and notably sensitive to glucose deprivation, relying on glucose for de novo nucleotide synthesis. Uridine, but not purine or pyrimidine, rescues pS6+ cells from glucose deprivation, highlighting uridine is essential for their survival. Active signaling in pS6+ cells drives uridine production through activating phosphorylation of carbamoyl phosphate synthetase (CAD), the enzyme catalyzing the initial steps of uridine synthesis. Inhibition of signaling abolishes glucose dependency and CAD phosphorylation in pS6+ cells. Primary pS6+ cells demonstrate high expression of uridine synthesis proteins, including dihydroorotate dehydrogenase (DHODH), the rate-limiting catalyst of de novo uridine synthesis. Gene expression demonstrates that increased expression of DHODH is associated with relapse and inferior event-free survival after chemotherapy. Further, the majority of B-ALL genomic subtypes demonstrate activity of DHODH. Inhibiting DHODH using BAY2402232 effectively kills pS6+ cells in vitro, with its IC50 correlated with the strength of pS6 signaling across 14 B-ALL cell lines and patient-derived xenografts (PDX). In vivo DHODH inhibition prolongs survival and decreases leukemia burden in pS6+ B-ALL cell line and PDX models. These findings link active signaling to uridine dependency in B-ALL cells and an associated risk of relapse. Targeting uridine synthesis through DHODH inhibition offers a promising therapeutic strategy for chemo-resistant B-ALL as a novel therapeutic approach for resistant disease.

Biography:

Dr. Yuxuan Liu received her Ph.D. from the University of Groningen in the Netherlands and completed her postdoctoral training at Columbia University in New York. She is currently a Senior Scientist at Stanford University. Dr. Liu's research focuses on uncovering the metabolic dependencies of chemo-resistant cells in B cell malignancies, utilizing cutting-edge single-cell technologies.

Impact of an early educational protocol on the oral language of children born preterm exhibiting phonological fragility: a multicenter randomized clinical trial

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We conducted a six-center, prospective, randomized, open-label trial to assess whether an early standardized educational protocol provided from 42 to 48 months of age improved the progression of oral language and phonological development in children born preterm. A total of 552 children with phonological fragility were included in this study. The children were randomized to receive the educational protocol (guided arm, n = 87) or not (non-guided arm, n = 78). In the guided arm, the oral language development used a short “say and do” type educational protocol designed to maintain visual attention and train the developmental phonology/lexicon/morphosyntax structural links. In contrast, a conservative approach was used in the non-guided arm. A total of 70 guided and 73 non-guided children completed the study. After 6 months, the educated children showed a non-significant increase in their phonology score (p = 0.37), while the variations in the scores of the expressive

lexicon (secondary endpoints) were significantly improved (p = 0.0008). We conclude that the short, standardized stimulation of the sensorimotor aspects of language in children born very preterm increased the expressive lexicon. This protocol improved the language of the premature children, especially those with minimal motor skills, with more significant improvement in the phonological scores.

Keywords: Prematurity, Oral Language Disorder, Language and Motor Skills, School-Age, Child Born Preterm, Phonology Score, Education Speaking

Novel truncating PPM1D variant in a dichorionic diamniotic (DCDA) twin with Jansen-de Vries syndrome. an updated perspective

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Jansen-de Vries syndrome (JDVD) is a rare autosomal dominant neurodevelopmental disorder that is caused by truncating variants in exons 5 and 6 of the PPM1D gene. The condition is characterised by intellectual disability, hypotonia, dysmorphic features, behavioural abnormalities, and systemic complications. Despite an increasing recognition of the disorder, JDVD remains underdiagnosed due to its phenotypic variability and symptom overlap with other neurodevelopmental disorders such as autism spectrum disorder (ASD) and attention-deficit/hyperactivity disorder (ADHD). This study presents a case of 6-year-old diamniotic and dichorionic twin brothers, both diagnosed with ASD and ADHD, but with distinct clinical manifestations. Trio-whole-exome sequencing (WES) revealed a de novo frameshift mutation (c.1411_1412del) in PPM1D in one twin, which was classified as likely pathogenic. In contrast to his unaffected twin, the proband exhibited delayed language acquisition, learning difficulties, behavioural problems, restrictive eating, and impaired autonomy. This case underscores the importance of genetic testing in differentiating JDVS from other neurodevelopmental conditions, particularly when clinical symptoms are ambiguous. The findings of this study highlight the expanding clinical spectrum of this syndrome and underscore the critical role of molecular diagnostics, particularly trio-WES, in improving diagnostic accuracy and facilitating early intervention. Further research is needed to refine genotype-phenotype correlations and develop standardised diagnostic and management guidelines for JDVS.

Biography:

He holds a PhD in Medicine and a PhD in Psychology. He is a specialist in Clinical Biochemistry and has a master's degree in Medicine. He is a professor of Biochemistry and Genetics at the Faculty of Nursing at the University of Malaga. He is the author of numerous books and articles. He currently works in the Genetics Department at the Regional University Hospital of Malag.

Association of Screen Time with Behavioral Risk in Children Under Five: A Cross- Sectional Study across Central India

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Excessive screen time is associated with developmental (mainly socio-cognitive and language) and behavioral problems more in children below five years than in older children and adolescents. The present study was conducted to determine the prevalence and pattern of exposure to excessive screen time in children below five years of age. It was also aimed at determining the prevalence of the risk of behavioral problems in these children in relation to their average daily screen time.

This study was conducted as an observational descriptive study on children attending Outpatient Department (OPD) in the Department of Pediatrics of a tertiary care medical college in central India over the study period of 12 months. Parents were enquired about their socio-demographic details, the daily average screen time of their child, details regarding screen time and exposures, associated factors, sleeping and eating habits, average time spent by parents with their children, and also the behavioral issues according to the preschool pediatric symptom checklist (PPSC). All these questions were asked of the parents and duly filled out by a single doctor. The IAP (Indian Academy of Pediatrics) screen time recommendation was used as a cut-off value for excessive screen time definition, which states screen time recommendations for children below two years and two to five years old.

The daily average screen time greater than one hour (exceeding the recommended guidelines) was found in 57.7% of children. Twenty-six percent of children were exposed to screen time before their first birthday. Among the study participants, 37.7% (n=95) of children were found to be at risk of behavioral problems as per PPSC score, with a mean PPSC score of 6.7 ± 3.3 in these children. The daily average screen time of one to three hours was found in 39.3% of children, three to five hours in 13% of children, and more than five hours in 5.3% of children. The PPSC score was found to be significantly higher in children with higher average screen time, children using multiple devices as compared to single devices, children who preferred electronic devices or devices with toys as preferred sources of entertainment, children becoming restless when devices are not given, children having feeding and sleep issues, and children who spend less time with parents ($p < 0.05$).

Multivariate analysis revealed higher odds of behavioral problems in children having more screen time (1-3 hours OR: 1.536; 95% CI: 1.03-7.237; $p = 0.035$, 3-5 hours OR: 2.022; 95% CI: 1.477-

8.570; $p = 0.029$, and > 5 hours OR: 9.793; 95% CI: 2.065-46.447; $p = 0.004$), children using multiple devices as compared to single devices (OR: 2.43; 95% CI: 1.194-4.95; $p = 0.014$), and children whose parents spend less time with them (< 3 hours OR: 1.311; 95% CI: 1.02-2.77; $p = 0.012$).

The present study found that almost all children below five years are exposed to digital devices, with the majority of children exceeding the recommended screen time guidelines, and they mostly use it for entertainment purposes. During this period, exposure of children to screens is significantly associated with the risk of behavioral problems. It was observed that excessive screen time, less time spent by parents with their children, and use of multiple devices are significant predictors of increasing risk of behavioral problems in children below five years.

Evaluation of Accidental and Intentional Pediatric Poisonings: Retrospective Analysis of Emergency Medical Service Interventions in Wroclaw, Poland

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Poisonings among children are a major cause of morbidity and mortality worldwide and present a significant challenge for emergency medical services (EMS). The aim of this study was to analyze the types of substances causing poisonings and the intention of poisonings in children, providing detailed information on the most common causes of poisonings in different age groups.

Methods: A retrospective study was conducted analyzing medical records of pediatric patients under the care of emergency medical services (EMS) in Wroclaw, Poland, between 2015 and 2017. The study included all patients under 18 years old diagnosed with poisoning. Data collected included age, sex, cause of poisoning, location of the incident, neurological status (GCS), and the type of healthcare facility.

Results: The study included 484 patients, with a mean age of 13 years. The largest age group was 16–18 years (44%), and the majority were female (58%). The most common causes of poisonings were alcohol (29.3%), medications (26%), and intoxicants (24.8%). Over half of the incidents occurred at home (52.5%). Intentional poisonings constituted 75% of cases, particularly involving alcohol (38.6%), intoxicants (32%), and medications (26.7%). Accidental poisonings were mainly due to household chemicals (28.7%) and carbon monoxide (27%). The differences in causes and locations of poisonings were statistically significant ($p < 0.001$).

Conclusions: Our study showed that the most common causes of poisonings among children were alcohol, medications, and intoxicants. Poisonings most frequently occurred at home, especially with medications, while outside the home, alcohol and intoxicants were predominant. This study was not registered.

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