www.jpnim.com Open Access eISSN: 2281-0692
Journal of Pediatric and Neonatal Individualized Medicine 2025;14(1):e140101
doi: 10.7363/140101
Published online: 2025 Jan 20

Selected Abstracts of the 20th International Workshop on Neonatology and Pediatrics

CHILDREN'S HEALTH: MESSAGES FROM THE FUTURE

CAGLIARI (ITALY) • OCTOBER 23RD-26TH, 2024

The Workshop has been organized with the patronage of Autonomous Region of Sardinia, City of Cagliari, Azienda Ospedaliera di Cagliari, University of Cagliari, Board of Physicians of the Province of Cagliari, Italian-Albanian Pediatric Society, AMBO (Alleanza per un Ambiente a misura di Bambino: alliance for a child-friendly environment), European Project Better4u, European Project Life Milch, National Confederation of Pediatrics Heads (CONAPP), Italian Federation of Family Pediatricians (FIMP), Italian-Arabian Pediatric Society (IAPS), Italian-Romanian Pediatric Society (IRPS), Norman Group of Neonatal and Pediatric Nephrology, International Academy of Perinatal Medicine, Italian Society of Pediatric Allergology and Immunology (SIAIP), Italian Society of Clinical Biochemistry (SIBioC), Italian Society of Gynecology of Infancy and Adolescence (SIGIA), Italian Society of Neonatology (SIN), Italian Society of Hospital Pediatricians (SIPO), Association SIPO in the World, Italian Society of Developmental Origins of Health and Disease (SI-DOHaD), Alfred Nobel International Study Center Sanremo, Italian Society of Pediatricians (SIMPE), Italian Society of Pediatric Psychology (S.I.P.Ped.), Romanian Society of Pediatrics, Italian Society for Research on Essential Oils (SIROE), Italian Society of Neonatal Nursing (SIN INF), Union of European Neonatal and Perinatal Societies (UENPS).

PRESIDENTS

Vassilios Fanos (Cagliari, Italy), Antonia Charitou (Athens, Greece), Georgina Kuli Lito (Tirana, Albania), Michele Mussap (Mestre and Cagliari, Italy)

EXECUTIVE SCIENTIFIC SECRETARIAT (CAGLIARI, ITALY)

Viviana Marinelli

SCIENTIFIC SECRETARIAT (CAGLIARI, ITALY)

Vittoria Annis, Alessandra Atzei, Francesca Birocchi, Federico Cannas, Francesca Cioglia, Karolina Kopeć, Donatella Gariel, Danila Manus, Maria Antonietta Marcialis, Paola Neroni, Antonio Noto, Cristina Pintus, Denis Pisano, Melania Puddu, Anna Bruna Zanda

ORGANIZING SECRETARIAT (CAGLIARI, ITALY)

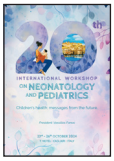
Alice Bosco, Giulia Concas, Elisabetta Coni, Elena Curridori, Cristina Loddo, Michele Loi, Valentina Masile, Roberta Mazzitti, Marta Melis, Roberta Pintus, Gaia Serrau, Claudia Spiga

ADMINISTRATIVE SECRETARIAT (CAGLIARI, ITALY)

Patrizia Baire

FACULTY

Massimo Agosti (Varese, Italy), Bartolomeo Allegrini (Monopoli, Italy), Osama Al Jamal (Cagliari, Italy), Vittoria Annis (Cagliari, Italy), Roberto Antonucci (Sassari, Italy), Enrico Arletti (Bologna, Italy), Pietro Armento (Florence, Italy), Alessandra Atzei (Cagliari, Italy), Luigi Atzori (Cagliari, Italy), Patrizia Baire (Cagliari, Italy), Marta Balzarini (Cagliari, Italy), Susanna Barella (Cagliari, Italy), Pier Paolo Bassareo (Dublin, Ireland), Luca Bernardo (Milan, Italy), Sergio Bernasconi (Parma, Italy), Enrico Bertino (Turin, Italy), Francesca Birocchi (Cagliari, Italy), Gianni Bona (Novara, Italy), Alice Bosco (Cagliari, Italy), Giuseppe Buonocore (Siena, Italy), Georgeta Burlea (Iași, Romania), Francesco Saverio Camoglio (Sassari, Italy), Alessandro Canetto (Cagliari, Italy), Federico Cannas (Cagliari, Italy), Marco Carotenuto (Naples, Italy), Valeria Casotti (Bergamo, Italy), Luigi Cataldi (Rome, Italy), Flaminia Cesare Marincola (Cagliari, Italy), Antonia Charitou (Athens, Greece), Gabriella Cherchi (Cagliari, Italy), Massimo Chessa (Milan, Italy), Iolanda Chinellato (Castellaneta, Italy), Francesca Cioglia (Cagliari, Italy), Anamaria Ciubara (Galați, Romania), Antonio Colangelo (Trento, Italy), Francesco Comisi (Cagliari, Italy), Giulia Concas (Cagliari, Italy), Elisabetta Coni (Cagliari, Italy), Giovanni Corsello (Palermo, Italy), Marcello Cossu (Cagliari, Italy), Maria Antonia Cossu (Sassari, Italy), Giangiorgio Crisponi (Cagliari, Italy), Antonio Cualbu (Nuoro, Italy), Sebastiano Curreli (Arborea, Italy), Elena Curridori (Cagliari, Italy), Spiro Damo (Tirana, Albania), Antonio D'Avino (Naples, Italy), Sonia Deantoni (Turin, Italy), Francesco De Luca (Catania, Italy), Angelica Dessì (Cagliari, Italy), Alice Di Carlo (Cagliari, Italy), Elena Esposito (Cagliari, Italy), Susanna Esposito (Parma, Italy), Gavino Faa (Cagliari, Italy), Claudio Fabris (Turin, Italy), Vassilios Fanos (Cagliari, Italy), Grazia Fenu Pintori (Sassari, Italy), Gabriele Finco (Cagliari, Italy), Thomas Foiadelli (Pavia, Italy), Laura Folgori (Milan, Italy), Anna Maria Fulghesu (Cagliari, Italy), Donatella Gariel (Cagliari, Italy), Eloisa Gitto (Messina, Italy), Monica Greco (Turin, Italy), Eleftheria Hatzidaki (Heraklion, Greece), Alketa Hoxha-Qosja (Tirana, Albania), Tatyana Itova (Ruse, Bulgaria), Pramod Jog (Aundh, Pune, India), Karolina Kopeć (Krakow, Poland, and Cagliari, Italy), Gentiana Koroveshi-Gora (Tirana, Albania), Georgina Kuli Lito (Tirana, Albania), Alberto Lai (Cagliari, Italy), Emanuela Lanfranchi (Urbino, Italy), Marie Leiner (El Paso, Texas, USA), Gianluca Lista (Milan, Italy), Cristina Loddo (Cagliari, Italy), Michele Loi (Cagliari, Italy), Georgios Loudianos (Cagliari, Italy), Rita Luciano (Rome, Italy), Antonio Macciò (Cagliari, Italy), Valerio Mais (Cagliari, Italy), Valeria Manca (Cagliari, Italy), Fabio Mancini (Milan, Italy), Danila Manus (Cagliari, Italy), Matteo Marchi (Modena, Italy), Maria Antonietta Marcialis (Cagliari, Italy), Monica Marica (Cagliari, Italy), Viviana Marinelli (Cagliari, Italy), Gianluigi Marseglia (Pavia, Italy), Mina Martini (Pisa, Italy), Carla Marzetti (Bologna, Italy), Luigi Mascia (Cagliari, Italy), Valentina Masile (Cagliari, Italy), Giuseppe Masnata (Cagliari, Italy), Matteo Mauri (Cagliari, Italy), Roberta Mazzitti (Cagliari, Italy), Marta Melis (Cagliari, Italy), Gianfranco Meloni (Sassari, Italy), Mirela Meta (Fier, Albania), Francesco Mola (Cagliari, Italy), Francesca Mondello (Rome, Italy), Giovanni Monni (Cagliari, Italy), Fabio Mosca (Milan, Italy), Sandro Muntoni (Cagliari, Italy), Michele Mussap (Cagliari and Mestre, Italy), Paola Neroni (Cagliari, Italy), Simona Nava (Milan, Italy), Antonio Noto (Cagliari, Italy), Erminia Maria Oppedisano (Cagliari, Italy), Enrica Paderi (Oristano, Italy), Paola Palanza (Parma, Italy), Umberto Pelosi (Cagliari, Italy), Diego Peroni (Pisa, Italy), Domenico Perri (Aversa, Italy), Giovanna Perricone (Palermo, Italy), Raida Petrela (Tirana, Albania), Silvia Petza (Cagliari, Italy), Enrica Pezzoni (Fontanellato, Italy), Giulia Picari (Cagliari Italy), Andrea Pietrobattista (Rome, Italy), Roberta Pintus (Cagliari, Italy), Cristina Pintus (Cagliari, Italy), Cristina Piras (Cagliari, Italy), Denis Pisano (Cagliari, Italy), Elisabetta Piu (Cagliari, Italy), Concetta Polizzi (Palermo, Italy), Carmela Porcu (Cagliari, Italy), Melania Puddu (Cagliari, Italy), Alessandra Reali (Cagliari, Italy), Serge Rezzi (Epalinges, Switzerland), Maria Luisa Ricci (Rome, Italy), Domenico Romeo (Milan, Italy), Martino Ruggeri (Catania, Italy), Simone Rugolotto (Rovigo, Italy), Giuseppe Sanna (Cagliari, Italy), Matilde Sassi (Trieste, Italy), Salvatore Savasta (Cagliari, Italy), Claudia Sbaffi (Cagliari, Italy), Manuel Scarano (Cagliari, Italy), Consolata Soddu (Cagliari, Italy), Roberto Solinas (Cagliari, Italy), Claudia Spiga (Cagliari, Italy), Marcus Stanton (Lübeck, Germany), Maria Elisabeth Street (Parma, Italy), Liborio Stuppia (Chieti, Italy), Gianfranco Temporin (Rovigo, Italy), Giorgio Terziani (La Spezia, Italy), Gianfranco Trapani (Sanremo, Italy), Camilla Trenta (Cagliari, Italy), Effrossine Tsekoura (Athens, Greece), Attilio Varricchio (Naples, Italy), Francesco Velardi (Rome, Italy), Salvatore Vendemmia (Aversa, Italy), Katya Wulhfard (Turin, Italy), Anna Bruna Zanda (Cagliari, Italy), Maurizio Zanda (Cagliari, Italy), Massimiliano Zonza (Cagliari, Italy), Gian Vincenzo Zuccotti (Milan, Italy)



Organizing secretariat
Kassiopea group srl

How to cite

[Abstracts's authors]. [Abstracts's title]. In: Selected Abstracts of the 20th International Workshop on Neonatology and Pediatrics; Cagliari (Italy); October 23-26, 2024. J Pediatr Neonat Individual Med. 2025;14(1):e140101. doi: 10.7363/140101.

ABS 1

EXAMINATION OF CBD PRODUCT SAFETY MESSAGING FOR CHILDREN IN SOCIAL MEDIA PLATFORMS: A COMPARATIVE STUDY

I. Ham[1], O.S. Khilji[2], M. Leiner[2]

[1]Paul L. Foster School of Medicine, El Paso, Texas, USA

[2]Department of Pediatrics, Texas Tech University Health Sciences Center El Paso, El Paso, Texas, USA

The use of cannabidiol (CBD) products, particularly in children, has become a topic of increasing concern as the industry continues to expand rapidly. Despite the legalization of CBD in all fifty states in the United States and the approval of Epidiolex® by the FDA for pediatric seizure disorders [1], there remains a lack of concrete evidence regarding the efficacy, safety, and recommended dosages of CBD products. Concerns of consumer safety have been on the rise as traces of tetrahydrocannabinol (THC) were discovered in these unregulated products. In 2022, Johnson et al. [2] examined 80 unregulated products and found that 52 of the samples had detectable THC levels. Additionally, 5 of the 21 products labeled "THC free" were found to have detectable THC levels.

This study aimed to compare the messaging surrounding CBD product safety for children as portrayed in social media, particularly YouTube video clips from a preview study conducted in 2020 by Ohmed et al. [3] with new information in 2024. The initial study, after analyzing 43 YouTube video clips promoting CBD products, concluded that the information presented suggested that CBD products are safe for children and do not require professional advice. To further explore this issue, a comparative qualitative study was conducted, reviewing the 43 videos from the 2020 study and selecting 31 new videos in YouTube. The videos were re-analyzed based on previous factors such as view count, scientific content, disorder cure or improvement claims, and safety measures. The results indicated that while some videos from 2020 had been removed or were unavailable, there was an increase in subscriber and view count. More videos from 2020 study had children shown in the videos (32 of 43) with 16 out of the 43 using products, the newer videos only had 12 out of 31 videos showing children with 6 out of 31 using the products. Additionally, few newer videos showed content created by professionals with more emphasis on the research and science behind CBD oil, including warnings against unregulated CBD use. Warnings about the use and child proof caps continues to be very low or inexistent and 15/31 of the newer videos claims CBD is safe for children. Pediatric emergency visits due to CBD gummies are now included in the videos.

The findings of this study highlight the ongoing challenges in ensuring accurate and responsible messaging regarding CBD product safety for children in social media platforms [3]. As the market for CBD products continues to expand, there is a pressing need to address the potential risks associated with misinformation and inadequate regulation. Social media and advertising play a crucial role in shaping attitudes and behaviors towards CBD use in children, underscoring the importance of promoting evidence-based information and professional guidance. This research contributes to the broader conversation on pediatric CBD product safety and calls for increased awareness, regulation, and education to safeguard children.

REFERENCES

[1] Patel AD, Mazurkiewicz-Bełdzińska M, Chin RF, Gil-Nagel A, Gunning B, Halford JJ, Mitchell W, Scott Perry M, Thiele EA, Weinstock A, Dunayevich E, Checketts D, Devinsky O. Long-term safety and efficacy of add-on cannabidiol in patients with Lennox-Gastaut syndrome: Results of a long-term open-label extension trial. Epilepsia. 2021;62(9):2228-39.

[2] Johnson E, Kilgore M, Babalonis S. Cannabidiol (CBD) product contamination: Quantitative analysis of Δ9-tetrahydrocannabinol (Δ9-THC) concentrations found in commercially available CBD products. Drug Alcohol Depend. 2022;237:109522.

[3] Khilji O, Leiner M, Pathak I. Social Media Information About Cannabidiol (CBD) Products Use Among Children: Are the Messages Presented Suggesting They Are Safe for Children? Pediatrics. 2021;147(3):110-1.

ABS 2

L1CAM IS SECRETED THROUGH EXOSOMES IN THE DEVELOPING HUMAN BRAIN

F. Cau[1], F. Coghe[2], G. Faa[3,4]

[1]Department of Medical Sciences and Public Health, University of Cagliari, Cagliari, Italy

[2]Laboratory of Chemical-Clinical and Microbiological Analysis, AOU Cagliari, Cagliari, Italy

[3]Professor Emeritus, Department of Medical Sciences and Public Health, University of Cagliari, Cagliari, Italy
[4]Department of Biology, College of Science and Technology, Temple University, Philadelphia, Pennsylvania, USA

INTRODUCTION

Exosomes are nanovesicles that originate from cells, being released when multivesicular bodies fuse with the surface of the cell. They have the ability to activate target cells' signaling pathways and transfer lipids, proteins, and nucleic acids for intercellular communication. Exosomes have the potential to contribute to cell growth, immune response regulation, prevention of the epithelial-mesenchymal transition and stimulation of angiogenesis [1, 2]. Exosomes exhibit unique properties, including the ability to migrate to distant locations, modulating the microenvironment during human development [3]. Moreover, in recent years, exosomes have been shown to play a functional role in cancer development and metastasis [4]. Here we show that exosomes derived from brain cells in the developing human spinal cord, carry L1 cell adhesion molecule (L1CAM) towards neurons, participating actively to the development of the human central nervous system.

MATERIALS AND METHODS

Five human spinal cord samples, comprising three embryos and two fetuses of varying gestational ages, spanning from 8 to 12 weeks, were subjected to immunohistochemistry for L1CAM.

RESULTS

In each of the five spinal cords, L1CAM expression was found. The great majority of cells contained the adhesion molecule. The areas of the spinal cord where sensory and motor fibres are located, on the outskirts of the developing organs, exhibited the highest levels of L1CAM immunoreactivity. The positivity for L1CAM, which was mostly expressed in axons, was characterized by a reticular pattern in the alar and basal columns. Extracellular vesicles were also seen to exhibit strong L1CAM reactivity. The ability of L1CAM to mediate the transmission of extracellular signals that assist axon development may be indicated by its extracellular localization.

CONCLUSIONS

Our discovery that L1CAM localizes extracellularly lends credence to the theory that this adhesion molecule plays a significant role in nervous system development by promoting connections between cells, axons, and the substrate. Previous research on L1CAM's capacity to stimulate axon sprouting and branching in the developing nervous system is supported by the high reactivity of L1CAM in the axons of growing neurons in the fetal spinal cord. According to this work, L1CAM, whose expression is highly expressed in the growing neuronal and glial progenitors, is a new player in the intricate area of human spinal cord development.

REFERENCES

[1] Liu J, Ren L, Li S, Li W, Zheng X, Yang Y, Fu W, Yi J, Wang J, Du G. The biology, function, and applications of exosomes in cancer. Acta Pharm Sin B. 2021;11(9):2783-97.
[2] Gomes DE, Witwer KW. L1CAM-associated extracellular vesicles: A systematic review of nomenclature, sources, separation, and characterization. J Extracell Biol. 2022;1(3):e35.
[3] Cau F, Fanni D, Manchia M, Gerosa C, Piras M, Murru R, Paribello P, Congiu T, Coni P, Pichiri G, Piludu M, Van Eyken P, Gibo Y, La Nasa G, Orrù G, Scano A, Coghe F, Saba L, Castagnola M, Faa G. Expression of L1 Cell Adhesion Molecule (L1CAM) in extracellular vesicles in the human spinal cord during development. Eur Rev Med Pharmacol Sci. 2022;26(17):6273-82.
[4] Sinha D, Roy S, Saha P, Chatterjee N, Bishayee A. Trends in Research on Exosomes in Cancer Progression and Anticancer Therapy. Cancers (Basel). 2021;13(2):326.

ABS 3

LANGUAGE DEFICIENCIES IN CHILDHOOD – INTERDISCIPLINARY APPROACHES

S.L. Burlea[1], G. Burlea[2], V.V. Lupu[3]

[1]Public Health and Management Department, Faculty of Dental Medicine, "Grigore T. Popa" University of Medicine and Pharmacy, Iași, Romania
[2]Helicomed Clinic, Iași, Romania
[3]Pediatrics, Faculty of Medicine, "Grigore T. Popa" University of Medicine and Pharmacy, Iași, Romania

The child's language, being the basis of all mental development and being a process that can be influenced by education, must be constantly monitored, so that, in the event of disturbances, prompt intervention can be made through appropriate strategies. One of the language disorders encountered in speech therapy case studies and which calls for the initiation of an interdisciplinary approach is organic dyslalia. Diseases of the oral cavity can cause pronunciation disorders consisting in the impossibility of correctly emitting one or more sounds. These language disorders have serious consequences on learning, school success, social adaptability, on the entire personality of the child with such deficiencies. The identification of any disturbance in the articulation of phonemes requires the prompt initiation of appropriate remedial measures. The therapeutic approach in

organic pronunciation disorders requires a complex approach, which involves teamwork, with the active participation of all the specialists involved. In the team, a primary diagnosis and a treatment plan will be formulated. One of the key factors of the effectiveness of the application of the intervention programs is the concrete way of involving the parents in this approach, especially through the emotional and motivational support offered to the child. Through its corrective role, this therapeutic intervention will ensure the prevention of school failure, the equalization of training, education and integration opportunities.

ABS 4

LONG-TERM EFFECTS OF EARLY VOCAL CONTACT, BY PARENTS' BOOK-READING, FOR PRETERM NEWBORNS IN NICUs

C. Peila, S. Deantoni, L. Riboldi, M. Mainardi, E. Bertino, A. Coscia

Neonatology Unit, Department of Public Health and Pediatrics, University of Turin, Turin, Italy

BACKGROUND
Preterm birth (< 37 weeks of gestational age [GA]) is associated with an increased risk of atypical neurodevelopment in cognitive, motor, behavioral, and language domains, leading to difficulties that emerge in preschool age and often persist into adolescence. One of the most recent good care practices, hoping to reduce this risk, is early vocal contact (EVC) in Neonatal Intensive Care Units (NICUs), mediated by parents reading of children's books. The present study wants to investigate the long-term effects of the proposal of this practice to parents of preterm infants in NICUs, focusing firstly on its frequency of implementation and secondly on children's neurodevelopment.
METHODS
From March 2023 to May 2023, hospitalized preterm newborns with a GA < 34 weeks and/or birth weight < 1,500 kg, considered stable by the care team and whose parents could be regularly present in the ward, were recruited in the NICU of the Neonatology Unit of the City of Science and Health of Turin. Parents were explained the importance of EVC and were supported to read to their babies, choosing among selected books in the NICU library. They were also recommended to carry on this practice after discharge. Reading frequency, liking, and other qualitative variables were analyzed through three questionnaires given to parents during the first three follow-up visits (40 weeks GA, 3 months, and 6 months of corrected age); neurodevelopmental assessment, including language assessment, was carried out through Griffiths III Mental Development Scales during the third follow-up visit, at around 6 months of corrected age.
RESULTS
18 infants were enrolled, of whom 2 were lost during follow-up and 1 was transferred to another hospital. The final study population consisted in 15 infants, including 9 males and 6 females, with an average GA of 30 weeks, an average birth weight of 1.32 kg, an incidence of small for GA (SGA) of 20% and an incidence of twins of 40%. 77% of children who were exposed to reading in the NICU were also found to be exposed at 6 months of corrected age; 67% of children were regularly exposed to reading throughout the entire evaluation period. Parental satisfaction was high and the children average neurodevelopmental performance was good. Regarding learning foundations and language domains, children who were exposed to reading more regularly and more frequently over time reported higher scores than children who were exposed less regularly and less frequently. Twinning, low birth weight and parent level of education did not appear to have adversely affected neurodevelopment.
CONCLUSIONS
Reading in the NICU is a safe practice, appreciated by parents and easily continued after discharge. This study confirms the existence of dose-dependent beneficial effects on neurodevelopmental skills, including language and communication skills.

ABS 5

RESPIRATORY DISTRESS IN A FEMALE INFANT WITH PRIMARY CILIARY DYSKINESIA. A CASE REPORT

E. Tsianaka[1], M. Karypidou[1], A. Loutsidi[1], K. Angelou[2], A. Charitou[1]

[1]*Neonatal Intensive Care Unit, Rea Maternity Hospital, Athens, Greece*
[2]*Alexandra General Hospital, Athens, Greece*

INTRODUCTION
Primary ciliary dyskinesia (PCD) is an autosomal recessive disorder with over 45 currently identified causative genes. The clinical features are chronic wet cough, drainage from the paranasal sinuses,

chronic otitis media with hearing impairment as well as infertility. Almost 50% of patients have a *situs inversus totalis*.

Neonatal respiratory distress occurs in more than 80% of patients. The overlapping symptoms with other causes of neonatal respiratory distress leads often to a delayed diagnosis, so that a work up for PCD should be performed in the newborn with characteristic clinical features.

A case report of a female infant with PCD, *situs inversus* and respiratory distress is reported below.

CASE REPORT

A female infant with gestational age 38^{+4} weeks was delivered by C-section due to cephalopelvic disproportion. The maternal medical history revealed the presence of hypothyroidism treated with thyroxine. 7 hours after delivery, the infant was transported to the Neonatal Intensive Care Unit due to respiratory distress. Due to severe respiratory distress syndrome symptoms and radiological findings of right upper lobe atelectasis, the infant was intubated and supported with a mechanical ventilation for 4 days. Respiratory support with high-flow nasal cannula was necessary until the 14th day, as well as oxygen therapy until the 13th day. Furthermore, the infant received a complementary treatment with inhalative salbutamol and fluticasone. Anteroposterior chest radiograph showed dextrocardia and a right-sided gastric bubble indicating *situs inversus*. This could be confirmed through echocardiography and abdominal ultrasound. The infant was treated initially with i.v. antibiotics and after 8 days with oral amoxicillin and was discharged home after 20 days in good clinical condition.

Due to the clinical and radiological findings, a genetical examination (next generation sequencing) was performed.

RESULT

Genetic diagnosis was compatible with PCD 14.

Genetic result

Chromosomal deletion on 3q26.33 part of the *CCDC39* gene. Furthermore, homozygous pathogenic variant c.2061del p.(Ala688LeufsTer8) of the *CCDC39* gene.

Heterozygous variant of uncertain significance on c.3532G>A p.(Asp1178Asn) part of the *CFAP74* gene.

CONCLUSION

PCD typically causes respiratory distress in the neonate. It is possible that the initial transient nature of the symptoms poses a missed opportunity for diagnosis. Some helpful features include the later onset of symptoms, the fact that the patient is typically a term infant without other obvious risk factors for respiratory distress, and furthermore the fact that the chest imaging often shows lobar atelectasis. Moreover, many PCD patients have *situs inversus totalis*.

As a conclusion, a work up for PCD such as genetic test panels, ciliary biopsy or brush biopsy culture for transmission electron microscopy (TEM) and nasal nitric oxide measurement should be initiated in the neonate with respiratory distress and compatible clinical and radiological features.

REFERENCES

- Machogu E, Gaston B. Respiratory Distress in the Newborn with Primary Ciliary Dyskinesia. Children (Basel). 2021;8(2):153.
- Mullowney T, Manson D, Kim R, Stephens D, Shah V, Dell S. Primary ciliary dyskinesia and neonatal respiratory distress. Pediatrics. 2014;134(6):1160-6.
- Peng B, Gao YH, Xie JQ, He XW, Wang CC, Xu JF, Zhang GJ. Clinical and genetic spectrum of primary ciliary dyskinesia in Chinese patients: a systematic review. Orphanet J Rare Dis. 2022;17(1):283.

ABS 6

WHEN CLINICAL FINDINGS REVEAL THE DIAGNOSIS: A CASE OF WILLIAMS SYNDROME

E. Esposito[1], G. Sanna[1], F. Comisi[1], M. Marica[2], S. Savasta[1]

[1]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

[2]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", Cagliari, Italy

G. was born at 36 weeks of pregnancy by an urgent cesarean section, performed due to a non-reassuring cardiotocographic trace and intrauterine growth restriction, following a pregnancy conceived through IVF. The mother has chromosomal mosaicism (45 X0, 46 XX), and the father carries mutations for cystic fibrosis and glycogen storage disease type II. During pregnancy, the Non Invasive Prenatal Test (NIPT) was performed and it was in the normal range. At birth, the baby weighed 2,040 g and had an Apgar score of 8 at the 1' minute and 8 at the 5' minute. On physical examination, a cardiac murmur was noted, along with facial dysmorphisms characterized by low-set ears, hypertelorism, a depressed nasal bridge, and anteverted nares. She required respiratory support with nasal CPAP and oxygen supplementation up to 30% for the first week of life. A comprehensive cardiological evaluation, including a color Doppler echocardiogram, revealed

hypoplasia and stenosis of the right branch of the pulmonary artery, hypoplasia of the aortic root and arch, a restrictive ventricular septal defect, and a patent foramen ovale. Due to feeding difficulties, a nasogastric tube was placed to administer, through gavage, the portion of milk feed that she could not take orally. Upon discharge, a speech therapy program was recommended to strengthen the perioral muscles, along with a referral to a specialized rare disease genetics clinic. Genetically, the clinical picture immediately raised suspicion for the specialist, who confirmed the diagnosis of Williams syndrome by performing karyotype analysis and DNA microarray, which identified a deletion at 7q11.23 [1].

Williams syndrome has a population incidence of 1:7,500 and is characterized by typical cardiac defects, supravalvular aortic stenosis, feeding difficulties and poor growth, psychomotor delay, behavioral disorders, hypotonia, ligamentous laxity, dental enamel hypoplasia, star-shaped iris, full lips with an everted lower lip, and a long face. Affected individuals may also present, particularly in childhood, with idiopathic hypercalcemia, which can contribute to irritability, muscle cramps, hypercalciuria and nephrocalcinosis [2].

In this case, the clinical finding of aortic stenosis, associated with facial dysmorphisms, immediately raised suspicion of Williams syndrome. The rapid diagnosis ensured that the infant received optimal care, including follow-up and supportive therapy, to detect and manage potential complications associated with the condition, at an early stage.

REFERENCES

[1] Morris CA, Braddock SR; Council on Genetics. Health Care Supervision for Children With Williams Syndrome. Pediatrics. 2020;145(2):e20193761.

[2] Sindhar S, Lugo M, Levin MD, Danback JR, Brink BD, Yu E, Dietzen DJ, Clark AL, Purgert CA, Waxler JL, Elder RW, Pober BR, Kozel BA. Hypercalcemia in Patients with Williams-Beuren Syndrome. J Pediatr. 2016;178:254-60.e4.

ABS 7

NON-FATAL DROWNING: A CASE REPORT

V. Carosio[1], C. Cocco[1], R. Basile[2], G. Cherchi[2]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Department of Pediatric Urgent Care, Brotzu Hospital, Cagliari, Italy*

BACKGROUND

Drowning, defined as a process leading to respiratory impairment from submersion or immersion in a liquid medium, is classified as either fatal or non-fatal, with or without morbidity. Drowning is a major global health issue, responsible for over 2.5 million preventable deaths in the last decade and ranking as the third leading cause of injury-related death in individuals under 14 years. Despite its prevalence, drowning remains poorly understood, with limited scientific evidence guiding emergency treatment, particularly in pediatric cases. This case presents a non-fatal drowning incident in a 4-year-old boy with no residual morbidity.

CASE PRESENTATION

A 4-year-old male child presented to the Pediatric Emergency Department following a non-fatal drowning incident. While snorkeling, he inhaled seawater after losing his snorkel. Upon resurfacing, he experienced dyspnea and cough, with white foam observed from his mouth. He remained conscious throughout the event and received oxygen therapy (6 L/min via mask) during transport. Initial evaluation revealed: moderate signs of respiratory distress with intermittent jugular and intercostal retractions; a respiratory rate of 35/min; oxygen saturation at 95%; hypotransmitted vesicular murmur throughout the lung fields with widespread coarse rales. Blood gas analysis showed mild acidosis (pH 7.31, BE -7.0 mmol/L). The chest X-ray indicated accentuation of interstitial markings. Treatment included oxygen therapy (2 L/min), oral betamethasone, and salbutamol via aerosol. The child's clinical picture remained stable with oxygen saturation around 95-96%, although he exhibited occasional desaturation to 92-93%, leading to his admission to the Pediatric Department. During the hospitalization he fully recovered and was discharged 48 hours later.

CONCLUSION

Drowning in children is a serious public health issue that requires attention on two fronts: prevention and standardization of emergency treatments. Preventative measures include constant supervision around water, using physical barriers to prevent children from accessing water, ensuring that children wear life jackets or floatation device, teaching swimming skills early, educating parents and caregivers on basic life support (BLS) and awareness of risk factors like epilepsy, channelopathies and neuropsychiatric disorders. Additionally, developing local, evidence-based guidelines for pediatric drowning emergencies is necessary. Ongoing research is crucial to enhance both prevention strategies and emergency treatment protocols.

ABS 8

CLEIDOCRANIAL DYSPLASIA

B.M. Trapani

Hôpital de Sion CHVR, Sion, Switzerland

A young couple, both in good health, bring their 16-month-old daughter to a medical appointment for a growth and psychomotor development check-up. The girl was still unable to stand, she ate very little and reluctantly, preferring only breast milk, as her mother was still breastfeeding her. In the family history, the father is 1.85 m tall, while the mother is 1.36 m tall. The child appears small and shows a slight psychomotor delay. During the clinical examination, psychomotor delay is confirmed, along with difficulty standing, delayed speech, and refusal to eat foods other than breast milk. The child is referred for further tests at the regional referral hospital. During pregnancy, fetal long bones were below the 5th percentile during biometric morphology scans. Molecular analysis of the *FGFR3* gene and *SHOX* gene in the mother's blood were negative. The baby was born at term by cesarean section due to fetopelvic disproportion, with no neonatal complications. At birth, she was small for her gestational age, with a head circumference of 33 cm (10th percentile) and wide fontanelles. A postnatal brain ultrasound was normal. Subsequently, various transfontanellar ultrasounds and neurobehavioral evaluations were performed, suggesting the presence of skeletal dysplasia. The child was then hospitalized for further investigation in the context of macrocephaly and wide fontanelles. During hospitalization, psychomotor developmental immaturity was noted. A brain and spinal MRI revealed a midline dysmorphism and a low conus medullaris, which was surgically treated with spinal cord detethering. Cleidocranial dysplasia is a rare skeletal dysplasia characterized by autosomal dominant inheritance. It often presents with delayed closure of the fontanelles, dental anomalies, and scoliosis, with potentially severe manifestations such as absent clavicles. Dental, hearing, and respiratory complications are also common, as is short stature in some cases. There is significant intrafamilial variability in stature and growth patterns. Cognitive development does not appear to be significantly affected. An individualized evaluation and genetic counseling for affected families are necessary, along with appropriate follow-up care.

REFERENCES

• Callea M, Fattori F, Bertini ES, Yavuz I, Bellacchio E, Avendaño A, Araque D, Lacruz-Rengel MA, Da Silva G, Cammarata-Scalisi F. [Clinical and molecular study in a family with cleidocranial dysplasia]. [Article in Spanish]. Arch Argent Pediatr. 2017;115(6):e440-4.

• Medina O, Muñoz N, Moneriz C. [Cleidocranial dysplasia: a case report]. [Article in Spanish]. Rev Chil Pediatr. 2017;88(4):517-23.

• Ramos Mejía R, Rodríguez Celin M, Fano V. Clinical, radiological, and auxological characteristics of patients with cleidocranial dysplasia followed in a pediatric referral hospital in Argentina. Arch Argent Pediatr. 2018;116(4):e560-6.

ABS 9

PANAYIOTOPOULOS SYNDROME: A CASE REPORT

L. Calandriello[1], V. Ferasin[2], S. Bellonzi[1], L. Dell'Anna[2], C. Lorenzetto[1], F. Marino[1], G. Passarella[1], F. Rigon[1], E. Fiumana[1], A. Mussari[1], L. Saggioro[3], F. Sansone[1], S. Rugolotto[1]

[1]*Division of Pediatrics, Rovigo Hospital, Rovigo, Italy*

[2]*School of Pediatrics, University of Padua, Padua, Italy*

[3]*School of Pediatrics, University of Verona, Verona, Italy*

INTRODUCTION

Panayiotopoulos syndrome (PS) is a benign focal seizure disorder occurring in early and middle childhood [1]. It represents the most common cause of afebrile nonconvulsive status epilepticus (SE) in children [2]. PS is characterized by seizures, often prolonged, usually nocturnal, with predominantly autonomic symptoms (emetic symptoms, cardio-respiratory and thermoregulatory alterations), and urinary incontinence. Unusual manifestations include ictal syncope, headache, unilateral eye and head deviation, ictal nystagmus, and motor automatisms. Consciousness is usually preserved at seizure onset with subsequent unresponsiveness in most patients [2]. 1/3 of seizures end in hemi- or generalized convulsions. The condition appears to be associated with a diffuse cortical hyperexcitability at electroencephalography (EEG) (multifocal, high-voltage sharp waves or spike-and-wave complexes typically over the posterior regions) [1] while neuroimaging (NI) studies do not show significant alterations [3].

CASE REPORT

Our patient was a healthy 10-year-old boy, with unremarkable personal and family history. The day before our evaluation, he suddenly complained of nausea associated with headache and followed by vomiting, pallor, and right deviation of the eyes. Body

temperature was 37.8°C. After being unresponsive for some minutes, he gradually recovered after a few hours of sleep. The next morning, he woke up with left hand automatism, then he presented vomiting, urinary incontinence, and unresponsiveness, so he was taken to the hospital. At our evaluation, he was found with tonic-clonic seizure of the arms, consciousness impairment, fixed gaze, left head deviation, desaturation, and low-grade fever. The intra-ictal EEG showed slow bilateral discharges and independent left-temporal and right-occipital spike-waves. SE was eventually interrupted by IV midazolam (MDZ). NI studies were normal, while cerebrospinal fluid (CSF) analysis showed a slight increase in white blood cell (WBC) count, in the absence of other data compatible with central nervous system infection. A few days later, the boy had an episode of headache followed by generalized tonic-clonic seizure with associated hypertonia, sialorrhea, and eyeballs revulsion, which ended spontaneously. Levetiracetam was prescribed as daily therapy, and oral MDZ as rescue therapy.

DISCUSSION AND CONCLUSION

PS is a self-limited condition; it usually remits within 1-2 years from onset. Seizures are infrequent in most patients. Prolonged seizures do not appear to result in residual neurological damage [1]. Education about the nature and prognosis of the syndrome is the cornerstone of correct management. Minor elevations in CSF WBC count can occur after SE of any type, nonetheless pleocytosis should not be attributed to SE alone unless other causes have been eliminated, as seen in our case. Regular antiepileptic treatment is probably most appropriately reserved for children in whom seizures are unusually frequent. Prolonged seizures in PS can reasonably be treated with "rescue" benzodiazepines as for febrile seizures [1].

REFERENCES

[1] Ferrie C, Caraballo R, Covanis A, Demirbilek V, Kivity S, Koutroumanidis M, Martinovic Z, Oguni H, Verrotti A, Vigevano F, Watanabe K, Yalcin D, Yoshinaga H. Panayiotopoulos syndrome: a consensus view. Dev Med Child Neurol. 2006;48(3):236-40.

[2] Semprino M, Galicchio S, Espeche A, Cersosimo R, Chacon S, Gamboni B, Adi J, Fasulo L, Fortini S, Cachia P, Gallo A, Caraballo RH. Panayiotopoulos syndrome: Unusual clinical manifestations, Epilepsy Behav. 2022:128:108552.

[3] Graziosi A, Pellegrino N, Di Stefano V, Raucci U, Luchetti A, Parisi P. Misdiagnosis and pitfalls in Panayiotopoulos syndrome, Epilepsy Behav. 2019;98(Pt A):124-8.

ABS 10

MEASURING KNOWLEDGE OF PARENTS AND CHILDREN WITH CONGENITAL HEART DISEASE: ADAPTATION AND VALIDATION OF THE "LEUVEN KNOWLEDGE QUESTIONNAIRE FOR CONGENITAL HEART DISEASE"

S. Flocco[1], F. Crò[1], C. Arrigoni[2], G. Conte[1], G. Zaffino[1], S. De Vecchi[1], L. Schianchi[1], G. Campioni[3], M. Chessa[4], R. Caruso[1,5], A. Magon[1]

[1]Health Professions Research and Development Unit, IRCCS Policlinico San Donato, San Donato Milanese, Milan, Italy

[2]Department of Public Health, Experimental and Forensic Medicine, Section of Hygiene, University of Pavia, Pavia, Italy

[3]Italian Guch Association (AICCA), San Donato Milanese, Milan, Italy

[4]Pediatric and Adult Congenital Heart Disease Center, IRCCS Policlinico San Donato, San Donato Milanese, Milan, Italy

[5]Department of Biomedical Sciences for Health, University of Milan, Milan, Italy

BACKGROUND

Congenital heart disease (CHD) is one of the most common congenital anomalies, affecting approximately 1% of live births worldwide. Advances in medical and surgical treatments have significantly improved the survival rates of children with CHD. However, the complexity of CHD and its management necessitates ongoing care and attention, which extends beyond the healthcare setting into the family environment. Parents play a pivotal role in the care of children with CHD. They are not only primary caregivers but also critical partners in managing the child's health. Effective parental involvement is associated with better adherence to treatment regimens, timely recognition of symptoms, and improved psychosocial outcomes for both the child and the family. Despite the importance of parental involvement, studies show that parents of children with CHD often have suboptimal levels of knowledge regarding the disease, its management, and potential complications. This knowledge gap can lead to inadequate care at home, increased anxiety, and poor health outcomes. To address this knowledge gap effectively, it is crucial to have reliable and valid tools to assess parental knowledge. The Leuven Knowledge Questionnaire for Congenital Heart Disease (LKQCHD) is a validated instrument designed to evaluate caregivers and patients knowledge across four key areas: a) the disease and its treatment; b) prevention of complications; c) physical activities; and d) reproductive issues. Although extensively tested in several languages, the LKQCHD has not yet been adapted for the Italian-speaking population. This study aims to translate and adapt the LKQCHD

into Italian and evaluate its content validity and psychometric properties specifically for parents and children with CHD.

METHODS

The study will follow a multi-phase, multi-method design consisting of: a) cultural and linguistic translation; b) content validity; c) construct validity; and d) reliability assessment. Each phase will be conducted according to COSMIN methodology, ensuring a rigorous and standardized approach to validating the tool. The cultural and linguistic translation phase involves forward- and back-translation procedures with bilingual experts to ensure accuracy and consistency. Content validity will be assessed by a panel of experts to ensure relevance and comprehensiveness in addressing parents and children knowledge gaps. Construct validity will involve testing the questionnaire with a sample of parents and patients with CHD, and reliability will be assessed using statistical measures. Data collection will be carried out at IRCCS Policlinico San Donato (San Donato Milanese, Milan, Italy).

RESULTS

In January 2024, the cultural and linguistic translation phase was completed. This phase involved three authors fluent in both Italian and English. The process included an initial translation from English to Italian, followed by a back-translation to ensure accuracy, and a final forward-translation to confirm consistency with the original version. The agreement among five experts on the initial draft of the Italian version was moderate (Fleiss' Kappa = 70.56% [95% CI 0.50-0.72]). This moderate agreement indicates initial reliability and sets the stage for subsequent phases of validation to further refine and confirm the tool's effectiveness in addressing the knowledge gaps among parents and children with CHD.

CONCLUSIONS

Validating the LKQCHD in Italian will enable healthcare providers to assess and address the knowledge levels of parents and children with CHD in a national context. This validation is crucial for improving the effectiveness of parents and patients education and ensuring better management of CHD.

ABS 11

NEW PERSPECTIVES ON NEONATAL LUNG ASSESSMENT

S. Carnazzo[1], S. Nasikas[2], F.F. Comisi[3]

[1]Department of Clinical and Experimental Medicine, University of Catania, Catania, Italy

[2]Department of Neonatology, Armand Trousseau Hospital, Sorbonne University, Paris, France

[3]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

INTRODUCTION

Ultrasound (US) use in diagnosing pulmonary diseases began in the 1960s, adapted from veterinary medicine. Over time, US role in pulmonary pathology has expanded, requiring a universal terminology (e.g., A-lines, B-lines, pleural sliding). In Neonatology, thoracic US is adopted for its accessibility, non-ionizing radiation, and image quality, complementing existing diagnostic tools. This study reviews thoracic US in Neonatology as a reliable, non-invasive diagnostic tool in diagnosing conditions like transient tachypnoea of the newborn (TTN), respiratory distress syndrome (RDS), meconium aspiration syndrome, and pneumothorax.

METHODOLOGY

Various probes are used empirically for pulmonary US evaluation. Linear probes (7.5-18 MHz) provide high resolution for pleural morphology. Convex probes (3.5-5 MHz) explore consolidations and pleural effusions. Sector probes (2-3.5 MHz) are mainly for cardiac but also thoracic evaluations. US images are real-time reconstructions, relying on artifacts like the hyperechoic pleural line. A-lines indicate a well-aerated lung; B-lines suggest partial aeration loss, and their coalescence forms "white lung." Lung anatomy is explored in anterior, lateral, and posterior segments.

FINDINGS IN NEONATES

Minutes after birth, A-lines and B-lines are visible in neonates' lungs. Cesarean-delivered newborns have more B-lines due to lack of thoracic compression. A-lines appear normal by 36 hours post-birth. Persistent B-lines beyond 24 hours may indicate chronic lung disease.

SPECIFIC CONDITIONS

Meconium aspiration syndrome can be diagnosed by US, showing consolidations with bronchograms, irregular pleural lines, scattered B-lines, and reduced A-lines. US monitors bronchoalveolar lavage, showing improved lung aeration.

TTN is a common neonatal respiratory distress cause, diagnosed by US with alveolar-interstitial syndrome, irregular pleural lines, "white lung," and the "double lung point sign."

RDS affects premature infants due to surfactant deficiency. US shows consolidation with air

bronchograms, compact B-lines, and irregular pleural lines. Scores like Lung Ultrasound Score (LUS) predict surfactant needs.
In pneumothorax, US shows absence of lung sliding, B-lines, lung pulse, and lung point, proving superior to radiography.

CONCLUSION

Lung US is a valuable method for diagnosing neonatal conditions, offering easy reproducibility and non-ionizing radiation, with adequate specificity and sensitivity. Poerio et al. demonstrated its predictive value for NICU admission in newborns with TTN or RDS after cesarean delivery [1]. Blank and colleagues showcased its effectiveness in monitoring postnatal respiratory adaptation [2]. Hedstrom et al. confirmed its role in enhancing RDS diagnosis when combined with the Silverman score [3]. Pulmonary US could complement neonatologists in the Delivery Room, emerging as a promising, versatile technology for neonatal respiratory assessment and management.

REFERENCES

[1] Poerio A, Galletti S, Baldazzi M, Martini S, Rollo A, Spinedi S, Raimondi F, Zompatori M, Corvaglia L, Aceti A. Lung ultrasound features predict admission to the neonatal intensive care unit in infants with transient neonatal tachypnoea or respiratory distress syndrome born by caesarean section. Eur J Pediatr. 2021;180(3):869-76.

[2] Blank DA, Rogerson SR, Kamlin COF, Fox LM, Lorenz L, Kane SC, Polglase GR, Hooper SB, Davis PG. Lung ultrasound during the initiation of breathing in healthy term and late preterm infants immediately after birth, a prospective, observational study. Resuscitation. 2017;114:59-65.

[3] Hedstrom AB, Faino AV, Batra M. Utility of lung ultrasound and respiratory severity score for detection of respiratory distress syndrome in the delivery room. Acta Paediatr. 2021;110:1683.

ABS 12

THE ODYSSEY OF CHRONIC PAIN

G. Sanna[1], F.F. Comisi[1], E. Esposito[1], C. Soddu[2], S. Savasta[1]

[1]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

[2]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", Cagliari, Italy

INTRODUCTION

Research from Johns Hopkins Hospital reveals that 40% to 80% of chronic pain patients receive an incorrect diagnosis, primarily due to inappropriate test selection and incomplete patient history collection. Poor understanding of sensitivity and specificity of commonly used medical tests, as well as improper use of anatomical testing to determine the physiological origins of pain, are common diagnostic errors. With a prevalence ranging from 11% to 38%, young adults and adolescents frequently experience chronic pain. When the underlying cause of pain cannot be identified, diagnoses like "chronic pain" or "amplified musculoskeletal pain disorder" are commonly employed as catch-all classifications. Chronic pain is multifaceted, making diagnosis more difficult; additionally, untreated pain can exacerbate mental distress and cause central sensitization. Non-steroidal anti-inflammatory drugs, corticosteroids, antiepileptics, opioids, and cannabinoids are only a few of the pharmacological drugs that are frequently rotated or combined for symptomatic treatment, despite the risk of addiction.

CASE REPORT

The case of two siblings, ages 18 and 21, who had chronic pain under investigation for five years, provides an example of this complexity. Both recounted experiencing sporadic, scorching pain in their limbs and chest that started in childhood; they also experienced significant cutaneous hyperemia, hyperhidrosis, and hyposthenia. Over time, these episodes, lasting from minutes to days, were further complicated by weakness and impaired mobility. Symptomatic treatment provided only partial relief and was accompanied with drowsiness and confusion. A previous diagnosis of juvenile-onset fibromyalgia had been made for one sibling. All tests were normal despite extensive investigations, which included complete body magnetic resonance imagings, chest radiographs, abdominal ultrasounds, infectious serology, liver, kidney, and thyroid function tests, autoantibody screening, metabolic studies, complete cerebrospinal fluid examinations, somatosensory evoked potentials, visual evoked potentials, electromyography, electroneurography, electrocardiogram, echocardiograms, and skin biopsies. Genetic testing using specific gene panels also revealed no abnormalities. Through clinical exome sequencing, a mutation in the *CLCN1* gene was discovered during an evaluation at the University of Cagliari's Pediatric Clinic, where the "microscopic" origins of altered pain sensitivity transmission have been examined. This mutation is suggestive of a rare form of myotonia congenita; both patients showed significant improvement in symptomatology and quality of life with an important decrease in the frequency and intensity of painful episodes after starting mexiletine, an etiology-specific drug.

CONCLUSIONS

By utilizing all available tests to find potential etiology-specific treatments, this case emphasizes the importance of a systematic, comprehensive diagnostic approach; this will help minimize diagnostic delays and the use of poor symptomatic treatments.

REFERENCES

- Hendler N. Why Chronic Pain Patients are Misdiagnosed 40 to 80% of the Time? J Recent Adv Pain. 2016;2(3):94-8.
- Stunnenberg BC, LoRusso S, Arnold WD, Barohn RJ, Cannon SC, Fontaine B, Griggs RC, Hanna MG, Matthews E, Meola G, Sansone VA, Trivedi JR, van Engelen BGM, Vicart S, Statland JM. Guidelines on clinical presentation and management of nondystrophic myotonias. Muscle Nerve. 2020;62(4): 430-44.
- Tanna V, Heathcote LC, Heirich MS, Rush G, Neville A, Noel M, Pate JW, Simons LE. Something Else Going On? Diagnostic Uncertainty in Children with Chronic Pain and Their Parents. Children (Basel). 2020;7(10):165.

ABS 13

PROPHYLAXIS IN CHILDHOOD IN THE REPUBLIC OF BULGARIA

T. Itova

University Hospital Medica Ruse Ltd., Ruse, Bulgaria
University of Ruse "Angel Kanchev", Ruse, Bulgaria

Childhood prevention has the task of ensuring health well-being for children and preventing disease and disability among them. The preventive measures that are carried out by the medical specialists in the Republic of Bulgaria are regulated in the "Children's Health" program. It includes mandatory preventive activities to monitor the growth and development of children from 0 to 18 years.

During the early neonatal period, all newborns undergo hearing screening and screening for hypothyroidism, phenylketonuria, and congenital adrenal insufficiency.

Parents choose a general practitioner to monitor their child's development and carry out regular immunizations. In the first year of life, monthly examinations are carried out to assess the state of health, from 2 to 7 years the examinations are every 3 months, and then annually – until the age of 18. Physical development is assessed monthly until the 12th month, twice a year until the age of 7 and then once a year until reaching adulthood. Neuropsychological development is assessed monthly during the first year, then once a year until 18 years of age.

During the first 12 months, children are subject to two screenings of vision and hearing, screening for dysplasia of the hip joints, prophylactic examination of blood indicators, albuminuria. Non-ultrasound screening for congenital anomalies of the excretory system is performed at 6 months of age.

For the Republic of Bulgaria, the mandatory immunization plan includes the administration of vaccines against tuberculosis, hepatitis B, diphtheria, tetanus, pertussis, *Haemophilus influenzae*, poliomyelitis, *Pneumococcus*, measles, mumps, rubella. At the request of parents, vaccines against rotavirus infection and human papillomavirus are administered.

Children with chronic diseases (including inflammatory bowel diseases, chronic diseases of the digestive system, tuberculosis, congenital syphilis, chronic hepatitis, AIDS, parasitic diseases, malignant and benign formations, cardiovascular diseases, diabetes, hypothyroidism and other deviations in thyroid gland function, congenital adrenal insufficiency, chronic anemias, congenital immunodeficiency states, congenital errors of metabolism, deviations in neuropsychological development, neurological diseases such as cerebral palsy and epilepsy, arthropathies, chronic arthritis, eye and ear diseases, etc.) are subject to dispensation. With the skillfully conducted pediatric consultation and dispensation of many chronically ill children, their quality of life has improved and is approaching that of their peers.

REFERENCES

- Ministry of Health of the Republic of Bulgaria. [Ordinance No. 8 of 3 November 2016 on prophylactic examinations and medical examinations]. [Ordinance in Bulgarian].
- Mumdzhiev N. [Fundamentals of Pediatrics]. [Book in Bulgarian]. Plovdiv: Raykov, 2001.
- Bobev D. [Pediatrics – a textbook for medical students]. [Book in Bulgarian]. Sofia: MI "Arso", 2000.

ABS 14

LYSOZYME IS HIGHLY EXPRESSED IN MATERNAL MILK STEM/PROGENITOR CELLS

G. Faa[1,2], G. Pichiri[3], P. Coni[3], M. Piras[3], Y. Gibo[4], A. Reali[5], V. Fanos[5]

[1]*Professor Emeritus, Department of Medical Sciences and Public Health, University of Cagliari, Cagliari, Italy*
[2]*Department of Biology, College of Science and Technology, Temple University, Philadelphia, Pennsylvania, USA*
[3]*Department of Medical Sciences and Public Health, University of Cagliari, Cagliari, Italy*

[4]Department of Gastroenterology, University of Matsumoto, Matsumoto, Japan
[5]Neonatal Intensive Care Unit, Department of Surgical Sciences, University of Cagliari, Cagliari, Italy

BACKGROUND AND AIMS

Lysozyme is present in maternal human milk, its content ranging from 3 up to 3,000 μg/ml, with a mean concentration of about 200-400 μg/ml. Lysozyme is an important modulator of innate immunity, participating in the immune response against bacteria and viruses [1]. Moreover, in weanling pigs, lysozyme can modulate the inflammatory response, playing a crucial role in preventing intestinal inflammation in the perinatal period [2]. For these reasons, recent data indicate that lysozyme could be used as a good alternative to antibiotic treatment [3]. Since few data are available on lysozyme expression in milk cells, and particularly in milk stem/progenitor cells, this study was aimed at investigating lysozyme immunoreactivity in the cell compartment of human colostrum.

MATERIALS AND METHODS

To this end, samples of human maternal colostrum were centrifuged and the pellet fixed in thin prep solution. Milk stem cells were isolated. Immunohistochemistry for lysozyme was performed using a routine method.

RESULTS

Lysozyme was strongly expressed in the vast majority of milk cells, including stem/progenitor cells. Lysozyme expression was detected in the cytoplasm and inside the nuclei of milk stem/progenitors.

DISCUSSION

Considering that human breastmilk contains a heterogeneous cell population, prevalently composed of stem and immune cells, we may hypothesize that lysozyme, as other stem and immune markers previously described by our group (CD44, CD33, ISL1, L1CAM), could be considered as an alternative signal of breast milk hierarchy and "plasticity." The fact that lysozyme reactivity was almost always present in the nucleus underlines this hypothesis. It remains to be established if lysozyme detected in our immunohistochemical experiments is produced by the reactive cells, or, alternatively, is a consequence of the selective milk protein diffusion. Further studies will reveal the significance of the high levels of lysozyme detected in our study inside milk stem cells.

REFERENCES

[1] Ragland SA, Criss AK. From bacterial killing to immune modulation: Recent insights into the functions of lysozyme. PLoS Pathog. 2017;13(9):e1006512.
[2] Long Y, Lin S, Zhu J, Pang X, Fang Z, Lin Y, Che L, Xu S, Li J, Huang Y, Su X, Wu D. Effects of dietary lysozyme levels on growth performance, intestinal morphology, non-specific immunity and mRNA expression in weanling piglets. Anim Sci J. 2016;87(3):411-8.
[3] Ferraboschi P, Ciceri S, Grisenti P. Applications of Lysozyme, an Innate Immune Defense Factor, as an Alternative Antibiotic. Antibiotics (Basel). 2021;10(12):1534.

ABS 15

TRIMETHYLAMINURIA

B.M. Trapani

Hôpital de Sion CHVR, Sion, Switzerland

A 43-year-old mother brings her fifth child for a check-up. The 8-year-old boy appears in excellent condition during the clinical examination, with height, weight, and body mass index appropriate for his age. His blood tests are perfect, and when the pediatrician asks if there are any other issues besides the routine check-up, the mother bursts into tears. She reports that the boy emits a very strong odor under certain conditions, such as when he eats fish and shellfish, cruciferous vegetables, and sometimes dairy (especially organic dairy bought from farmers), legumes, and soy. In these circumstances, he faces significant relational problems, being excluded from games and sports by his peers, which causes him great emotional distress. In the family, she is labeled as phobic and obsessive, and the child is seen as unable to interact with his peers. She asks the doctor for help.

The diagnostic hypothesis led to the fish odor syndrome, or primary trimethylaminuria. The child was referred to a specialized center outside the region, and the clinical diagnosis was confirmed. In fact, the boy was immediately subjected to a urine test, which, under normal dietary conditions, revealed more than 10% trimethylamine (TMA) in the urine, and the genetic test identified a homozygous mutation in the *FMO3* gene, responsible for the variant associated with trimethylaminuria. The deficiency in the enzyme flavin monooxygenase 3 (FMO3), which normally breaks down TMA, was confirmed. The genetic test indicates that the child has this variant in homozygosity, meaning he inherited the mutation from both parents. This

condition confirms that the psychosocial problems caused by the persistent odor are not related to the child or the mother but are the main issue of this disease. It is important that the pediatrician knows about it to avoid confusing a clinical problem with a psychological one.

Investigations are ongoing to determine whether he has one of the common variants, such as Glu158Lys and Glu308Gly, which are more frequent in patients, especially in the homozygous state. Given the patient's young age, it is crucial to implement dietary and personal hygiene measures to improve the situation before puberty, when the symptoms are expected to worsen. The patient should avoid fish (marine fish), eggs, red meat, and organ meats, legumes (beans, peas, lentils, soy), cabbage and broccoli, nuts and seeds, and soy-based products (all foods rich in choline, a precursor of TMA). Additionally, he should avoid dairy products such as aged cheese and butter.

The pediatrician must be aware of trimethylaminuria to diagnose the condition early, guide the parents in managing it with appropriate dietary measures and treatments, avoid unnecessary therapies, and provide psychological support to the child, preventing social and emotional issues. Furthermore, the pediatrician can offer genetic counseling to the parents to assess the risk of passing the condition on to future children.

REFERENCES

- Ferreira F, Esteves S, Almeida LS, Gaspar A, da Costa CD, Janeiro P, Bandeira A, Martins E, Teles EL, Garcia P, Azevedo L, Vilarinho L. Trimethylaminuria (fish odor syndrome): genotype characterization among Portuguese patients. Gene. 2013;527(1):366-70.
- Kloster I, Erichsen MM. Trimethylaminuria. Tidsskr Nor Laegeforen. 2021;141.
- Phillips IR, Shephard EA. Primary Trimethylaminuria. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A (Eds.). GeneReviews®. Available at: https://pubmed.ncbi.nlm.nih.gov/20301282/, date of publication: October 2007, last update: November 2020, last access: October 2024.

ABS 16

STEVENS-JOHNSON SYNDROME IN CHILDHOOD: TWO CASE REPORTS

F. Marino[1], L. Dell'Anna[2], V. Ferasin[2], F. Rigon[1], S. Bellonzi[1], L. Calandriello[1], F. Sansone[1], C. Lorenzetto[1], E. Fiumana[1], G. Passarella[1], A. Mussari[1], L. Saggioro[3], S. Rugolotto[1]

[1]*Division of Pediatrics, Rovigo Hospital, Rovigo, Italy*

[2]*School of Pediatrics, University of Padua, Padua, Italy*

[3]*School of Pediatrics, University of Verona, Verona, Italy*

Stevens-Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN) are rare, severe, CD8+ T-cell-mediated mucocutaneous reactions, characterized by blisters and skin detached, triggered by drugs or infections. They differ in the extent of the skin detached area and have high mortality (1-5% for SJS, 25-40% for TEN). Ocular sequelae, as blinding for corneal scarring, may occur. The diagnosis is mainly clinical, skin biopsy is not required. Rapid withdrawal of the culprit drug/infection and intensive supportive care are the basis of treatment. The use of systemic corticosteroids and intravenous immunoglobulins (IVIGs) is still controversial and no guidelines are available for the treatment for either adults or children.

CASE REPORTS

Case report 1

An 11-year-old boy, healthy, came to the Emergency Room (ER) for persistent fever and mucocutaneous rash which appeared after clarithromycin for pneumonia's suspicion. Physical examination revealed bullous, erythematous eruption in skin and oral, ocular, genital mucous membranes. The Nikolsky sign was positive. Microbiological tests showed active *Mycoplasma pneumoniae* infection. Clarithromycin was precautionally stopped and levofloxacin IV was started. Suspecting *Mycoplasma*-induced SJS, supportive care (fluid replacement, nutritional assessment, pain relief), methylprednisolone (1 mg/kg for 3 days) and IVIGs (3 g/kg in 3 days) were administered. After an initial improvement, the child showed deterioration of conditions with fever and oxygen deficiency. Clindamycin and ceftriaxone were added and steroid therapy was resumed. The patient was referred to a third level center and was discharged after 19 days of hospitalization without sequelae.

Case report 2

A 15-year-old boy, trisomy 21, in therapy with levotiroxin and allopurinol, came to the ER for fever, cough, feeding difficulties and conjunctivitis for 3 days and blistering of skin and oral mucous membranes for 1 day. Acyclovir was started but was discontinued after negative herpes microbiological tests. The patient's home drugs list was reviewed and allopurinol was found to have been started 2 weeks before the rash. Diagnosis of allopurinol-induced SJS was established, the causative drug was discontinued and supportive care (monitoring of fluid and electrolyte balance, nutritional support and empiric antibiotic therapy) was administered. Methylprednisolone (1 mg/kg for 2 days) and IVIGs (1 g/kg in 2 days) were started, with gradual

improvement. The boy was discharged without sequelae after 13 days of hospitalization.

CONCLUSIONS

Limited evidence is available to guide the treatment of SJS/TEN. Immunosuppressive treatment is still controversial; in the acute phase it is considered to be suitable, since a strong inflammation-like "cytokine storm" occurs in the patient, but during wide skin detachment, it may avoid re-epithelization and increase the risk of infection. Recently, the beneficial effects of cyclosporine and TNF-α antagonists have been explored, but further studies are necessary to define optimal management.

REFERENCES

- Jacobsen A, Olabi B, Langley A, Beecker J, Mutter E, Shelley A, Worley B, Ramsay T, Saavedra A, Parker R, Stewart F, Pardo Pardo J. Systemic interventions for treatment of Stevens-Johnson syndrome (SJS), toxic epidermal necrolysis (TEN), and SJS/TEN overlap syndrome. Cochrane Database Syst Rev. 2022;3(3):CD013130.
- Ramien M, Goldman JL. Pediatric SJS-TEN: Where are we now? F1000Res. 2020;9:F1000FacultyRev-982.

ABS 17

KAWASAKI DISEASE OR MIS-C?

S. Vendemmia[1], I. Pezone[2], M. Vendemmia[3]

[1]Emeritus Chief of Pediatrics and Neonatology, Hospital of Aversa, Aversa, Italy

[2]Division of Pediatrics and Neonatology, "San Giuseppe Moscati" Hospital, Aversa, Italy

[3]Neonatal Intensive Care Unit, University Federico II Naples, Naples, Italy

INTRODUCTION

Kawasaki disease is a complex and peculiar form of vasculitis that occurs mainly in infancy. It can be found in infants and children between 1 and 8 years of age. About 80% of the affected persons have, generally, less than 5 years. It appears to be prevalent in males. The peak ranges between 18 and 24 months of age. Cases between adolescents and adults are very rare. It is manifested by high temperature, 39-40°C, lasting at least 5-7 days, associated with irritability, intermittent abdominal pain, lethargy.

Suspicion of diagnosis can be raised by simultaneous presence of bilateral non-exudative conjunctivitis, lesions to the lips, presence of oral mucosa edema, hyperemia, cracking and dryness, reddened tongue with "strawberry or raspberry" appearance. Almost always a polymorphic, erythematous and macular exanthema is noted, sometimes orticarioid, morbilliform or scarlatiniform, more evident in the trunk, often accentuated in the perineal region. The rash can also occur on the face and limbs. Often obvious alterations at the extremities of the limbs: desquamation, erythema, edema. The edema is peculiar: taut, hard without the mark of the fovea. After the first week, usually, a particular periungual, palmar, plantar, perineal desquamation begins, and sometimes the superficial skin layer detaches into large flaps, highlighting a normal underlying skin. From a symptomatologic point of view, 30-40% of cases may have arthritis, arthralgia, thrombocytosis. Early manifestations are acute myocarditis with heart failure, arrhythmias, endocarditis, pericarditis. Coronary aneurysms may subsequently form. Dangerous is the giant coronary artery aneurysm that, although rare, can cause cardiac tamponade, thrombosis and heart attack.

Inflammation may affect the upper airways, pancreas, and bile ducts, kidneys, mucous membranes, extravascular tissue, lymph nodes.

CLINICAL CASE

Male subject, 15 years old, temperature 39-40°C for 7 days, dehydrated, with raspberry tongue, reddened and edematous oral mucosa, dry and slightly fissured lips, bilateral conjunctivitis, mild drowsiness, headache, muscle and joint pain. Massive maculo-papulous exanthema on the entire body surface, irregular bowel function with intermittent abdominal pain. No desquamative marks at the extremities. Systolic puff 1-2/6 on the mesocardium, no meningeal signs, to the chest extension of the expiratory phase. Neurological reflexes in the normal, left humeral pressure 90/40. Laboratory tests: high erythrocyte sedimentation rate, C-reactive protein, antinuclear antibody and anti-smooth muscle antibody, rheumatoid factor test was normal, leukopenia, neutropenia, mild lymphocytosis, immature cell enlargement. Serological evidence of elevated COVID-19 infection. Negative cultures, moderate thrombocytosis, high lactate dehydrogenase, slightly altered liver and pancreatic enzymes. Echocardiogram within limits. The therapy practiced was symptomatic: hydration, antipyretics, symbiotic. After 10 days the patient was in good condition. The diagnosis of multisystemic inflammatory syndrome related to COVID-19 (MIS-C) was then formulated. This pathology should be kept in mind. It has been rarely observed and described as a complication of SARS-CoV-2 infection. Symptoms in children and adults are remarkably similar to those of Kawasaki disease.

CONCLUSIONS

In such a case, it is worth being careful and prudent, as the symptoms are extremely misleading. It is advisable to carefully follow the symptomatologic evolution, to carry out all useful laboratory tests and the necessary instrumental investigations to exclude a Kawasaki disease. The positive results from the tests for SARS-CoV-2 infection and further examinations and observations of the patient will make us place the diagnosis of MIS-C.

REFERENCES

- Burney JA, Roberts SC, DeHaan LL, Shimizu C, Bainto EV, Newburger JW, Dominguez S, Jone PN, Jaggi P, Szmuszkovicz JR, Rowley AH, Samuy N, Scalici P, Tremoulet AH, Cayan DR, Burns JC; KIDCARE Study Investigators. Epidemiological and Clinical Features of Kawasaki Disease During the Covid-19 Pandemic in the United States. JAMA Netw Open. 2022;5(6):e2217436.
- Darby JB, Jackson JM. Kawasaki Disease and Multisystem Inflammatory Syndrome in Children: An Overview and Comparison. Am Fam Physician. 2021;104(3):244-52.

ABS 18

RISK FACTORS FOR RECOARCTATION OF THE AORTA AFTER SURGICAL REPAIR IN PEDIATRIC PATIENTS

G. Concas[1], E. Esposito[2], A. Atzei[1], P. Neroni[1], A. Giamberti[3], M. Chessa[4], V. Fanos[1]

[1]Neonatal Intensive Care Unit, AOU Cagliari, Cagliari, Italy

[2]School of Pediatrics, University of Cagliari, Cagliari, Italy

[3]IRCCS Policlinico San Donato, Department of Congenital Cardiac Surgery, San Donato Milanese, Milan, Italy

[4]IRCCS Policlinico San Donato, Department of Pediatric and Adult Congenital Cardiology, San Donato Milanese, Milan, Italy

BACKGROUND

Aortic coarctation is the fifth most common congenital heart disease, accounting for 5-8% of all congenital heart defects [1]. Over the years, the survival of pediatric patients who have undergone corrective surgery has improved considerably. However, recoarctation after surgery is still a major problem [2]. Considering this aspect, we have reviewed our experience at "IRCCS Policlinico San Donato" (San Donato Milanese, Milan, Italy) over the last 19 years.

METHODS

Demographic and clinical data were collected on all patients who underwent cardiac surgery for aortic recoarctation at the "IRCCS Policlinico San Donato," from August 2004 to March 2023. A total of 195 patients aged 0-14 years were recorded and subsequently analyzed.

RESULTS

The incidence of recoarctation in our population was 20% (39/195). At the time of surgery, in the total study population, the median age was 30 days and the median weight was 3.8 kg; in the group of patients who presented recoarctation, the median age was 10 days and the median weight was 3.3 kg. Among the parameters considered, younger age ($p = 0.00016$) and lower weight ($p = 0.00033$) at the time of surgery were found to be risk factors for recoarctation. The other considered factors (sex, time of diagnosis, presence of associated defects, choice of surgical technique, type of surgical approach, development of arterial hypertension after surgery, presence of arterial pulses in the lower extremities and signs of ventricular hypertrophy on electrocardiogram) showed no significant differences between the total study population and the group of patients who presented recoarctation.

CONCLUSIONS

Our data are in line with the current literature showing that age and weight at the time of surgery appear to be key factors in determining the likelihood of recoarctation in patients undergoing cardiac recoarctation surgery in childhood. This study underlines that a close follow up will be necessary in all patients but a closer follow up should be envised for ones at highest risk, like younger and smaller patients. In the future, it is advisable to identify other possible risk factors by analyzing larger cohorts of patients with long-term follow-up.

REFERENCES

[1] Mitchell SC, Korones SB, Berendes HW. Congenital heart disease in 56,109 births. Incidence and natural history. Circulation. 1971;43(3):323-32.

[2] Minotti C, Scioni M, Castaldi B, Guariento A, Biffanti R, Di Salvo G, Vida V, Padalino MA. Effectiveness of Repair of Aortic Coarctation in Neonates: A Long-Term experience. Pediatr Cardiol. 2022;43(1):17-26.

ABS 19

PRIMARY LYMPHEDEMA: TWO CASE REPORTS IN CHILDREN

L. Dell'Anna[1], F. Marino[2], F. Rigon[2], S. Bellonzi[2], E. Fiumana[2], L. Calandriello[2], F. Sansone[2], C. Lorenzetto[2], G. Passarella[2], V. Ferasin[1], L. Saggioro[3], A. Mussari[2], C. Bertolin[4], S. Rugolotto[2]

[1]School of Pediatrics, University of Padua, Padua, Italy

[2]Division of Pediatrics, Rovigo Hospital, Rovigo, Italy

[3]School of Pediatrics, University of Verona, Verona, Italy
[4]Clinical Genetics Unit, Department of Women's and Children's Health, University of Padua, Padua, Italy

INTRODUCTION

Lymphedema (LE) is an abnormal accumulation of fluid that causes limb swelling, buildup of fibrous connective tissue and chronic inflammation. It can be primary, caused by a developmental abnormality of lymphatic system, or secondary, caused by damage to the lymphatic system (radiation, surgery, tumors). In children it is more often primary and can be associated with various genetic syndromes (Noonan's and Turner's syndromes, etc.) or can be idiopathic. According to the age of onset, primary LE can be congenital, praecox or tarda. This chronic disease predisposes to infections and causes a reduction in quality of life. The management of primary LE generally involves conservative measure. Surgery may be considered as an alternative option.

CASE REPORTS

Case report 1

A 10-year-old girl, healthy, with a body mass index of 28, accessed the Emergency Room (ER) for swelling of both legs accompanied by inability to use usual footwear for a week. She denied trauma, fever or previous surgery; no family history of edema. Physical examination revealed non-pitting edema of both foot, leg and thigh. Skin temperature was normal, pulses were palpable and there was no joint involvement. There were no clinical features of hypercortisolism and Tanner's scale was: PH1, B2, I1. Renal, liver and thyroid function tests were normal. Doppler ultrasound ruled out venous insufficiency or thrombosis and abdominal-pelvic magnetic resonance imaging excluded secondary LE caused by a compressive mass. Lymphoscintigraphy showed the absence of tracer uptake into the inguinal nodes indicating lymphatic dysfunction. The girl was referred to a third level center with the diagnosis of primary LE praecox associated with pubertal onset.

Case report 2

A 10-year-old boy presented to ER with suspected left ankle's soft-tissue infection. History revealed surgery for distichiasis and family history of LE (mother and maternal grandmother). Physical examination revealed warm and erythematous edema in left foot, non-pitting edema in right foot. Ultrasound found out imbibition of soft tissues without signs of joint inflammation; the patient underwent drainage for purulent collection in the left ankle and antibiotic therapy. Suspecting soft-tissue infection secondary to hereditary LE, lymphoscintigraphy was performed and showed a delayed transit time to the regional lymph nodes and collateral lymphatic channels of both lower limbs. The identification of a heterozygous *FOXC2* pathogenic variant (c.595dupC) by molecular genetic testing confirmed the diagnosis of LE-distichiasis syndrome. The patient was referred to a third level center.

CONCLUSIONS

Primary LE is rare, affecting 1/100,000 children, and can be difficult to identify. Diagnosis is often clinical and requires exclusion of secondary causes. Lymphoscintigraphy can be a helpful adjunct; it is 100% specific and 96% sensitive, and it is the gold standard for diagnosis. The literature on treating primary LE is limited and, for better case management, it is important to address patients to a specialized center.

REFERENCES

• Colmant C, Turpin S, Lambert R, Wong N, Ondrejchak S, Lapointe C, Powell J, Dubois J, McCuaig C. Pediatric Lymphedema: Study of 180 Patients Referred to a Tertiary Lymphedema Clinic. J Cutan Med Surg. 2022;26(5):502-11.
• Vignes S, Albuisson J, Champion L, Constans J, Tauveron V, Malloizel J, Quéré I, Simon L, Arrault M, Trévidic P, Azria P, Maruani A; French National Referral Center for Primary Lymphedema. Primary lymphedema French National Diagnosis and Care Protocol (PNDS; Protocole National de Diagnostic et de Soins). Orphanet J Rare Dis. 2021;16(1):18.

ABS 20

A COMMON PRESENTATION, YET A RARE DIAGNOSIS: INSIGHTS INTO MRFACD SYNDROME

F.F. Comisi[1], E. Esposito[1], G. Sanna[1], M. Marica[2], C. Soddu[2], S. Savasta[1]

[1]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy
[2]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", Cagliari, Italy

INTRODUCTION

MED13L-related intellectual disability/MRFACD (mental retardation and distinctive facial features with or without cardiac defects) syndrome is an autosomal dominant disorder caused by *de novo* mutations in the *MED13L* gene [1, 2]. Key features include motor delay, intellectual disability, and speech delay. Characteristic facial dysmorphisms

consist of frontal bossing, up-slanting palpebral fissures, a broad or depressed nasal root, macrostomia, macroglossia, and ear anomalies [1, 2]. Cardiac defects range from none to patent foramen ovale, septal defects, or transposition of the great arteries [1]. Although common among syndromic intellectual disabilities, only about 100 cases have been reported [2]. Herein, we further delineate the phenotypic spectrum of this syndrome by presenting two cases of affected individuals, each showing distinct clinical features associated with MRFACD.

CASE REPORTS

Case report 1

The first patient is a 4-year-old male, born full term to non-consanguineous parents. The perinatal period involved respiratory distress and hypotonia. Examination revealed persistent axial hypotonia, brachycephaly, frontal bossing, low-set ears, epicanthal folds, telecanthus, right-sided ptosis, a bulbous nasal tip, and micrognathia. At age 1, he developed drug-resistant seizures. Magnetic resonance imaging showed corpus callosum abnormalities. By age 2, conical dental anomalies appeared. Initial genetic tests, including single nucleotide polymorphism array (SNP-array) and *FMR1* analysis, were negative. Next-generation sequencing (NGS) identified a heterozygous c.700A>G (Lys234Glu) missense variant in the *MED13L* gene, confirming MRFACD syndrome.

Case report 2

The second patient is an 11-year-old girl, born at 42 weeks to consanguineous parents. The perinatal period was complicated by cyanosis due to meconium aspiration. Examination revealed frontal bossing, hypertelorism, epicanthal folds, amblyopia, strabismus, preauricular tags, depressed nasal bridge, macrostomia, macroglossia, bifid uvula, pterygium, winged scapulae, lumbar scoliosis, fetal pads, coxa valga, and toe clinodactyly. At age 4, she showed oppositive and obsessive behaviors with episodes of self-harm. She also had dysphagia and urinary incontinence. Genetic testing, including comparative genomic hybridization array (CGH-array), *FMR1* analysis, and tests for Pompe and Gaucher diseases, was negative. NGS identified a heterozygous c.1768C>T (p.Gln590) nonsense variant, confirming MRFACD syndrome.

CONCLUSIONS

These cases show the clinical variability of MRFACD syndrome. Both patients had typical facial dysmorphisms and developmental delays, but also unique features like drug-resistant seizures, corpus callosum abnormalities, and behavioral issues. This variability underscores the importance of advanced genetic testing, such as NGS, for accurate diagnosis in syndromic intellectual disabilities. Understanding the full spectrum of *MED13L* mutation effects plays a key role in diagnosis, management, and genetic counseling. Further case studies are needed to enhance knowledge of genotype-phenotype correlations and improve patient care.

REFERENCES

[1] Adegbola A, Musante L, Callewaert B, Maciel P, Hu H, Isidor B, Picker-Minh S, Le Caignec C, Delle Chiaie B, Vanakker O, Menten B, Dheedene A, Bockaert N, Roelens F, Decaestecker K, Silva J, Soares G, Lopes F, Najmabadi H, Kahrizi K, Cox GF, Angus SP, Staropoli JF, Fischer U, Suckow V, Bartsch O, Chess A, Ropers HH, Wienker TF, Hübner C, Kaindl AM, Kalscheuer VM. Redefining the MED13L syndrome. Eur J Hum Genet. 2015;23(10):1308-17.

[2] Bessenyei B, Balogh I, Mokánszki A, Ujfalusi A, Pfundt R, Szakszon K. MED13L-related intellectual disability due to paternal germinal mosaicism. Cold Spring Harb Mol Case Stud. 2022;8(1):a006124.

ABS 21

ADMINISTERING DRUGS IN NEONATAL POPULATION THROUGH IN-LINE FILTERS: RESULTS OF FILTERABILITY STUDIES

C. Capetti[1], E. Maggiora[1,2], M. Capitanio[1,2], S. Deantoni[1], C. Quarti[3], C.G. Catona[3], F. De Matteis[2], S. Cosimi[2], M. Luciano[2], E. Bertino[1], A. Coscia[1,2], F. Cresi[1,2]

[1]*Neonatal Intensive Care Unit, Department of Public Health and Pediatrics, University of Turin, Turin, Italy*

[2]*Città della Salute e della Scienza di Torino, Turin, Italy*

[3]*Pall Medical part of Cytiva, Buccinasco, Milan, Italy*

BACKGROUND AND AIMS

The use of in-line filtration for intravenous infusions protects patients against inadvertent particulates, air and microbial contamination, but evidence in newborns is still lacking. To verify the effectiveness of in-line filtration, the multicenter randomized controlled trial "Intravenous Neonatal Central Access Safety" (INCAS trial) was designed. The aim of this preparatory study for the INCAS trial was to evaluate the filterability of the 15 most commonly used drugs in neonatal intensive care. Below are the filterability results of 5 of these drugs.

METHODS

Compatibility for doses and concentrations between 90 drugs and filtration has already been validated in pediatric and adult studies. The filterability of dopamine, dobutamine, amikacin and recombinant

human (rH) insulin was known for adults only; for dexmedetomidine no data were available. These 5 drugs were analyzed for filterability through Pall NEO96E filters in neonatal condition ("worse case" in terms of drug infusion therapy for low concentration and low volume) before and after passage through the filter by ultraviolet spectrophotometry and high performance liquid chromatography (HPLC). The drug concentration was measured upstream and downstream of the filter to determine whether any drug binding occurred.

RESULTS

There was negligible binding for 4 of the 5 drugs tested. rH-insulin was completely adsorbed by the filter when used at concentrations and infusion rates suitable for infusion in newborns.

CONCLUSION

The compatibility between drugs and filtration validated in adults can be assumed for newborns, but exceptions may exist. Under the testing conditions described above, no binding was detected on 4 of the 5 drugs tested. rH-insulin in the neonatal population is not compatible with filtration.

ABS 22

A RETROSPECTIVE ANALYSIS OF UPJO CLINICAL BURDEN IN SARDINIAN CHILDREN

A. Bertini[1], M. Cossu[1], M.E. Murru[1], M. Gamberoni[1], V. Manca[2], E. Flore[2], F. Zurrida[2], G. Masnata[2]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy*

Ureteropelvic junction obstruction (UPJO) is a CAKUT (congenital anomaly of the kidney and urinary tract): it is the most prevalent obstructive uropathy. Its optimal management remains a topic of debate. Nonetheless, there is a growing trend towards non-surgical management. If not detected and treated properly, UPJO evolves in significant kidney damage or even kidney loss. UPJO can be detected early during pregnancy through ultrasound (US) in the second trimester [1]. Compression at the ureteropelvic junction can be caused by both congenital and acquired factors (fibrosis, retroperitoneal lymphadenopathy, retroperitoneal masses, ectopic kidneys, ureteral tumors or iatrogenic causes). Congenital causes are the most common; they include: a peristaltic segment of the ureter, high insertion of the ureter into the renal pelvis or an abnormal polar vessel (APV) that crosses and compresses the ureter. APV may originate from the renal vessels, aorta, vena cava, or iliac vessels [2]. Intermittent griping pain, vomiting, recurrent urinary tract infections and abdominal masses are UPJO symptoms. UPJO is more prevalent in males and often affects the left side [3]. All patients with suspected UPJO should undergo a thorough evaluation of glomerular filtration rate, urine test and renal-bladder US. Most patients can resolve their UPJO and hydronephrosis without surgery. Anteroposterior (A-P) renal pelvic diameter is a significant factor in predicting hydronephrosis' progression: functional damage is absent below 12 mm, rare between 12 and 20 mm, and frequent and rapidly progressive above 40 mm. Surgical intervention may be necessary in a patient with deteriorated renal function (evaluated with angiography) and increased A-P pelvic diameter. We conducted a retrospective analysis on 216 patients with hydronephrosis followed up by our Unit. Our analysis revealed that 130 of these patients presented with primary UPJO and 11 with an APV. Our analysis focused on the A-P grade of pelvic dilation at the surgery time in both groups. Patients with UPJO who underwent surgery demonstrated a significant prevalence of IV grade dilation (76%) and the remaining patients had III grade dilation. Notably, no patients with lower dilation grade underwent surgical intervention; instead, conservative management was chosen. Remarkably, only 35% of the surgical patients exhibited symptoms. Parallel findings were observed in operated APV patients: 73% presented a IV grade dilation and the remaining 27% had a III grade dilation. No patients with lower grades were operated. In contrast to the earlier group, a significantly higher proportion of APV patients (73%) presented symptoms before surgical intervention. Regarding the side involved in these two groups, we observed that APV exclusively affected the left side, while UPJO demonstrated a more balanced distribution, with cases of both bilateral and unilateral involvement.

These data demonstrate that a follow-up is essential to identify and treat UPJO before the onset of irreversible kidney damage. A multidisciplinary team, comprised of pediatric nephro-urologist, radiologist, primary care physician and gynecologist (essential for prenatal diagnosis) is indispensable for optimal patient management.

REFERENCES

[1] Jackson L, Woodward M, Coward RJ. The molecular biology of pelviureteric junction obstruction. Pediatr Nephrol. 2018;33(4):553-71.

[2] Al Aaraj MS, Badreldin AM. Ureteropelvic Junction Obstruction. In: StatPearls [Internet]. Available at: https://www.ncbi.nlm.nih.gov/books/NBK560740/, last update: July 2023, last access: October 2024.

[3] Grasso M, Caruso RP, Phillips CK. UPJ Obstruction in the Adult Population: Are Crossing Vessels Significant? Rev Urol. 2001;3(1):42-51.

ABS 23

CLINICAL VARIABILITY IN SSADHD

M. Gamberoni[1], C. Sbaffi[1], G. Sanna[1], M. Asunis[2], M. Balzarini[3], E. Flore[3], D. Pruna[2], G. Masnata[3]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Pediatric Neurology and Epileptology Unit, Pediatric Department, ARNAS Brotzu/ASL, Cagliari, Italy*

[3]*Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy*

Succinic semialdehyde dehydrogenase deficiency (SSADHD) is a rare autosomal recessive disorder of γ-aminobutyric acid (GABA) catabolism caused by variants in the *ALDH5A1* gene. Around 450 patients have been diagnosed worldwide [1]. This disease has a nonspecific, variable clinical presentation with a slowly progressive or static course. Presenting symptoms include: developmental delay; hypotonia; ataxia; movement, neuropsychiatric and sleep disorders; generalized seizures and sudden unexpected death in epilepsy [1]. SSADHD suspicion can arise from clinical presentation, altered biochemical findings (accumulation of γ-hydroxybutyric acid in body fluids, easily detected as organic aciduria) and magnetic resonance imaging findings (hyperintense, symmetric, bilateral lesions in the globus pallidus) that leads to metabolic and genetic testing to confirm the diagnosis [1].

CASE REPORTS

We report two unrelated female patients of Sardinian heritage with different clinical outcomes. E., 9 years old, was evaluated at 16 months for mild hypotonia in the upper and lower limbs with joint laxity, clubfoot, global development delay and non-evocable deep tendon reflexes (DTR) in the lower limbs. Altered electroencephalogram preceded the development of absence seizures at 2 years of age. In later years, E. developed ataxia, hands stereotypies, sleep apnea, dorsal-lumbar scoliosis and rounded facies. After specific behavioral therapy there has been an improvement in her abilities, mostly regarding speech.

N., 10 years old, at 6 months was evaluated for poor head control, sucking difficulty, drowsiness, poor weight gain, marked axial and upper limb hypotonia, neurodevelopmental delay, bite malocclusion with dental malposition, diffuse joint hyperlaxity, hypoevocable DTR in the upper limbs. She emitted vocalizations at 5 months in absence of words and subsequently a reduction in social interaction has been reported, associated with vocal and motor stereotypes. Walking onset was at 30 months of age, showing ataxia and intra-rotated left foot. In the following years, N. developed nonverbal autism spectrum disorders (ASD), behavioral pollakiuria and avoidant restrictive food intake disorder (ARFID), that persist nowadays.

In both cases, genetic testing for *ALDH5A1* highlighted the presence of nonsense likely pathogenic variant in exon 10, c.1429C>T, p.(Gln474Ter), confirming the diagnosis.

CONCLUSIONS

Although both patients had the same variant and similar initial presentation, their clinical outcomes were considerably different. The genetic and epigenetic factors underlying the wide clinical variability remain to be elucidated.

Current treatment of SSADHD is only symptomatic, targeting neurological and psychiatric manifestations. Ongoing research focus on enzyme replacement therapy utilizing viral vectors [2] and gene therapy (gene editing, e.g., CRISPR-Cas9) as valid options for the future [3].

REFERENCES

[1] Sergi C, Parayil Sankaran B. Succinic Semialdehyde Dehydrogenase Deficiency. In: StatPearls [Internet]. Available at: https://www.ncbi.nlm.nih.gov/books/NBK560724/, last update: August 2023, last access: October 2024.

[2] Lee HHC, Latzer IT, Bertoldi M, Gao G, Pearl PL, Sahin M, Rotenberg A. Gene replacement therapies for inherited disorders of neurotransmission: Current progress in succinic semialdehyde dehydrogenase deficiency. J Inherit Metab Dis. 2024;47(3):476-93.

[3] Afshar-Saber W, Chen C, Teaney NA, Kim K, Yang Z, Gasparoli FM, Ebrahimi-Fakhari D, Buttermore ED, Pin-Fang Chen I, Pearl PL, Sahin M. Generation and characterization of six human induced pluripotent stem cell lines (hiPSCs) from three individuals with SSADH Deficiency and CRISPR-corrected isogenic controls. Stem Cell Res. 2024;77:103424.

ABS 24

AN UNSUPERVISED LEARNING TOOL FOR STEM CELL BURDEN ASSESSMENT IN THE PRETERM KIDNEY

M. Fraschini[1], V. Fanos[2,3], L. Didaci[1], G. Faa[4,5]

[1]*Department of Electrical and Electronic Engineering, University of Cagliari, Cagliari, Italy*

[2]*Neonatal Intensive Care Unit, AOU Cagliari, Cagliari, Italy*

[3]*Department of Surgical Sciences, University of Cagliari, Cagliari, Italy*

[4]*Professor Emeritus, Department of Medical Sciences and Public Health, University of Cagliari, Cagliari, Italy*

[5]*Department of Biology, College of Science and Technology, Temple University, Philadelphia, Pennsylvania, USA*

BACKGROUND AND AIM

Human kidney development is a complex process requiring interactions between pluripotential stem cells, undifferentiated mesenchymal cells of the metanephros, and both epithelial and mesenchymal components, eventually leading to the insurgence of the complex renal architecture [1]. Given this complexity, interpreting renal histology in the newborn can be challenging and often requires expert histopathologists. The evaluation of the maturation stage of the neonatal kidney, as well as the assessment of the stem cell compartment, indicating the ability of a single kidney to generate new nephrons, is a significant challenge in clinical practice [2]. In recent years, some machine learning models have been proposed as new tools for rapid, automated glomerular annotation and assessment of all renal cell types involved in the complex kidney architecture [3]. This study aimed to evaluate the ability of an unsupervised learning algorithm to speed up the measurement of pluripotential renal cells for the evaluation of their residual nephrogenesis potential in a newborn kidney.

PATIENTS AND METHODS

One neonatal kidney was routinely processed for this study. One micron-thick section was H&E-stained and digitalized using the PrimeHisto XE scanner (Pacific Image Electronics Co., Ltd., New Taipei City 221, Taiwan). The subcapsular area occupied by mesenchymal pluripotential cells (appearing as a "blue strip") was evaluated using an unsupervised learning clustering approach based on the k-means clustering algorithm.

RESULTS

The proposed tool allows pathologists to visually inspect and interpret the segmented regions within the whole-slide image. Using segmentation techniques and machine-learning models, we have identified and highlighted different tissue types and structures in the original raw image. On the quantitative side, our approach allows quantifying the segmented regions' extensions within the original raw image. Furthermore, the proposed tool successfully allowed a fast evaluation of the stem cell compartments.

CONCLUSIONS

Machine learning models may represent a new important tool for analyzing of the complex architecture of newborn kidneys. Further studies are needed to extend the use of supervised approaches capable of analyzing the interglomerular stroma of the renal cortex and the interstitial cells of the medulla, with the aim of advancing neonatal nephrology.

REFERENCES

[1] Faa G, Gerosa C, Fanni D, Monga G, Zaffanello M, Van Eyken P, Fanos V. Morphogenesis and molecular mechanisms involved in human kidney development. J Cell Physiol. 2012;227(3):1257-68.

[2] Chevalier RL, Charlton JR. The Human Kidney at Birth: Structure and Function in Transition. In: Faa G, Fanos V (Eds.). Kidney Development in Renal Pathology. New York: Springer, 2014.

[3] Simon O, Yacoub R, Jain S, Tomaszewski JE, Sarder P. Multi-radial LBP Features as a Tool for Rapid Glomerular Detection and Assessment in Whole Slide Histopathology Images. Sci Rep. 2018;8(1):2032.

ABS 25

MOVEMENT-DEPENDENT MODULATION OF MULTISENSORY INTEGRATION EARLY IN LIFE: THE DEVELOPMENTAL CONTEXT OF A SPATIAL BODILY-SELF REPRESENTATION

C. Peila[1], S. Deantoni[1], L. Riboldi[1], G. Serra[1], A. Rossi Sebastiano[2], I. Ronga[2,3], K. Poles[2], S. Russo[4], A. Comanducci[5], A. Coscia[1], E. Bertino[1], A. Pigorini[4], F. Garbarini[2]

[1]*Neonatal Unit, Department of Public Health and Pediatrics, University of Turin, Turin, Italy*

[2]*Manibus Lab, Department of Psychology, University of Turin, Turin, Italy*

[3]*BIP Research Group, Department of Psychology, University of Turin, Turin, Italy*

[4]*Department of Biomedical, Surgical and Dental Sciences, University of Milan, Milan, Italy*

[5]*IRCCS Fondazione Don Carlo Gnocchi Onlus, Milan, Italy*

BACKGROUND

Compelling evidence shows that immediately after birth the neural mechanism underlying multisensory integration (MSI) of stimuli occurring on the body or within the peripersonal space is modulated by body proximity: boosted responses are elicited whether they are presented near rather than far from the body surface. The spatial modulation of MSI can be considered as an implicit physiological marker of an early ability to distinguish the own body as a

separate entity from the environment and to define bodily boundaries, then leading to the construction of a mental functional bodily-self representation, already present early in life. Early motor experience may normally represent the developmental context allowing to encode multisensory stimuli in a body-centered reference frame, if synchronous in time and congruent in space, by anchoring visual, auditory, and tactile signals to the body through proprioceptive inputs.

METHODS

Based on these premises, it has been addressed whether MSI is modulated by the directionality of movements performed with the own hand, that is, whether distinct patterns of MSI can be measured following movements directed either to the self-body or to the external environment. Therefore, an audio-tactile paradigm able to capture this possible effect has been devised and submitted to a population of 18 healthy term newborns soon after birth and 18 healthy infants at 4/5 months of postnatal life. Tactile stimuli were delivered on the dorsum of the right hand, either alone or concomitant with sounds played by a loudspeaker placed near the body midline. Crucially, before each trial, a postural manipulation was leveraged by dragging the hand in two positions: toward the body midline (self-directed) or away from it (externally directed). While administering the sensory stimuli, which could be both bimodal or unimodal, the electroencephalography (EEG) signal has been recorded.

RESULTS

From the analysis of evoked related potential (ERP) responses, it has been proven that the postural manipulation induced a body-centered modulation of multisensory processing. In other words, not only the EEG showed greater responses in the bimodal stimulation, but irrespectively of the position of the loudspeaker, over a centroparietal channel cluster, greater MSI responses were observed following self-directed, as compared to externally-directed movements. Already in newborns and later in infants, the evoked responses followed an adult-like pattern with an enhanced amplitude of ERPs after the passive movement of the hand aimed toward the self.

CONCLUSIONS

This finding suggests that newborns are already capable of encoding the reciprocal localization between the self-body, in a trunk-centered reference frame, and specific body districts, such as the hand, necessary to direct voluntary and oriented movements.

ABS 26

BASIC FAMILY-CENTERED CESAREAN SECTION OUTCOMES: A RETROSPECTIVE COHORT STUDY

L. Saggioro[1], F. Marino[2], L. Dell'Anna[3], V. Ferasin[3], F. Rigon[2], S. Bellonzi[2], L. Calandriello[2], F. Sansone[2], C. Lorenzetto[2], E. Fiumana[2], G. Passarella[2], A. Mussari[2], S. Rugolotto[2]

[1]School of Pediatrics, University of Verona, Verona, Italy

[2]Division of Pediatrics, Rovigo Hospital, Rovigo, Italy

[3]School of Pediatrics, University of Padua, Padua, Italy

BACKGROUND

Family-centered cesarean section (FCS), also called gentle cesarean, natural cesarean or mother-centered cesarean, allow parents to be close to the birth through cesarean delivery as it happens during vaginal delivery.

Standard of care may differ between institutions.

Mainly, we can observe three models: "basic," "advanced" and "plus."

In the "basic" model, the father is in the Operating Room; the newborn, after cord clamping and section, is dried up and given to the mother for skin-to-skin contact (STS) and early breastfeeding (EB); neonatal body temperature and SpO_2 are controlled by the neonatologist and a nurse. In the "advanced" model, umbilical cord is intact, the neonate is given directly to the mother for STS and EB while the surgical drape is pulled down (or up). In the "plus" model, the father will also cut the umbilical cord.

OBJECTIVE

Evaluate safety in basic FCS and its impact on breastfeeding.

MATERIALS AND METHODS

An observational retrospective cohort study was designed. Population of study: all basic FCSs performed in the hospitals of Rovigo and Adria from March 2024 (when the program was started) to August 2024. Standard elective singleton term cesarean sections (CSs) performed from January to August 2024 were chosen as comparison group. Safety was defined as incidence of complications requiring admission to the Neonatal Unit. Breastfeeding was defined as both exclusive maternal and maternal plus artificial lactation at hospital discharge; artificial lactation wasn't counted if prescribed for maternal pathology. T-Student test and Fisher exact test were used.

RESULTS

20 FCSs were performed, with mean gestational age 38^{+6}, mean birth weight 3,266 g and mean Apgar index was 9/10. The comparison group counted 43 standard CSs and showed the previous mentioned parameters to be statistically homogeneous within the study group. The study group didn't report more complications than the control group ($p = 0.4151$). One complication occurred in the study group and was transient tachypnoea that didn't required oxygen or ventilation and recovered in some hours; in the comparative group, 6 complications occurred: 5 respiratory distresses and 1 early neonatal sepsis. There was no difference between the two groups in terms of breastfeeding ($p = 0.3624$). In the study group, mean axillary temperature at 5-15 minutes from birth was 36.5°C; at 30 minutes it was 36.4°C. Nineteen out of 20 completed 30 minutes of STS.

CONCLUSIONS

Unlike other studies on FCS, we didn't experience more admissions due to low SpO_2.

None in the FCS group underwent hypothermic event, showing that combination of maternal STS, newborn's hat and metallic drape used in our institution guarantees satisfactory thermoregulation.

We didn't observe an increase in maternal breastfeeding, suggesting that the Operating Room setting still hinders early lactation, as monitoring devices worsen maternal comfort during STS and there is poor intimacy.

REFERENCES

- Armbrust R, Hinkson L, von Weizsäcker K, Henrich W. The Charite cesarean birth: a family orientated approach of cesarean section. J Matern Fetal Neonatal Med. 2016;29(1):163-8.
- Narayen IC, Mulder EEM, Boers KE, van Vonderen JJ, Wolters VERA, Freeman LM, Te Pas AB. Neonatal Safety of Elective Family-Centered Caesarean Sections: A Cohort Study. Front Pediatr. 2018;6:20.
- Smith J, Plaat F, Fisk NM. The natural caesarean: a woman-centred technique. BJOG. 2008;115(8):1037-42; discussion 1042.

ABS 27

FEBRILE SEIZURES AS A MANIFESTATION OF ENCEPHALITIS

M. Cossu[1], M. Olla[1], A. Falchi[1], A. Ghigino[1], A. Bertini[1], S. Cossu[2], D. Pruna[2], B. Cancedda[3], M. Tiddia[3], G. Masnata[4], M. Zanda[3]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Pediatric Neurology and Epileptology Unit, Pediatric Department, ARNAS Brotzu/ASL, Cagliari, Italy*

[3]*Department of Pediatrics, Brotzu Hospital, Cagliari, Italy*

[4]*Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy*

INTRODUCTION

Herpes simplex virus type 1 (HSV-1) encephalitis is a severe central nervous system infection, predominantly affecting the temporal lobes due to the virus's tropism for these regions. It is among the most common forms of viral encephalitis in developed countries and may lead to devastating consequences, including permanent neurological deficits or death if not treated promptly. The most common clinical manifestations include fever, altered mental status, convulsions, and focal neurological deficits. Diagnosis is based on clinical presentation, imaging examinations, electroencephalogram (EEG), and analysis of cerebrospinal fluid by polymerase chain reaction (PCR) for HSV. Early treatment with acyclovir is essential to improve prognosis [1, 2].

CASE REPORT

This case report describes a 14-month-old female patient who presented to the hospital due to seizures lasting over 15 minutes, accompanied by a high fever (39.6°C), vomiting, diarrhea, and loss of appetite for 3 days. The seizures were characterized by altered consciousness, ocular deviation, and limb tremors, with no response to intra-rectal diazepam. Upon admission, the child was in a postictal state with a mildly impaired general condition. Brain computed tomography revealed mild hypodensity in the left parahippocampal region, while EEG demonstrated slow activity in the left temporal regions and localized seizures. After lumbar puncture, empirical therapy with ceftriaxone was initiated, later switched to intravenous acyclovir (100 mg/kg/day) following confirmation of HSV-1 infection by cerebrospinal fluid PCR. Seizures were initially controlled with phenobarbital, and maintenance therapy with carbamazepine was subsequently administered. In the following days, febrile episodes gradually subsided, and the patient's clinical condition improved significantly: she became alert, regained motor functions, and began interacting with her surroundings, with no evident neurological deficits.

CONCLUSION

Complex febrile seizures, as observed in this case, can be an early clinical manifestation of encephalitis, especially in pediatric patients. Given the frequency of febrile seizures in children, distinguishing between benign febrile seizures and those indicative of a serious underlying condition, such as a central nervous system infection, is crucial. According

to LICE guidelines, patients with complex febrile seizures should always undergo investigation to determine the cause of the fever, including routine laboratory tests, an EEG, neuroimaging, and lumbar puncture. Early recognition and appropriate management are key to reducing the risk of long-term neurological complications [1-3].

REFERENCES

[1] Hardarson HS, Messacar K. Acute viral encephalitis in children: Clinical manifestations and diagnosis. Available at: https://www.uptodate.com/contents/acute-viral-encephalitis-in-children-clinical-manifestations-and-diagnosis, last update: December 2022, last access: October 2024.

[2] Kliegman RM, St Geme JW, Blum NJ, Shah SS, Tasker RC, Wilson KM (Eds.). Nelson Textbook of Pediatrics. 21st ed. Philadelphia, PA: Elsevier, 2020.

[3] Capovilla G, Mastrangelo M, Romeo A, Vigevano F. Recommendations for the management of "febrile seizures": Ad Hoc Task Force of LICE Guidelines Commission. Epilepsia. 2009;50(Suppl 1):2-6.

ABS 28

ASSESSING EARLY PREDICTORS OF 12-MONTH NEURODEVELOPMENT IN INFANTS WITH CONGENITAL CYTOMEGALOVIRUS INFECTION: A SINGLE-CENTER EXPERIENCE

C. Deantoni[1,2], A. Spadavecchia[1], C. Rubino[1], A. Leone[1], C. Terzi[1], C. Angieri[1], E. Maggiora[1], C. Peila[1], E. Bertino[1], F. Cresi[1], A. Coscia[1]

[1]Neonatal Unit, Department of Public Health and Pediatrics, University of Turin, Turin, Italy

[2]Neonatal Intensive Care Unit and Pediatric Department, Santa Croce Hospital, Moncalieri, Turin, Italy

INTRODUCTION

Congenital cytomegalovirus (cCMV) infection is a major cause of neurodevelopmental disability and hearing loss in infants, worldwide [1]. Although most infants with cCMV are asymptomatic at birth, a significant subgroup develops long-term neurological and sensory complications [2]. Predicting neurodevelopmental outcomes in infected infants is of clinical relevance and currently under research. Our study aims to identify early predictors of favourable neurodevelopmental outcomes in infants diagnosed with cCMV.

METHODS

Sixty-six infants diagnosed with cCMV at birth and followed up at the Neonatal Intensive Care Unit of St. Anna University Hospital (Turin, Italy) from 2005 to 2023 were included in this retrospective study. We collected data on maternal infection, clinical, laboratory and neuroimaging neonatal presentation, audiological and neurological follow-up. Logistic regression models were used to identify associations between early clinical indicators and outcomes at 12 months of age.

RESULTS

Among the 66 enrolled infants (46.9% females, 53.1% males; 81.8% born at term), 86.3% were asymptomatic at birth. Logistic regression analysis highlighted that a normal absolute neutrophil count (ANC), defined as values > 1,000 μl^{-1}, and normal hearing instrumental assessment at birth were significant predictors of normal neurodevelopmental outcomes at 12 months of age ($p < 0.05$). Although not statistically significant, trends suggested that a normal platelet count and normal findings on brain magnetic resonance imaging were also associated with better neurodevelopmental outcomes. Additionally, normal hearing instrumental assessment at birth was significantly associated with normal auditory outcomes at 1 year of age.

CONCLUSION

This study identifies normal ANC and hearing assessment at birth as significant predictors of favorable neurodevelopmental outcomes in cCMV-infected infants. These findings emphasize the importance of comprehensive clinical and audiological evaluations at birth to identify infants at risk of long-term sequelae, potentially guiding early therapeutic interventions. Further prospective studies with larger cohorts are needed to validate these findings and refine prognostic models for cCMV.

REFERENCES

[1] Leruez-Ville M, Chatzakis C, Lilleri D, Blazquez-Gamero D, Alarcon A, Bourgon N, Foulon I, Fourgeaud J, Gonce A, Jones CE, Klapper P, Krom A, Lazzarotto T, Lyall H, Paixao P, Papaevangelou V, Puchhammer E, Sourvinos G, Vallely P, Ville Y, Vossen A. Consensus recommendation for prenatal, neonatal and postnatal management of congenital cytomegalovirus infection from the European congenital infection initiative (ECCI). Lancet Reg Health Eur. 2024;40:100892.

[2] Salomè S, Corrado FR, Mazzarelli LL, Maruotti GM, Capasso L, Blazquez-Gamero D, Raimondi F. Congenital cytomegalovirus infection: the state of the art and future perspectives. Front Pediatr. 2023;11:1276912.

ABS 29

CASE REPORT: INFANT WITH PNEUMOCOCCAL MENINGITIS AND SEPSIS

A. Falchi[1], A. Ghigino[1], V. Sanneris[1], C. Sbaffi[1], D. Manunza[2], D. Congiu[2], G. Masnata[3], M. Zanda[2]

[1]School of Pediatrics, University of Cagliari, Cagliari, Italy

[2]Department of Pediatrics, Brotzu Hospital, Cagliari, Italy

[3]Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy

INTRODUCTION

We present the clinical case of an infant referred to our Pediatric Unit from Olbia Hospital due to sepsis, fast worsening of general conditions and suspected meningitis. Bacterial meningitis is a neurological emergency characterized by inflammation of the meninges in response to a bacterial infection. *Streptococcus pneumoniae* is the commonest cause of pediatric bacterial meningitis in the USA and Europe [1].

CASE REPORT

A 7-month-old male presented poor general conditions, fever, bulging fontanel, lethargy, irritability, respiratory distress, poor feeding, positive meningeal signs and increased inflammatory indices. Ceftriaxone had already been administered few hours before admission to our Department. On admission, oxygen therapy and intravenous rehydration therapy were initiated. Urgent cardiologic evaluation revealed a wide interatrial defect with signs of right heart overload. To rule out contraindications to spinal tap, a head computed tomography scan was performed, which identified bilateral otomastoiditis. Turbid, pressure-increased cerebrospinal fluid (CSF) samples were collected by lumbar puncture; chemical-physical and microscopic examination revealed the presence of numerous cells and proteins and low glucose levels; polymerase chain reaction (PCR) identified *Streptococcus pneumoniae*; while microbiological culture was negative. No pathogens were detected on pharyngeal swab or blood culture. Prophylaxis with dexamethasone was immediately started to avoid neurosensory damage. A wide spectrum antibiotic therapy with ceftriaxone and vancomycin was set for respectively 14 and 10 days. After 4 days, following cardiological evaluation, furosemide therapy was initiated to target edema of the face and extremities. Neurological evaluation with electroencephalography, brain magnetic resonance imaging, otorhinolaryngology and ophthalmogy consultations were performed to exclude neurological sequelae and damages: all resulted negative. The patient's clinical-laboratoristic conditions progressively improved until full recovery.

CONCLUSION

This clinical case highlights the importance of early diagnosis and prompt treatment of bacterial meningitis; in fact, although advances in medical management and vaccinations have changed its epidemiology, this condition is still associated with high morbidity and mortality. Antibiotics are the mainstay of bacterial meningitis' therapy: empirical antibiotic treatment should be administered immediately, possibly after lumbar puncture, to reduce chances of morbidity and mortality [2]. Moreover, corticosteroids use in acute bacterial meningitis significantly reduces neurological sequelae [3]. CSF culture remains the diagnostic gold standard, albeit it may be altered if performed after the administration of antibiotic therapy, causing its sterilization. Future studies should clarify the role of non-culture diagnostic methods, such as multiplex PCR and metagenomic sequencing [1].

REFERENCES

[1] Alamarat Z, Hasbun R. Management of Acute Bacterial Meningitis in Children. Infect Drug Resist. 2020;13:4077-89.

[2] [No authors listed]. Evidence review for antibiotics for bacterial meningitis before or in the absence of identifying causative infecting organism in older infants and children. Meningitis (bacterial) and meningococcal disease: recognition, diagnosis and management. London: National Institute for Health and Care Excellence (NICE), 2024.

[3] Brouwer MC, McIntyre P, Prasad K, van de Beek D. Corticosteroids for acute bacterial meningitis. Cochrane Database Syst Rev. 2015;2015(9): CD004405.

ABS 30

UNMASKING THE OVERLOOKED: RECOGNIZING BEALS-HECHT SYNDROME

G. Sanna[1], E. Esposito[1], F.F. Comisi[1], C. Soddu[2], S. Savasta[1]

[1]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

[2]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", Cagliari, Italy

INTRODUCTION

Congenital contractural arachnodactyly (CCA), also known as Beals-Hecht syndrome, is a rare connective tissue disorder with autosomal dominant transmission and variable expression. A Marfanoid habitus, crumpled ears, arachnodactyly, multiple joint contractures, progressive kyphoscoliosis, and thoracic abnormalities are among the typical findings. The most severe complication is scoliosis, which frequently calls for ongoing observation and surgery. There are also uncommon "severe" types that include gastrointestinal and/or cardiac abnormalities. The connective tissue protein fibrillin-2 is encoded by the *FBN2* gene, and

mutations in this gene change the protein shape, altering connective tissue and causing CCA. CCA can be distinguished from Marfan syndrome, despite their similarities, by the presence of various joint contractures, crumpled ears, and a lack of considerable aortic dilatation. Due to its overlap with Marfan syndrome, the prevalence of CCA is difficult to determine, and its incidence is unknown. Clinical diagnosis and/or the discovery of an *FBN2* gene mutation may be the basis for the diagnosis. Following diagnosis, scheduling ophthalmologic, cardiologic, orthopedic, and orthodontic evaluations is crucial. Additionally, physical treatment should be initiated to improve joint mobility, and occupational therapy should be started to correct camptodactyly.

CASE REPORT

A 5-year-old kid was assessed for ligamentous hyperlaxity at University of Cagliari's Pediatric Clinic (Cagliari, Italy). His medical history showed low staturoponderal growth and a delay in reaching psychomotor developmental milestones. Upon physical examination, he presented with an elongated face, crumpled ears, micrognathia, muscle hypotrophy, ligamentous hyperlaxity, and arachnodactyly. His head circumference and height fell between the 25th and 50th percentiles, and his weight was at the 3rd percentile. Following a general assessment, a heterozygous c.5054 A>C mutation in the *FBN2* gene was found using the "joint hypermobility" panel in next-generation sequencing (NGS) molecular analysis; his father also carried this mutation. Over time, after diagnosis of CCA, camptodactyly and mild scoliosis have been noted in physiatric and orthopedic assessments. No cardiac, respiratory, or ophthalmologic issues have been detected. Additionally, an orthodontic evaluation has been scheduled to address a dental malocclusion.

CONCLUSIONS

Early orthopedic and physiatric intervention made possible by an accurate differential diagnosis greatly enhances patients' quality of life. Even though clinical grading systems have been established and only 25-75% of patients with clinically confirmed CCA have an identifiable *FBN2* pathogenic mutation, genetic investigation is crucial due to the complex clinical presentation. Therefore, it is important to know and look for the disease's distinctive characteristics and make use of genetic panels that have the *FBN2* gene.

REFERENCES

• Tunçbilek E, Alanay Y. Congenital contractural arachnodactyly (Beals syndrome). Orphanet J Rare Dis. 2006;1:20.

• Meerschaut I, De Coninck S, Steyaert W, Barnicoat A, Bayat A, Benedicenti F, Berland S, Blair EM, Breckpot J, de Burca A, Destrée A, García-Miñaúr S, Green AJ, Hanna BC, Keymolen K, Koopmans M, Lederer D, Lees M, Longman C, Lynch SA, Male AM, McKenzie F, Migeotte I, Mihci E, Nur B, Petit F, Piard J, Plasschaert FS, Rauch A, Ribaï P, Pacheco IS, Stanzial F, Stolte-Dijkstra I, Valenzuela I, Varghese V, Vasudevan PC, Wakeling E, Wallgren-Pettersson C, Coucke P, De Paepe A, De Wolf D, Symoens S, Callewaert B. A clinical scoring system for congenital contractural arachnodactyly. Genet Med. 2020;22(1):124-31.

ABS 31

RENAL INVOLVEMENT IN TWO PEDIATRIC CASES OF SEPSIS

F. Zanco[1], V. Sanneris[1], M. Cossu[1], M. Olla[1], F. Zurrida[2], V. Manca[2], M. Balzarini[3], L. Foschini[3], M. Zanda[3], G. Masnata[2]

[1]School of Pediatrics, University of Cagliari, Cagliari, Italy

[2]Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy

[3]Department of Pediatrics, Brotzu Hospital, Cagliari, Italy

INTRODUCTION

We report two pediatric cases of sepsis with renal involvement, negative cultures and unremarkable abdominal ultrasound (US), both showing positive computer tomography (CT) findings.

CASE REPORTS

Case report 1

A 6-years-old male presented with fever unresponsive to paracetamol, increased inflammatory markers, and abdominal pain, suggesting sepsis. Abdominal X-ray and US were performed: results excluded any surgical urgencies. Later urine exam, blood and urine cultures were taken, resulting negative, and the patient started an antibiotic intravenous therapy with ceftriaxone. Thorax X-ray showed interstitial thickening. Finally, chest-abdomen CT was performed, identifying a small areola of parenchymal thickening in the lower lobe of the left lung and a volumetric increase of the left kidney, with multiple hypodense areas located in the cortical-medulla (mostly in the superior pole) compatible with pyelonephritic outbreaks. Therefore, antibiotic therapy was switched to ceftazidime, teicoplanin and gentamicin, the latter suspended after 8 days, for a total of 14 days of antibiotic therapy. To exclude other disease localization, positron emission tomography (PET)-CT was performed, with metabolic capture exclusively in the upper pole of the left kidney. The patient showed progressive

improvement, both clinical and of the inflammation markers.

Case report 2

A 15-years-old girl presented with fever, lower back pain, high inflammation markers. Urine exam, blood and urine cultures, abdominal US and chest X-ray were negative. On abdominal CT, several hypodense areas were found bilaterally in the renal parenchyma, the largest in the right lower pole, as for pyelonephritis. Therapy with ceftazidime and teicoplanin was promptly started and continued for 14 days, with full recovery.

DISCUSSION

Sepsis is one of the main causes of death in children; it must be defined according to the Phoenix Sepsis Score criteria, that evaluate the respiratory, cardiovascular, coagulation and nervous systems, when the score is at least 2 [1]. Cultures may result negative. However, it is important to start antibiotic therapy and monitoring the infectious foci with radiological exams [2]. The CT should be performed when US is non diagnostic, also allowing to stage the disease. PET-CT may be useful in the assessment of infection [3].

CONCLUSION

The case series highlights the importance of considering renal involvement in sepsis, even when cultures are negative, and the need to evaluate the dissemination and involvement of multiple organs for therapeutic and follow-up purposes through radiological exams. Finally, PET-CT might be relevant in evaluating disease dissemination.

REFERENCES

[1] Schlapbach LJ, Watson RS, Sorce LR, Argent AC, Menon K, Hall MW, Akech S, Albers DJ, Alpern ER, Balamuth F, Bembea M, Biban P, Carrol ED, Chiotos K, Chisti MJ, DeWitt PE, Evans I, Flauzino de Oliveira C, Horvat CM, Inwald D, Ishimine P, Jaramillo-Bustamante JC, Levin M, Lodha R, Martin B, Nadel S, Nakagawa S, Peters MJ, Randolph AG, Ranjit S, Rebull MN, Russell S, Scott HF, de Souza DC, Tissieres P, Weiss SL, Wiens MO, Wynn JL, Kissoon N, Zimmerman JJ, Sanchez-Pinto LN, Bennett TD; Society of Critical Care Medicine Pediatric Sepsis Definition Task Force. International Consensus Criteria for Pediatric Sepsis and Septic Shock. JAMA. 2024;331(8):665-74.

[2] Tamburrini S, Lugarà M, Iannuzzi M, Cesaro E, De Simone F, Del Biondo D, Toto R, Iulia D, Marrone V, Faella P, Liguori C, Marano I. Pyonephrosis Ultrasound and Computed Tomography Features: A Pictorial Review. Diagnostics (Basel). 2021;11(2):331.

[3] Servaes S. Imaging infection and inflammation in children with (18)F-FDG PET and (18)F-FDG PET/CT. J Nucl Med Technol. 2011;39(3):179-82.

ABS 32

BASSEL-HAGEN DISEASE AND ARTHRALGIA: IS THE *EXT* GENE TO BLAME?

F.F. Comisi[1], C. Cerruto[2], E. Esposito[1], G. Sanna[1], S. Carnazzo[3], S. Savasta[1], R. Mura[4]

[1]Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

[2]Pediatrics and Neonatology Department, "R. Guzzardi" Hospital, Vittoria, Italy

[3]Department of Clinical and Experimental Medicine, University of Catania, Catania, Italy

[4]Emato-Oncology Unit, Brotzu Hospital, Cagliari, Italy

INTRODUCTION

Hereditary multiple exostoses (HME) is an autosomal dominant disorder characterized by multiple exostoses, mainly found at the epiphyses of long bones. HME exhibits genetic heterogeneity, with at least three associated loci: *EXT1* on chromosome 8q24.1, *EXT2* on chromosome 11p11-p13, and *EXT3* on chromosome 19p [1]. The increased risk of malignant transformation of osteochondromas into secondary chondrosarcomas is well established. In contrast, the association with acute leukemia is exceedingly rare, with only two reported cases, an 8-year-old girl with acute myeloblastic leukemia [2] and a 14-month-old boy with pre-B-cell precursor acute lymphoblastic leukemia [3]. Herein, we report the third documented case of HME associated with leukemia.

CASE REPORT

A 14-year-old girl was admitted with acute onset of fever and lumbosacral pain. Physical examination was significant only for severe pain, bilateral deformity of the forearms and brachydactyly of the left second toe. Pedigree revealed multiple exostoses in some relatives, father and brother. Whole-body X-ray detected bilateral deformities of the ulnar ends, multiple exostoses of the upper and lower extremities, along with degenerative changes in the lumbosacral spine. A preliminary, clinical diagnosis of Bassel-Hagen disease/HME was established. Laboratory investigations revealed mild anemia, leukopenia, and elevated inflammatory markers. The patient was started on broad-spectrum antibiotics and analgesics. During the following days, serial complete blood counts were monitored, showing progressive deterioration of anemia and leukopenia, after an initial transient improvement. Spinal magnetic resonance imaging revealed osteonecrosis of the lumbar spine. Fluorodeoxyglucose positron emission tomography (FDG-PET) imaging demonstrated diffuse heterogeneous radiotracer uptake in extensive regions of the osteo-medullary tissue. Bone marrow aspiration

and immunophenotyping confirmed a diagnosis of acute lymphoblastic leukemia (CD10+). Treatment was started in accordance with the AIEOP-BFM ALL 2017 protocol [4].

CONCLUSIONS

Mutations in either the *EXT1* or *EXT2* gene are implicated in most HME cases. The development of exostoses is due to the loss of tumor suppressor functions of these genes [2]. The EXT1 protein is essential for the biosynthesis of heparan sulphate glycosaminoglycans, which play a critical role in regulating tumorigenesis, tumor progression, and metastasis [2]. To date, only two cases of HME associated with leukemia have been reported. Nakane et al. [3] proposed a multistep carcinogenesis model involving *EXT1*, although this remains hypothetical at present. The loss of tumor suppressor function in these genes may predispose to chondrosarcoma or other malignancies such as leukemia. Further evidence is required to confirm this association.

REFERENCES

[1] Philippe C, Porter DE, Emerton ME, Wells DE, Simpson AH, Monaco AP. Mutation screening of the EXT1 and EXT2 genes in patients with hereditary multiple exostoses. Am J Hum Genet. 1997;61(3):520-8.

[2] Gözdaşoğlu S, Uysal Z, Kürekçi AE, Akarsu S, Ertem M, Fitöz S, Ikincioğullari A, Cin S. Hereditary multiple exostoses and acute myeloid leukemia: an unusual association. Pediatr Hematol Oncol. 2000;17(8):707-11.

[3] Nakane T, Goi K, Oshiro H, Kobayashi C, Sato H, Kubota T, Sugita K. Pre-B-cell acute lymphoblastic leukemia in a boy with hereditary multiple exostoses caused by EXT1 deletion. Pediatr Hematol Oncol. 2014;31(7):667-9.

[4] Schrappe M. Treatment Protocol for Children and Adolescents With Acute Lymphoblastic Leukemia – AIEOP-BFM ALL 2017. Identifier NCT03643276. Available at: https://clinicaltrials.gov/study/NCT03643276, last access: October 2024.

ABS 33

A CAT-ASTROPHIC CASE OF FUO!

E. Esposito, F. Comisi, G. Sanna, S. Campus, S. Savasta

Pediatric Clinic and Rare Diseases, Microcitemico Hospital "A. Cao", University of Cagliari, Cagliari, Italy

The most common presentation of *Bartonella* infection is cat scratch disease, with subacute regional lymphadenitis, but rarely the Gram-negative bacillus is responsible for a systemic disease. We present an insidious case of fever of unknown origin (FUO) [1]. The patient in remote pathological anamnesis has a suspicion of asymptomatic congenital infection by *Toxoplasma*; a post-infectious arthritis contracted at age 3, initially labeled as juvenile idiopathic arthritis and placed on oral steroid therapy and methotrexate; Shonlein Henoch's purpura complicated by renal involvement, at age 7. He was observed by a pediatric rheumatology specialist at the age of 13 for an unconfirmed suspicion of dactylitis on the 1st toe, bilaterally. Diagnosis of vitiligo at 15 years. At the age of 17, episodes of serotin fever appeared, as high as 40°C, with shaking chills and sweating. Therefore, he was hospitalized, with investigations including: blood chemistry tests showing blood count in the norm, CRP 6.93 mg/dL, VES 47 mm, IgA, IgG, IgM in the norm, antibody panel for major autoimmune diseases negative. Coproculture, urine culture, blood culture and bone marrow aspiration were negative, cardiological examination was normal. A negative chest X-ray was performed; a complete abdominal ultrasound showed evidence of splenomegaly and a small accessory spleen; in addition, the presence of a non-homogeneous area of parenchyma with irregular contours was documented at the lower third, measuring approximately 17 x 14 mm. An abdominal computed tomography (CT) confirmed the presence of a spleen lesion compatible with hemangioma. Blood samples were sent to the zooprophylactic institute. The patient was discharged after 10 days of hospitalization, apyretic, in good general clinical conditions, after antibiotic therapy with ceftriaxone and amikacin, pending investigations to determine the cause of the fever. After 12 days, the patient presented again episodes of fever, with acme at 41°C, and the hospitalization was arranged, during which the determination of lymphocyte subpopulations was carried out. A positron emission tomography (PET)-CT [2] was performed, which documented a finding located at the lower splenic pole not compatible with angiomatous formation, as previously interpreted, but typical of abscess. In light of this new evidence, an infectious as a cause of fever was hypothesized, to the detriment of possible onco-hematological or rheumatological etiology [3]. Further confirmation was the positive antibody for IgG and IgM against *Bartonella*, titration 1:80. Antibiotic therapy with azithromycin and cotrimoxazole was started, with good results. The boy continued regular follow-up in outpatient regime to monitor the evolution of the splenic abscess and to modulate home therapy. One year later, the boy was completely asymptomatic, he performed a control magnetic resonance imaging, which documented a clear reduction of the splenic lesion.

REFERENCES

[1] Zangwill KM. Cat scratch disease and other Bartonella infections. Adv Exp Med Biol. 2013;764:159-66.

[2] Barat M, Hoeffel C, Aissaoui M, Dohan A, Oudjit A, Dautry R, Paisant A, Malgras B, Cottereau AS, Soyer P. Focal splenic lesions: Imaging spectrum of diseases on CT, MRI and PET/CT. Diagn Interv Imaging. 2021;102(9):501-13.

[3] Sarno M, Rosanio FM, De Brasi D, Santoro C, Lo Vecchio A, Esposito F, Siani P, Vajro P, Mandato C. Systemic Cat-Scratch Disease: a "Troublesome" Diagnosis. Pediatr Infect Dis J. 2021;40(3):e117-9.

ABS 34

NON-OBSTETRIC SURGERY DURING PREGNANCY: A CASE REPORT AND A NEONATOLOGY PERSPECTIVE

V. Ferasin[1], S. Bellonzi[2], L. Calandriello[2], L. Dell'Anna[1], C. Lorenzetto[2], F. Marino[2], G. Passarella[2], F. Rigon[2], E. Fiumana[2], A. Mussari[2], L. Saggioro[3], F. Sansone[2], S. Rugolotto[2]

[1]School of Pediatrics, University of Padua, Padua, Italy

[2]Division of Pediatrics, Rovigo Hospital, Rovigo, Italy

[3]School of Pediatrics, University of Padua, Padua, Italy

INTRODUCTION

Non-obstetric surgery not rarely occurs during pregnancy. The most common surgical interventions are: appendectomy (incidence 1/1,500 pregnancies), cholecystectomy (1/1,500-1/3,000), bowel obstruction, traumatology, neurosurgery, tumorectomy (breast tumor), gynecologic laparoscopy. Annually, in the USA, 1-2% of pregnant women undergo surgery unrelated to pregnancy (about 75,000 procedures/year). In cases of abdominal surgery, hypoxic events with short-term or long-term sequelae are documented. We report on a 38-week pregnant woman who underwent orthopedic surgery due to a tibial plateau fracture.

CASE REPORT

A 38-year-old woman reported a lateral tibial plateau fracture during biking. She went to the Emergency Room where, after X-ray confirmation, a joint arthrocentesis after 3 days was performed. The woman was gravida 6 para 3 at 36 weeks of gestation, and the case was discussed among gynecologists, orthopedics, and anesthesiologists. An arthroscopic-assisted fracture reduction with internal fixation was planned after 15 days, with regional anesthesia and minimal sedation. Fetal cardiotocography was added during intervention, with on-site midwife presence. Also, an isolette with full resuscitation equipment and a neonatologist were available close to the Operating Room (OR), in case of emergent delivery. The patient remained in the OR for 4 hours and then returned to the ward. She was discharged the day after.

DISCUSSION

Non-obstetric surgery in pregnancy is a challenge especially for anesthesiologists and obstetricians. Three situations must be considered. The first situation is elective surgery (delayed until postpartum). The second situation is emergency surgery, with anesthetics modified according to maternal physiologic changes and fetal well-being; intraoperative and postoperative fetal and uterine monitoring must be provided, along with perinatologist and obstetrician consultation. The third situation is essential surgery, which might occur in the 1st or in the 2nd/3rd trimester. During the 1st trimester, an option to delay until mid-trimester has to be considered if it is possible without increased maternal risk; if surgery cannot be delayed, it has to be done following all the recommendations of the emergency surgery. If surgery occurs during the 2nd/3rd trimester, it has to be done following all the recommendations of emergency surgery. Current American College of Obstetricians and Gynecologists (ACOG) recommendations state that any pregnant woman should receive medically necessary surgery or should delay surgery if there are risks for her or for her fetus; that corticosteroids should be offered since some surgery can cause preterm delivery, gestational age considered; that every pregnant woman should be screened for thromboembolism. Specific guidelines are given by ACOG for fetus heart rate (HR) monitoring or for HR and contraction monitoring. The obstetrician, after a multidisciplinary analysis, should discuss all the pregnancy-specific issues regarding non-obstetric surgery with the pregnant woman, so that a fully informed plan of action is offered. Neonatologists as well should be aware of the risks of these procedures.

REFERENCES

- [No authors listed]. ACOG Committee Opinion No. 775 Summary: Nonobstetric Surgery During Pregnancy. Obstet Gynecol. 2019;133(4):844-5.
- Bonnet MP. Sedation and anaesthesia for non-obstetric surgery. Anaesth Crit Care Pain Med. 2016;35(Suppl 1):S35-41.
- McCurdy RJ. Intraoperative Fetal Monitoring for Nonobstetric Surgery. Clin Obstet Gynecol. 2020;63(2):370-8.
- Rasmussen AS, Christiansen CF, Uldbjerg N, Nørgaard M. Obstetric and non-obstetric surgery during pregnancy: A 20-year Danish population-based prevalence study. BMJ Open. 2019;9(5):e028136.

ABS 35

BODY DYSMORPHIC DISORDER: SOCIAL MEDIA IMPACT

S. Petza

Neonatal Intensive Care Unit, AOU Cagliari, Cagliari, Italy

In recent decades, body image has become a dominant phenomenon, permeating culture and social interaction, particularly due to the growing prevalence of social media. These media have become a continuous showcase of bodies, often idealized, which contribute to the formation of a frequently distorted body image [1]. Body dysmorphic disorder (BDD) is a condition characterized by an excessive and obsessive preoccupation with perceived defects in physical appearance, often non-existent or minimal, which can result in significant adverse effects on mental health and quality of life. The primary comorbidities associated with this disorder are depression and eating disorders. In recent years, there has been a growing interest in the correlation between BDD and the use of social media, particularly among adolescents and young adults. Current scientific evidence suggests that the excessive use of social media may be a contributory factor in the formation of distorted body perceptions and an increase in the incidence of BDD. Indeed, social media, particularly visual platforms such as Instagram and TikTok, through the provision of a continuous stream of idealized images and the use of filters and retouching, can intensify insecurities related to physical appearance, thereby contributing to the onset or exacerbation of this disorder [2]. In particular, individuals with BDD may engage in constant comparison of their appearance with unrealistic standards presented on social media, which can contribute to feelings of insecurity and body dissatisfaction. Furthermore, the pursuit of approval and validation through likes and comments may intensify anxiety and obsession with perceived physical defects [2]. This is closely related to the possibility of image editing offered by some platforms such as Instagram or TikTok, which allow users to manipulate their physical appearance. This contributes to the generation of a constant social comparison that has a profound impact on the construction of identity, pushing many people to seek, by artificial means, the attainment of an unrealistic ideal of beauty that increases the discrepancy between the real and perceived body [1]. Nevertheless, social media can also have a beneficial impact on body image. This is evidenced by the existence of campaigns that promote diversity, fostering a more inclusive and authentic body image. These initiatives seek to challenge the prevalence of unrealistic beauty ideals, promoting instead self-acceptance and reducing social pressure and the risk of developing body image disorders [3].

REFERENCES

[1] Vandenbosch L, Fardouly J, Tiggemann M. Social media and body image: Recent trends and future directions. Curr Opin Psychol. 2022;45:101289.

[2] Vendemia MA, DeAndrea DC. The effects of engaging in digital photo modifications and receiving favorable comments on women's selfies shared on social media. Body Image. 2021;37:74-83.

[3] Laughter MR, Anderson JB, Maymone MBC, Kroumpouzos G. Psychology of aesthetics: Beauty, social media, and body dysmorphic disorder. Clin Dermatol. 2023;41(1):28-32.

ABS 36

BROWN RECLUSE SPIDER BITE IN CHILDREN: A CASE REPORT

M. Rossi[1], G. Pianese[1], M.R. Angelo[1], G. Cherchi[2]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Department of Pediatric Urgent Care, Brotzu Hospital, Cagliari, Italy*

CASE REPORT

A 9-year-old child presented a reddened lesion following a probable spider bite, associated with onset of fever (maximum temperature 38.6°C). The mother would report seeing the arachnid and identifying it as a fiddler spider (i.e., brown recluse spider).

ABCDE assessment:

A. Airways pervious.
B. No signs of respiratory distress. Respiratory rate within range. O_2Sat: 98%. Normal breath sounds.
C. Regular rate and rhythm. No murmurs. Heart rate: 81 bpm. Skin warm, dry, with good turgor. Symmetrical peripheric pulses.
D. Glasgow Coma Scale: 15. Normal neurological assessment.
E. Temperature: 36.6°C. Examination of ears, nose and throat: normal. Abdomen is soft, symmetric, and non-tender, without distention. Bowel sounds are present and normoactive in all four quadrants. No masses, hepatomegaly, or splenomegaly are noted. Forehead skin lesion of about 1 cm x 1.5 cm, characterized by four erythematous-papular elements with abraded bleeding roof, with surrounding erythema and edema, and absence of necrosis.

The history and clinical features of the skin lesion allowed us to confirm the diagnosis of fiddle spider bite. As there were no ongoing systemic symptoms, the performance of laboratory investigations was not necessary. He was discharged with the following

prescriptions: local disinfection and application of topic antibiotic therapy on the lesion, associated with oral antibiotic therapy (azithromycin 10 mg/kg/day in 1 dose) for 3 days. It was also recommended to avoid sun exposure and refrain from bathing in salt water.

DISCUSSION

Fiddler spider bites result in a clinical presentation characterized by necrotic skin lesions with a distinctive gravitational spread pattern. The bite is initially painless. Pain sets in after 2-8 hours. Small signs at the bite site, erythema and swelling are common. This is followed by the formation of a blister, surrounded by an area of ischemic tissue, and the blister may become hemorrhagic. During the subsequent week, the central zone thickens to form an eschar [1]. In some cases, systemic manifestations, including fever, vomiting, joint pain, petechiae and hemolysis, can occur. Young pediatric patients have the greatest risk of these reaction. While rare, severe cases can result in kidney failure, and death [2]. Treatment of local symptoms includes wound care, analgesics, and antipruritic drugs as necessary. There is no antivenom available. Prophylactic antibiotics are not indicated, but should be used as appropriate if a secondary bacterial infection develops. Patients with an expanding necrotic lesion or symptoms of systemic loxoscelism should be admitted to a medical facility.

CONCLUSION

The Italian Society of Pediatrics suggests measures to prevent these type of bites [3]. If it occurs: first, it is important to disinfect the affected area. Children who do not have systemic symptoms do not need to be hospitalized. However, as hemolysis has been described up to 7 days after a spider bite, in case of systemic symptoms it is important to re-evaluate the findings quickly.

REFERENCES

[1] Goddard J. Physician's guide to arthropods of medical importance. Boca Raton, FL: CRC Press, 2013.

[2] Shahi M, Shahi A, Khademi Z, Zamani AR, Nakhaii AR, Rafinejad J. Loxoscelism: a case report from Bandar Abbas in south of Iran. Hormozgan Med J. 2014;18(5):467-73.

[3] Bozzola E. Ragno violino, 6 cose da sapere. Available at: https://sip.it/2024/08/28/ragno-violino-6-cose-da-sapere/, last access: October 2024.

ABS 37

NIGHT AND DAY URINARY INCONTINENCE

A. Bertini[1], M. Cossu[1], M. Gamberoni[1], V. Manca[2], E. Flore[2], F. Zurrida[2], G. Masnata[2]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy*

INTRODUCTION

Urinary incontinence (UI) is the involuntary loss of urine that can lead to social discomfort, particularly in young individuals. A differential diagnosis is essential to determine the specific type of UI – such as stress, urge, overflow, or functional incontinence – and to identify any related medical conditions. UI, especially in infants, may be caused by ureteral duplication [1].

CASE REPORT

We report the case of a 14-year-old girl who presented at our hospital in March 2022 with suspected dysfunctional voiding. Since turning 6, she had been complaining of "day and night" UI, often associated with physical activity and preceded the previous year by a poorly described vaginal disorder. Urine cultures and urinalysis were always normal, with regular bowel habits, adequate fluid intake. She had menarche at the age of 12, which was associated with dysmenorrhea and menometrorrhagia. During hospitalization for abdominal pain in 2021, ultrasound revealed modifications in the upper pole of the right kidney, including decreased parenchymal thickness, cystic formation, and visualization of the ureter. If ultrasound shows a cyst in the upper pole, the possibility of a duplex kidney should be considered. It is essential to perform a pad test to reevaluate the volume and the frequency of incontinence. Blood tests showed normal renal function, with a mild functional predominance of the left kidney. Physical examination did not reveal any pathological findings. The diagnostic work-up was completed with a DTPA renal scintigraphy, and a pad test. Scintigraphy revealed a hypofunctioning area in the upper pole of the right kidney. The pad test showed constant urine leakage (2-3 mL/15 min). A magnetic resonance imaging scan was performed and showed the presence of a second ureter just below and parallel to the ipsilateral (non-dilated) orthotopic ureter, with a terminal morphology resembling a "rat's tail." The distal part of the abnormal ureter was not visualized. After a collegial discussion, it was decided to proceed with right ureteral reimplantation surgery, based on the clinical suspicion that the ectopic ureteral orifice was located in the vaginal area. During the procedure, the connection between the ectopic ureter and the vagina was confirmed, and the symptoms resolved following the surgery. UI is a common presenting

symptom of a duplex ureter with an ectopic orifice, and it is usually treated with corrective surgery. The majority of people presenting with UI have an ectopic orifice that is extravesical. This abnormality is seen in females [2].

CONCLUSION

Accurate imaging is essential for a correct diagnosis. Management of this condition should be tailored. Upper pole nephroureterectomy is usually performed for non-functioning ureter segments, while ureteropielostomy or ureteral reimplantation can be considered for functioning segments. Although the results are generally satisfactory, long-term follow-up is required [3].

REFERENCES

[1] Aoki Y, Brown HW, Brubaker L, Cornu JN, Daly JO, Cartwright R. Urinary incontinence in women. Nat Rev Dis Primers. 2017;3:17042.

[2] Morhason-Bello IO, Adebayo SA, Abdusalam RA, Bello OO, Odubamowo KH, Lawal OO, Olapade-Olaopa EO, Ojengbede OA. Bilateral double ureters with bladder neck diverticulum in a nigerian woman masquerading as an obstetric fistula. Case Rep Urol. 2014;2014:801063.

[3] Peng HC, Chen HC. Surgical management of complete ureteric duplication abnormalities in children. Zhonghua Yi Xue Za Zhi (Taipei). 2000;63(3):182-8.

ABS 38

SEGMENTAL *PIK3CA*-RELATED OVERGROWTH SYNDROME: DIAGNOSIS AND NEW THERAPEUTIC STRATEGIES

C. Sbaffi[1], M. Gamberoni[1], M. Manca[1], E. Flore[2], V. Setzu[3], S. Murru[4], M. Balzarini[5], G. Masnata[2]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy*

[3]*Unit of Pediatric Orthopedics, Sant'Elena Hospital, Quartu Sant'Elena, Italy*

[4]*Laboratory of Genetics and Genomics, Microcitemico A. Cao Hospital, ASL8, Cagliari, Italy*

[5]*Department of Pediatrics, Brotzu Hospital, Cagliari, Italy*

INTRODUCTION

PIK3CA-related overgrowth syndromes (PROS) are a spectrum of conditions caused by mutations in *PIK3CA* gene, mostly mosaicisms. These rare conditions have a variable phenotype, including fibroadipose, vascular, lymphatic, lipomatous, central nervous system overgrowth; skeletal abnormalities; megalencephaly [1].

CASE REPORT

We report a case of 12-year-old female, born with macrodactyly of the 2nd toe of right foot, surgically corrected at 9 years of age. Unremarkable family history. Referred meningoencephalitis at 7 months of age. Regular growth and neurodevelopment. At our first evaluation in 2021, physical examination highlighted foot scar of debulking surgery and facial dysmorphism: low-set ears, wide mouth, mandibular prognathism, low-set hairline. Cardiac evaluation diagnosed patent ductus arteriosus, non-hemodynamically significant. We searched for DNA variants in genes *PTEN* and *PIK3CA*, after DNA extraction from the tissue biopsied during surgical resection. The c.3140A>G, p.His1047Arg variant was detected by sequence analysis in exon 20 of *PIK3CA* gene, in the heterozygous state and at a low grade (20-30%). This pathogenetic variant was absent in DNA extracted from peripheral blood and buccal swab. In 2022, brain magnetic resonance imaging (MRI) and abdominal ultrasound were performed: both showed absence of significant findings. Since January 2023, patient referred painful resumption of growth of the 2nd toe of right foot; right foot MRI showed synostosis of basal and intermediate phalanx; hypertrophy of distant phalanx, II metatarsal bone and soft tissues, mostly adipose tissue. Longitudinal, dorsoplantar and transversal diameters were 60 x 48 x 44 mm. Future debulking surgery was planned. In 2024 brain MRI was repeated: no significant findings; physical examination revealed initial signs of scoliosis, confirmed at X-ray.

CONCLUSION

In 2022, an international consensus on *PIK3CA*-related disorders was published; however, no consensus on surgical treatment, management of infections, hemorrhages, pain nor indications for pharmacological trials was reached [1]. Different drugs targeting the mTor pathway, already used in cancer treatment, are being studied for applications in PROS. In 2018, a study described the use of BYL719 (alpelisib), an orally bioavailable PI3Kα inhibitor, to successfully treat patient with PROS. 6/17 studied patients had the same mutation as ours: all benefit from the treatment [2]. In 2023, alpelisib was approved by the Food and Drug Administration (FDA) to treat severe *PIK3CA*-related overgrowth [3]. The European Medicines Agency (EMA) has not yet approved this drug; however, multiple trials are ongoing across the globe to further test efficiency and side effects of alpelisib.

Currently, our patient does not meet including criteria for ongoing clinical trials but could be eligible for compassionate use or on label use in case of future EMA approval. World collaboration is needed to work toward innovative therapies for rare diseases.

REFERENCES

[1] Douzgou S, Rawson M, Baselga E, Danielpour M, Faivre L, Kashanian A, Keppler-Noreuil KM, Kuentz P, Mancini GMS, Maniere MC, Martinez-Glez V, Parker VE, Semple RK, Srivastava S, Vabres P, De Wit MY, Graham JM Jr, Clayton-Smith J, Mirzaa GM, Biesecker LG. A standard of care for individuals with PIK3CA-related disorders: An international expert consensus statement. Clin Genet. 2022;101(1): 32-47.

[2] Venot Q, Blanc T, Rabia SH, Berteloot L, Ladraa S, Duong JP, Blanc E, Johnson SC, Hoguin C, Boccara O, Sarnacki S, Boddaert N, Pannier S, Martinez F, Magassa S, Yamaguchi J, Knebelmann B, Merville P, Grenier N, Joly D, Cormier-Daire V, Michot C, Bole-Feysot C, Picard A, Soupre V, Lyonnet S, Sadoine J, Slimani L, Chaussain C, Laroche-Raynaud C, Guibaud L, Broissand C, Amiel J, Legendre C, Terzi F, Canaud G. Targeted therapy in patients with PIK3CA-related overgrowth syndrome. Nature. 2018;558(7711):540-6.

[3] Canaud G, Lopez Gutierrez JC, Irvine AD, Vabres P, Hansford JR, Ankrah N, Branle F, Papadimitriou A, Ridolfi A, O'Connell P, Turner S, Adams DM. Alpelisib for treatment of patients with PIK3CA-related overgrowth spectrum (PROS). Genet Med. 2023;25(12):100969.

ABS 39

MISSING A PINCH OF SALT IN A NEONATE

F. Sansone[1], V. Ferasin[2], S. Bellonzi[1], L. Dell'Anna[2], C. Lorenzetto[1], F. Marino[1], G. Passarella[1], F. Rigon[1], E. Fiumana[1], A. Mussari[1], L. Saggioro[3], L. Calandriello[1], S. Rugolotto[1]

[1]Division of Pediatrics, Rovigo Hospital, Rovigo, Italy

[2]School of Pediatrics, University of Padua, Padua, Italy

[3]School of Pediatrics, University of Verona, Verona, Italy

INTRODUCTION

The syndrome of inappropriate antidiuretic hormone secretion (SIADH) is a condition characterized by hypotonic and euvolemic hyponatremia along with urinary hyperosmolarity, resulting from antidiuretic hormone (ADH) release from hypophysis in the absence of adequate stimuli. ADH binds to V2 receptors in the renal collector duct increasing the synthesis and exhibition of aquaporin-2 water channels, which facilitate water reabsorption and urine concentration [1]. Inappropriate ADH secretion results in a reduction of plasma osmolality, causing cerebral edema. Symptoms range from mild and nonspecific (weakness or nausea) to severe and life-threatening (seizures or coma), depending on the severity of hyponatremia and rapidity of onset [1]. The etiology includes malignancies (para-neoplastic syndrome), pulmonary disorders, and central nervous system pathology [1].

CASE REPORT

A 24-days-old breastfed newborn presented to the ER for hyporeactivity and pallor following a 2-days history of increasing respiratory difficulty, cough and feeding refusal. The examination revealed a plaintive cry, minimal subcostal retractions and reduced air entry bilaterally with fine lung crepitations. During the assessment he presented 6 episodes of apnea and seizures. Blood gas analysis showed respiratory acidosis and severe hyponatremia (Na 115 mmol/L). Lung ultrasound showed a bilateral interstitial pattern and the nasopharyngeal swab PCR was positive for Parainfluenza 3 virus. SIADH was confirmed by detection of low plasma osmolality, increased urine osmolality and high urine sodium concentration. Hyponatremia was treated by intravenous 0.9% sodium chloride administration and fluid restriction, reaching normalization in 24 hours. Apneas necessitated endotracheal intubation and ventilation for 1 day. Non-invasive respiratory support with CPAP was performed for the following 48 hours.

DISCUSSION

Pulmonary diseases can trigger an excessive secretion of ADH in different ways. A ventilation-perfusion mismatch gives rise to compensatory hypoxic pulmonary vasoconstriction, leading to inadequate filling of the left atrium (volume receptors) and consequent ADH secretion. Alternative mechanisms include the release of pro-inflammatory cytokines (such as IL-6), which could directly stimulate the non-osmotic release of ADH [2]. Thus, routine sodium concentration detection is recommended in children with pulmonary infection to detect SIADH. In the absence of severe symptoms requiring urgent intervention, fluid restriction is the first-line treatment. Symptomatic hyponatremia should be treated by bolus of intravenous hypertonic saline (NaCl 3%), obtaining an initial sodium increase of 4-6 mmol/L within 1-2 hours. Thereafter sodium correction should be achieved without exceeding the limit of 8-10 mmol/L within the first 24 hours in order to prevent central pontine myelinolysis, characterized by progressive neurologic deterioration leading ultimately to coma and death [3].

REFERENCES

[1] Mentrasti G, Scortichini L, Torniai M, Giampieri R, Morgese F, Rinaldi S, Berardi R. Syndrome of Inappropriate Antidiuretic Hormone Secretion (SIADH): Optimal Management. Ther Clin Risk Manag. 2020;16: 663-72.

[2] Fajri M, Essafti M, Aloua R, Mouaffak Y. Severe case of COVID -19 pneumonia complicated by SIADH. Ann Med Surg (Lond). 2022;73:103153.

[3] Warren AM, Grossmann M, Christ-Crain M, Russel N. Syndrome of inappropriate antidiuresis: from pathophysiology to management. Endocr Rev. 2023;44(5):819-61.

ABS 40

SYDENHAM'S CHOREA: CORTICOSTEROIDS' ROLE IN QUICK SYMPTOM CONTROL

M. Manca[1], G. Sanna[1], F. Zanco[1], V. Sanneris[1], S. Cossu[2], S. Dettori[2], D. Pruna[2], E. Flore[3], C. Angioni[4], S. Marras[4], M. Zanda[4], G. Masnata[3]

[1]School of Pediatrics, University of Cagliari, Cagliari, Italy

[2]Pediatric Neurology and Epileptology Unit, Pediatric Department, ARNAS Brotzu/ASL, Cagliari, Italy

[3]Department of Complex Diseases and Pediatric Nephrourology, ARNAS Brotzu Hospital, Cagliari, Italy

[4]Department of Pediatrics, Brotzu Hospital, Cagliari, Italy

INTRODUCTION

Rapid, erratic and uncontrollable movements, frequently accompanied by hypotonia and motor problems, are the hallmarks of Sydenham's chorea, a neurological manifestation of acute rheumatic fever. This disorder mostly affects pediatric and adolescent patients and is caused by an autoimmune reaction in the central nervous system to group A streptococcal infections. The illness can persist for months without treatment, increasing the risk of neurological problems and negatively affecting the patient's quality of life. Corticosteroid-treated patients typically have a shorter and milder course of the disease than non-treated patients, and they also have a lower chance of relapses and long-term consequences. On the other hand, non-treated patients may have a persistence of the active symptomatic period, increasing the risk of long-term harm and postponing the return to normality.

CASE REPORT

C., a 9-year-old girl, presented to our attention with sudden onset of continuous involuntary movements affecting the right side of her body, along with difficulties in performing fine motor tasks and speech. Symptoms had started 1 week before hospitalization and became worse over time. She was in generally good condition upon admission and blood tests revealed no signs of infection. After neurological consultation, an urgent brain and spinal MRI with and without contrast revealed mild T2 signal hyperintensity in the left caudate nucleus and ipsilateral lentiform nucleus. Lumbar puncture for cerebrospinal fluid analysis was negative. ASLO levels were 760, and therefore diagnosis of Sydenham's chorea was made. Treatment was immediately started with methylprendisolone sodium succinate 20 mg/kg/day for 5 days and amoxicillin 900 mg twice a day for 10 days. Neurological improvement was rapid: from the first day of treatment, C. regained better control of her movements, and her speech became progressively more fluent. In this case, rheumatic chorea presented as an isolated and singular manifestation; cardiac evaluation was normal, with no mitral valve involvement. C. was discharged after 9 days of hospitalization, with a tapering course of oral corticosteroids and a prophylactic antibiotic regimen.

CONCLUSIONS

The effectiveness and safety of corticosteroid therapy for Sydenham's chorea are confirmed by this case. Corticosteroids are a vital treatment option for controlling the disease because of their capacity for quickly reducing symptoms and enhancing results when used early in the treatment process. Early and aggressive corticosteroid therapy is therefore essential to optimize prognosis in affected patients.

REFERENCES

• Cappellari AM, Rogani G, Filocamo G, Petaccia A. Corticosteroid Treatment in Sydenham Chorea: A 27-Year Tertiary Referral Center Experience. Children (Basel). 2023;10(2):262.

• Eyre M, Thomas T, Ferrarin E, Khamis S, Zuberi SM, Sie A, Newlove-Delgado T, Morton M, Molteni E, Dale RC, Lim M, Nosadini M; Sydenham's Chorea Systematic Literature Review Working Group. Treatments and Outcomes Among Patients with Sydenham Chorea: A Meta-Analysis. JAMA Netw Open. 2024;7(4):e246792.

ABS 41

A NEW CHALLENGE: BITTER TASTE RECEPTORS

B.M. Trapani[1], G. Trapani[2]

[1]Hôpital de Sion CHVR, Sion, Switzerland

[2]Alfred Nobel Study Center, Sanremo, Italy

Bitter taste receptors, type 2 (TAS2Rs), belong to the G protein-coupled receptor (GPCR) family. Till now, 25 functional TAS2Rs have been identified in humans, each detecting different bitter compounds from various sources. These receptors are found not only in taste cells on the tongue but also in the respiratory, gastrointestinal, and reproductive tracts, as well as in certain types of cancer cells [1]. In the respiratory system, TAS2Rs play a role

in regulating intracellular calcium and the innate immune response. When bacterial infections are detected, they activate communication via chemical signaling, coordinating collective behaviors based on population density (quorum sensing). Specifically, TAS2R38 can modulate the production of inflammatory cytokines and influence bronchial contraction in response to bacterial signals [1].

In the gastrointestinal tract, TAS2Rs regulate the secretion of hormones such as glucagon-like peptide-1 (GLP-1) and cholecystokinin (CCK), which are involved in appetite control and digestion. Activation of these receptors modulates intestinal motility and enhances the metabolic response to nutrient intake [2]. Beyond bitter taste perception, TAS2R activation has clinical implications. In respiratory diseases like chronic sinusitis and cystic fibrosis, genetic variations in TAS2R38 influence susceptibility to infections and disease progression through mechanisms such as nitric oxide production, which has bactericidal effects [3]. TAS2Rs found in airway smooth muscle cells can induce bronchodilation, suggesting a potential therapeutic role in conditions like asthma and chronic obstructive pulmonary disease (COPD), where specific drugs could stimulate these receptors to achieve clinical benefits [3]. In the gastrointestinal tract, TAS2Rs also modulate glucose metabolism, with polymorphisms in receptors like TAS2R9 being linked to impaired glucose homeostasis and increased risk of metabolic disorders, including type 2 diabetes [3].

These findings suggest potential for new pharmacological treatments. Some existing drugs, such as antibiotics and analgesics, activate TAS2Rs, offering new therapeutic paths, particularly in respiratory diseases like asthma and COPD. Activation of TAS2Rs in the lungs has shown bronchodilatory effects, which could improve respiratory function in affected patients. Bitter taste receptors, particularly TAS2Rs, have roles beyond the oral cavity, contributing to immune, metabolic, and respiratory regulation. Further research is needed to explore their therapeutic potential.

REFERENCES

[1] Jalševac F, Terra X, Rodríguez-Gallego E, Beltran-Debón R, Blay MT, Pinent M, Ardévol A. The Hidden One: What We Know About Bitter Taste Receptor 39. Front Endocrinol (Lausanne). 2022;13:854718.

[2] Taruno A, Nomura K, Kusakizako T, Ma Z, Nureki O, Foskett JK. Taste transduction and channel synapses in taste buds. Pflugers Arch. 2021;473(1):3-13.

[3] Jeruzal-Świątecka J, Fendler W, Pietruszewska W. Clinical Role of Extraoral Bitter Taste Receptors. Int J Mol Sci. 2020;21(14):5156.

ABS 42

FETAL AND NEONATAL ARRYTHMIAS: PERSONAL EXPERIENCE WITH 167 CASES

M.R. Scanu[1], A. Atzei[2], P. Neroni[2], V. Fanos[2,3]

[1]*School of Pediatrics, University of Cagliari, Cagliari, Italy*

[2]*Neonatal Intensive Care Unit, AOU of Cagliari, Cagliari, Italy*

[3]*Department of Surgical Sciences, University of Cagliari, Cagliari, Italy*

INTRODUCTION

Cardiac arrhythmia is an alteration of the heart rhythm, resulting in an irregular heartbeat. Fetal and neonatal arrhythmias represent a critical challenge for any physician who must manage them, including obstetricians, neonatologists, and pediatric cardiologists. The difficulty in management is not primarily due to the underlying pathophysiological mechanism but rather to the high heart rates and the patient's inability to communicate the symptoms, leading to diagnostic delays and potentially fatal outcomes.

STUDY DESCRIPTION

The study was conducted at the Neonatal Intensive Care Unit and the Pediatric Cardiology Clinic of the University Hospital of Cagliari (Cagliari, Italy). A retrospective analysis was performed on patients diagnosed with fetal and neonatal arrhythmia between January 2014 and April 2024. For each patient, the following data were considered: sex, gestational age, mode of delivery, birth weight, any positive history of fetal arrhythmia, presence of maternal or gestational disease, presence of comorbidities such as congenital heart disease, severity of the clinical condition, any ventricular dysfunction with subsequent signs of heart failure, acute treatment to interrupt the arrhythmia, and the type of antiarrhythmic drugs used in maintenance therapy.

The study included 167 neonates who experienced an arrhythmia episode either during fetal life or within the first month of life. Electrocardiograms performed during the arrhythmia episodes showed characteristics indicative of benign arrhythmia (premature atrial contractions and premature ventricular contractions) in 134 out of 167 patients (80.24%) and non-benign arrhythmias (atrioventricular reentrant tachycardia [AVRT], atrial ectopic tachycardia [AET], permanent junctional reciprocating tachycardia [PJRT], multifocal atrial tachycardia [MAT], atrial flutter, and ventricular tachycardia [VT]) in 33 out of 167 patients (19.76%).

The incidence of diagnosed benign neonatal arrhythmias was 6%, while the incidence of non-benign neonatal arrhythmias was 1.49%. Supraventricular extrasystole, which occurred in 74.25% of patients, was the most frequent arrhythmia observed in the sample. The other identified arrhythmias included ventricular extrasystole in 3.60% of patients, concurrent presence of supraventricular and ventricular extrasystole in 2.40% of patients, AVRT in 14.40%, AET in 1.80%, MAT in 0.60%, PJRT in 1.20%, atrial flutter in 1.20%, and VT in 0.60% of patients.

CONCLUSIONS

Cardiac arrhythmias are an important cause of infant morbidity and a possible cause of infant mortality if undiagnosed and untreated. In most cases, they occur in neonates with structurally normal hearts. Early diagnosis and accurate distinction between benign and more severe arrhythmias are essential to promptly initiate treatment, which, in most situations, proves to be effective.

REFERENCES

• [No authors listed]. [Fetal arrhythmias. The general concepts]. [Article in Portuguese]. Acta Med Port. 1993:6(Suppl 1):I41-2.

• Bücking C, Michaelis A, Markel F, Weidenbach M, Dähnert I, Gebauer RA, Paech C. Evaluation of Clinical Course and Maintenance Drug Treatment of Supraventricular Tachycardia in Children During the First Years of Life. A Cohort Study from Eastern Germany. Pediatr Cardiol. 2022;43(2):332-43.

• Capponi G, Belli G, Giovannini M, Remaschi G, Brambilla A, Vannuccini F, Favilli S, Porcedda G, De Simone L. Supraventricular tachycardias in the first year of life: what is the best pharmacological treatment? 24 years of experience in a single centre. BMC Cardiovasc Disord. 2021;21(1):137.

ABS 43

NOT JUST HYPERGLYCINEMIA…

I. Rulli[1], A.M. Carcione[2], E. Gitto[1]

[1]*AOU G. Martino, Emergency Department Time Dependent, Messina, Italy*

[2]*School of Pediatrics, University of Messina, Messina, Italy*

BACKGROUND

Metabolic diseases' management often result challenging, even more when two mutations combine and clinical picture is worsened by acute events.

CASE REPORT

A 15-month-old boy was diagnosed with non-ketotic hyperglycinemia, requiring sodium benzoate therapy. An older brother died of undiagnosed metabolic disease and an older sister needed intensive care for severe pneumoniae. The child maintained a good metabolic compensation until he developed seizures preceded by fever, so levetiracetam therapy was administered, unsuccessfully. He was taken to the Emergency Department, where a severe non-ketotic hypoglycemia was detected and corrected with intravenous (IV) therapy; after that, he was admitted to the Pediatrics Department. Clinical conditions deteriorated to coma, so admission to the Pediatric Intensive Care Unit was needed. Therapy was administered via nasogastric tube and IV rehydration was started. On the first day of hospitalization, inflammatory indices resulted increased, with positivity for *Haemophilus influenzae* on nasal swab, so IV ceftazidime was started. A metabolic screening was sent to the Metabolic Diseases Center, that revealed normal glycine levels, but also a beta-oxidation deficit of medium-chain fatty acids (MCDA); so, prolonged fasting was avoided and carnitine was prescribed. During hospitalization, seizures occurred; EEG showed severe abnormalities of cerebral activity on the left hemispheric derivations, so dextromethorphan was started. A week later, a metabolic re-evaluation was conducted, revealing normal glycine levels and improvement of the MCDA deficit. In addition, the child was subjected to a brain MRI, revealing findings typical in non-ketotic hyperglycinemia.

CONCLUSIONS

This case suggests that coexistence of two metabolic diseases in the same patient, although rare, is possible, especially in presence of suspicious deaths in family. A case in the literature [1] showed similar metabolic pattern. Suggestive symptoms and associated factors, like fasting or intercurrent diseases, should lead to a re-evaluation of the case. More than ever, children like these require careful follow-up by a reference center for metabolic diseases.

REFERENCE

[1] Stanley CA, Hale DE, Coates PM, Hall CL, Corkey BE, Yang W, Kelley RI, Gonzales EL, Williamson JR, Baker L. Medium-chain acyl-CoA dehydrogenase deficiency in children with non-ketotic hypoglycemia and low carnitine levels. Pediatr Res. 1983;17(11):877-84.

ABS 44

EXTERNAL CEPHALOHEMATOMA WITH ATYPICAL CLINICAL COURSE

A. D'Apuzzo[1], M. Vendemmia[2]

[1]*Emeritus Chief of Pediatrics and Neonatology, Hospital of Castellammare di Stabia, Naples, Italy*

[2]Neonatal Intensive Care Unit, University Federico II Naples, Naples, Italy

INTRODUCTION

External cephalohematoma is a subperiosteal blood collection due to rupture of a vessel during prolonged labor, or after application of forceps or suction cup, or even in the absence of traumatic events. It generally involves only one of the cranial bones, mostly the parietal, and does not extend beyond the suture lines (where the periosteum inserts). This characteristic demarcation, in addition to its elastic consistency on palpation, allows it to be differentiated from the delivery tumor, which is an edematous tumefaction of the soft parts corresponding to the part of the skull presented in the second stage of delivery. The external cephalohematoma sometimes persists for months, without, however, causing any damage.

CASE REPORT

We describe an atypical case of the course of an external cephalohematoma in a 4-month-old infant, who came to our observation for a convulsive episode.

Baby M. was born at term from a normal conducted pregnancy and eutocic delivery. The mother has temporal epilepsy and is being treated with carbamazepine. The pathological history is completely negative until the age of 4 months, at which time he is admitted to our division for a seizure of limb hypertonia; ocular retroversion with negative neurological examination in the postcritical period. An initial electroencephalographic tracing shows nothing pathological. During the hospital stay, new seizures recur throughout the day, with a rhythm of 5-6 episodes/24 h, configuring a state of epileptic illness, which is controlled with the intravenous infusion of levetiracetam. A second and third electroencephalographic tracing is performed, which show asymmetry of rapid sleep spindles, voltage asymmetry with high voltage slow elements in right central front. Therapy with sodium valproate at a dose of 25 mg/kg/day is instituted. Seizures recur despite continued therapy and normal sodium valproate blood range. Brain CT scan without contrast medium shows a ventricular system in place, with no definite changes. Extensive basilar and Sylvian cistern as well as pericerebral spaces in the frontal region and inter-hemispheric scissure. Absence of parenchymal densitometric changes. In right high parietal region, prominence and thickening of the right parietal draft is appreciated, below which there is a linear calcified image with which the brain surface is in contact. This appearance is compatible with an organized and partly calcific cephalic hematoma. Transferred to the neurosurgical setting, the baby was operated on for removal of the hematoma that had disrupted the bony structure and caused cerebral compression. Follow-up was normal; 6 months later he was revisited: negative neurological examination, no more seizures, normal EEG.

REFERENCES

- Barkovich AJ, Truwit CL. Brain damage from perinatal asphyxia: correlation of MR findings with gestational age. AJNR Am J Neuroradiol. 1990;11(6):1087-96.
- Levene MI, Bennett MJ, Purit J. Fetal and neonatal neurology and neurosurgery. London: Churchill Livingstone, 1988.
- Vendemmia S, Vendemmia M. Atlante di Pediatria e Neonatologia per immagini. Naples: Cuzzolin, 2022.

ABS 45

ATP: A POTENTIAL WEAPON AGAINST HCAI IN THE NICU

G. Simoni[1], M. Melis[2], V. Fanos[2,3]

[1]University of Cagliari, Cagliari, Italy
[2]Neonatal Intensive Care Unit, AOU of Cagliari, Cagliari, Italy
[3]Department of Surgical Sciences, University of Cagliari, Cagliari, Italy

Healthcare-associated infections (HCAI) represent one of the greatest challenges for global healthcare and pose a particular threat to vulnerable patients, such as neonates. Sepsis is the third leading cause of death in the neonatal population, with the most vulnerable patients being preterm and low birth weight infants (VLBW, ELBW) [1]. Incubators provide the environment where neonates spend a postnatal period of varying length. They must replicate conditions such as temperature and humidity necessary for the neonate's survival, which would naturally be provided by the mother's womb. However, these characteristics, along with possible contamination by healthcare workers and parents, as well as the presence of the newborn itself, contribute to the potential risk of infection that the patient may encounter. In preventing HCAI, rapid adenosine triphosphate (ATP) detection could prove useful for improving current surface sanitation procedures, particularly for incubators.

ATP is a molecule used as an intracellular energy source. It is also produced by bacterial cultures, where it is released into the extracellular space in amounts proportional to the growth rate. It has been highlighted in cultures of various pathogens, such

as *E. coli*, *Pseudomonas spp.*, *Salmonella spp.*, *Staphylococcus spp.*, etc. [2]. For these reasons, ATP is used as a reliable indicator of biological contamination.

Meizheng Pure Trust™ is an intelligent fluorescence detector that uses the luciferase-luciferin reaction system to quantify ATP present on a swab through a highly sensitive photomultiplier. Based on the amount of ATP detected, we can classify surfaces as "dirty," "hygienically clean," "clinically clean," and "sterile."

We have used Meizheng Pure Trust™ in our facility to assess the hygiene of hot spots surfaces: porthole of a sanitized incubator, porthole of an in-use incubator, handle of a sanitized incubator, handle of an in-use incubator, mattress support of a sanitized incubator, mattress support of an in-use incubator, water tray of a sanitized incubator, water tray of an in-use incubator, milk pasteurization filter, work surface for milk preparation. At each site, we performed a rapid ATP test and a culture swab simultaneously. The comparative results were consistent, demonstrating the potential of this technique.

Rapid ATP detection on surfaces is a technique widely used in the pharmaceutical, cosmetic, and food industries that could prove to be of great importance in healthcare surveillance as well. Using rapid swabs on incubators at the end of sanitation procedures, before their use in the ward, can help reduce the risk of infection for the neonates.

REFERENCES

- Attia Hussein Mahmoud H, Parekh R, Dhandibhotla S, Sai T, Pradhan A, Alugula S, Cevallos-Cueva M, Hayes BK, Athanti S, Abdin Z, Basant K. Insight Into Neonatal Sepsis: An Overview. Cureus. 2023;15(9):e45530.
- Mempin R, Tran H, Chen C, Gong H, Kim Ho K, Lu S. Release of extracellular ATP by bacteria during growth. BMC Microbiol. 2013;13:301.

ABS 46

MAGNET INGESTION IN CHILDREN: A CASE REPORT

M. Marsan[1], M. Brutti[1], R. Cacace[1], G. Amato[2], F. Sau[3], G. Cherchi[3]

[1]School of Pediatrics, University of Cagliari, Cagliari, Italy

[2]Department of Pediatric Surgery, Brotzu Hospital, Cagliari, Italy

[3]Department of Pediatric Urgent Care, Brotzu Hospital, Cagliari, Italy

CASE REPORT

C., a 6-year-old boy, presented at 01:30 a.m. with abdominal pain and one episode of vomit after having accidentally ingested a single piece of magnet (dimension approximately 2 x 2 cm^2) at 9:30 p.m. The patient's brother had previously tried to identify the position of the object by moving another magnet in front of his abdomen and had drawn an "X" on the skin.

ABCDE assessment:

A. Patent airways.
B. No signs of respiratory distress. Respiratory rate within range. O_2 Sat: 98%. Normal breath sounds.
C. Regular rate and rhythm. No murmurs. Heart rate 98 bpm. Well perfused and hydrated skin. Symmetrical peripheric pulses.
D. Glasgow Coma Scale: 15. Normal neurological assessment.
E. Afebrile. Skin warm, dry, with good turgor. Examination of ears, nose and throat normal. Abdomen is soft, symmetric, and non-tender without distention. Bowel sounds are present and normoactive in all four quadrants. No masses, hepatomegaly, or splenomegaly are noted.

Biplane radiography of the abdomen (with and without contrast) showed foreign body in gastric fundus. Pediatric Surgery consult opted for urgent removal, which was done endoscopically, and it revealed that the foreign body had already caused a minor ulcer in the gastric wall.

DISCUSSION AND CONCLUSION

Foreign body ingestion in children is common cause of acute referral to the Emergency Room. Type, size, time since ingestion, last meal, symptoms and any other pre-existing condition must be noted, as they are fundamental in determining the urgency level of removal and its method [1]. Biplane radiographs (with or without contrast, based on the suspected foreign body) are recommended, especially suspecting ingestion of magnets, as multiple items may appear as one [2]. Timing of removal may be divided into emergent (< 2 hours), urgent (< 24 hours), elective (> 24 hours). If the ingestion was either witnessed by a caregiver or it is clearly shown in radiographs and the child is symptomatic, the magnet has to be removed. In case of an asymptomatic child, the number of magnets must be determined: multiple magnets ingestion requires urgent removal, as the powerful attraction may cause ischemia, perforation, fistulas and volvulus in the bowel wall; this also applies in case of ingestion of a single magnet and a metallic object [2]. On the other hand, removal of a single magnet in the absence of symptoms is usually not necessary, and the child can undergo prolonged observation with daily radiographs; if the foreign

body does not progress in 24 hours, an endoscopic removal should be discussed [2, 3]. Moreover, it is important to educate the parents on avoiding having other metallic objects in the close vicinity, in order to avoid accidental ingestion and to prevent magnet adherence to the abdominal wall [3].

REFERENCES

[1] Oliva S, Romano C, De Angelis P, Isoldi S, Mantegazza C, Felici E, Dabizzi E, Fava G, Renzo S, Strisciuglio C, Quitadamo P, Saccomani MD, Bramuzzo M, Orizio P, Nardo GD, Bortoluzzi F, Pellegrino M, Illiceto MT, Torroni F, Cisarò F, Zullo A, Macchini F, Gaiani F, Raffaele A, Bizzarri B, Arrigo S, De' Angelis GL, Martinelli M, Norsa L; Italian Society of Pediatric Gastroenterology Hepatology and Nutrition (SIGENP); Italian Association of Hospital Gastroenterologists and Endoscopists (AIGO). Foreign body and caustic ingestions in children: A clinical practice guideline. Dig Liver Dis. 2020;52(11):1266-81.

[2] Altokhais T. Magnet Ingestion in Children Management Guidelines and Prevention. Front Pediatr. 2021;9:727988.

[3] Kramer RE, Lerner DG, Lin T, Manfredi M, Shah M, Stephen TC, Gibbons TE, Pall H, Sahn B, McOmber M, Zacur G, Friedlander J, Quiros AJ, Fishman DS, Mamula P; North American Society for Pediatric Gastroenterology, Hepatology, and Nutrition Endoscopy Committee. Management of ingested foreign bodies in children: a clinical report of the NASPGHAN Endoscopy Committee. J Pediatr Gastroenterol Nutr. 2015;60(4):562-74.

ABS 47

INTERNATIONAL STANDARDS FOR TWIN NEWBORN WEIGHT, LENGTH, AND HEAD CIRCUMFERENCE BY GESTATIONAL AGE AND SEX: THE INTERGROWTH-21ST CROSS-SECTIONAL STUDY

F. Giuliani[1], S. Deantoni[2], S. Gandino[2], A. Coscia[2], I. Stura[3], S.H. Kennedy[4,5], A.T. Papageorghiou[5,6], E. Bertino[2], G. Migliaretti[6], J. Villar[5,6] for the International Fetal and Newborn Growth Consortium (INTERGROWTH-21st)

[1]Neonatal Special Care Unit, Regina Margherita Children Hospital, Città della Salute e della Scienza, Turin, Italy

[2]Neonatal Intensive Care Unit, Department of Sciences of Public Health and Pediatrics, University of Turin, Italy

[3]Rita Levi Montalcini Neuroscience Department, University of Turin, Italy

[4]Nuffield Department of Women's & Reproductive Health, Green Templeton College, University of Oxford, Oxford, UK

[5]Oxford Maternal and Perinatal Health Institute, Green Templeton College, University of Oxford, Oxford, UK

[6]Department of Sciences of Public Health and Pediatrics, University of Turin, Italy

INTRODUCTION

The need for anthropometric neonatal charts specific for twins is under debate. In order to advance this, the availability of specific twin charts would allow evaluation of differential effects of pregnancy-related morbidity beyond the physiological adaptation induced by twinning itself, hence avoiding over diagnosis of small for gestational age (SGA).

MATERIALS AND METHODS

INTERGROWTH-21st was a population-based, international project aimed to produce prescriptive growth standards for fetuses and newborns, using the same prescriptive conceptual framework used for the construction of the WHO international Child Growth Standards. The project was implemented in eight geographically defined populations chosen based on meeting health and nutritional needs for mothers, adequate antenatal care and lack of environmental constraints on growth. The population-based Newborn Cross-Sectional Study (NCSS) collected information on weight, length and head circumference from all births from these sites. A low-risk subsample was selected including all pregnancies and live births meeting the low-risk population characteristics above as well as strict individual eligibility criteria, meaning pregnancies were at low risk of impaired growth. From this population, the international standards for singleton births for weight, length, and head circumference, by gestational age and sex were developed, complementing the WHO Child Growth Standards. In the present study, international cross-sectional standards specific for twins were constructed using the same population, methodology, equipment and conceptual framework. This was achieved by selecting all twin births in the same population used for the creation of the INTERGROWTH-21st standards for singletons and applying the same strict eligibility criteria. Additional twin-specific exclusion criteria were evidence of twin-to-twin transfusion or twin birth weight discordance above 25%. Three smoothing techniques have been evaluated: fractional polynomials, cubic spline and penalized spline. After evaluation of the goodness-of-fit, fractional polynomials assuming a skewed t distribution were used to estimate the fitted centiles.

RESULTS

From the 59,137 deliveries enrolled in NCSS, there were 1,002 twin pregnancies. After exclusion according to selection criteria, a final sample of 894 twin newborns was obtained. Twin charts were constructed based on these data. We did sensitivity analyses for maternal age and assisted reproduction to assess the effect on the centiles of the remaining pooled samples of removing a single variable

at a time, no substantive effect was recorded. Anthropometric measures of each individual twin newborn were plotted against INTERGROWTH-21st standards for singletons, showing that twins plot on lower centiles. Comparisons of twins' and singletons' centiles showed an increasing divergence along gestational ages towards term age for all anthropometric variables but less marked for the head circumference.

CONCLUSIONS

International twin specific anthropometric standards for weight, length, and head circumference for sex and gestational age at birth have been constructed. Their availability is of relevance for clinical practice to allow evaluation of twin newborns worldwide and for research applications such as comparison of single and twin growth.

ABS 48

ACUTE LYMPHOBLASTIC LEUKEMIA AND GANGLIONEUROBLASTOMA: CASE REPORT

F. Gigliotti[1], C. Fortugno[1], E. Galea[1], R. Cantaffa[1], D. Loiacono[1], V. Talarico[2], M. Ceravolo[2], A. Pugliese[1], G. Raiola[2], M.C. Galati[1]

[1]*Department of Pediatric Onco-Hematology, Renato Dulbecco University Hospital, Catanzaro, Italy*

[2]*Department of Pediatric, Renato Dulbecco University Hospital, Catanzaro, Italy*

BACKGROUND

Ganglioneuroblastoma is an uncommon malignant tumor of the sympathetic nervous system, which is considered a disease of children with the majority of cases in patients less than 4 years old; it rarely occurs in adults. In addition, the most common sites of origin are the adrenal medulla (about 35%), extra adrenal retroperitoneum (30-35%), and posterior mediastinum (20%) [1-3]. We encountered a very unusual case of an 8-year-old female patient affected by synchronous acute lymphoblastic leukemia (ALL) and ganglioneuroblastoma. The management of the clinical case from diagnosis to treatment will be described below.

CASE REPORT

An 8-year-old female patient shortly after the diagnosis of ALL presents with abdominal pain and mass. In November 2016, during induction chemotherapy treatment for ALL, the patient presented abdominal pain, constipation with severe intestinal hypotony, and the abdominal ultrasound examination showed a mass between the left iliac vessels and the lower left renal pole. The chest-abdomen CT scan with contrast medium defined the presence of a neoformation in the left lower abdominal area. FDG PET and 123I-MIBG scintigraphy showed non-homogeneous uptake of the radionuclide at the lower left abdomen neoformation. Percutaneous biopsy was performed and the patient continued chemotherapy treatment until the consolidation phase when, between the second and third administration of high-dose methotrexate, she underwent surgery. The histological examination described a composite tumor characterized by the presence of a "Schwannian-stroma-dominant" peripheral neuroblastic tumor, with overall aspects of a maturing ganglioneuroma, and a c-Myc and ALK negative undifferentiated neuroblastoma nodule. The patient completed treatment for ALL in October 2018 and went on to follow-up, but in October 2020 there was a locoregional recurrence of ganglioneuroblastoma. Therefore, the patient enrolled in the LINES protocol, unresectable neuroblastoma risk group, and in December 2020 began chemotherapy with carboplatin and etoposide for two cycles followed by radiation treatment and a further two cycles of carboplatin and etoposide. Since April 2021, the patient had negative oncological follow-up until now.

CONCLUSIONS

At the onset of ALL, the patient did not present abdominal masses, which were instead observed later. We believe that ganglioneuroblastoma cells were present at onset, but not detectable, as the immune surveillance exerted its tumoricidal action by creating a balance between the immune system and neoplastic ganglioneuroblastoma cells. However, the state of immunosuppression induced by ALL induction chemotherapy undermined this balance, conferring a proliferative advantage to ganglioneuroblastoma cells. Our case suggests that the appearance of new masses should not be considered as a leukemic progression with organ infiltration; rather, it is necessary to biopsy any new onset mass.

REFERENCES

[1] Lam AK. Update on Adrenal Tumours in 2017 World Health Organization (WHO) of Endocrine Tumours. Endocr Pathol. 2017;28(3): 213-27.

[2] Lonergan GJ, Schwab CM, Suarez ES, Carlson CL. Neuroblastoma, ganglioneuroblastoma, and ganglioneuroma: radiologic-pathologic correlation. Radiographics. 2002;22(4):911-34.

[3] Brossard J, Bernstein ML, Lemieux B. Neuroblastoma: an enigmatic disease. Br Med Bull. 1996;52(4):787-801.

ABS 49

ANTITUMOR ACTIVITY OF LAROTRECTINIB IN GLIOBLASTOMA MULTIFORME WITH *NTRK* GENE AMPLIFICATION CASE REPORT

F. Gigliotti[1], C. Fortugno[1], E. Galea[1], R. Cantaffa[1], R.L. Fabiano[1], V. Talarico[2], M. Ceravolo[2], G. Raiola[2], M.C. Galati[1]

[1]Department of Pediatric Onco-Hematology, Renato Dulbecco University Hospital, Catanzaro, Italy

[2]Department of Pediatric, Renato Dulbecco University Hospital, Catanzaro, Italy

BACKGROUND

Advances in genomics have increased the molecular knowledge underlying cancer development and growth. In recent years, great importance has been given to *NTRK* gene fusions which codes for the tropomyosin kinase receptor (TRK) [1]. Entrectinib and larotrectinib are currently approved by the FDA and EMA in *NTRK* translocated tumors [2]. Below we will describe a case report of a patient with *NTRK* amplified glioblastoma multiforme who received targeted therapy with larotrectinib with complete disease response.

CASE REPORT

In October 2021, a 16-year-old boy accessed the Emergency Department due to the onset of nausea and vomiting subsequently accompanied by loss of consciousness; he performed brain CT and MRI, which showed a right temporal-parietal-occipital brain mass, which was biopsed with a definitive diagnosis of glioblastoma multiforme. The patient underwent surgery and the mass was surgically removed two times. At the third recurrence, the boy started first-line treatment according to concomitant radio-chemotherapy from the end of January 2022; however, the disease progressed. For disease progression and *NTRK2* and *NTRK3* amplifications, in August 2022, the patient began second-line treatment with larotrectinib. Currently, the patient is continuing therapy with larotrectinib, maintaining a complete disease response, free from drug-related toxicities.

CONCLUSIONS

The immunohistochemical profile of the solid tumor is an expression of the genetic alterations underlying the tumor birth and growth. In this case report, we show how a simple technique such as immunohistochemistry can be sufficient to start an effective target therapy when it showed amplification of *NTRK*2 and *NTRK*3.

The use of a complete immunohistochemistry, specifically for *NTRK*, enabled the selection of a targeted therapy that led to a morphological and functional complete disease response. This case suggests that the *NTRK3* amplification immunochemistry is enough to start larotrectinib treatment. Furthermore, our clinical case demonstrates the effectiveness of larotrectinib in *NTRK* amplified tumors; therefore, we believe that new clinical trials are necessary to demonstrate the effectiveness of the treatment in *NTRK* amplified patients.

REFERENCES

[1] Ardini E, Bosotti R, Borgia AL, De Ponti C, Somaschini A, Cammarota R, Amboldi N, Raddrizzani L, Milani A, Magnaghi P, Ballinari D, Casero D, Gasparri F, Banfi P, Avanzi N, Saccardo MB, Alzani R, Bandiera T, Felder E, Donati D, Pesenti E, Sartore-Bianchi A, Gambacorta M, Pierotti MA, Siena S, Veronese S, Galvani A, Isacchi A. The TPM3-NTRK1 rearrangement is a recurring event in colorectal carcinoma and is associated with tumor sensitivity to TRKA kinase inhibition. Mol Oncol. 2014;8(8): 1495-507.

[2] Vaishnavi A, Le AT, Doebele RC. TRKing down an old oncogene in a new era of targeted therapy. Cancer Discov. 2015;5(1):25-34.

ABS 50

ABERNETHY TYPE II MALFORMATION IN CHILDREN: AN UNUSUAL PRESENTATION. A CASE OF ABERNETHY MALFORMATION

V. Talarico[1], A. Nicoletti[1], G. Guarascio[2], A. Viscomi[2], G. Stranieri[3], A. Gigliotti[4], M.C. Galati[4], D. Alberti[5], G. Raiola[1]

[1]Department of Pediatrics, Renato Dulbecco Hospital, Catanzaro, Italy

[2]Department of Radiology, Renato Dulbecco Hospital, Catanzaro, Italy

[3]Department of Pediatric Surgery, Renato Dulbecco Hospital, Catanzaro, Italy

[4]Department of Pediatric Onco-Hematology, Renato Dulbecco Hospital, Catanzaro, Italy

[5]Pediatric Surgery, University of Brescia, Brescia, Italy

BACKGROUND

Abernethy syndrome is a congenital vascular anomaly in which the portal blood completely or partially bypasses the liver through a congenital porto-systemic shunt. This condition was first reported by Abernethy in 1793 and is named after him. The diagnosis of this condition has increased with routine newborn screening for galactosemia and advances in imaging techniques.

CASE REPORT

A 7-year-old boy presented to our Pediatric Clinic for persistent soft tissue edema in the right foot. Perinatal history was characterized by: premature birth at 33rd week of gestation, hospitalization in Neonatal Intensive Care Unit, respiratory distress syndrome, bilateral hydrothorax, hypoglycemia, hypocalcemia, anemia, sepsis and congenital heart disease (cuspid aorta, aortic and mitral stenosis). Physical examination was normal, except for a grade 1-2/6 systolic ejection murmur at the cardiac examination and edema of the right ankle, not painful at typing pressure, with four-limb blood pressure measurement normal. Radiological imaging (ultrasound and X-ray) of right foot were not significant, so for the persistence of the edema and its increase only in orthostatic position we performed abdominal CT and subsequent RMN that showed anomalous vascular connection between the wider hepatic vein and portal vein, typical of Abernethy type II malformations. He underwent intravascular shunt occlusion with intravenous plug positioning. After 1 year, he showed reappearance of edema of right foot. He performed lymphoscintigraphy, which highlighted severe delay of lymphatic drainage on the lower right limb, with hypertrophy of the lymph node stations. He started conservative treatment without beneficial effect, so he underwent lymphatic-venous anastomosis surgery at the level of the right forefoot, with reduction of foot edema. The postoperative course was regular, with reduction of foot edema. If the edema should appear, the need for local lymph node transplantation will be evaluated.

CONCLUSION

Although the number of recognized and reported cases is gradually increasing, Abernethy syndrome is still a rare disease entity, with an estimated prevalence between 1 per 30,000 and 1 per 50,000 cases. The extrahepatic congenital portosystemic shunt (CEPSh), belonging to the group of congenital portosystemic shunts (CPSS), can be extrahepatic and intrahepatic, with the latter being more commonly reported in the literature. Morgan and Superina classified CEPSh into two types based on the vascular anatomy of the exhepatic shunts: termino-lateral or latero-lateral. They can have various clinical features, affecting almost all organ systems and presenting at any age, that make a very structured diagnostic work-up and surgical approach necessary.

REFERENCES

• Franchi-Abella S, Branchereau S, Lambert V, Fabre M, Steimberg C, Losay J, Riou JY, Pariente D, Gauthier F, Jacquemin E, Bernard O. Complications of congenital portosystemic shunts in children: therapeutic options and outcomes. J Pediatr Gastroenterol Nutr. 2010;51(3):322-30.

• Kim MJ, Ko JS, Seo JK, Yang HR, Chang JY, Kim GB, Cheon JE, Kim WS. Clinical features of congenital portosystemic shunt in children. Eur J Pediatr. 2012;171(2):395-400.

• Schaeffer DF, Laiq S, Jang HJ, John R, Adeyi OA. Abernethy malformation type II with nephrotic syndrome and other multisystemic presentation: an illustrative case for understanding pathogenesis of extrahepatic complication of congenital portosystemic shunt. Hum Pathol. 2013;44(3):432-7.

ABS 51

A STUDY OF CORD BLOOD ALKALINE PHOSPHATASE AS A PREDICTOR OF NEONATAL HYPERBILIRUBINEMIA IN A TERTIARY HOSPITAL

V.B. Sonawane, R. Agarwal, M. Maksud, A. Saxena, V. Kotrashetti

Department Of Pediatrics, D. Y. Patil University, School of Medicine, Nerul, Navi Mumbai, Maharashtra, India

BACKGROUND

Neonatal hyperbilirubinemia is one of the most prominent and problematic illnesses that affect newborns. Neonatal jaundice is a common condition that affects up to 80% of pre-term infants and 60% of term infants. This study aimed to evaluate the role of cord blood alkaline phosphatase (ALP) levels as a predictor of hyperbilirubinemia in full-term neonates.

METHODS

This hospital-based prospective observation study was conducted on newborns for 1.5 years in Dr. D.Y. Patil Medical College, Hospital and Research Centre Navi Mumbai (Navi Mumbai, Maharashtra, India). Cord blood ALP levels upon delivery and serum bilirubin levels at 72 hours of life were measured.

RESULTS

Out of 200 cases, we found that 122 cases were having cord ALP level > 314 IU/L and remaining 78 cases were having cord ALP level < 314 IU/L. The mean cord ALP level among the neonates was 315.7 + 139.6 IU/L. During follow up, out of those 122 cases, 15 developed hyperbilirubinemia, and out of those 78 cases, 2 cases developed hyperbilirubinemia, which was confirmed by serum bilirubin level > 13 mg/dL at 72 hours after birth. Here it shows the predictive role of cord ALP levels in diagnosing neonatal hyperbilirubinemia: there was a sensitivity of 88.2%, specificity of 41.5%,

PPV of 12.3%, NPV of 97.4%, and accuracy of 45.5%. This infers that cord ALP level estimation during early stages helps in diagnosing the neonatal hyperbilirubinemia. The ROC curve also depicts the same, with area under curve of 0.64.

CONCLUSION

There was a significant correlation between cord blood ALP levels and serum bilirubin levels at 72 hours of life, with high sensitivity and low specificity. Therefore, cord blood ALP may be considered as a predictor for hyperbilirubinemia in neonates.

ABS 52

PERCEIVED FAMILY FUNCTION, PERCEIVED SOCIAL SUPPORT AND DEPRESSION OF NEW PARENTS UNDER COVID-19 PANDEMIC: A DYADIC ANALYSIS

T. Kokkinaki[1], K. Koutra[2], O. Michopoulou[3], L. Chaziraki[4], P. Kokarida[5], E. Hatzidaki[3]

[1]Laboratory of Applied Psychology, Department of Psychology, University of Crete, Rethymnon, Greece

[2]Addiction Psychology Laboratory, Department of Psychology, University of Crete, Rethymnon, Greece

[3]Department of Neonatology/Neonatal Intensive Care Unit, University Hospital of Heraklion, School of Medicine, University of Crete, Heraklion, Greece

[4]Department of Obstetrics – Gynecology, General Hospital of Chania, Chania, Greece

[5]Department of Psychology, University of Crete, Rethymnon, Greece

Limited evidence regarding the role of family functioning and social support on both parents' mental health during the COVID-19 pandemic – although focused at the individual level of analysis – showed that: a) in Portugal, postpartum parents who described a marked impact of COVID-19 on their intimate relationships reported higher depression and scored lower on perceived social support [1]; and b) in China, family function fully mediated the relationship between social support and depression [2].

The present study aimed to investigate in a dyadic context the mediating role of family cohesion and flexibility in the association between social support and postnatal depression in new parents during the COVID-19 crisis in Crete, Greece.

Two hundred and twenty-two new parents, nested in 111 heterosexual couples, completed self-report questionnaires within 8 weeks after birth. Perceived social support was assessed with the Multidimensional Scale of Perceived Social Support (MSPSS), perceived family functioning with the Family Adaptability and Cohesion Evaluation Scales IV Package (FACES IV) and maternal/paternal postnatal depression with the Edinburgh Postnatal Depression Scale (EPDS). The Actor-Partner Interdependence Model (APIM) and the Actor-Partner Interdependence Mediation Model (APIMeM) were used to analyze intra- and interpersonal effects.

We found an actor-actor effect indicating that both mothers' and fathers' own perceived social support decreases or improves their depressive symptoms via their own perception of family cohesion and flexibility. Additionally, the actor-partner effect indicates that both mothers' and fathers' own perceived social support decreases or improves their partners' depressive symptoms via their own perception of family cohesion and flexibility.

These findings confirm the crucial role of perceived family function in the association between perceived social support and postnatal depression and the shared interdependence of new parents' experiences of the pandemic crisis. We highlight the need for healthcare professionals to focus on evaluating family function constantly across the perinatal period and provide partner-inclusive and couple-based interventions in order to promote new parents' well-being.

REFERENCES

[1] Tavares IM, Fernandes J, Moura CV, Nobre PJ, Carrito ML. Adapting to Uncertainty: A Mixed-Method Study on the Effects of the COVID-19 Pandemic on Expectant and Postpartum Women and Men. Front Psychol. 2021;12:688340.

[2] Huang Y, Liu Y, Wang Y, Liu D. Family function fully mediates the relationship between social support and perinatal depression in rural Southwest China. BMC Psychiatry. 2021;21(1):151.

ABS 53

EVALUATING THE PREVALENCE OF POSTNATAL GROWTH FALTERING OF PRETERM INFANTS USING TWO GROWTH CHARTS AND ITS RELATION TO CLINICAL AND NUTRITIONAL PARAMETERS

I. Kakatsaki[1], N. Anagnostatou[1], T. Roumeliotaki[2], S. Papanikolaou[1], E. Hatzidaki[1]

[1]Department of Neonatology/Neonatal Intensive Care Unit, University General Hospital of Heraklion, School of Medicine, University of Crete, Heraklion, Greece

[2]*Clinic of Preventive Medicine and Nutrition, Division of Social Medicine, School of Medicine, University of Crete, Heraklion, Greece*

BACKGROUND

Postnatal growth faltering (PGF) negatively impacts preterm neonates, leading to compromised neurodevelopment and an increased risk of metabolic syndrome and cardiovascular disease [1, 2]. However, comparing studies on PGF is challenging due to the absence of a standardized definition and the considerable variation in growth assessment charts [3].

AIM

Our study aimed to compare the prevalence of PGF in preterm neonates using two commonly used growth charts, Fenton2013 and INTERGROWTH-21st, and to identify associated nutritional and clinical factors.

METHODS

This retrospective study, conducted at a level III Neonatal Intensive Care Unit, analyzed 650 preterm neonates born before 33 weeks of gestation. PGF and severe PGF were defined as weight loss exceeding one or two standard deviations, respectively, between birth and discharge.

RESULTS

The mean gestational (GA) and postmenstrual age (PMA) at discharge was 30 weeks and 37.1 weeks, respectively. The mean birth weight (BW) of the infants was 1,475.9 grams. The Fenton2013 growth charts showed a higher prevalence of PGF (43%) and severe PGF (14.6%) compared to the INTERGROWTH-21st (24.5% and 10.3%, respectively). Perinatal factors such as lower GA and lower BW were associated with higher PGF risk across both growth charts. Additionally, being small for gestational age (SGA) was significantly associated with a lower risk of PGF in both growth charts ($p < 0.001$), suggesting a protective effect. Earlier initiation of enteral feeding and a more rapid establishment of full enteral feeds were strongly associated with reduced PGF prevalence in both growth charts ($p < 0.001$), as was shorter hospitalization. A declining trend in PGF prevalence over the study period was noted, likely due to the implementation of updated nutritional guidelines.

CONCLUSION

The differences in PGF prevalence between the Fenton2013 and INTERGROWTH-21st growth charts underscore the need for further research to identify the most suitable growth chart for evaluating and monitoring PGF in preterm neonates. Establishing standardized PGF definitions, while incorporating associated nutritional and clinical factors, is crucial for optimizing nutritional interventions aimed at improving growth, neurodevelopment, and long-term health outcomes in this vulnerable population.

REFERENCES

[1] Zozaya C, Díaz C, Saenz de Pipaón M. How Should We Define Postnatal Growth Restriction in Preterm Infants? Neonatology. 2018;114(2):177-80.

[2] Ordóñez-Díaz MD, Pérez-Navero JL, Flores-Rojas K, Olza-Meneses J, Muñoz-Villanueva MC, Aguilera-García CM, Gil-Campos M. Prematurity With Extrauterine Growth Restriction Increases the Risk of Higher Levels of Glucose, Low-Grade of Inflammation and Hypertension in Prepubertal Children. Front Pediatr. 2020;8:180.

[3] Fenton TR, Cormack B, Goldberg D, Nasser R, Alshaikh B, Eliasziw M, Hay WW, Hoyos A, Anderson D, Bloomfield F, Griffin I, Embleton N, Rochow N, Taylor S, Senterre T, Schanler RJ, Elmrayed S, Groh-Wargo S, Adamkin D, Shah PS. "Extrauterine growth restriction" and "postnatal growth failure" are misnomers for preterm infants. J Perinatol. 2020;40(5): 704-14.