www.jpnim.com Open Access eISSN: 2281-0692
Journal of Pediatric and Neonatal Individualized Medicine 2026;15(1):e150102
doi: 10.7363/150102
Published online: 2025 Nov 30

Selected Abstracts of the 21st International Workshop on Neonatology and Pediatrics

EVOLUTION AND REVOLUTION IN NEONATOLOGY AND PEDIATRICS

CAGLIARI (ITALY) · OCTOBER 22ND-25TH, 2025

The Workshop has been organized with the patronage of the Autonomous Region of Sardinia, City of Cagliari, Azienda Ospedaliera di Cagliari, University of Cagliari, Board of Physicians of the Province of Cagliari, Albanian-Italian Pediatric Society (AIPS), AMBO (*Alleanza per un Ambiente a misura di Bambino*: alliance for a child-friendly environment), Board of Directors of Italian Pediatrics Schools, European Project Better4u, European Project Life Milch, National Coordination of Heads of Pediatric Departments (CONAPP), Italian Federation of Family Pediatricians (FIMP), Italian-Arabian Pediatric Society (IAPS), Italian-Romanian Pediatric Society (IRPS), Italian-Swedish Cultural Association (ACIS), Norman Group of Neonatal and Pediatric Nephrology, International Academy of Perinatal Medicine (IAPM), 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), "SIPO in the World" Association, Italian Society of Pediatric Psychology (S.I.P.Ped.), Italian Society for Research on Essential Oils (SIROE), Italian Society of Neonatal Nursing (SIN INF), "*Pediatria Più*" Association, Italian Society of Developmental Origins of Health and Disease (SI-DOHaD), Italian Society of Pediatricians (SIMPE).

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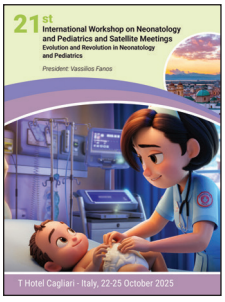

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How to cite

[Abstract's authors]. [Abstract's title]. In: Selected Abstracts of the 21st International Workshop on Neonatology and Pediatrics; Cagliari (Italy); October 22-25, 2025. J Pediatr Neonat Individual Med. 2026;15(1):e150102. doi: 10.7363/150102.

ABS 1

PLATELET TRANSFUSIONS IN NEONATES: IMPACT ON COAGULATION AND IMMUNOLOGICAL EFFECTS

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Prophylactic platelet transfusion is considered to reduce the risk of bleeding in preterm sick thrombocytopenic neonates. Variable transfusion strategies have been recorded among institutes. The results of the Platelets for Neonatal Transfusion Study 2 (PlaNeT-2) demonstrated that neonates with the highest baseline risk for bleeding or mortality, according to gestational age and clinical status, benefited from transfusions based on a restrictive threshold of platelets at 25,000 PLTs/μL. Conversely, liberal platelet transfusions are associated with increased morbidity and mortality among preterm neonates admitted to the Neonatal Intensive Care Unit. Additionally, the recent literature highlights the harmful effects of platelet transfusions in premature neonates regarding neurodevelopmental impairment, while the indicative platelet count for transfusion varies among neonatal groups. Platelets are fundamental components of hemostasis, but they are equally involved in immunological pathways. Regarding the hemostatic functions of neonatal platelets, it is well known that neonatal platelets exhibit hypo-responsiveness to multiple platelet agonists compared to adult platelets; however, little is known regarding differences in platelet immune function across development. The detrimental effects of platelet transfusions in neonates might be associated with disturbances in the neonatal hemostatic balance or aberrant immune responses induced by the transfused adult platelets.

ABS 2

PATTERNS AND ASSOCIATED FACTORS OF CONGENITAL ANOMALIES AMONG NEONATES IN 14 YEMENI GOVERNORATES (2021-2023): A CASE-CONTROL STUDY

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BACKGROUND
Long-term disability and a reduced quality of life are often associated with congenital anomalies, which present as structural, functional, or metabolic defects. This study offers a comprehensive overview of neonatal congenital anomalies in 14 Yemeni governorates, a significant yet often overlooked public health concern. The current study aimed to determine the patterns and associated factors of congenital anomalies in 14 Yemeni governorates between 2021 and 2023.
METHODS
An unmatched case-control 1:2 design was employed using secondary data collected from various health facilities across 14 Yemeni governorates between 2021 and 2023. The sample size was calculated, and the data were analyzed using Epi Info™ version 7.2, comprising 612 neonates with a documented diagnosis of congenital anomalies and 1,224 healthy neonates. Binary and multiple logistic regression analyses were used to identify factors associated with congenital anomalies, in conjunction with the chi-square test.
RESULTS
The majority of the congenital anomalies identified were located in Al Hudaydah (34%), Ibb (17.2%), and Sana'a (13.1%). Most were isolated (518 [84.64%]), whereas 94 (15.36%) were multiple. The predominant system was the nervous system (33.9%), followed by the skeletal system (14.8%) and orofacial anomalies (10.6%). Furthermore, strong associations were found with positive consanguinity (OR = 28.82), low socioeconomic status (OR = 10.70), maternal age ≥ 35 years old (OR = 7.66), stress (OR = 4.95), acute diseases (OR = 3.56), gestational age < 37 weeks (OR = 3.32), maternal age < 20 years old (OR = 2.32), positive family history (OR = 1.74), low birth weight (OR = 1.27), grand-multiparity (OR = 0.71) and male sex (OR = 0.10).
INTERPRETATION
This broad research identified significant patterns, maternal and neonatal associations, and protective variables for congenital anomalies. These results can help inform national interventions and policies aimed at preventing and improving neonatal care.

FUNDING

This study was self-funded by the authors and did not receive any external funding or any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

ABS 3

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

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The need for anthropometric neonatal charts specific to twins is under debate. Assessing the size of twins at birth using charts developed for singletons may lead to overdiagnosis of small for gestational age in this subpopulation. In contrast, the availability of specific twin charts would enable the evaluation of differential effects of pregnancy-related morbidity beyond the physiological adaptation induced by twinning itself.

INTERGROWTH-21st was a population-based, international project aimed at producing prescriptive growth standards for fetuses and newborns, adopting the same prescriptive conceptual framework used for the construction of the WHO International Child Growth Standards. This longitudinal observational study, conducted in 8 geographically diverse settings, prospectively collected data from May 2009 to August 2013 on healthy pregnant women and their newborns. All the women, in addition to the underlying population characteristics of low perinatal risk, met strict individual criteria for a population at low risk of impaired fetal growth. Newborn weight, length and head circumference measures were collected independently in duplicate by 2 trained anthropometrists within 12 hours of birth using identical equipment and protocols at all sites.

From this population, international standards for singleton births in terms of 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 to twins were constructed using the same population, methodology, equipment, and conceptual framework. The twin standards were created by selecting all twin births from the same population used to develop 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 assessing the goodness of fit, fractional polynomials were used to estimate the fitted centiles, assuming a skewed t-distribution.

From 1,034 multiple pregnancies, after exclusions of conditions such as smoking, maternal diseases, high maternal BMI, and congenital malformations, the final sample was 864 twin newborns. Most of the twins fall below the 50th centile of the INTERGROWTH-21st standards for singletons. Comparisons of twins' and singletons' centiles showed an increasing divergence along gestational ages towards term age for all anthropometric variables, but this divergence was less marked for head circumference.

We present international newborn size normative charts specifically tailored for twins. Their availability is relevant for clinical practice, enabling the evaluation of twin newborns worldwide, and for research applications, such as comparing growth rates between single and twin births.

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ABS 4

IMPACT OF MATERNAL SARS-CoV-2 INFECTION ON NEONATAL URINARY METABOLOME: A LONGITUDINAL STUDY OVER THE FIRST MONTH OF LIFE

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Maternal SARS-CoV-2 infection during the perinatal period may influence neonatal development through immune, nutritional, and microbial pathways, potentially altering maternal physiology, breast milk composition, and the perinatal environment [1]. While vertical transmission is rare, the neonatal metabolic imprint of maternal infection remains poorly understood [2]. Neonatal urine metabolomics provides a non-invasive window into early-life systemic changes during a critical period for developmental programming [3].

In this study, we investigated the impact of maternal SARS-CoV-2 infection on the neonatal urinary metabolome across the first month of life. An untargeted NMR-based metabolomics approach was applied to urine samples collected at three postpartum timepoints: T0 (week 1), T1 (week 2), and T2 (weeks 4-5). Infants were grouped according to maternal infection status and timing: Control, mothers with no history of SARS-CoV-2 infection (n = 27); PosNeg, mothers testing positive at delivery and during the first postpartum week (n = 19); PosPos, mothers testing positive at delivery and remaining positive up to 15 days postpartum (n = 8). Multivariate and univariate statistical analyses were performed to assess metabolic alterations, including principal component analysis (PCA), Kruskal-Wallis tests with Dunn's post-hoc comparisons, differential correlation analysis, timepoint-specific linear regression, and linear mixed-effects models. This comprehensive approach aimed to characterize dynamic metabolic changes potentially associated with maternal infection status and to identify early biomarkers of altered neonatal physiology.

PCA highlighted timepoint-driven shifts in metabolomic profiles, with T1 showing the most apparent separation. Discriminant metabolites included acetoacetate, formate, succinate, myo-inositol, glycine, and 4-hydroxyphenylacetate. In PosNeg neonates, disrupted correlation patterns were observed, particularly involving citrate, betaine, and TCA cycle intermediates. Linear mixed models revealed group-specific effects, with PosNeg infants at T1 exhibiting distinct alterations in metabolites associated with energy metabolism, methylation, and microbial co-metabolism.

Maternal SARS-CoV-2 infection appears to be associated with transient metabolic changes in neonates, with more pronounced differences observed during the second week postpartum. While these findings suggest a potential impact of maternal infection on early neonatal metabolism, further studies are necessary to confirm these observations and clarify their physiological significance.

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ABS 5

ARTIFICIAL INTELLIGENCE IN NEONATOLOGY: WHERE ARE WE NOW?

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Artificial intelligence (AI) has rapidly advanced from experimental applications to clinically relevant tools, with neonatology emerging as one of the

most promising fields of impact. Neonatal Intensive Care Units generate vast amounts of physiological, imaging, and laboratory data, making them particularly suitable for AI-driven analysis. Current applications of AI in neonatology span neurological monitoring, respiratory support, imaging analysis, and risk stratification for major complications of prematurity.

One of the earliest and most successful domains has been neonatal neurology. Automated electroencephalogram classifiers and seizure detection algorithms have been developed to support clinicians in real time, addressing the challenge of continuous monitoring and expert interpretation [1]. Similarly, deep learning models have demonstrated accuracy comparable to that of human experts in brain magnetic resonance imaging segmentation, aiding in the assessment of white matter development and prognosis of neurodevelopmental disorders [2].

In respiratory medicine, AI has been applied to predict the risk and severity of conditions such as neonatal respiratory distress syndrome and bronchopulmonary dysplasia. Models that integrate spectroscopy, biomarker analysis, and chest imaging offer opportunities for earlier diagnosis and the development of personalized treatment strategies. These tools aim to reduce delays in intervention and to optimize long-term respiratory outcomes [3].

Beyond single-disease applications, AI also holds promise for integrating multimodal data, supporting precision medicine in neonatology. Predictive analytics may enable individualized care pathways, while automation can alleviate clinician workload and standardize interpretation across centers. Nevertheless, significant challenges remain, including the need for large, high-quality datasets, external validation, and transparent algorithms that can be trusted in clinical decision-making. Ethical considerations such as data privacy, algorithmic bias, and equitable access must also be addressed before widespread implementation.

Currently, AI in neonatology is transitioning from proof-of-concept studies to early clinical translation. While most tools remain in validation stages, the trajectory suggests that AI will soon become an integral component of neonatal care, supporting clinicians in providing safer, more accurate, and personalized treatment for the most vulnerable patients.

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ABS 6

A TWISTED TIBIA IN A PRETERM NEONATE

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INTRODUCTION

Procurvatum tibia is a rare condition characterized by an abnormal curvature of the tibia, usually associated with talo-valgus foot deformity [1, 2].

CASE REPORT

A preterm newborn at 31^{+6} weeks' gestation, delivered by cesarean section, was transferred to the Neonatal Intensive Care Unit due to respiratory distress. During hospitalization, a deformity of the tibia was observed, accompanied by a left talo-valgus deformity of the foot. Radiographic examination of the lower limb, performed in anteroposterior and lateral views, confirmed the diagnosis of procurvatum tibia with talo-valgus foot, without evident focal lesions. Subsequently, an orthopedic evaluation confirmed the malformation and recommended a regimen of physiotherapeutic manipulations, along with an early ultrasound assessment of the hips, which proved normal for the patient's age.

The newborn completed a cycle of 20 physiokinesitherapy sessions, showing partial improvements in mobility and postural alignment. An orthopedic follow-up was scheduled at a specialized Pediatric Orthopedic Center to monitor the condition's progression, as the most frequently reported outcome in similar cases in the literature is limb length discrepancy. This phenomenon requires close monitoring during follow-up. It is also essential to rule out the possibility that this anomaly may be part of a more complex syndromic picture [1, 2].

CONCLUSIONS

The prognosis for a preterm newborn with procurvatum tibia is generally favorable, with resolution of the deformity within the first 2 to 3 years of life. However, continuous orthopedic follow-up is crucial for monitoring the condition's evolution and preventing complications such as limb length discrepancies, fractures, joint instability, altered plantar support, or pseudoarthrosis [1, 2]. A timely and multidisciplinary approach is essential to optimize long-term functional outcomes [3].

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ABS 7

SCHRÖDINGER'S FETUS

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The moral status of the embryo has been a subject of contention among scientists, philosophers, and religious groups. A fetus is actually called a person only after a living birth. However, determining when, how, or under which conditions the moral status is established and taken into consideration can be proven challenging.

According to Schrödinger's experiment regarding quantum superposition, a cat kept in a sealed box can be considered at the same time dead and alive, since there is no way to "see" the reality inside the box. The only way to define its condition is to open the box and determine its actual state. As a paradox of this idea, recently the moral status of an embryo has been described as an unknown condition until we open this box and see its actual future. In the classical approach, a fetus is considered to have a moral status from the moment of its existence. At the same time, it is believed that a standard fetus that will be born has a moral status, but a fetus that will not be born does not.

However, this theory demands one vital precondition to be accepted, the future does not belong to today, so it cannot be used as a reference for a definition of today's condition. A crucial event must occur to signal the beginning of morality, the moment when a fetus is transformed into a person, both legally and ethically. In literature, events like this are the beginning of human life and the establishment of consciousness. Each one of these events serves as the opening of the box, which will reveal the real status of the fetus. Nevertheless, neither the first nor the second precondition can be totally defined by science today.

The beginning of human life has been a subject of great debate since antiquity, a battle in which medicine, philosophy, and religion have crossed swords endlessly. It has been claimed that life begins at conception, as the unique DNA of a person is formed at this point. But is it truly unique? Many developmental biologists believe that life starts after gastrulation, which occurs around the 14th day after fertilization. Their argument is simple: before that day, distinct twins can emerge, so if life begins at conception, then life can be split in two. However, many experts support the opinion that life itself begins when consciousness is established, which is defined as the emergence of a functional electroencephalogram, typically around 22 weeks of gestation, when thalamocortical radiations start to develop. As they claim, the end of life is defined as the absence of central nervous system signals, so their emergence must determine the beginning. Finally, some believe, including representatives in the legal field, that life begins at birth.

The theory of Schrödinger's fetus might be helpful to determine the moral status of an early embryo, especially in the field of the interruption of pregnancy. This twofold nature of the embryo might resolve many disputes, although a scientific consensus regarding the significant event that will determine when the moral status is established must be established.

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ABS 8

UNEXPLAINED HYPOTONIA IN THE NEWBORN: A CASE THAT CHALLENGES FIRST IMPRESSIONS

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INTRODUCTION

Congenital myopathies are a heterogeneous group of neuromuscular disorders typically presenting in the neonatal period with hypotonia, muscle weakness, and respiratory impairment. The *TNNC2* gene encodes troponin C type 2, a calcium-binding protein expressed in fast-twitch skeletal muscle fibers, essential for excitation-contraction coupling. While heterozygous pathogenic variants have been linked to autosomal dominant forms, recent studies suggest that biallelic variants may cause more severe autosomal recessive presentations. We report the first neonatal case with compound heterozygous *TNNC2* variants, contributing to the expanding clinical and molecular spectrum of *TNNC2*-related disease [1, 2].

CASE REPORT

A term neonate was born to non-consanguineous parents via emergency cesarean section for fetal bradycardia, following an uneventful pregnancy. At birth, the infant presented with respiratory distress requiring non-invasive support (nCPAP and supplemental oxygen). The patient was admitted to the Neonatal Intensive Care Unit due to persistent desaturations and feeding difficulties. Neurological examination revealed axial hypotonia, distal tremors, and weak sucking. Feeding was associated with episodes of bradycardia, cyanosis, trunk arching, and nasal regurgitation of milk. A tracheoesophageal fistula was initially suspected but excluded through contrast-enhanced imaging and multidisciplinary evaluation. Chest MRI demonstrated a pulmonary opacity; angio-CT confirmed an active inflammatory focus consistent with aspiration pneumonia, which resolved with oral cefotaxime. A 24-hour pH-impedance study revealed pathological gastroesophageal reflux (102 episodes; Symptom Index: 25%), prompting the initiation of esomeprazole (1 mg/kg/day) and an anti-reflux formula, resulting in significant clinical improvement. Despite initial clinical concerns, the brain MRI and EEG results were unremarkable. At discharge, the infant showed improved feeding and stable respiratory function, although mild hypotonia and joint hyperlaxity persisted. Trio-based exome sequencing identified two *TNNC2* variants in compound heterozygosity: c.314+1G>C (paternal, pathogenic) and c.326G>A (maternal, likely pathogenic). This genotype is consistent with a recessive congenital myopathy, similar to a recently reported case involving complete loss of functional transcript [1].

CONCLUSIONS

This case expands the phenotypic understanding of *TNNC2*-related congenital myopathies, showing that compound heterozygous variants may manifest with a mild yet diagnostically challenging neonatal presentation. It highlights the value of early genomic testing in neonates with unexplained hypotonia to enable accurate diagnosis, personalized follow-up, and family counseling. While no targeted therapies are currently available, emerging approaches involving calcium sensitizers may hold promise for the future in *TNNC2*-associated disorders [2].

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ABS 9

ACUTE MONOARTHRITIS IN A PEDIATRIC PATIENT: A CASE OF *SALMONELLA*-INDUCED REACTIVE ARTHRITIS

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INTRODUCTION

Reactive arthritis (ReA) is a sterile joint inflammation triggered by a previous gastrointestinal or genitourinary infection, typically occurring days to a few weeks prior, and primarily affecting individuals between 15 and 35 years of age [1]. *Campylobacter*, *Salmonella*, and *Shigella* are the three enteric pathogens most commonly associated with ReA, while *Chlamydia* is the primary pathogen linked to genitourinary infections [2]. The clinical picture of ReA is predominantly mono- or oligoarthritis of the lower extremities; however, synovitis of smaller joints has also been described (e.g., hands, feet, sacroiliac joints). Duration of diarrhoea is significantly longer in patients who develop ReA, suggesting that the persistence of bacteria in the mucosa may trigger an exaggerated immune response. Antibiotic therapy for enteritis appears to have no preventive effect on the development of ReA [3].

CASE REPORT

Three days after eating raw eggs, a 13-year-old boy began experiencing headache, abdominal pain, and watery stools, some with traces of blood. One week later, he developed arthralgia in the left knee, accompanied by significant warmth, swelling, limited range of motion, and fever. He was then admitted to our Pediatric Unit. Blood tests revealed a slight elevation of inflammatory markers (CRP: 3.1 mg/dL, PCT: 0.06 ng/mL). Both stool PCR and stool culture identified *Salmonella* group B. Ultrasound examination of the knee revealed a marked joint effusion of anechoic fluid (thickness of 2 cm) and synovial thickening. Due to significant swelling, arthrocentesis was performed, yielding 90 mL of turbid, citrine-yellow synovial fluid. Analysis showed pH 8, SG 1,036, protein 53 g/L, 61% PMNs, 16% lymphocytes, 23% macrophages, and 1,000-2,000 RBCs/μL. Cultures from both blood and synovial fluid were negative, supporting a reactive rather than septic origin of the arthritis. Anti-inflammatory treatment with oral ibuprofen was initiated (400 mg, administered 3 times daily for 10 days), resulting in clinical improvement and recovery of joint mobility over the subsequent days. After discontinuing ibuprofen, the boy was reassessed due to the recurrence of arthralgia and limping. The ultrasound showed an anechoic joint effusion with a maximum thickness of 1.2 cm, so ibuprofen therapy was resumed, administered at regular intervals for 10 days. Tests were performed to exclude an autoimmune etiology for the condition (ESR, C3, C4, ANA, RF), all of which were negative. The boy has steadily improved and has not shown any further relapses.

DISCUSSION

This case confirms the link between *Salmonella* infection and ReA in pediatric patients. Prompt diagnosis and anti-inflammatory therapy led to rapid clinical improvement and prevented long-term complications. Arthrocentesis was performed due to significant joint distension and proved essential in definitively ruling out an infectious origin of the condition.

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ABS 10

NUTRITIONAL MANAGEMENT OF THE NEWBORN WITH NECROTIZING ENTEROCOLITIS: A CASE REPORT

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INTRODUCTION

Necrotizing enterocolitis (NEC) is a common disease in premature infants, with a multifactorial etiology and a complex interaction among nutrition, abnormal bacterial colonization, and the inflammatory response of the intestinal epithelium [1, 2]. Nutrition, particularly timing, type of milk and feeding practices, plays a crucial role as a major modifiable risk factor for NEC [1, 3]. Exclusive breastfeeding is a protective factor, although it does not entirely prevent the disease [1, 2]. After NEC diagnosis, enteral nutrition (EN) is suspended, and parenteral nutrition (PN) and antibiotic therapy are initiated. The need for surgical intervention is subsequently evaluated [2, 3]. The resumption of

EN is characterized by gradual increases, preventing the complications of prolonged PN [3].

CASE REPORT

A preterm infant, fed with human milk and formula from birth, developed abdominal distention associated with gastric residuals stained with bile and later with blood at 17 days of age. Physical examination, abdominal ultrasound, and blood tests raised the suspicion of NEC, leading to the initiation of antibiotic therapy and discontinuation of EN. Serial abdominal X-rays and ultrasounds, as well as repeated surgical consultations, were performed until he was 25 days old. As clinical conditions did not improve, despite the apparent absence of perforation, the surgeon recommended abdominal exploration, which revealed two large perforations in the last ileal loop. Ileal resection and distal and proximal ileostomy were performed. The patient remained fasting for 26 days (18 days post-surgery). EN was gradually reintroduced, achieving complete EN after 21 days, with excellent tolerance. After 49 days, intestinal recanalization surgery was performed, followed by successful reintroduction of EN from the third postoperative day.

DISCUSSION

Nutritional management of NEC is an essential aspect in both prevention and treatment [2, 3]. In this clinical case, in accordance with the literature [1-3], nutrition with human milk did not completely prevent the disease. However, suspending EN when NEC was suspected, along with a slow and gradual approach to resuming feeding after surgery, ensured a good recovery.

CONCLUSION

This case highlights the importance of nutritional management in infants with NEC. Additionally, adjusting the refeeding schedule based on the patient's tolerance is essential to promote optimal clinical recovery.

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ABS 11

POSTERIOR REVERSIBLE ENCEPHALOPATHY SYNDROME IN PEDIATRIC AGE: A CASE REPORT

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INTRODUCTION

Posterior reversible encephalopathy syndrome (PRES) is a severe clinical and neuroradiological syndrome with a rare occurrence among children and a good prognosis when treated [1]. The most common symptoms are headache, seizures, visual disturbances and systemic hypertension.

The MRI pathognomonic sign is a reversible vasogenic brain edema located in the parietal and occipital regions. Although the pathophysiology of PRES has yet to be fully clarified, multiple etiologies can lead to endothelial dysfunction, the main feature of PRES, including renal failure, arterial hypertension, autoimmune conditions, and oncological and hematological diseases.

The disease usually resolves when the cause is identified and treated [2]. A study found an increased incidence of underlying hypertension in the renal disease group compared to the group without renal disease (68.8% vs. 42.8%); additionally, antihypertensive therapy tended to be more effective in the group without renal disease [3].

CASE REPORT

A 9-year-old child was admitted to the ER for a focal onset seizure, which shortly after became a generalized tonic-clonic seizure. Laboratory tests revealed leukocytosis, thrombocytosis, and negative inflammatory markers. Microbiological tests on blood, stool, and CSF fluid were negative, while hypoproteinorrachia in CSF was found.

A contrast-enhanced brain MRI revealed bilateral hyperintensity of the white matter in the posterior regions, consistent with a posterior vasogenic edema, along with a slight shrinking of the affected gyri. EEG initially showed slow-wave activity in the temporo-parietal level bilaterally; when repeated after a week during the waking state, the abnormalities migrated to the temporo-parieto-occipital regions. Simultaneously, hypertension was detected in repeated measurements. Secondary hypertension was ruled out through multiple examinations, including ECG, cardiac ultrasound, ophthalmoscopy, and laboratory tests (RAS system, thyroid profile, cortisol, ACTH, antistreptolysin O-titer, C3, C4, IgA, ANA, ENA, and urinary

catecholamines). Therefore, hypertension was due to a non-renal disease; nocturnal awakenings and a mild mood deflection were reported only.
Antihypertensive and antiepileptic therapy was started with amlodipine and levetiracetam.
After 1 month, a contrast-enhanced brain MRI showed regression of the vasogenic edema in the posterior occipital regions, a finding that confirmed the hypothesis of PRES. The antihypertensive therapy was stopped after pressure monitoring and a few hypotensive episodes. After 2 months, clinical and neuroradiological improvement was confirmed, and antiepileptic treatment was also stopped.

DISCUSSION AND CONCLUSION

This case shows a parallel resolution of imaging abnormalities along with idiopathic hypertension and PRES symptoms when treatment was started. Although in our patient the final etiology of hypertension and PRES was not clear, prompt treatment was the key to a good prognosis.

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ABS 12

INFANTILE COLIC AND INTESTINAL INFLAMMATION

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INTRODUCTION

Infantile colic, characterized by inconsolable crying and irritability, is often associated with subclinical intestinal inflammation, which can be documented by an increase in fecal calprotectin [1]. Recent studies have shown that specific probiotics, in particular *Bifidobacterium longum ES1*, and butyric acid can modulate the mucosal immune response and reduce inflammatory markers [2, 3].

AIM

The aim is to describe the clinical and laboratory course of 3 children with infantile colic treated with butyric acid and *Bifidobacterium longum ES1* in combination with a controlled Mediterranean diet.

CASES REPORT

Three children were observed (2 females aged 1 year and 5 months and 2 years and 6 months, and 1 male aged 2 years and 9 months), all in good general health, with no anemia or other associated allergic conditions. Preliminary tests (fecal occult blood, urine, celiac panel including total IgA, anti-gliadin, anti-endomysium, and anti-transglutaminase) were negative. The children initially had elevated fecal calprotectin levels and recurrent colic.
Treatment consisted of butyric acid (1 tablet/day crushed and dissolved in water) and *Bifidobacterium longum ES1* (1 sachet/day) for 5 days a week, combined with a strict ethical Mediterranean diet, free of sugary drinks and ultra-processed foods, with attention to fiber, glycemic index, and the absence of foods containing advanced glycation end products (AGEs), combined with microbiota modulation interventions.
A progressive and significant reduction in fecal calprotectin levels was observed (from week 1 to week 4): Child 1: 800 → 300 → 60 µg/g; Child 2: 900 → 300 → 70 µg/g; Child 3: 850 → 200 → 30 µg/g. Fecal occult blood remained negative. At the same time, the clinical picture improved, with a reduction in the frequency and intensity of colic, more regular sleep patterns, and enhanced food tolerance. No adverse events were recorded.
The combination of butyric acid and *Bifidobacterium longum ES1*, along with an ethical and balanced Mediterranean diet, resulted in a marked clinical and laboratory improvement in 3 children with infantile colic and hyperexpression of fecal calprotectin.

CONCLUSION

Although limited by the small number of cases, these results support the hypothesis of a link between subclinical intestinal inflammation and colic, and suggest the usefulness of an integrated approach combining dietary therapy and microbiota modulation. Further prospective and controlled studies are needed to confirm these observations.

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ABS 13

A STUDY IN CYCLES: CLINICAL CLUES FROM CHILDHOOD VOMITING

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INTRODUCTION

First described in France (1806) and later in England (1882), cyclic vomiting syndrome (CVS) is a functional disorder characterized by recurrent, acute episodes of severe nausea and vomiting alternating with symptom-free intervals. The condition shows a slight female predominance (55:45), typically begins in early childhood, often resolves during adolescence, and is frequently followed by migraine. The differential diagnosis is wide, and NASPGHAN guidelines recommend targeted evaluation when alarm features are present, including abdominal pain or bleeding, metabolic triggers such as fasting or high-protein intake, and focal neurological deficits [1, 2].

CASE REPORT

An 8-year-old female with a ventriculo-peritoneal shunt placed at 7 months of age presented with vomiting episodes occurring every 3 to 4 weeks, each lasting about 72 hours. On the third day, the episodes invariably ended in a syncopal event with transient hypertonia, adding a neurological dimension to what had long been considered CVS. Physical examination and ancillary investigations excluded common organic causes of vomiting. The autonomic pattern led to an initial suspicion of Panayiotopoulos syndrome/self-limited epilepsy with autonomic seizures (SeLEAS), in which ictal vomiting is the hallmark. Electroencephalography instead showed rare epileptiform abnormalities during sleep, with biphasic spikes in the left centro-parietal region. These features were not typical of SeLEAS but pointed to an age-related focal epilepsy, confirming the coexistence of two distinct conditions: CVS and epilepsy. Antiseizure therapy improved overall clinical stability and supported the revised diagnosis.

CONCLUSIONS

This case highlights the complexity of evaluating recurrent vomiting in children. Although CVS is a recognized diagnosis, episodes that are prolonged, highly stereotyped, or associated with neurological manifestations, such as syncope or tonic posturing, need prompt re-evaluation. While autonomic epilepsies like SeLEAS remain an essential differential diagnosis, clinicians should recognize that vomiting may also coexist with an independent epileptic disorder [3]. Awareness of this overlap is necessary, as failure to consider epilepsy can delay appropriate treatment, prolong diagnostic uncertainty, and expose patients to repeated and unnecessary investigations. A comprehensive, multidisciplinary approach that integrates gastroenterological and neurological expertise plays a crucial role in everyday pediatric practice, facilitating timely diagnoses and optimizing patient outcomes.

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ABS 14

WHEN BREATHING FAILS, BLAME THE HEART

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INTRODUCTION

Neonatal atrial flutter (AFL) is a rare supraventricular tachyarrhythmia characterized by rapid and regular atrial contractions, typically ranging from 280 to 500 beats per minute [1]. It can follow a severe clinical course and lead to potentially life-threatening complications such as heart failure and hydrops fetalis [2]. Electrical cardioversion is the first-line treatment. Digoxin may be considered in cases that are refractory or recurrent [3].

METHODS

We describe a suggestive clinical case and conduct a systematic review of the literature on the clinical features and management of fetal/neonatal AFL.

CASE REPORT

A male newborn was delivered at term via induced labor (Apgar 8-9-9). The pregnancy was complicated by unrecognized gestational diabetes. At birth, the infant was admitted to the Neonatal Intensive Care Unit due to respiratory distress and abnormal neurological examination; therapeutic hypothermia was initiated. At approximately 4 hours of life, due to worsening respiratory status, he was intubated and treated with surfactant.

At the same time, due to occasional episodes of tachycardia, an ECG was performed, which revealed supraventricular tachycardia. He was then administered increasing boluses of adenosine for diagnostic purposes to differentiate between AVRT and AFL. After the third bolus of adenosine (0.25 mg/kg), an AV block 4:1 with a ventricular rate of 100 bpm was observed. The appearance of the AV block pattern confirmed the suspicion of AFL. Given the patient's hemodynamic stability, amiodarone therapy was initiated. Due to refractoriness to medical treatment, electrical cardioversion was performed, restoring sinus rhythm after the third attempt (1 J/kg).

Subsequent cardiac evaluations were regular. There was progressive improvement in respiratory status, with a return to spontaneous breathing by the 8th day. EEG during hypothermia was normal, while the following two were poorly organized for age. CUS and brain MRI were normal. The neurological examination revealed a disorganized motor pattern and paratonia. Due to dysmorphic features (microretrognathia, dolichocephaly, narrow palpebral fissures), a genetic consultation and trio-array CGH were performed, with negative results. Extended metabolic screening was regular. The patient remains under follow-up care at the Rare Diseases Outpatient Clinic.

DISCUSSION

Neonatal AFL is often a prenatal condition, but its diagnosis *in utero* is challenging due to variable degrees of atrioventricular conduction, which may result in a fetal heart rate within normal limits. Moreover, CTG monitoring does not detect fetal heart rates exceeding 200 bpm [1]. As a result, at birth, varying degrees of heart failure may present as neonatal respiratory distress, and the regular rhythm of AFL may be misinterpreted as sinus tachycardia [3].

CONCLUSIONS

In term neonates, respiratory distress warrants investigation for underlying causes. In this case, despite neurological complexity, continuous monitoring and multidisciplinary collaboration between the neonatologist and pediatric cardiologist led to the diagnosis of AFL with early cardiac decompensation as the primary cause of distress.

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ABS 15

UNUSUAL LOCALIZATION OF *BARTONELLA HENSELAE*: SPONDYLODISCITIS IN A FIVE-YEAR-OLD CHILD

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INTRODUCTION

Spondylodiscitis (SD) is a rare condition in children, with an incidence of 0.3 cases per 100,000, compared to 2.4 cases per 100,000 in adults, representing only 3% of osteoarticular infections

in children. Typically, it is primarily bacterial and acute, caused by pyogenic bacteria (*S. aureus*, *K. kingae*, etc.), and less often subacute/chronic, caused by non-pyogenic, granulomatous infections (*M. tuberculosis*, *Brucella spp.*, and *B. henselae* [BH]). Bone involvement in cat-scratch disease (CSD) due to BH is exceedingly rare, occurring in only 0.17-0.27% of cases. Vertebrae are the most common bone localizations of infection, reached by hematogenous spread.

CASE REPORT

A 5-year-old girl came to the Emergency Room (ER) with a 4-month history of intermittent back pain, initially irradiated to the abdomen, then worsening until total refusal to walk/to stand/to sit. The fever began 3 days before any injury or previous infectious event. During the 2 previous ER visits, gastroenteritis and myalgia were reported after a journey in Morocco 2 years before.

The examination revealed forced posture with legs bent on the trunk, no spinal deformities, and no neurological signs. Lab tests were remarkable for low levels of C-reactive protein and non-pathological leucocyte count. A lumbosacral MRI showed subacute-chronic findings of spondylodiscitis, in particular a widespread alteration of the STIR signal of the trabecular bone in L1-L2 vertebral bodies with marked thinning of the intervertebral disc.

Abdominal sonography and chest X-ray were normal. Intravenous cefazolin and morphine were started. After 8 days of admission, a CT-guided needle biopsy was planned but not performed due to the report of the following BH serology: positive IgM and IgG. Since all other infectious tests were negative, we promptly stopped cefazolin and started azithromycin and rifampicin orally. A corset was fitted to the patient to immobilize the spine. With the new treatment and spine immobilization, the patient showed a rapid clinical improvement. After 13 days of admission, the patient was discharged on oral antibiotics to be continued for 3 weeks.

After 8 weeks, MRI showed marked regression of inflammatory changes and a substantial reduction in disease activity. Serology testing was repeated with evidence of seroconversion. No neurosurgical intervention was necessary; a corset was used until the following neuroradiological control.

DISCUSSION AND CONCLUSION

Pediatric SD is rare, is hardly diagnosed and treated due to the non-specificity of clinical presentation and laboratory investigations, difficulty in etiologic identification, and a lack of management guidelines. In our case, the diagnosis of BH-induced SD was supported by serology and MRI, and non-invasive diagnostic tests were performed. In complicated forms of CSD, such as bone infection, the appropriate choice of drug is critical to achieve a cure. In our case, the synergic effect of azithromycin and rifampicin allowed us to eradicate the infection.

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ABS 16

CLAVICLE FRACTURES AT BIRTH: EXPERIENCE FROM TWO CASE REPORTS

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INTRODUCTION

Birth-related clavicle fractures are among the most frequent neonatal injuries, typically associated with large birthweight infants and complicated vaginal deliveries, particularly in the setting of shoulder dystocia. In most cases, the fracture is incomplete and heals spontaneously [1]. However, exceptions may occur in preterm or asphyxiated infants, where clavicular fractures can present even in the absence of macrosomia [2].

CASE REPORTS

Case 1: a preterm infant born at 32 weeks by precipitous labor with difficult extraction presented with reduced spontaneous upper limb movement. Clinical examination revealed crepitus at the medial third of the clavicle, and radiography confirmed a displaced fracture. The arm was initially immobilized with a Desault bandage, but this was removed shortly after due to worsening respiratory distress. Pain was managed with paracetamol as needed.

Case 2: a term infant (38 weeks), delivered asphyxiated and subsequently treated with systemic hypothermia,

was found to have a displaced fracture of the medial third of the clavicle. Crepitus was detected on palpation, and radiography confirmed the diagnosis. No immobilization was performed, as the newborn was sedated with fentanyl during hypothermia treatment, which also provided adequate analgesia.

In both cases, management was conservative. Ultrasonographic follow-up at 7-10 days demonstrated callus formation, with restoration of spontaneous limb mobility.

DISCUSSION

Although most clavicle fractures at birth are associated with macrosomia and shoulder dystocia, these cases highlight atypical presentations in a preterm infant and in a term asphyxiated newborn, both appropriate for gestational age. The fractures involved the medial third of the clavicle and were displaced, a morphology that may warrant careful evaluation for concomitant brachial plexus injury. Diagnosis was based on clinical examination, palpable crepitus and reduced limb use, confirmed by radiography. Ultrasonography proved effective for follow-up, enabling the assessment of callus formation while avoiding repeated radiation exposure.

CONCLUSION

Neonatal clavicle fractures, although usually linked to large birthweight and difficult vaginal delivery, may also occur in preterm or asphyxiated infants. Conservative management remains the treatment of choice, with ultrasonographic follow-up providing a safe and reliable tool to monitor healing. These findings emphasize the importance of considering clavicle fractures in atypical perinatal contexts and the value of systematic evaluation for associated brachial plexus injury.

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ABS 17

HIDDEN IN PLAIN SIGHT: WHEN HERPES ISN'T THE ANSWER

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INTRODUCTION

Mycoplasma pneumoniae is a frequent cause of community-acquired pneumonia in children, with extrapulmonary complications occurring in up to one quarter of cases. Among these, *Mycoplasma pneumoniae*-induced rash and mucositis (MIRM) has been recognized as a distinct clinicopathological entity, defined by predominant mucosal involvement and relatively limited skin disease. Since its systematic description in 2015, MIRM has remained underdiagnosed and is frequently mistaken for erythema multiforme, Stevens-Johnson syndrome (SJS), or toxic epidermal necrolysis (TEN) [1-3]. Accurate differentiation is essential, as the clinical course and prognosis of MIRM are substantially more favorable.

CASE REPORT

A 13-year-old previously healthy female was admitted with severe painful mucositis after 5 days of chest pain, dyspnea, and lip swelling, unsuccessfully treated with corticosteroids and antihistamines. On admission, she was apyretic and in fair general condition. Physical examination revealed multiple vesiculocrusted lesions of the lips, palate, and gingiva with marked gingival erythema and cervical lymphadenopathy, with no relevant skin involvement. Laboratory investigations showed slight leukocytosis with neutrophilia, elevated ESR, and normal renal and hepatic function. Serology confirmed *Mycoplasma pneumoniae* infection, while HSV1/2 and HHV6/7 PCR were negative. The combination of severe mucosal involvement, minimal skin disease, serological evidence of recent *Mycoplasma pneumoniae* infection, and exclusion of alternative viral etiologies led to the diagnosis of MIRM. The patient was treated with oral azithromycin and supportive measures, with progressive improvement of mucosal lesions, pain relief, and recovery of oral intake. Within 1 week, the mucosal lesions had completely resolved, and the patient achieved a full recovery.

CONCLUSIONS

This case highlights the defining features of pediatric MIRM, including extensive mucosal involvement, minimal skin manifestations, and microbiological evidence of *Mycoplasma pneumoniae*. The exclusion of alternative viral etiologies reinforced the diagnosis. Early recognition and timely administration of antibiotics, in combination with supportive care, resulted in a complete recovery.

Awareness of MIRM among pediatricians and dermatologists is essential to prevent misdiagnosis as SJS/TEN and to ensure appropriate treatment and follow-up [1-3].

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ABS 18

SEPSIS-ASSOCIATED ENCEPHALOPATHY IN A SIX-MONTH-OLD INFANT

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INTRODUCTION

Sepsis-associated encephalopathy (SAE) is a diffuse brain dysfunction without a direct brain infection [1]. Symptoms include impaired consciousness, cognitive deficiency, convulsions, or deep coma, with possible long-term neurological sequelae. Disruption of the blood-brain barrier and extensive inflammatory activation during bacterial sepsis (caused by *S. agalactiae*, *S. aureus*, *S. pyogenes*, etc.) are key factors in the pathogenesis [2]. The most commonly applied diagnostic tools include EEG, neuroimaging, and biomarker detection [2, 3]. Treatment mainly focuses on managing underlying conditions and supportive therapy [2].

CASE REPORT

A previously healthy 6-month-old infant was admitted to a peripheral hospital with fever, tachycardia, weak pulse, diarrhea, cough, contraction of diuresis, inappetence and irritability. Blood tests showed leukopenia, neutropenia, a significant elevation of inflammatory indices, and hyponatremia. In the suspicion of sepsis, a first dose of IV antibiotics was given, and he was transferred to our hospital, where he appeared alert but poorly responsive, irritable, with no neurological deficits. Here, blood cultures were taken and turned up negative after 5 days. CSF at spinal tap was found to be turbid, with no protein, mild hypoglycorrhachia, and WBC 18 U/mcL (neutrophils 5.6%, monocytes 94.4%). Microbiological tests on the CSF were negative, as were PCR Film-Array tests on stool, upper tract swab Film-Array, and testing for West Nile. IgM and IgG antibodies for CMV were found to be positive with intermediate avidity. Viral PCR on blood was positive for CMV, with a low viral load. The EEGs initially showed diffuse slowing of background activity compatible with diffuse distress; later, the background trace improved, but epileptiform abnormalities appeared in the right posterior and anterior regions. Brain MRI showed an area of restriction at the level of the juxtacortical white matter of the right rolandic operculum and an alteration of the outer fibers of the corpus callosum in the proper paramedian position. Since the start of antibiotic therapy with ceftriaxone (stopped after 2 weeks), the baby showed a progressive clinical and biochemical response, except for persistent moderate neutropenia.

DISCUSSION AND CONCLUSIONS

MRI images were compatible with SAE. The etiology of sepsis remained uncertain, likely due to the inappropriate timing of blood cultures. The baby was followed up at our hospital with neurological examinations, EEG, and blood chemistry tests. Currently, he exhibits normal neurological development and is showing progressive improvement in his EEG. A follow-up MRI scan is scheduled for the coming months. The initial hypothesis that CMV infection had caused neutropenia, which subsequently contributed to sepsis, has been challenged by the persistence of low neutrophil count. Therefore, specialist investigations are underway to determine the possibility of a primary immunological deficiency that may have predisposed the child to such a severe infection and the resulting SAE.

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ABS 19

PREVENTING AND FACING OBESITY THROUGH THE BETTER4U EU-FUNDED PROJECT

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BETTER4U is a project funded by the European Union's Horizon Europe programme (grant agreement No. 101080117) that addresses the rising rates of obesity and weight gain. Obesity is a chronic non-communicable disease (NCD) and a significant risk factor for other chronic NCDs, including type 2 diabetes, cardiovascular and renal diseases, and several cancers.

The project conducts comprehensive research to elucidate the interplay of genetic, lifestyle, and environmental determinants of weight gain. This knowledge will be used to develop, evaluate, and promote sustainable, personalised lifestyle interventions aimed at preventing and managing obesity across the life course.

BETTER4U develops a preventive intervention methodology for weight-gain prevention, leveraging artificial intelligence (AI) techniques informed by insights from a pilot study conducted across 7 European countries. The effectiveness of the BETTER4U methodology will be tested in a clinical trial with technology-enabled, real-time monitoring. The project will also produce obesity-prevention guidelines grounded in a people-centred care approach to ensure relevance and adaptability across diverse populations.

The initiative's goal is to help people live longer, healthier lives. Harokopio University of Athens coordinates the project and involves 28 international partners. The University of Cagliari (UNICA), through its Metabolomics Laboratory led by Prof. Vassilios Fanos and Prof. Luigi Atzori, is primarily engaged in Work Packages (WPs) 4 and 7.

WP4 focuses on the study of the interplay between psychosocial, biological, and behavioural factors, as obesity and weight change are not solely driven by biological risk (e.g., genetics, metabolomics), but are also strongly modulated by psychosocial and contextual factors. Preliminary evidence, including transcript discussions, suggests that sex and gender play specific roles in pathways to weight gain and barriers to weight loss.

UNICA is already conducting a meta-analysis of data from targeted proton nuclear magnetic resonance (^{1}H-NMR) metabolomics, utilizing datasets generated using the Nightingale Health, Metaboneer, and Biocrates platforms from an extensive multi-cohort study. The aims are to (i) evaluate the association between comprehensive metabolic profiles and continuous body mass index (BMI) across diverse European cohorts and (ii) assess the influence of geographical clustering on these associations. This work aligns with WP4's broader objective to develop a refined lifestyle risk score that captures not only diet and physical activity but also upstream determinants.

WP7 is a multicentre clinical trial focused on implementation, monitoring, and evaluation.

WP7 will design and deliver a multicenter, open-label, parallel-group, randomized controlled trial across 7 sites. The arms are the control group, which is based on standard counselling by dietitians/nutritionists, and the intervention group, in which personalized counseling will be performed via the BETTER4U platform, integrating AI-based causal models and polygenic risk scores. The primary outcome will be to observe a change in BMI after 6 months. Whereas, the secondary outcome will be a weight loss ≥ 5% at 6 months and maintenance at 12 months; changes in biomarkers (metabolomics [UNICA team], lipidomics, adipokines, cardiometabolic and inflammatory markers, gut microbiota); blood pressure; lifestyle parameters; quality of life; psychosocial measures (anxiety, depression, motivation); and BMI changes in offspring/family members.

The sample size is approximately 1,022 participants (~146 per site), with balanced recruitment by sex.

In particular, the UNICA team will oversee biospecimen handling and perform NMR-based metabolomics analyses at 2 time points (baseline and 6-month follow-up). The team will also contribute to trial implementation, real-time monitoring of participants and processes, and evaluation of effectiveness and adherence/feasibility to inform iterative improvements to the programme.

ACKNOWLEDGMENTS

The BETTER4U project (https://better4u.eu/) has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement No. 101080117.

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ABS 20

SUBSTANCES THAT MAY INCREASE THE RISK OF PRECOCIOUS PUBERTY IN CHILDREN

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INTRODUCTION

In addition to well-known endocrine disruptors, various everyday substances may also contribute to the onset of early puberty in children. A significant correlation has been demonstrated between the use of sweeteners, commonly found in foods and beverages, and early puberty. The products studied are: aspartame, sucrose, glycyrrhizin, and added sugars.

DISCUSSION

According to a recent study, the use of these substances, especially in genetically predisposed individuals, increases the risk of early puberty. These data were recently presented at ENDO 2025, the Endocrine Society's annual meeting (held in San Francisco, CA, USA). Yang-Ching Chen, M.D., Ph.D., of Taipei Municipal Wan Fang Hospital and Taipei Medical University (Taipei, Taiwan), was one of the speakers who highlighted this risk. According to Chen, previous studies had already indicated that some sweeteners could react with hormones and intestinal microbiota, thereby influencing the immune system and the biological mechanisms of puberty. It has been shown that acesulfame K activates exceptional taste receptors and stimulates the production of hormones associated with puberty. Glycyrrhizin can alter the intestinal flora and modify the gene activity involved in sexual development. Prof. Chen is convinced that, by resorting to genetic screening and careful control of sweetener consumption, early puberty and its effects can be prevented or reduced. These studies have also highlighted significant differences between males and females in their response to different sweeteners. The use of sucralose is associated with an increased risk of precocious puberty in males. In females, the risk increases in the presence of glycyrrhizin, sucralose, and added sugars.

CONCLUSIONS

It is important to present this new knowledge because precocious puberty occurs very often after the age of 8 in females and 9 in males, and can cause adverse physical, behavioral and psychological effects: reduced stature, emotional distress, etc. This research and these results highlight that the problem exists, is serious, and poses a danger. Therefore, careful prevention will be necessary to promote healthier and more harmonious development of our children.

ABS 21

LOCKED EYE, PAINFUL HEAD: A RARE PEDIATRIC NEUROPATHY

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INTRODUCTION

Recurrent painful ophthalmoplegic neuropathy (RPON), previously known as ophthalmoplegic migraine, is classified in the third edition of the International Classification of Headache Disorders (ICHD-3) [1]. The diagnostic criteria for RPON require at least two attacks characterized by unilateral headache associated with ipsilateral paresis of one, two, or all three ocular motor nerves. Appropriate investigations must exclude orbital, parasellar, or posterior fossa lesions, and the condition should not be better accounted for by another ICHD-3 diagnosis [2]. Although brain MRI with gadolinium frequently reveals thickening of the affected nerve, this finding is not included among the formal diagnostic criteria, as imaging may be standard in a proportion of cases [2].

CASE REPORTS

Case 1

A 6-year-old female patient was first evaluated at our Center at 16 months of age for eyelid ptosis

associated with an abduction deficit of the right eye. Family history was positive for migraine. MRI revealed a hyperintense signal with thickening along the emergence and course of the right III cranial nerve. After treatment with corticosteroids, the patient showed clinical improvement and was discharged with a diagnosis of RPON. During follow-up, a recurrence with a similar clinical presentation was observed at 3 years of age. At 6 years, a third episode was recorded, less intense than previous ones; in the following months, slight residual exophoria persisted. MRI did not show further signal variations compared to prior examinations. Corticosteroid therapy led to the resolution of clinical symptoms.

Case 2

An 11-year-old male had a history of recurrent migraine, right eyelid ptosis and exotropia. Family history was negative. MRI revealed neuritis of the right III cranial nerve. The patient presented again at the age of 11 with headache accompanied by nausea, vomiting, and photophobia, together with ptosis and ophthalmoplegia of the right eye. Diagnosis of RPON was confirmed. The patient was treated with methylprednisolone, followed by maintenance therapy with indomethacin and flunarizine, with substantial clinical improvement. MRI of the cranial nerves showed enhancement in the right III nerve root entry zone.

CONCLUSIONS

RPON represents a diagnostic challenge, particularly in sporadic forms or during the first episode, when diagnostic uncertainty is high. Differential diagnosis with other neurological and ophthalmological conditions (such as tumors, malformations, or hematomas) is essential, as these can mimic ophthalmoplegic migraine. Follow-up is recommended to detect possible recurrences, as recurrent episodes are common in children [3].

In the cases described, careful clinical history, thorough, accurate neurological evaluation, and neuroradiological investigations with exclusion of other pathologies allowed us to identify this rare pathology.

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ABS 22

WHEN SINUSITIS TURNS DANGEROUS: A PEDIATRIC CASE OF POTT'S PUFFY TUMOR WITH INTRACRANIAL EXTENSION

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INTRODUCTION

Osteomyelitis of the frontal bone with the development of a subperiosteal abscess is the hallmark of Pott's puffy tumor (PPT), an uncommon but serious complication of frontal sinusitis. It still happens in pediatric populations, where the diploic venous system predisposes to intracranial dissemination, although it has been rare since the start of the antibiotic era. There is a considerable danger of long-term neurological consequences if diagnosis and treatment are postponed. The *Streptococcus anginosus* group, especially *Streptococcus intermedius*, has been strongly linked to deep-seated abscesses and involvement of the central nervous system in children [1, 2]. Early detection of clinical warning signs, such as frontal swelling, together with rapid imaging, broad-spectrum intravenous antibiotics, and surgical drainage, is crucial to improving outcomes [3].

CASE REPORT

We report the case of a previously healthy 10-year-old child who developed sinusitis. After 10 days of corticosteroids and antibiotics as part of his initial outpatient treatment, he came to San Francesco Hospital (Nuoro, Italy) with fever, headache, and a peculiar, tender swelling of the frontal region that looked "unicorn-like". Frontal sinusitis with posterior wall erosion, subcutaneous collection, and dural thickening was discovered by cranial CT with contrast. *Streptococcus intermedius* was isolated from the soft tissue abscess via needle aspiration. After being moved to a tertiary care facility, the patient's MRI revealed intracranial extension with no focal neurological issues. He required urgent functional endoscopic sinus surgery to remove purulent debris from his frontal and maxillary sinuses. Imaging after surgery revealed

increasing resolution. Ceftriaxone, vancomycin, and metronidazole were given intravenously for 3 weeks, as advised by infectious disease consultants. The child was afebrile, neurologically intact, and his frontal swelling had gone away at the time of discharge. An MRI and otolaryngological examinations were planned as follow-ups.

CONCLUSIONS

This case highlights the need for increased clinical vigilance in pediatric sinusitis characterized by frontal swelling, a key indicator of PPT. With a higher morbidity than other pathogens, *Streptococcus intermedius* is becoming more widely acknowledged as a major contributor to cerebral suppurative problems [1, 2]. Research highlights that the risk of meningitis, cerebral abscess, and dural venous sinus thrombosis is significantly increased when detection and treatment are delayed [3]. The successful outcome of our patient emphasizes the value of early imaging, aggressive surgical and medical therapy, and interdisciplinary teamwork. PPT is still a potentially fatal condition even in the age of antibiotics. If a child with sinusitis exhibits a noticeable swelling of the forehead, immediate evaluation and treatment are necessary to avoid serious consequences.

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ABS 23

BOTULISM IN CHILDREN AND ADULTS

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INTRODUCTION

The *Clostridiaceae* are Gram-positive bacilli, spore-forming and obligate anaerobes, motile due to the presence of peritrichous flagella. They are capable of producing ovoid or spherical endospores, which, depending on their position, can deform the bacterial body, allowing us to divide *Clostridia* into two groups. In the first group, the spore is located in a central or paracentral position; in the second group, the spore is located in a terminal or paraterminal position.

Clostridia are ubiquitous and therefore found in soil, in the sediments of fresh or salt water, and in the intestines of humans and animals. Numerous different forms of *Clostridia* have been identified, currently numbering more than 50.

MAIN DISEASES

Specific diseases attributable to a single species of *Clostridium* are caused by *C. botulinum*, *C. difficile*, and *C. tetani*.

However, most *Clostridia* can also cause severe wound infections that take the form of simple infection, anaerobic cellulitis, or gas gangrene. The latter is contracted through the implantation of spores present in the soil. In such cases, the following *Clostridia* can be isolated from the lesions: *C. perfringens*, *C. ramosum*, and *C. bifermentans*.

Infection with *C. botulinum* manifests as neuroparalysis caused by an extremely dangerous and lethal exotoxin. It is a protein substance that interferes with the release of acetylcholine, acting on the neuromuscular junctions and blocking neuronal transmission at the level of the cholinergic synapses and presynaptic terminals. Classic *C. botulinum* infection is foodborne, caused by the ingestion of preformed toxins in contaminated food. There is also an infantile infection where the toxin is produced *in vivo* in the intestine of the child, colonized by *C. botulinum*, due to the ingestion of spores. This infection can cause sudden death in individuals aged from 1 month to 1 year. The first symptoms of infection are nausea, dizziness, dry mouth, abdominal pain, diarrhea, or constipation. These symptoms usually appear 12-36 hours after ingestion of contaminated food. This prodromal phase may be followed by mydriasis, diplopia, dysphagia, dysarthria, ptosis, respiratory paralysis, and risk of death. *C. botulinum* toxin can be isolated

from serum, gastric juice, vomit, and feces, while the germ can be isolated from food, feces, and infected tissues. It is essential to know that *C. botulinum* toxins are the most potent toxins known to exist in nature.

DATA ON CASES AND MORTALITY

In Italy, 20-30 cases of *C. botulinum* infection are identified and recorded annually. Italy is currently one of the European countries with the highest number of reported cases.

INFANTILE BOTULISM

Infantile botulism is caused by the ingestion of *C. botulinum* spores that colonize the large intestine, producing toxins *in vivo*. Symptoms vary, from initial constipation to neuromuscular paralysis. The literature reports cases that have affected infants in the first month of life and children up to 12 months.

Infantile infection with *C. botulinum* is caused by the ingestion of spores and not by preformed toxin. Many hypothesize that the ingestion of honey could contain spores of *C. botulinum*.

At its onset, the infantile form must be suspected based on clinical signs, and the mere suspicion should prompt us to prescribe an urgent stool test.

It is certainly essential to be alarmed in the presence of neuromuscular paralysis that begins in the cranial nerves and progresses to the peripheral and respiratory muscles.

Signs of cranial nerve deficits are: weak crying, poor sucking, ptosis of the eyelids, paralysis of the extraocular muscles, reduction of the pharyngeal reflex, loss of oral secretions, reduction of muscle tone (flaccid child!), expressionless face, and respiratory failure.

Be careful not to confuse infant botulism with sepsis, spinal muscular atrophy, benign congenital hypotonia, or congenital muscular dystrophy.

A diagnostic confirmation is provided by the detection of toxins or *C. botulinum* in the stool.

THERAPY

Children must be hospitalized to receive ventilatory support therapy if necessary.

If the diagnosis is certain, specific treatment is carried out with human anti-botulinum immunoglobulin (BabyBIG®). This antitoxin is derived from human donors who have high titers of antitoxin A and/or B. The prescribed dose is 50 mg/kg IV, injected slowly. Antibiotics are not recommended because they can cause lysis of *C. botulinum* in the intestine, increasing the amount of toxin available.

BabyBIG® is available from the Infant Botulism Treatment and Prevention Program (IBTPP) of the California Department of Public Health (IBTPP website, or by calling the phone number 1-510-231-7600).

CONCLUSIONS

Italy is undoubtedly one of the European countries with the highest number of annual cases of botulism. Less severe cases may escape our attention.

Of particular concern is the infant form, which can affect children in the first months of life. In fact, the symptoms can be confused with other diseases (sepsis, spinal muscular atrophy, hypothyroidism, etc.), partly because botulism is rarely considered, and specific tests are not requested.

Finding the toxin or microorganisms of *C. botulinum* in the stool confirms the suspected diagnosis of infantile botulism.

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ABS 24

ANISAKIASIS

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INTRODUCTION

Helminths are obligate parasites, prevalent in nature, and they can infect all vertebrates. Helminths have common biological and morphological characteristics. They almost always have a smooth body, lacking motor and sensory organs. The exception is Nematodes, which have a digestive system. All helminths have a high reproductive ca-

pacity and are hermaphrodites, except for Cestodes and Schistosomes. Helminths enter humans in the form of eggs or larvae, through ingestion of infected meat or fish, unwashed vegetables, or polluted water. Transcutaneous penetration in the larval stage has rarely been described, and most likely also transplacental penetration (*S. japonicum*, *T. spiralis*). The helminths are eliminated in larval or egg form via the rectal, urinary, or cutaneous routes, and in the latter case exclusively in the larval stage, either spontaneously or through the action of blood-sucking insects.

ANISAKIS

Anisakis, which is a Nematode, grows in the marine environment with a cycle that involves marine mammals, including dolphins, seals, whales, and other similar species. The adult parasites nest in the stomach and intestines as definitive hosts and are expelled with the feces. The first intermediate hosts are small crustaceans that form the "krill" where the larva is in its first stage (L1). The krill is eaten by a second intermediate host, namely a fish or a mollusk, where the larvae pass from a second to a third larval stage (L2-L3). Suppose the infected mollusk or fish is eaten by a marine mammal, in the stomach or intestine of this new host: in that case, the Anisakis larva becomes an adult worm and completes the reproductive cycle. Humans become infected by eating undercooked, raw, or marinated mollusks or fish that contain L3 stage larvae. After ingestion, the larvae often die without causing any symptoms. In cases where Anisakis causes symptoms, the live larvae invade the gastric and intestinal mucosa, causing gastrointestinal Anisakiasis. The adult worm is pinkish-white and easily visible to the naked eye.

EPIDEMIOLOGY

Anisakis is a tiny parasite found especially in countries where raw fish is eaten and raw fish restaurants are typical. Infection is common among people who eat raw, pickled, or salted fish, particularly in regions such as Scandinavia, Japan, the Netherlands, and along the Pacific coast of South America. The parasite is also present in the Mediterranean area. Recent data show that this parasite is present in the central Adriatic Sea, with percentages ranging from 3.1% for sardines to 4.1% for anchovies. It spreads through the consumption of raw or marinated anchovies, crustaceans, mackerel, or swordfish (the latter two account for over 70% of infestations in fish). Fortunately, in Italy, recorded cases are sporadic, and from 1996 to 2011, there were only 54 confirmed cases of human infestation. Unfortunately, we do not have more recent data, and it is possible that any decrease in cases in our country is attributable to the introduction and application of European regulations on the freezing of fish, starting in 1987 with subsequent amendments in 1996. In fact, producers, restaurateurs, and fishmongers must freeze it at a temperature between -20° and -40°C. According to the *Istituto Superiore di Sanità* (Italian National Institute of Health), the larvae that infect humans die without becoming adult parasites. Therefore, humans do not eliminate eggs, feeding the parasite's cycle. For this reason, human-to-human transmission is not possible; infection occurs only through the ingestion of live larvae in intermediate hosts (fish, mollusks).

SYMPTOMS

The acute form, the gastric form, has the following symptoms: fever, vomiting, leukocytosis, diarrhea, nausea, cramps, and abdominal pain. Rarely, the larvae perforate the gastrointestinal mucosa, causing hemorrhages, and very infrequently, they insinuate themselves into the mesentery. They can also cause urticaria, conjunctivitis, and anaphylactic shock.

DIAGNOSIS AND DIFFERENTIAL DIAGNOSIS

Anisakis can be confused with ulcers, Crohn's disease, intestinal obstruction, and other gastro-intestinal conditions. A thorough medical history and careful clinical examination are essential, but above all, extensive experience is necessary. In doubtful cases, a prick test and immunoCAP can be performed to detect specific IgE immunoglobulins for Anisakis. In many cases, endoscopic examination is essential for a definitive and exact diagnosis. The endoscopic examination can also be curative, offering us the possibility of extracting the larvae present in the host. In severe cases, such as appendicitis, peritonitis, or intestinal obstruction, surgery is necessary. According to the Italian Ministry of Health, treatment with albendazole could provide an effective therapeutic result.

WHAT PREVENTION?

Above all, proper freezing of fish and shellfish is the safest and most effective way to prevent such infestation. Fish must be frozen at -18°C for at least 96 hours. Only after this treatment can raw fish, sushi, sashimi, carpaccio, smoked fish, or marinated fish be consumed. To ensure the larvae are dead, we must cook the fish to a temperature above 60°C for more than 10 minutes. There is a European regulation that requires the blast chilling of raw fish. It is Regulation (EC) No. 853/2004, adopted on April 29, 2004. This regulation, in application of protection against parasites such as Anisakis,

requires that raw fish be subjected to thermal blast chilling or kept at a temperature of at least -20°C for a minimum period of at least 24 hours before being served.

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ABS 25

MRI BEYOND CT IN PEDIATRIC SPINAL TRAUMA: A CASE REPORT

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INTRODUCTION

Pediatric vertebral fractures are usually the result of high-energy trauma, such as motor vehicle accidents or falls from significant heights. Vertebral fractures in children account for 1-3% of all pediatric fractures. The incidence of pediatric spinal injuries peaks in two age groups: children under 5 years and children older than 10 years [1]. Imaging plays a crucial role in the diagnostic workup of pediatric spinal trauma. Computed tomography (CT) and conventional radiography (X-ray) are widely used as primary imaging modalities. However, their use is limited by the risk of ionizing radiation exposure in a still-developing spine. Magnetic resonance imaging (MRI) represents a radiation-free alternative with high sensitivity for both osseous and soft tissue injuries.

CASE REPORT

We report on the case of a 12-year-old boy admitted to the Pediatric Emergency Department of San Francesco Hospital (Nuoro, Italy) after being immobilized on a spinal board and transported by ambulance because of a lumbar trauma sustained from a fall onto rocks after diving from a height of 3 meters. No loss of consciousness or vomiting was reported. Pain was rated as 7/10 on the VAS scale, localized to the lumbar region. On admission, spinal and pelvic X-rays and CT were performed, showing traumatic compressive collapse fractures at L1, L2, and L3. The following day, an MRI was performed and reported by the same radiologist who had read the CT scan. MRI findings included: inter-spongious bone marrow edema and compression fractures at T4 and T5; bone marrow edema without fracture from T9 to L3 with confirmed fractures of L1 and L2; a fracture of the fourth sacral vertebra; and importantly, no evidence of spinal cord or intradural nerve root injury. In the absence of spinal injuries, the neurosurgical management was conservative, consisting of the use of braces and donut cushions while sitting, along with a 1-month restriction on weight-bearing.

CONCLUSIONS

A retrospective review conducted by Franklin et al. [2] compared CT and MRI findings in pediatric thoracolumbar compression fractures. CT was shown to have high sensitivity in determining the presence or absence of vertebral fractures compared with MRI. Although variability exists between the two modalities in identifying the exact number of affected spinal levels, the final treatment and clinical outcome were not altered by the addition of MRI [2]. This case highlights how MRI has a higher sensitivity than CT for identifying spinal trauma lesions, especially when it comes to identifying edema, soft tissue, disc, and ligament injury [3]. Additionally, MRI is important for ruling out spinal cord and intradural damage, which has a direct impact on clinical confidence in conservative treatment choices [3]. To ensure a comprehensive evaluation, MRI should be taken into consideration in pediatric spinal trauma cases when subtle or further injuries are suspected or to confirm that there is no spinal cord involvement.

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ABS 26

SYSTEMIC LUPUS ERYTHEMATOSUS IN A PEDIATRIC PATIENT: A CASE WITH ONSET

COMPLICATED BY CONCOMITANT VISCERAL LEISHMANIASIS

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INTRODUCTION

Systemic lupus erythematosus (SLE) is a chronic, multisystem autoimmune disease that can present during childhood and adolescence with heterogeneous clinical manifestations, ranging from mild to severe forms. Diagnosis may be challenging, particularly in the presence of concomitant systemic infections.

CASE REPORT

A 15-year-old female, with a history of psoriasis and recent diagnosis of ADHD with mild autism spectrum disorder, presented with a 7-month history of asthenia, headache, diffuse arthralgia, and persistent low-grade fever. Clinical examination revealed a malar rash and arthritis of the small joints of the hands with "puffy fingers." Laboratory tests showed leukopenia, thrombocytopenia, hypocomplementemia, and ANA positivity (1:640), along with anti-histone++++, anti-dsDNA, anti-Scl70, and anti-ribosome antibodies. These findings allowed the diagnosis of SLE according to the 2019 EULAR/ACR criteria, with high disease activity based on the SLEDAI index. In addition, abdominal ultrasound revealed hepatosplenomegaly. Before initiating corticosteroid therapy, a bone marrow aspirate was performed, showing foamy histiocytes and hemophagocytosis figures suggestive of early macrophage activation syndrome (MAS), as well as an amastigote form consistent with visceral leishmaniasis (VL), later confirmed by positive serology. The patient was first treated with intravenous methylprednisolone pulses (1 mg/kg/day for 3 days) followed by oral prednisone; liposomal amphotericin B was later added, with hydroxychloroquine (5 mg/kg/day) administered throughout, achieving rapid clinical and laboratory improvement. As part of the diagnostic workup, we evaluated any potential organ involvement, with particular focus on renal function, where intermittent proteinuria was detected. Consequently, nephrology follow-up was started to assess the appropriateness of implementing immunosuppressive therapy (intravenous mycophenolate or cyclophosphamide).

CONCLUSIONS

This case highlights the diagnostic complexity of SLE in adolescence, especially in the presence of concomitant infections. Literature reports describe overlapping clinical and laboratory features between VL and SLE, making it difficult to discern whether the presentation is due to infection, autoimmune onset, or both. Our patient exhibited signs consistent with both conditions: leishmania seropositivity and microscopic evidence, alongside immunological and clinical criteria for SLE, complicated by early MAS. This clinical picture required a combined therapeutic approach targeting both infection and autoimmunity. Such cases demonstrate how infections and SLE may mimic and interact with each other, influencing diagnostic and therapeutic strategies. Early diagnosis, combined with multidisciplinary and individualized management, is essential to prevent organ damage and improve prognosis.

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ABS 27

A (NOT SO) UNEXPECTED PERINATAL STROKE: WHY EARLY RISK IDENTIFICATION MATTERS

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INTRODUCTION

Perinatal ischemic stroke is defined as a focal or multifocal cerebrovascular event occurring between the 20th gestational week and the 28th postnatal day. It affects approximately 1 in 1,100 live births, mainly males [1]. The pathogenesis is multifactorial: maternal risk factors include thrombophilia, age ≥ 35 years, smoking during pregnancy, and infertility history; neonatal risk factors comprise polycythemia, low protein C and S levels, vacuum-assisted delivery, cephalohematoma, Apgar score < 7 and *MTHFR* homozygosity [1].

AIM

To furnish critical insights for healthcare workers involved in the diagnosis and prevention of perinatal stroke.

METHODS

We describe a suggestive clinical case and conduct a systematic review of the literature on perinatal stroke linked to placental anomalies, focusing on risk factors and possible preventive strategies.

CASE REPORT

A male patient was born at term via vacuum-assisted delivery with a nuchal cord (Apgar scores 6-7-9). Maternal history: two previous miscarriages, antithrombotic prophylaxis with low-dose aspirin until 36 weeks due to homozygous *MTHFR* gene mutation, and smoking. At birth, the child was transferred to the Neonatal Intensive Care Unit for mild respiratory distress and abnormal neurological exam, and underwent therapeutic hypothermia. Clinical conditions progressively improved. Cerebral ultrasound on the 1st day showed nonspecific bilateral hyperechogenicity of the periventricular white matter, and on the 4th day, hyperechogenicity of the right insular region too. Brain MRI on the 7th day revealed a focal parenchymal hemorrhage (12 x 8 mm) in the right frontal lobe affecting both gray and white matter, along with a large area of hypoxic-ischemic injury in the same region. Additionally, a small hemorrhagic lesion (10 x 3 mm) was found near the left periventricular area. Placental histopathology showed features suggestive of maternal vascular malperfusion and concurrent fetal vascular malperfusion with focal chronic villitis.

DISCUSSION

Fetal vascular malperfusion occur more often in the placentas of children with perinatal stroke than in controls (87% of cases) [2]. The precise mechanism linking placental abnormalities and perinatal stroke is unclear because the placenta is commonly discarded before the clinical presentation of stroke. It is known that it definitely involves malperfusion, both fetal and placental, or inflammation, given the strong association with villitis of unknown etiology [3]. Moreover, inherited or acquired thrombophilias would compound the physiologically hypercoagulable state of pregnancy and present a second hit that would increase the risk of stroke [2].

CONCLUSIONS

The multifactorial etiology of perinatal arterial stroke makes the early identification of risk factors necessary. It is essential to study the placental function using postpartum histopathology and new techniques during pregnancy – such as omics and advanced imaging – that may provide useful biomarkers for targeted therapies and more effective prevention strategies [3]. In this way, there will be a considerable reduction in the risk of perinatal stroke both during the current pregnancy and in any subsequent ones.

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ABS 28

PERIVIABILITY AND THE CINDERELLA EFFECT

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Classical medical theory of the last decades has been dominated by arbitrary horizontal thresholds, which group patients into large categories defined by diagnostic labels, without taking into consideration the patient as an autonomous entity. Gradually, this obsolete approach is being abandoned as personalized medicine emerges, with new protocols focusing on patients rather than diseases. Limits

of neonatal viability are among the most critical topics in everyday neonatal clinical practice, since extreme prematurity is a leading cause of mortality and morbidity in neonatology. Yet, many official guidelines use a single horizontal threshold, suggesting rigid cut-off points that dictate whether resuscitation should be attempted or not. Even if in many countries the official guidelines include a decision-making stratification system, the lowest limit is set at 22, 23 or 24 weeks of gestation, which serves as a wall separating viability and non-viability. This threshold creates what has been described as a "Cinderella effect": before the clock strikes midnight, the newborn is considered non-viable and receives only comfort care, but the moment gestational age passes a predefined threshold, active and aggressive treatment is offered. Yet, medicine rarely functions in such stark black-and-white contrasts, but most of the time it operates as a great spectrum of a grey palette. This arbitrary limit involves a large scale of uncertainty, and does not reflect the physiological evolution of the pregnancy and of the fetus.

First and foremost, it is not possible to accurately calculate the exact gestational age of a fetus with an ultrasound. Secondly, defining such a specific threshold works as a self-fulfilling prophecy; if resuscitation is not provided, survival is obligatory to be zero. Consequently, in recent literature, the term "limits of viability" seems to be replaced by the more appropriate "periviability". Traditionally, viability was defined as occurring between 22 and 25 weeks of gestation, in which the infant has a reasonable possibility, but perhaps not always a high probability, of survival in extrauterine life. Periviability encompasses a broader window from 20 to 25 weeks of gestation, reflecting the range of gestation where the chance of survival for the newborn ranges from 0%, as the lungs are too immature to be mechanically supported, to over 50%. The importance of this reframing lies in its recognition that all the related parameters must be considered, such as the birth weight, sex, medical history, corticosteroid administration, etc. Periviability constitutes a gray zone that encompasses the limits of stillbirth and viability. It is not a specific point in time but a component of all the factors that can make a newborn viable or not, which aligns far more closely with the principles of personalized medicine, since every neonate is unique.

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ABS 29

OSTEOMYELITIS OF THE CLAVICLE IN A PEDIATRIC PATIENT: A CASE REPORT

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INTRODUCTION

Pediatric osteomyelitis (OM) is an acute, subacute or chronic bone infection with a prevalence of approximately 1 in 5,000 children, primarily affecting the metaphysis of long bones. Clavicular involvement is rare (1-3% of cases) [1]. Clavicular OM can be primary from hematogenous spread or secondary from direct inoculation of the infecting organism, likely from a direct trauma to the clavicle. Diagnosis can be delayed because symptoms are frequently nonspecific, and initial radiographs may be normal. Magnetic resonance imaging (MRI) is considered the gold standard. Early antibiotic treatment is essential to prevent complications [2, 3].

CASE REPORT

An 11-year-old girl came to the Emergency Room with a 5-day history of fever (39°C), rhinitis, and productive cough associated with pain and swelling over the right clavicle for 2 days. In medical history, a recent hospitalization for an assault by a group of young girls, resulting in injuries from kicks and scratches to the head, neck, arms and abdomen. No radiological investigations were performed at that time.

Examination revealed mild swelling, redness, and intense pain upon pressure on the sternal end of the right clavicle, which extended to the supraclavicular fossa. No other skeletal sites were involved. Laboratory tests showed normal blood count, CRP 1.42 mg/dL, and PCT 0.29 µg/L. Radiographs were unremarkable. Based on clinical suspicion,

empiric intravenous therapy with cefazolin and clindamycin was started. Blood cultures performed before antibiotic treatment, MRSA nasal swab, viral panel, serologies, and *Bartonella* testing were all negative. Ultrasound showed soft tissue edema and reactive lymph nodes. A contrast-enhanced CT was performed because of worsening pain and new-onset torticollis, and it showed thickening of the sternocleidomastoid muscle and early osteolysis of the clavicle with minimal joint effusion. MRI confirmed T2 marrow hyperintensity with post-contrast enhancement, consistent with OM without abscess formation on possible microfracture. Pain, initially refractory to NSAIDs and mild opioids, required continuous intravenous morphine for 4 days, then tapered with improvement. After 10 days, the patient was discharged with marked symptom reduction and restored limb function. Oral cephalexin has been continued for a 6-week course. Clinical and radiological follow-up is planned.

DISCUSSION AND CONCLUSION

OM of the clavicle is a rare entity with a broad differential diagnosis and high potential for complications if not diagnosed promptly and treated appropriately. It should always be considered in children with localized bone pain and swelling, even if laboratory markers are only mildly elevated or initial radiographs appear normal. In our case, the bone infection was due to a likely undiagnosed post-traumatic clavicular microfracture, a known OM's risk factor. Careful clinical and radiological follow-up is essential to ensure complete resolution and prevent long-term sequelae.

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ABS 30

A COMPLEX CLINICAL CASE ON THE MANAGEMENT OF LATE PRETERM INFANT

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INTRODUCTION

In recent years, late prematurity has been increasing, identifying late preterm infants (LPIs) as a population at increased risk of developing short- and long-term complications [1]. Indeed, LPIs are at increased risk of developing respiratory (respiratory distress syndrome, transient tachypnea of the newborn, pulmonary hypertension), infectious, metabolic, feeding difficulties and neurological complications, which require multidisciplinary management [1, 2]. Furthermore, they may develop comorbidity complications, such as cerebral or pulmonary hemorrhages [2, 3]. Therefore, late prematurity has a high morbidity [1, 2].

CASE REPORT

An LPI transferred to the Neonatal Intensive Care Unit for neurological abnormalities and respiratory failure. He underwent noninvasive ventilation and chest-abdomen X-ray, which revealed severe respiratory distress syndrome. Consequently, he was intubated for surfactant administration, but it was observed that blood was rising through the endotracheal tube, suggesting pulmonary hemorrhage. Therefore, he was administered an additional dose of vitamin K, and his ventilation was switched to high-frequency invasive ventilation. To rule out active cerebral hemorrhage, a brain ultrasound was performed, which was negative. Evaluation for hypothermia treatment met the criteria, and it was initiated. Due to increased oxygen requirements, the newborn underwent cardiac examination, which identified a patent ductus arteriosus and pulmonary hypertension. A chest X-ray revealed pneumothorax and pneumomediastinum, and blood tests showed increased inflammatory markers. During hypothermia, he received antibiotic therapy and nitric oxide therapy. Serial assessments of inflammatory markers and cardiorespiratory parameters indicated gradual improvement, allowing for the stop of nitric oxide therapy and antibiotic therapy, and successful extubation after a few days.

DISCUSSION

This case report, in accordance with the literature [1-3], is an example of the complexity management of LPIs arising from the potential development of multiple comorbidities and their subsequent complications, as demonstrated by the pulmonary hemorrhage that the newborn presented.

CONCLUSIONS

This clinical case highlights how managing LPIs can be complex, underscoring the importance of addressing late prematurity as well.

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ABS 31

TRANSIENT ELEVATION OF FECAL CALPROTECTIN IN AN INFANT: CASE REPORT

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INTRODUCTION

We present the case of a 15-month-old infant who developed symptoms of persistent diarrhea in association with the administration of the MMRV (measles, mumps, rubella, and varicella) vaccine and the ACWY meningococcal vaccine.

CASE REPORT

Five days after immunization, the child developed fever (up to 39°C), irritability, and diarrhea with watery to pasty stools, approximately 6 times a day. The fever resolved within 72 hours, but the diarrhea continued for over 4 weeks, despite dietary changes and the administration of a probiotic containing *Lactobacillus rhamnosus GG*. In the fifth week, a booster dose of the meningococcal B vaccine was administered, with a new worsening of the intestinal condition: bowel movements increased to at least 4 per day, with variations in stool consistency throughout the day (from hard to semi-liquid), in the absence of abdominal pain or colic. Bacterial, viral, parasitic, and fungal stool cultures were negative, as was the test for occult blood in the stool. However, fecal calprotectin (FC) was markedly elevated (> 1,000 µg/g) at a check-up 2 weeks later. Such high FC in infants (> 1,000 µg/g) is rare but not always indicative of chronic intestinal disease. Although nonspecific, this value indicated intestinal inflammation, consistent with what is known in the literature about mucosal immune activation and dysbiosis in children [1]. Targeted probiotic therapy was initiated with *Clostridium butyricum*, *Bifidobacterium longum*, and *Bifidobacterium longum ES1*, in lactose- and gluten-free formulations, selected for their synergistic potential in restoring the balance of the intestinal microbiota. After 10 days, FC was reduced to 52 µg/g and the occult blood test was negative.

DISCUSSION

The case is consistent with evidence suggesting that some vaccines, in predisposed individuals, may temporarily alter intestinal homeostasis through immunological mechanisms and interactions with the microbiota [2]. It also highlights the usefulness of specific probiotics and postbiotics in the management of dysbiosis [3], through the modulation of IgA and the microbiota-immunity axis [4].

CONCLUSIONS

This case highlights the importance of including post-vaccination dysbiosis in the differential diagnosis of persistent diarrhea in infants. The determination of FC can help define the clinical picture and guide treatment. Early treatment targeting the intestinal microbiota can promote rapid resolution of symptoms and prevent the establishment of persistent alterations in the immune-intestinal interface. It is essential to emphasize that this manifestation does not represent a contraindication to the implementation and continuation of the vaccination schedule.

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ABS 32

PD-L1 EXPRESSION IN THE HUMAN PLACENTA AT THE MATERNAL-FETAL INTERFACE PROTECTS FETAL DEVELOPMENT, ESTABLISHING MATERNAL IMMUNOTOLERANCE

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BACKGROUND
Programmed cell death ligand 1 (PD-L1) is a member of the B7 family of immune molecules, also known as immune checkpoints, involved in the regulation of immune responses. Cells expressing PD-L1 on their surface can bind programmed cell death 1 (PD1) expressed on the cell surface of T-lymphocytes, blocking their activity and escaping immune response. The discovery of this molecular pathway of immunotolerance was at the basis of the development of novel antibodies against PD-L1, which can re-activate immune cells, allowing a proper immune response with significant improvement in anticancer therapy.
AIM
Here, we report the most important physiological role of PD-L1 during gestation: its expression on the surface of placental villi, at the interface between fetal and maternal blood, where PD-L1 plays a key role in establishing maternal immunotolerance.
METHODS
Thirty consecutive human placentas, ranging from 13 to 38 weeks of gestation, were included in this study. Placenta samples were formalin-fixed, routinely processed and paraffin-embedded. Five micron-thick sections were immunostained with a commercial antibody against PD-L1.
RESULTS
Immunoreactivity for PD-L1 was found in all the placentas analyzed in this study. PD-L1 was mainly expressed on the surface of syncytiotrophoblasts, covering the surface of terminal villi, at the maternal-fetal interface.
CONCLUSIONS
Our findings show that PD-L1 is highly expressed at the maternal-fetal interface, on the cell surface of syncytiotrophoblasts, throughout the whole gestation. Since PD1 is highly expressed in maternal T-lymphocytes, PD-L1 appears as the key molecule involved in maternal immune tolerance. Further studies are needed to correlate changes in PD-L1 expression at the maternal-fetal interface with pregnancy disorders, including maternal-fetal rejection reactions and pre-eclampsia.

ABS 33

METABOLOMICS INSIGHTS AND RESEARCH GAPS IN RUGBY: FROM ACUTE RESPONSES TO RECOVERY

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BACKGROUND
Rugby is a high-intensity, intermittent sport demanding both aerobic and anaerobic capacities, exposing athletes to substantial metabolic stress. Metabolomics, the systematic study of low-molecular-weight metabolites in biological fluids, offers a promising approach to monitor physiological responses, optimize training, and potentially prevent injuries.
AIM
This review examines current evidence on metabolomics profiling in rugby, focusing on acute responses, recovery, and long-term adaptations.
METHODS
We conducted a narrative review of studies analyzing metabolomics changes in rugby players during and after matches, highlighting biomarkers associated with energy metabolism, fatigue, inflammation, and oxidative stress. Both targeted and untargeted metabolomic approaches, using blood, urine, and saliva, were considered. Emphasis was placed on studies with repeated measurements and practical implications for training and performance monitoring.
DISCUSSION
Acute match-play induces marked changes in metabolites linked to glycolysis, anaerobic metabolism, amino acid turnover, and oxidative stress, reflecting high mechanical and physiological loads. Evidence regarding chronic adaptations and recovery is limited, with few studies integrating metabolomics with external load metrics, nutrition, or individualized recovery strategies. The current literature identifies potential biomarkers for performance monitoring and early detection of over-

training or injury risk, but highlights significant research gaps, with no studies conducted in pediatric populations.

CONCLUSION

Metabolomics provides valuable insights into acute physiological responses in rugby, yet the application for long-term adaptation and recovery remains underexplored. Future research should focus on longitudinal multi-omics studies, integration with external load data, and personalized approaches to enhance performance and injury prevention.

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ABS 34

WEST NILE AND USUTU VIRUSES

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INTRODUCTION

West Nile virus (WNV) disease and Usutu virus disease are very similar viral diseases transmitted by mosquitoes of the genus Culex. Favorable climatic conditions promote the proliferation of mosquitoes, in this case, Culex. The spread of the disease is favored by the presence of natural reservoirs in the area.

Currently, epidemiological and veterinary surveillance conducted in Italy during the current vector season has confirmed the circulation of the WNV and the Usutu virus on Italian territory. The following regions are most affected: Emilia-Romagna, Veneto, Piedmont, Lombardy, Campania, Puglia, and Sardinia. These arboviruses belong to the Flaviviridae family and are particularly prevalent in Africa, Western Asia, Europe, the Americas, and Australia. The virus is primarily transmitted to humans through the bite of a mosquito. The disease can also be transmitted in other ways: organ transplants, blood transfusions, and from mother to fetus during pregnancy. This disease is not transmitted through contact between a healthy person and an infected person. The WNV can also infect other mammals, including horses, dogs, cats, rabbits, and others. The incubation period after the bite varies from 2 to 14 days or more. In general, the patient may be asymptomatic. Symptomatic cases may present with fever, nausea, headache, skin rashes, and swollen lymph nodes. These symptoms last for a maximum of 1-2 weeks and gradually subside. In children, however, symptoms include mild to high fever, headache, conjunctivitis, and muscle pain. Severe symptoms are infrequent, generally affecting 1 person in every 150/200 cases, and manifest as high fever, severe headache, tremors, muscle weakness, disorientation, visual disturbances, numbness and convulsions, up to paralysis and coma. In the most severe cases (1 in 1,000), the virus can cause severe encephalitis. We therefore distinguish between various clinical forms: asymptomatic infection (80% or more of cases), paucisymptomatic infection (approximately 20% of cases), and neuroinvasive infection (< 1% of cases, with a 10% mortality rate). In this last form, we can go from few symptoms to an acute and progressive neurological syndrome, characterized by encephalitis, meningitis, clear cerebrospinal fluid, polyradiculoneuritis (Guillain-Barré type), and acute flaccid paralysis.

DIAGNOSIS

The presentation of cases with fever of unclear origin should validate the clinical suspicion of a WNV-type arbovirus infection. Still, it should also prompt us to carefully consider other arbovirus infections (Dengue, Chikungunya, Zika virus, Usutu virus) and other diseases transmitted by mosquitoes and other vectors (malaria, tick-borne viral encephalitis, Lyme disease, Toscana virus). The diagnosis of WNV fever is confirmed by a laboratory test (ELISA or immunofluorescence) per-

formed on serum or cerebrospinal fluid to detect IgM antibodies. These antibodies can persist and be detected for long periods, up to more than a year. Samples taken 8 days after the onset of symptoms may be negative; therefore, it is advisable to repeat the sample after a few weeks before ruling out the disease. The diagnosis can also be made through PCR or viral culture of serum or cerebrospinal fluid.

PREVENTION

As there is currently no vaccine available for prevention, it is advisable to reduce exposure to mosquito bites. To this end, it is advisable to use suitable repellents and wear long pants and long-sleeved shirts when outdoors, especially at dusk and dawn. Doors and windows should be fitted with mosquito nets. Also, avoid keeping containers and saucers containing stagnant water.

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ABS 35

SPORTOMICS: FOCUS ON BASKETBALL AND VOLLEYBALL

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Sportomics, a branch of metabolomics applied to the study of physical activity, is now an innovative tool for understanding how sport influences athletes' metabolic profiles. Several studies have shown that metabolites reflect not only the intensity and duration of activity, but also individual factors such as gender, role on the field, training status, and even sleep quality.

In basketball, studies by Khoramipour and colleagues have shown how metabolic profiles can vary over the four quarters of the game: in the first and third quarters, which are more characterized by explosive movements, the use of anaerobic energy systems prevails, while in the second and fourth quarters, due to fatigue, there is a greater reliance on aerobic and lipid metabolism [1]. Their further work compared the metabolic profiles and movement patterns between backcourt and frontcourt players, showing that the former mainly use aerobic metabolic pathways, while the latter show markers of greater involvement of anabolic and static pathways [2].

In the field of basketball, three studies conducted in Italy by the University of Florence have added an important piece to the puzzle:

- in the first study, they showed that female basketball players have higher levels of antioxidant capacity and lower levels of cortisol than sedentary controls [3];
- in the second study, specific gender-related differences in the use of lipids and amino acids were demonstrated, highlighting the importance of personalizing training programs [4];
- in the last study, they confirmed, through proteomic and salivary metabolomic analyses, the presence of common characteristics in exercise adaptation between males and females, but also gender-related differences, especially in the expression of inflammatory proteins and the use of amino acids [5].

Relevant data also emerge in volleyball. Akazawa and colleagues have shown that sleep quality influences urea cycle and Krebs cycle metabolites, as well as reaction times during intense exercise [6]; Zhou and colleagues showed that 2 weeks of strength-endurance training in adolescent volleyball players induces changes in energy, lipid, and amino acid metabolites, associated with an increase in cortisol and oxidative stress [7]. Finally, Oliveira et al.'s study of Brazilian professional volleyball players revealed a significant reduction in essential amino acids after playing a match, suggesting a possible link with glycogen stores and diet, and highlighting the need for nutritional compensation strategies [8].

Taken together, these studies demonstrate how sportomics allows us to capture the subtle metabolic variations induced by physical activity, providing information that cannot be obtained with traditional biochemical markers. It is therefore an approach that can guide the development of personalized training and nutrition programs, helpful for optimizing performance, reducing the risk of injury, and improving the health of athletes.

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ABS 36

EXPLORING UNCERTAINTY: LONG-TERM OUTCOMES AND NEUROPLASTICITY AFTER PRENATAL BRAIN HEMORRHAGE

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BACKGROUND

Prenatal intracranial hemorrhage is a rare event with highly variable neurological outcomes. While severe hemorrhages often predict poor prognosis, the correlation between lesion severity and long-term neurodevelopment remains unpredictable, highlighting the role of neuronal plasticity. We present a case of prenatal parenchymal hemorrhage with an apparently favorable short-term outcome, accompanied by a literature review on long-term outcomes in neonates.

CASE REPORT

A male neonate, born at 35^{+5} weeks via urgent cesarean section due to detection of an intracranial mass on the last prenatal ultrasound, was diagnosed with a right temporo-parietal parenchymal hemorrhage and associated subdural collections. Initial complications included moderate respiratory distress requiring noninvasive support, anemia requiring 2 transfusions, and transient sepsis. Brain MRI confirmed the hemorrhagic lesions without vascular malformations, and neurosurgical consultation excluded the need for intervention. EEG showed minor anomalies, and neurological examination at discharge was within normal limits.

DISCUSSION

Despite the significant hemorrhagic burden, the patient demonstrated no severe neurological deficits in the early postnatal period. Current literature shows that outcomes after prenatal parenchymal hemorrhage are heterogeneous and often unpredictable, ranging from severe impairment to near-normal development. This case raises questions about the contribution of neuroplasticity and the need for extended follow-up to explore long-term neurodevelopmental outcomes.

CONCLUSION

Prenatal parenchymal hemorrhage is associated with a wide spectrum of potential outcomes. Even in cases with extensive lesions, early neurodevelopment may appear relatively preserved, but the long-term neurological trajectory remains uncertain. Further research is needed to clarify the factors influencing neurodevelopment and the potential role of neural plasticity.

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ABS 37

THE SYNDROME WITH A THOUSAND FACES: A PEDIATRIC PRESENTATION OF SJÖGREN'S SYNDROME

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CASE REPORT

A 13-year-old female patient was referred to our clinic for the onset of acute neurological symptoms, including headache, tremors, sensory disturbances in the lower limbs, and gait difficulties. Her past medical history was notable for type 1 diabetes mellitus (diagnosed at age 9) and a history of recurrent parotitis since the age of 3, intermittently treated with NSAIDs and antibiotics. The patient also reported episodic musculoskeletal pain, xerostomia, and xerophthalmia (possibly following therapy). During hospitalization, the diagnostic workup included brain and spinal MRI – incidentally revealing a type I Arnold-Chiari malformation –, lumbar puncture, electromyography, nerve conduction studies, somatosensory evoked potentials, and blood tests with an extensive autoimmune panel, including anti-SSA and anti-SSB antibodies, as well as testing for autoantibodies in the cerebrospinal fluid, all of which were negative. Given the initial suspicion of Guillain-Barré syndrome, intravenous immunoglobulin therapy was administered, leading to gradual clinical improvement but without complete resolution of symptoms. Considering the patient's history of recurrent parotitis and suspecting Sjögren's syndrome, further evaluations were performed. Ophthalmologic examination was within normal limits (though a false negative due to conjunctivitis could not be excluded). A labial salivary gland biopsy revealed a focal lymphocytic sialadenitis with a focus score > 1, confirming the diagnosis of Sjögren's syndrome. Following diagnostic confirmation, off-label immunosuppressive treatment was initiated with periodic intravenous immunoglobulin infusions and mycophenolate mofetil, resulting in improvement of symptoms.

DISCUSSION

Primary Sjögren's syndrome is a systemic autoimmune disease characterized by exocrine gland dysfunction and multi-organ involvement. Central and peripheral nervous system involvement in Sjögren's syndrome is rarely reported but can be severe and heterogeneous [1]. Sensory neuropathy is described in approximately 15-20% of neuropathies observed in this condition. It is often the presenting feature, with negative serology, and may precede the diagnosis of Sjögren's syndrome by several years. For these reasons, a high index of suspicion is required, particularly in adolescent female patients with painful or ataxic sensory neuropathy or those with trigeminal sensory and autonomic involvement [2].

CONCLUSIONS

The frequency of neurological manifestations as an initial presentation of Sjögren's syndrome, combined with the high rate of negative biological markers, may hinder prompt diagnosis. It is crucial to maintain a high degree of clinical suspicion in similar cases to initiate timely and targeted therapy. The coadministration of intravenous immunoglobulin and mycophenolate mofetil was demonstrated to be beneficial and safe in a case of sensory neuronopathy associated with Sjögren's syndrome refractory to conventional immunosuppressive therapy [2].

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ABS 38

NEONATAL TRANSPORT FROM THE ISLANDS: A COMPARISON BETWEEN SARDINIA AND THE AZORES

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Neonatal transport represents a crucial element in ensuring the survival and proper management of critically ill newborns, particularly in island regions where access to highly specialized centers is limited. Sardinia constitutes a peculiar case in the Italian context: its insular condition, progressive depopulation, reduction of birth centers, and the absence of advanced neonatal surgical services make it necessary to rely on extraregional transfers systematically. These are carried out by the Italian Air Force, specifically the 31st Wing, through life-

saving flights that are indispensable yet burdensome in terms of both organization and cost.
This work analyzes neonatal air transfers from the University Hospital of Monserrato (Cagliari, Sardinia, Italy) to mainland referral centers between 2014 and 2024. Despite a marked decline in births, the number of transfers remained stable, with a proportional increase in the share of critical newborns. The main indications for transfer concerned complex neonatal conditions and surgical pathologies that cannot be managed regionally. The analysis of territorial origins highlighted inequalities related to difficulties in pregnancy monitoring, particularly in inland areas.
A comparative study was carried out with data from the Azores (Portugal), which documented 30 neonatal transfers during the same timeframe, representing roughly half the volume observed in our center. Distinctive features of the Azores that emerged from the comparison include the extensive use of inter-island transfers by helicopter (43% of cases), the practice of *in utero* transfer, and the deployment of specialized surgical teams on site, approaches that reduce both costs and clinical risks associated with urgent neonatal transport.
The findings confirm the strategic role of the Italian Air Force in Sardinia, while at the same time highlighting the need to reorganize the regional system, considering the implementation of alternative models already adopted in other insular contexts.

ABS 39

ECHOES IN THE CSF: METABOLOMICS MARKERS OF ACUTE IMMUNE NEUROPATHIES

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BACKGROUND
Guillain-Barré syndrome (GBS) comprises acute immune-mediated neuropathies with rapidly progressive weakness, areflexia, and albuminocytologic dissociation; major subtypes include acute inflammatory demyelinating polyneuropathy (AIDP), acute motor axonal neuropathy (AMAN), acute motor and sensory axonal neuropathy, and Miller Fisher syndrome (MFS), with geography-dependent distributions [1].
OBJECTIVE
To synthesize human metabolomics evidence on GBS for early diagnosis, subtype discrimination, and prognostic stratification.
METHODS
A PubMed search ("Guillain-Barré syndrome" AND "metabolomics") identified 2 human studies, using cerebrospinal fluid (CSF) or plasma and nuclear magnetic resonance (NMR), gas chromatography – mass spectrometry (GC-MS), or liquid chromatography – mass spectrometry (LC-MS) platforms. We extracted matrices, analytical methods, discriminatory metabolites, and clinical correlations.
RESULTS
In CSF, a targeted metabolite panel differentiated GBS from controls and separated AIDP, AMAN, and MFS variants; lower acetate, relevant to myelin lipid synthesis, and variant-specific lipid shifts correlated with disability. MFS showed higher CSF glucose; AMAN was enriched for sphingomyelins and fatty-acid esters of hydroxy fatty acids, whereas AIDP exhibited higher lysophosphatidylcholines (LysoPCs). In plasma, broad disturbances spanned lipids and amino acids in GBS versus multiple sclerosis and healthy controls, including lower phosphatidylcholines, LysoPCs, sphingomyelins, carnitines, aspartate, creatinine, serotonin, and taurine, with higher isoleucine, glucose, adenine, and hypoxanthine; lower lipid levels associated with greater severity and worse prognosis. Collectively, metabolomics fingerprints map onto myelin-lipid biology and electrophysiological subtypes [2, 3].
CONCLUSIONS
Despite small, heterogeneous cohorts, metabolomics shows promise for early triage, subtype-aware risk assessment, and treatment monitoring in GBS. Priorities include standardized pre-analytics, longitudinal sampling from onset through recovery, and integration with autoantibodies, electrophysiology, and imaging to build clinically deployable, minimally invasive biomarker panels.
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ABS 40

MATERNAL LIFESTYLES AND ENDOCRINE-DISRUPTING CHEMICALS: SARDINIAN COHORT IN THE EUROPEAN LIFE-MILCH PROJECT

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This study describes the Sardinian cohort enrolled in the European Life-MILCH project, which aims to evaluate maternal and infant exposure to endocrine-disrupting chemicals (EDCs) and to promote preventive public health strategies. A total of 36 healthy pregnant women between 36 and 41 weeks of gestation were recruited at the University Hospital of Monserrato (Cagliari, Sardinia, Italy) between June and December 2024. Inclusion criteria included willingness to breastfeed and absence of significant maternal or fetal pathology.

Participants completed detailed questionnaires on lifestyle, diet, environmental exposures, and personal care product use. Biological samples were collected, including maternal blood and urine at enrollment, breast milk, and infant urine at 1 month postpartum. The mean maternal age was 35 years; most participants had a normal pre-pregnancy weight, were nulliparous, and were of Italian nationality. Regarding occupational status, 28% were homemakers and 19% employed, with an average working time of 6.5 hours per day.

Clinical and behavioral data showed that 28% had pre-existing gynecological conditions, 67% had previously used hormonal contraception, and 50% engaged in outdoor physical activity. Daily use of personal care products, particularly toothpaste and facial creams, and frequent contact with plastic materials in food handling and clothing suggest potential EDC exposure sources. At the postpartum follow-up, a reduction in smoking exposure and some changes in food storage practices were observed, while physical activity remained stable.

These findings suggest that targeted educational campaigns can positively influence maternal behavior, thereby reducing early exposure to chemical contaminants. Future studies on larger Sardinian cohorts are needed to confirm these data and to develop targeted preventive interventions in this population.

ACKNOWLEDGEMENTS

The Life-MILCH (Mother and Infant dyads: Lowering the impact of endocrine disrupting Chemicals in milk for a Healthy Life) project (https://lifemilch.eu) has received funding from the European Union's LIFE Environment and Resource Efficiency program under grant agreement No. LIFE18 ENV/IT/000460.

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ABS 41

PERINATAL INFLAMMATION: A CHALLENGE FOR FETAL NEURODEVELOPMENT

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The "developmental origins of health and disease" hypothesis, also referred to as "perinatal programming," assumes that exposures and events occurring during prenatal and early postnatal life can have enduring influences on health and developmental trajectories across human life. Consequently, the response of a developing organism to specific challenges within critical windows of vulnerability, such as gestation, may qualitatively and/or quantitatively modify developmental processes, leading to stable phenotypic alterations. Among such challenges, perinatal inflammation (PI) has attracted increasing attention for its capacity to disrupt neuronal maturation and to induce hyperactivation of the immature innate immune system [1]. These mechanisms are thought to contribute to chronic maternal immune activation and to compromise the fetus's capacity to cope with subsequent insults. Immune-mediated perturbations of this kind are associated with an elevated risk of neurodevelopmental conditions,

including schizophrenia, autism spectrum disorder, anxiety, and depression. Experimental studies employing pro-inflammatory stimuli – such as lipopolysaccharide, polyinosinic:polycytidylic acid, and interleukin-6 – have consistently demonstrated alterations in fetal brain development, synaptic plasticity, and long-term behavioral phenotypes [2]. Specifically, PI may establish a chronic, low-intensity neuroinflammatory status marked by prolonged microglial activation and increased production of pro-inflammatory mediators [3].

Collectively, these findings reinforce the proposed link between early-life immune activation and atypical neurodevelopment. Nevertheless, translational evidence in humans remains limited. Future research is required to establish reliable biomarkers of PI, clarify long-term outcomes, and explore targeted preventive strategies.

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ABS 42

ASSOCIATION OF ENDOTHELIN-1 AND CYTOKINE LEVELS WITH HYPERFIBRINOLYSIS OF PREGNANT WOMEN WITH PREECLAMPSIA AND THE DEFICITS IN FIBRINOGEN AND EXTRINSIC PATHWAY FACTORS OF THEIR NEONATES

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INTRODUCTION

Rotational thromboelastometry (ROTEM) in pregnant women with preeclampsia (PE) reveals a hypercoagulable profile, while neonates of PE-women present a hypocoagulable profile compared to neonates of healthy controls.

METHODS

We compared cytokine and endothelin-1 (ET1) levels between 31 PE-women and 45 healthy pregnant controls and their neonates (34 and 47, respectively). We assessed their association with hyperfibrinolysis (clot-formation-time [CFT]-APTEM < CFT-EXTEM, maximum-clot-firmness [MCF]-APTEM > MCF-EXTEM, maximum-lysis [ML]-EXTEM > 15% with normal ML-APTEM), deficit in fibrinogen (MCF-FIBTEM < 13 mm), deficit in intrinsic (MCF-INTEM < 48 mm) and extrinsic (MCF-EXTEM < 45 mm) pathway factors. Neonatal samples were drawn within the 1st hour of life.

RESULTS

No differences were found in maternal and neonatal baseline characteristics.

PE-women presented higher interleukin-2 (IL2), IL6 and ET1 levels, while their neonates presented higher IL2 and tumor necrosis factor α levels.

PE-women presented higher rates of hyperfibrinolysis compared to healthy pregnant controls ($p = 0.002$). ET1 levels were higher in PE-women with hyperfibrinolysis compared to PE-women without hyperfibrinolysis ($p < 0.001$).

PE-neonates presented higher rates of deficit in fibrinogen and extrinsic pathway factors compared to controls ($p = 0.02$ and $p = 0.01$, respectively). ET1 levels were higher in PE-neonates with fibrinogen deficit compared to PE-neonates without fibrinogen deficit ($p = 0.03$). IL2 levels were higher in PE-neonates with extrinsic pathway factors deficit compared to PE-neonates without extrinsic pathway factors deficit ($p = 0.02$).

CONCLUSIONS

Hyperfibrinolysis of PE-women is associated with higher levels of ET1. During the 1st hour of life, the ROTEM hypocoagulable profile of PE-neonates is associated with ET1 and IL2, since ET1 and IL2 were elevated in PE-neonates with deficits in fibrinogen and extrinsic pathway factors, respectively.

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ABS 43

AMONG NEONATES BORN TO WOMEN WITH PREECLAMPSIA, BRONCHOPULMONARY DYSPLASIA IS ASSOCIATED WITH INTERLEUKIN 2 LEVELS AT BIRTH, WHILE NEONATAL THROMBOCYTOPENIA IS ASSOCIATED WITH MATERNAL INTERLEUKIN 2 AND ENDOTHELIN 1

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INTRODUCTION

Preeclampsia's (PE) endothelial dysfunction is characterized by endothelin-1 (ET1) overexpression and is mediated by interleukin-2 (IL2), IL6, IL8 and tumor necrosis factor α (TNFα). Fetuses of PE-mothers may experience storms of placenta-derived mediators, leading to analogous neonatal endothelial dysfunction.

METHODS

We compared cytokine and ET1 levels between 31 PE-women and 45 healthy pregnant controls, as well as their neonates (34 and 47, respectively). We assessed their association with neonatal thrombocytopenia at birth (platelets < 150 x 10^9/L), and bronchopulmonary dysplasia (BPD). Neonatal samples were drawn within the 1st hour of life.

RESULTS

No differences were found in maternal and neonatal baseline characteristics. PE-women presented higher IL2, IL6 and ET1 levels, while their neonates presented higher IL2 and TNFα levels. Among PE-neonates, neonatal thrombocytopenia at birth was associated with maternal IL2 (OR 1.75, p = 0.04) and ET1 (OR 7.38, p = 0.04). Maternal IL2 had an AUC value of 0.75 (sensitivity 67%, specificity 82%, cut-off point 4.81 pg/mL). Maternal ET1 had an AUC value of 0.75 (sensitivity 100%, specificity 54%, cut-off point 0.74 pg/mL). Among PE-neonates, BPD was associated with neonatal IL2 at birth (OR 1.24, p = 0.04), with an AUC value of 0.72 (sensitivity 86%, specificity 54%, cut-off point 4.81 pg/mL).

CONCLUSIONS

PE-neonates presented an altered inflammatory response during the 1st hour of life. Maternal ET1 and maternal IL2 presented a significant sensitivity and specificity, respectively, regarding the prediction of thrombocytopenia at birth among PE-neonates. Maternal PE may cause a neonatal IL2 increase within the 1st hour of life, leading probably to altered vascular remodeling and, thus, to a higher probability of BPD.

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ABS 44

PEDIATRIC ARTERIAL TORTUOSITY SYNDROME: CLINICAL AND GENETIC INSIGHTS FROM KOSOVO

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INTRODUCTION

Arterial tortuosity syndrome (ATS) is a rare connective tissue disorder caused by mutations in the *SLC2A10* gene, leading to vascular elongation, tortuosity, and stenosis. Clinical features often include hyperextensible skin, joint hypermobility, and characteristic facial dysmorphism. We present 2 pediatric cases from Kosovo, representing the first reported instances of ATS in the Albanian population.

CASES REPORT

Case 1

A 3-year-old boy presented with an incidental heart murmur and poor weight gain. Physical examination

revealed facial dysmorphism, scoliosis, spina bifida occulta, pectus excavatum, hypermobility, and an inguinal hernia. Echocardiography showed a bicuspid aortic valve, subaortic ventricular septal defect, and tortuous pulmonary arteries. CT angiography confirmed aortic coarctation and vascular tortuosity. Genetic testing revealed a homozygous pathogenic variant in *SLC2A10* (NM_030777.4: c.685C>T, p.R229*), annotated per Human Genome Variation Society (HGVS) guidelines. Surgical correction was successfully performed in Italy.

Case 2

A 6-year-old boy presented with poor weight gain and similar dysmorphic features. Examination revealed scoliosis, pectus excavatum, hypermobility, and a post-surgical scar from inguinal hernia repair. Imaging showed tortuous aortic and pulmonary arteries without intracardiac defects. Genetic testing confirmed the same homozygous *SLC2A10* mutation (NM_030777.4: c.685C>T, p.R229*).

DISCUSSION

ATS is characterized by systemic vascular involvement and connective tissue abnormalities. Cardiovascular complications may include aneurysms, stenosis, and ventricular hypertrophy. Diagnosis relies on imaging and genetic testing. These cases underscore the importance of considering ATS in children with unexplained vascular anomalies and connective tissue signs.

ATS has a prevalence of < 1 in 1,000,000, and fewer than 50 cases have been described, mainly in pediatric populations. Commonly affected vessels include the aorta, coronary, and pulmonary arteries, though mesenteric and peripheral arteries may also be involved. Intracranial aneurysms have been reported, typically alongside connective tissue features such as stretchable skin, joint laxity, and facial dysmorphism. Clinical manifestations vary depending on the affected arteries. Onset usually occurs in infancy or early childhood. Cardiovascular anomalies may lead to ventricular hypertension, respiratory symptoms, and cardiac failure. Patients are prone to aneurysm formation, dissection, and ischemic events. Typical features include a long face, hypertelorism, down-slanting palpebral fissures, beaked nose, sagging cheeks, high palate, and micrognathia. Other findings include soft, hyperextensible skin, cutis laxa, hernias, skeletal abnormalities, congenital contractures, keratoconus, and hypotonia. Although our patients had no history of consanguinity, both exhibited hallmark features of ATS. Imaging revealed elongated aortic arches and tortuosity of subclavian and mesenteric arteries. These findings, combined with genetic confirmation, support the diagnosis. CT angiography or cardiac catheterization is recommended when unexplained high ventricular pressure or peripheral artery stenosis is present.

ATS is caused by loss-of-function mutations in *SLC2A10* (20q13.12), which encodes the glucose/dehydroascorbic acid transporter GLUT10. At least 23 mutations have been identified in affected individuals.

CONCLUSIONS

ATS should be considered in pediatric patients with vascular tortuosity and connective tissue features. Genetic confirmation and imaging are essential for diagnosis and management. These cases contribute novel data from Kosovo to the global literature. Our patients demonstrated typical ATS features, including complex cardiovascular disease, inguinal hernias, and dysmorphic facial traits. Prognosis can improve with early intervention. One patient underwent successful cardiac surgery in Italy. Follow-up with echocardiography and CT/MRI every 3 years is recommended due to aneurysm risk. Routine ophthalmologic and orthopedic evaluations are also advised. ATS may be mistaken for other congenital heart or connective tissue disorders. Once suspected, genetic testing provides a definitive diagnosis. Proper identification is essential for monitoring and managing these complex patients. Although prognosis is variable, reported causes of death include pulmonary infection, myocarditis, and organ infarction.

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ABS 45

IRON-DEFICIENCY ANAEMIA IN PEDIATRIC CONGENITAL HEART DISEASE: A SIX-MONTH CLINICAL EVALUATION

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BACKGROUND

Iron-deficiency anemia (IDA) is the most common hematological disorder in infancy and childhood. Children with congenital heart disease (CHD), particularly those with cyanotic defects, are at increased risk due to chronic hypoxia, elevated metabolic demands, and nutritional challenges.

OBJECTIVE

To evaluate the prevalence and hematological patterns of IDA in children with CHD and assess the effectiveness of iron supplementation over 6 months.

METHODS

A retrospective study was conducted on 114 pediatric patients with CHD admitted to the Children's Clinic in Prishtina (Republic of Kosovo) in 2024. Hematological parameters, including hemoglobin, hematocrit, serum iron, and RBC count, were measured at admission and after 3 and 6 months of iron therapy.

RESULTS

At admission, 98 children had IDA. After 3 months of treatment, 103 showed improved hemoglobin and hematocrit levels, though 41 still had low serum iron. At 6 months, 76 children maintained normal hematological profiles, while 30 continued to show iron deficiency. Eight children required RBC transfusions, and 4 underwent hemodilution due to polycythemia.

CONCLUSION

IDA is highly prevalent in children with CHD. While most respond to iron supplementation, a subset requires more intensive interventions. Regular hematological monitoring and individualized treatment are essential for optimal outcomes.

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ABS 46

SUCCESSFUL MANAGEMENT OF PEDIATRIC PITYRIASIS RUBRA PILARIS WITH ETANERCEPT: A CASE REPORT FROM PRISHTINA

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Pityriasis rubra pilaris (PRP) is a rare, chronic, papulosquamous skin disorder of unknown etiology, particularly uncommon in children. We report the case of a 7-year-old male patient admitted to the Pediatric Clinic in Prishtina (Republic of Kosovo) with widespread erythema, scaling, and systemic symptoms. Initial management with topical corticosteroids and emollients yielded minimal improvement. Following histopathological confirmation of PRP, the patient was treated with etanercept, a tumor necrosis factor α inhibitor. Significant clinical improvement was observed within 4 weeks, with near-complete resolution by the 8th week. This case highlights the potential efficacy and safety of etanercept in treating pediatric PRP and underscores the importance of early diagnosis and biologic intervention in refractory cases. Etanercept may represent a valuable therapeutic option for pediatric PRP, particularly in cases unresponsive to conventional treatment. Its favorable safety profile and rapid clinical response make it a compelling choice for managing this rare and debilitating condition. Further studies and long-term follow-up are needed to establish standardized treatment protocols and assess sustained efficacy.

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TRUNCUS ARTERIOSUS: MANAGEMENT OF AN UNEXPECTED HEART

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INTRODUCTION

Truncus arteriosus (TA) is a rare congenital heart disease characterized by a ventricular septal defect (VSD), a single truncal valve, and a common ventricular outflow tract. Systemic and pulmonary venous blood mix at the VSD, and desaturated blood is ejected into the outflow tract. Definitive surgical correction may be performed as a single-stage repair within the first month of life. Pulmonary artery mobilization from the truncus to the right ventricle, conduit-based right ventricular outflow tract (RVOT) reconstruction, and VSD patch closure are performed within the same operation. Aortic arch abnormalities and the truncal valve are also fixed [1]. Repair of TA is associated with excellent survival beyond the first year after repair. However, reoperation rates remain high due to the use of a conduit for the RVOT [2].

CASE REPORT

In this study, we report a case of a newborn with a diagnosis of TA who underwent surgical repair. He was born at term by spontaneous vaginal delivery with an Apgar score of 9-10, after a physiological pregnancy. On the first day of life, the baby was transferred to the Neonatal Pathology Unit due to tachypnea and dysmorphic features (anteriorly placed anus, hypospadias, sacral dimple and calyceal-pelvic dilation). His general condition was poor: he had pale-gray skin, oxygen saturation (SpO_2) of 95% in ambient air, polypnea (respiratory rate of 100 breaths per minute), a 2/6 systolic murmur with a heart rate of 140 bpm and palpable femoral pulses bilaterally. Neurological examination revealed an incomplete Moro reflex and neck hypotonia. Echocardiographic evaluation revealed a single ventricular outflow tract with a normally functioning tricuspid truncal valve and a large outlet-type VSD due to malalignment. Pulmonary arteries originated from the postero-lateral portion of the common trunk with mild stenosis in the right pulmonary artery.

The baby was transferred to a tertiary-level Pediatric Cardiology and Cardiac Surgery Center with a diagnosis of TA type II. He underwent surgical repair, including VSD closure with a patch, plasty of the hypoplastic right pulmonary artery and placement of a 12-mm bovine valved conduit between the right ventricle and the pulmonary arteries. The postoperative course was complicated by the development of a pericardial effusion, and electrocardiographic monitoring revealed supraventricular and ventricular extrasystoles, for which propranolol therapy was initiated. Additionally, due to feeding difficulties, a fibrolaryngoscopy was performed, showing hypomobility of the left vocal cord.

For the complex clinical features and metabolic screening suggestive of methylmalonic acidemia with homocystinuria, the baby was sent to the local Rare Disease Center based on a suspected genetic condition.

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ABS 48

PEDIATRIC VOMITING: A CLINICAL CHALLENGE

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INTRODUCTION

Vomiting is a frequent reason for pediatric Emergency Department (ED) visits. Defined as oral forceful retrograde expulsion of gastrointestinal or esophageal contents, it represents a protective reflex mediated by the medullary vomiting center in response to stimuli from multiple organ systems. Although the majority of cases result from benign, self-limiting conditions, with gastrointestinal infections being the most common in the pediatric population, vomiting can conceal severe, time-critical, and potentially life-threatening disorders. In these cases, prompt recognition and timely intervention are crucial, as delays may lead to rapid clinical deterioration and compromise patient survival [1].

CLINICAL CASE

We report the case of a 4-year-old girl with no prior medical history, who presented to the ED with 3 hours of repeated vomiting, frontal headache, and lethargy, afebrile. On examination, vital signs were within normal limits; Glasgow Coma Scale was 10, Alert, Voice, Pain, Unresponsive (AVPU) Scale was P, and cervical flexion elicited pain. Laboratory tests revealed neutrophilic leukocytosis, mild thrombocytosis, and mild dehydration, with normal clinical chemistry, CRP, and procalcitonin. Contrast-enhanced brain CT showed a right periventricular lesion with hemorrhagic components, indeterminate from cavernoma and low-grade heteroplastic lesion, associated with a tetra-ventricular hemorrhage, necessitating urgent left frontal external ventricular drain placement. In the following days, her condition improved, with the regained full consciousness. Follow-up MRI confirmed the lesion, which remained indistinguishable from a cavernoma and a low-grade heteroplastic lesion. Subsequent histopathological analysis at a tertiary care center ultimately classified the lesion as a low-grade glioma.

DISCUSSION

Vomiting, while extremely common in childhood, can be an early and subtle sign of life-threatening conditions [1].

In our case, it was the onset manifestation of a brain tumor complicated by hemorrhage, in line with literature reporting vomiting as the onset symptom in up to 64% of pediatric cases. Pediatric intracranial hemorrhages, although rarely attributable to brain tumors, carry a mortality of up to 20% and frequently result in long-term sequelae among survivors [2].

These data highlight how the true challenge for the pediatric emergency physician lies in discriminating and identifying, within a broad spectrum of mostly benign conditions, the small subset of patients with severe and time-critical pathologies.

A systematic and rigorous approach is essential to guide timely diagnostic and therapeutic interventions, making the difference between favorable outcomes and serious complications [1].

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ABS 49

BEYOND THE HEART: A COMPLEX CLINICAL CASE

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INTRODUCTION

Atrial flutter presents with typical sawtooth waves. It is potentially fatal, but if recognized in time, it leaves no sequelae. It may be caused by congenital heart disease, fetal distress, hypoxia, myocarditis, metabolic disease, or the placement of an intracardiac venous catheter [1]. We present a complex clinical case of respiratory distress, hypoxic-ischemic encephalopathy, and atrial flutter.

CASE REPORT

Born at 37^{+5} weeks by spontaneous delivery, with Apgar 8-8 and blood gas in reasonable compensation, the newborn required ventilation after birth. The patient was transferred to the Neonatal Intensive Care Unit for respiratory distress and abnormal neurological examination. The umbilical vein was cannulated, and a chest X-ray was performed. Surfactant was administered at 200 mg/kg. After 30 minutes, due to a heart rate of 290 bpm and suspicion of supraventricular tachycardia, vagal maneuvers were attempted, without effect. After administration of 3 boluses of adenosine at increasing doses without benefit, and following the appearance of a 3:1 atrioventricular block, atrial flutter was diagnosed. A bolus of amiodarone (5 mg/kg) was given, followed by continuous infusion. Due to altered cerebral function monitoring, systemic hypothermia was initiated. For increased oxygen requirement, the newborn was intubated, and intermittent positive pressure ventilation with synchronization was started. At 10 hours of life, early signs of heart failure appeared, leading to electrical cardioversion (up to 3 joules, equal to 1 joule per kg) with restoration of sinus rhythm. At 14 hours of life, severe respiratory depression with miosis and apnea occurred after administration of naloxone. During hypothermia, due to hyperbilirubinemia, phototherapy was performed. At 72 hours of life, rewarming was initiated, and the infant was extubated. A progressive improvement in general conditions was observed, up to complete normalization.

CONCLUSIONS

If atrial flutter does not respond to medical therapy, electrical cardioversion is required. Maintenance antiarrhythmic therapy is reserved for the rare cases of recurrence [2].

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OCULAR FISHHOOK INJURY IN A 6-YEAR-OLD PATIENT

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INTRODUCTION

Fishhook injuries are a common presentation to Emergency Departments during the summer, predominantly affecting male patients with superficial lesions of the hands and arms. While these injuries are frequent, ocular involvement is rare, yet it can lead to devastating consequences such as vision loss, endophthalmitis, traumatic cataract, intraocular hemorrhage, corneal scarring, and retinal detachment [1, 2].

CASE REPORT

This report describes the clinical course and management of a severe penetrating ocular fishhook injury in a 6-year-old boy. The patient presented to our Emergency Room with a fishhook and attached lure embedded in his left eye. A comprehensive initial evaluation was difficult due to the patient's discomfort and lack of cooperation. Immediate consultations with anesthesiology and ophthalmology were requested, and midazolam was administered. The injury was identified as a penetrating open globe injury, in which a barbed hook had perforated the cornea and anterior chamber, entering from an inferior trajectory and anchoring in the superior eyelid [3]. Surgical removal was performed under general anesthesia. After thorough disinfection, the superior eyelid was incised to disengage the hook. The needle-cover method was then successfully employed to remove the hook, following an unsuccessful attempt with the advance-and-cut technique. The corneal laceration was repaired with 10/0 nylon sutures, and an intracameral injection of cefuroxime was administered at the conclusion of the procedure for endophthalmitis prophylaxis. The eye was then patched, and the patient started on a combination of topical and intravenous antibiotic therapy. In the postoperative period, the patient developed an incipient secondary cataract, amblyopia, astigmatism, and reduced visual acuity. One month after surgery, his corrected visual acuity in the injured eye was 3/10 (corrected to Cyl +3.50 ax 90°), in contrast to the 10/10 vision in his unaffected right eye. Further vision rehabilitation and potential future corrective surgery are indicated to improve the final visual outcome.

CONCLUSION

The dramatic consequences of such accidental traumatic lesions underscore the importance of prevention, which includes the consistent use of appropriate protective equipment and rigorous adult supervision during at-risk activities.

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LOOK BETTER TO SEE BEYOND: A DIAGNOSTIC CHALLENGE IN PEDIATRIC CHOROIDAL DETACHMENT

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INTRODUCTION

Vogt-Koyanagi-Harada syndrome (VKHS) is a chronic, bilateral granulomatous panuveitis characterized by exudative retinal detachment, which

may be associated with neurological abnormalities, hearing loss, and cutaneous manifestations such as vitiligo and poliosis [1]. Diagnosis in pediatric patients can be particularly challenging due to incomplete or atypical presentations.

CASE REPORT

We describe the case of a 12-year-old boy presenting with sudden unilateral visual loss, without a history of ocular trauma. Ophthalmologic evaluation revealed multifocal choroiditis and bilateral central serous neuroepithelial detachment. Fluorescein angiography showed macular and temporal areas of hyper- and hypofluorescence, suggestive of VKHS. To exclude an infectious etiology, TORCH serology tests, a zoonosis panel including *Brucella*, *Bartonella*, and *Toxocara*, and a tuberculosis test were performed and resulted in negative outcomes. Also, autoimmune pathogenesis was investigated, with negative autoimmunity screening. Audiometric testing was regular, excluding cochlear neuroepithelial involvement. Neurological examination was unremarkable, except for hyperactive deep tendon reflexes. Due to possible central nervous system involvement and taking into account the patient's systemic clinical condition, a brain MRI was scheduled. Indeed, given the patient's phenotype – short stature, microcephaly, diffuse skin lentigines, and neurodevelopmental disorders (ADHD, ODD, intellectual disability) – a genetic workup was initiated to investigate possible syndromic forms (CGH-array and RAS/MAPK pathway gene panel), currently pending [2]. As for the ocular picture, which was suggestive of VKHS, systemic corticosteroid therapy (IV methylprednisolone boluses followed by oral prednisone) was started, with recovery of 3 diopters after only 7 days of treatment [3].

CONCLUSIONS

Bilateral choroidal detachment is a rare condition in children and represents a diagnostic challenge, especially in the absence of trauma or known ocular disease. VKHS is associated with a CD4+ T-cell-mediated autoimmune response targeting melanocyte antigens (tyrosinase, Melan-A/MART-1). Genetic susceptibility related to *HLA-DRB1* has been described, and in our patient, HLA-DRB1*0102 was detected, although it is not the main allele involved [1]. Further investigations regarding the patient's complex condition are, as mentioned, ongoing. As no specific autoantibodies are currently available, diagnosis relies on clinical and imaging findings.

This case highlights the importance of early recognition and treatment to prevent irreversible visual complications such as sunset glow fundus, cataract, and glaucoma.

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CASE REPORT OF AN EARLY PRETERM NEWBORN WITH PRENATAL DIAGNOSIS OF SCHAAF-YANG SINDROME

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BACKGROUND

Schaaf-Yang syndrome (SYS) is a rare genetic disorder caused by a mutation in the paternally inherited copy of the *MAGEL2* gene, located on chromosome 15q11.2, which is subject to maternal imprinting. Approximately 50% of patients inherit a pathogenic variant from a clinically asymptomatic father, while the remaining cases result from a *de novo* mutation. SYS may manifest prenatally with reduced fetal movements, polyhydramnios, and arthrogryposis. In the neonatal period, hypotonia, respiratory distress and feeding difficulties are commonly observed. During childhood and adolescence, intellectual disability, autism spectrum disorders and endocrinological abnormalities, like central diabetes insipidus, may develop. To date, more than 250 individuals with SYS have been identified worldwide. Still, it is assumed that the actual prevalence is underestimated due to diagnostic complexity and limited access to genetic testing in many populations [1-3].

CASE REPORT

J. (a female) was born at 29 weeks + 1 days of gestational age, birth weight 1,390 g, via urgent cesarean section due to maternal sepsis and multiple organ failure.

Prenatal genetic testing, performed due to the US detection of bilateral clinodactyly and arthro-

gryposis, revealed an *ex-novo* frameshift variant c.1996dup in the *MAGEL2* gene, confirming the diagnosis of SYS.

In view of her early prematurity and the occurrence of hypotonia and respiratory distress at birth, which required immediate intubation, she was admitted to the Neonatal Intensive Care Unit.

During the first day of hospitalization, J.'s respiratory distress worsened as it was complicated by mild pneumothorax and respiratory acidosis due to hypercapnia (pCO_2 58.2 mmHg). Respiratory support consisted of high-frequency oscillatory ventilation. Additionally, the patient presented with hypotension and early signs of sepsis.

Throughout the following days, hypercapnia worsened (pCO_2 89.1 mmHg) and, after extubation on the third day, her respiratory support consisted of nasal intermittent positive pressure ventilation. Moreover, frequent episodes of apnea were registered.

For the first 10 days, immaturity of self-thermoregulation required continuous adjustment of the incubator's humidity and temperature.

The patient developed hypernatremia (highest value 151.1 mEq/L); central insipidus diabetes was excluded as urine output was persistently within the range of normality, as well as urinary and serum osmolality. Hypernatremia's therapeutic management involved the administration of high volumes of intravenous and oral fluids, with an inconsistent response to treatment. Furthermore, poor tolerance of parenteral feeding mandates continuous enteral feeding through a nasogastric tube.

To date, at 5 months of postnatal age, J. is now breathing spontaneously with a lower incidence of apnea episodes. Hypernatremia persists. She underwent a 30-day off-label treatment with intranasal oxytocin to improve suction and deglutition, in association with swallowing rehabilitation.

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FLOPPY INFANT: MORE THAN JUST WEAKNESS

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CASE REPORT

A 5-month-old female infant was referred to our Clinic for suspected neuromuscular disease. She presented with generalized hypotonia, absence of spontaneous movements, poor head control, fine tongue fasciculations, and a palmar grasp reflex. In contrast, the plantar grasp reflex and patellar reflexes were absent. She was born at term, with an unremarkable perinatal history. Neuromotor development was referred to as usual until the fourth month of life, when the ipothonia was noted. During hospitalization, laboratory tests and cardiology evaluation were within normal limits. Cranial ultrasound was normal except for a small cyst in the anterior portion of the right lateral ventricle choroid plexus. Electromyography showed diffuse subacute/chronic neurogenic motor impairment, consistent with an axonal/motoneuronal pattern. MLPA genetic research confirmed a homozygous deletion/conversion of the *SMN1* gene with 2 copies of the *SMN2* gene, consistent with the diagnosis of spinal muscular atrophy (SMA). Both parents were found to be carriers of the same mutation in the *SMN1* gene. The patient's maternal grandmother reported that two of her sisters had died at 5 months of age and had previously been diagnosed with SMA, suggesting a possible familial recurrence of the disease. The patient remained clinically stable throughout hospitalization. Based on the genetic findings, the presence of 2 copies of the *SMN2*, the age (> 2 months), treatment with risdiplam was initiated at a dose of 0.20 mg/kg/day. No adverse reactions were observed. Sierologic testing for anti-AAV9 antibodies, before gene therapy, returned negative, confirming eligibility for onasemnogene abeparvovec administration. Consequently, gene therapy has been scheduled.

DISCUSSION

Classic SMA is an autosomal recessive disorder caused by mutations in the *SMN1* gene on chromosome 5, leading to reduced SMN protein and degeneration of motor neurons. This results in

progressive muscle weakness affecting limbs, trunk, bulbar, and respiratory muscles [1]. Diagnosis relies on MLPA; 95% of patients show homozygous *SMN1* deletions. Determining *SMN2* copy number is important for prognosis and treatment planning [1]. SMA is classified into types 0-4 based on age of onset and severity. Type 1 is the most severe and a leading genetic cause of infant death [2]. Treatments like risdiplam and gene therapy (onasemnogene abeparvovec) are most effective if started before significant motor neuron loss.

CONCLUSION

This case highlights the need for an early neonatal diagnosis and timely, personalized treatment in SMA. Preconception carrier screening, prenatal diagnosis, and advanced reproductive technologies may help reduce SMA incidence globally.

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