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ABS 1

FROM ENGAGEMENT TO IMPACT: EVALUATING A COMMUNITY-DRIVEN MODEL FOR DIGITAL HEALTH INNOVATION

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OBJECTIVE
As digital health interventions become increasingly integrated into healthcare, approaches that actively engage communities are essential for developing solutions that are relevant, acceptable, and sustainable. Design Studios for Health (DSH) is a human-centered, community-driven, iterative framework that promotes collaboration, trust, and shared decision-making to co-design digital health interventions with end users. This study evaluated the effectiveness of DSH across multiple digital health initiatives, including pediatric and family-centered applications.
METHODS
Eleven DSH sessions conducted over a two-year period were evaluated across five digital health initiatives: Turbo Word (word recognition for children/students with dyslexia), Smart Start (type 2 diabetes management), Heat Mind (heat-related health risk mitigation), Walk Your Heart to Health (WYHH), and Maternal Health (group-based walking for mental and physical health). These initiatives represented different stages of development, ranging from implemented interventions to projects in design and pilot testing. Mixed-methods data were collected through post-session surveys, open-ended responses, and qualitative debrief discussions. Outcomes included collaboration, facilitator effectiveness, participant cohesion, trust, perceived support, and engagement in the co-design process.
RESULTS
Participants consistently reported high levels of engagement, psychological safety, and empowerment in contributing to intervention design. Successive DSH sessions demonstrated improvements in collaboration, group cohesion, and trust, reflecting the iterative nature of the framework. Across all projects, DSH strengthened researcher-community partnerships, facilitated shared ownership of intervention development, and generated contextually relevant digital health solutions tailored to community needs. The framework proved particularly valuable for engaging children, families, educators, healthcare professionals, and community stakeholders in the co-design of interventions addressing pediatric learning and chronic disease management.
CONCLUSIONS
DSH provides an effective community-engaged framework for co-designing digital health interventions across diverse health contexts, including pediatric and family-centered care. By fostering trust, collaboration, and meaningful stakeholder participation, DSH enhances the relevance, acceptability, and potential adoption of digital health innovations. This scalable approach can support the development and implementation of patient- and family-centered interventions for children, adolescents, and other underserved populations while strengthening community partnerships essential for sustainable health improvement.

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ABS 2

ASYMPTOMATIC TOXIGENIC *CLOSTRIDIOIDES DIFFICILE* COLONIZATION AS A POTENTIAL MARKER OF GUT DYSBIOSIS AND FECAL CALPROTECTIN IN RSV BRONCHIOLITIS: CLINICAL EVIDENCE FROM THE GUT-LUNG AXIS

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BACKGROUND
The gut-lung axis represents a bidirectional communication network through which the intestinal

microbiota may influence immune homeostasis and host responses to respiratory infections. During early infancy, the gut microbiota undergoes dynamic maturation and is strongly influenced by early-life exposures, including mode of delivery and feeding practices. In this context, asymptomatic *Clostridioides difficile* colonization, which is common during infancy and distinct from *C. difficile* infection, may represent a marker of an altered or immature gut microbial ecosystem. In parallel, fecal calprotectin may provide complementary information on intestinal mucosal immune activation. However, the relationship between these intestinal markers and the clinical severity of respiratory syncytial virus (RSV) bronchiolitis remains poorly characterized.

OBJECTIVE

To investigate whether asymptomatic toxigenic *C. difficile* colonization, considered as a potential surrogate marker of altered gut microbial ecology, and elevated fecal calprotectin concentrations are associated with the severity of RSV bronchiolitis, thereby exploring the potential involvement of the gut-lung axis in the clinical heterogeneity of RSV infection.

METHODS

This prospective, multicenter observational study was conducted between October 2024 and April 2026 across 3 pediatric hospitals in Sicily, Italy. A total of 140 children younger than 24 months hospitalized for a first episode of bronchiolitis with molecularly confirmed RSV infection were included. Stool samples were collected within 24 hours of hospital admission and again before discharge. Toxigenic *C. difficile* was detected using molecular testing, and fecal calprotectin concentrations were quantitatively measured. Associations with bronchiolitis severity, length of hospital stay, and respiratory support requirements were investigated. Factors independently associated with severe disease were assessed using multivariable logistic regression, while the discriminative performance of fecal calprotectin was evaluated using receiver operating characteristic (ROC) curve analysis.

RESULTS

Among the 140 patients, 56 (40.0%) had severe bronchiolitis. Asymptomatic toxigenic *C. difficile* colonization was detected in 50.0% of the overall cohort and was significantly more frequent among patients with severe disease. Colonized infants were also more frequently delivered by cesarean section and formula-fed, two early-life factors associated with different trajectories of gut microbiota maturation, and showed significantly higher admission fecal calprotectin concentrations than non-colonized infants. Patients with severe bronchiolitis had significantly higher admission fecal calprotectin concentrations than those with non-severe disease (800.0 μg/g [IQR 700.0-982.5] vs 544.5 μg/g [IQR 450.0-662.0]; $p < 0.001$). Admission fecal calprotectin was positively correlated with length of hospital stay ($\rho = 0.686$; $p < 0.001$). In multivariable analysis, both fecal calprotectin (adjusted OR 1.016 per μg/g; 95% CI: 1.009-1.024; $p < 0.001$) and toxigenic *C. difficile* colonization (adjusted OR 4.105; 95% CI: 1.232-13.678; $p = 0.021$) remained independently associated with severe bronchiolitis. Fecal calprotectin showed excellent discriminative performance (AUC = 0.906); a cut-off of 669 μg/g yielded a sensitivity of 89.3% and a specificity of 76.2%.

CONCLUSIONS

Asymptomatic toxigenic *C. difficile* colonization and elevated fecal calprotectin concentrations were independently associated with greater severity of RSV bronchiolitis. Rather than suggesting a direct pathogenic role for *C. difficile*, our findings support the hypothesis that asymptomatic colonization may represent a surrogate marker of an altered or immature gut microbial ecosystem associated with intestinal immune activation and a more severe respiratory phenotype. These findings provide clinical evidence supporting the potential involvement of the gut-lung axis in RSV bronchiolitis. Future studies integrating longitudinal gut microbiome profiling, metabolomics, and immune phenotyping are warranted to validate this hypothesis and clarify the underlying biological mechanisms.

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ABS 3

LACTOFERRIN BENEFITS: NEW INSIGHTS INTO NEWBORN AND INFANT HEALTH

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INTRODUCTION
Lactoferrin is an anti-infective whey protein present in both colostrum and mature breast milk, with an important role in the immune response. Numerous *in vivo* and *in vitro* studies have demonstrated that lactoferrin exerts antimicrobial and anti-inflammatory effects, modulates both innate and adaptive immune responses, influences cytokine production, and affects the proliferation and migration of immune cells.
DISCUSSION
Lactoferrin exerts its antimicrobial effects through several mechanisms. First, it can interact with porins located in the outer membrane of Gram-negative bacteria, disrupting bacterial membrane integrity. Second, its ability to sequester iron deprives bacteria of an essential nutrient required for growth and proliferation. Third, lactoferrin exerts important effects at the intestinal level. In particular, iron-free lactoferrin (apo-lactoferrin) has been shown to attenuate epithelial inflammatory signaling, including the production of TNF-α, IL-6, and IL-1β, through modulation of the NF-ϰB and PPAR signaling pathways. Bovine lactoferrin (BLF), which shares approximately 77% structural homology with human lactoferrin, has attracted considerable clinical interest and has become the focus of several randomized controlled trials. Its potential as a nutritional supplement is particularly relevant in preterm infants, whose immature intestinal and immune systems place them at increased risk of late-onset sepsis. An Italian randomized controlled trial in low-birth-weight infants demonstrated that BLF supplementation reduced the incidence of sepsis from 17% to 6%, with a particularly pronounced effect against Gram-negative organisms and *Candida spp.* The study also reported a marked reduction in severe retinopathy of prematurity. These findings are being further evaluated in a larger placebo-controlled trial involving 414 preterm infants in Lima, Peru. This study also investigates whether the biological activity of lactoferrin translates into measurable neurodevelopmental benefits, assessed using standardized developmental scales at 12, 18, and 24 months of age. A December 2025 real-world study involving 111 infants found that lactoferrin-fortified formula was associated with enhanced linear growth and a more favorable gut microbiome profile, characterized by increased production of short-chain fatty acids and enrichment of *Bifidobacterium breve*, compared with standard formula.
CONCLUSION
Lactoferrin represents a promising preventive strategy in neonatal care, with potential antimicrobial, immunomodulatory, and neuroprotective effects. Further large-scale randomized controlled trials are needed to establish its efficacy and clarify its role in preventing infection and improving long-term outcomes in preterm infants.

ABS 4

ARTIFICIAL INTELLIGENCE-DRIVEN MODELS FOR THE DIAGNOSIS AND MANAGEMENT OF RARE DISEASES

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Rare diseases comprise a highly heterogeneous group of disorders affecting millions of individuals worldwide. Despite major advances in genetics, molecular medicine and diagnostic technologies, achieving a timely diagnosis remains a major clinical challenge. Patients frequently experience a prolonged "diagnostic odyssey", characterized by multiple specialist consultations, repeated investigations, misdiagnoses and delayed access to appropriate care [1]. This problem is particularly relevant in children, in whom early signs may be subtle, nonspecific or shared by different disorders. Reducing diagnostic delay therefore represents one of the major unmet needs in rare disease medicine.
Artificial intelligence (AI) offers a unique opportunity to transform this scenario by shifting the diagnostic paradigm from recognition of established disease toward earlier identification of individuals at risk. Machine-learning and deep-learning approaches can analyze large and heterogeneous datasets, integrating clinical manifestations, electronic health records, laboratory findings, medical imaging, his-

topathology, genomic and other omics data. By identifying complex patterns and disease signatures that may escape conventional assessment, AI-based systems could support clinicians in recognizing rare disorders at earlier stages and prioritizing patients for targeted specialist evaluation and genetic or molecular testing [2].

The potential of AI is particularly relevant to rare diseases because their phenotypic heterogeneity, variable expressivity and overlap with more common conditions frequently complicate conventional diagnostic pathways. Multimodal AI models may help connect apparently unrelated clinical findings, identify previously unrecognized genotype-phenotype associations and generate individualized diagnostic probabilities. AI may also facilitate patient stratification, differential diagnosis and the identification of subjects with atypical or incomplete phenotypes.

However, the application of AI to rare diseases presents major challenges, including limited and fragmented datasets, small patient populations, data imbalance, lack of standardized phenotyping, algorithmic bias, limited interpretability and insufficient external validation. Data sharing and multicenter international collaboration will therefore be essential for developing robust and generalizable models.

AI should not replace clinical expertise, but rather act as a decision-support system capable of augmenting human diagnostic reasoning. The ultimate goal is not simply to automate diagnosis, but to shorten the diagnostic odyssey and enable earlier, personalized interventions. Properly validated and ethically implemented AI models may represent a major step toward a future in which rarity of disease no longer necessarily translates into rarity of diagnosis [3].

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ABS 5

BEYOND BELL'S PALSY: LYME NEUROBORRELIOSIS PRESENTING AS FACIAL PALSY IN A CHILD

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INTRODUCTION

Lyme neuroborreliosis (LNB) is a common extracutaneous manifestation of Lyme borreliosis in European children, usually occurring as early disseminated disease with peak incidence among children aged 5-9 years. Peripheral facial nerve palsy is a typical presentation, while erythema migrans may be absent [1]. Diagnosis relies on exposure history, clinical findings, serum two-tier serology and, when indicated, cerebrospinal fluid (CSF) analysis, in accordance with for IDSA/AAN/ACR criteria [2].

CASE REPORT

A previously healthy Swedish child developed low-grade fever after a tick bite in the right retroauricular region, followed by acute right-sided peripheral facial palsy with lagophthalmos. No meningeal signs or erythema migrans were present, although mild irritability was reported. Inflammatory markers and hepato-renal function were normal. Due to possible meningeal involvement, lumbar puncture was performed. CSF analysis showed lymphocytic pleocytosis, elevated protein and normal glucose, and intrathecal synthesis of *Borrelia*-specific antibodies; serum two-tier serology was strongly positive, confirming LNB. Intravenous ceftriaxone was started, with complete clinical recovery.

DISCUSSION

This case highlights the diagnostic approach to pediatric LNB presenting with facial palsy. CSF pleocytosis with intrathecal *Borrelia*-specific antibody production strongly supports CNS involvement; however, the antibody index may be negative early in the disease course, and a normal result does not exclude LNB [2]. The need for lumbar puncture in children with isolated facial palsy and no meningeal features remains debated. Current guidelines recommend individualizing CSF examination, since treatment of LNB may be the same whether meningitis is present or not [2]. Oral doxycycline and intravenous ceftriaxone are

both recommended options for Lyme-associated meningitis or cranial neuropathy, with route of administration guided by individual clinical factors rather than efficacy [2]. In this patient, irritability increased concern for meningeal involvement and supported CSF evaluation. Pediatric LNB generally has a favorable outcome after appropriate treatment, although persistent facial nerve dysfunction may occur and follow-up is recommended [3]. A high index of suspicion during peak tick season remains crucial for timely diagnosis and optimal outcome.

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ABS 6

"DON'T FORGET YOUR INHALER!" — IMPROVING INHALER TECHNIQUE ASSESSMENT IN A PAEDIATRIC RESPIRATORY CLINIC USING SMS REMINDERS

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BACKGROUND

Inhaler technique is an important component of effective asthma management, with national recommendations supporting regular assessment and review. However, assessment may not be possible when patients do not bring their inhaler and spacer to clinic. This quality improvement project aimed to improve inhaler availability for assessment and increase the proportion of patients receiving inhaler technique review in a paediatric respiratory clinic.

METHODS

A baseline audit was undertaken among randomly selected patients attending a paediatric asthma clinic over a 4-week period. Three months later, the audit was repeated over a similar period following implementation of an additional SMS reminder intervention. Patients continued to receive the standard clinic appointment letter. In addition, parents/carers received an SMS reminder the day before the appointment confirming the date and time and specifically reminding them to bring the patient's inhaler and spacer to clinic.

RESULTS

At baseline, 7/23 patients (30.4%) brought their inhaler to clinic and 6/23 (26.1%) received an inhaler technique review. Following introduction of the SMS intervention, 10/16 patients (62.5%) brought their inhaler to clinic, representing a 32 percentage-point increase. The proportion receiving documented inhaler technique assessment increased to 10/16 (62.5%), a 36 percentage-point increase. The SMS intervention was delivered to all patients in the re-audit cohort (16/16, 100%). Among patients assessed, the proportion requiring correction of inhaler technique increased from 2/6 (33%) to 4/10 (40%). The increased number of assessments also identified equipment-related problems, including cracked or inappropriate spacers and device leaks, alongside technique errors such as failure to shake the inhaler and incorrect cleaning.

CONCLUSION

Addition of a simple SMS reminder to the existing clinic appointment letter increased the proportion of patients bringing their inhaler to clinic from 30.4% to 62.5% and enabled a corresponding increase in documented inhaler technique assessment. The intervention improved the reliability of the assessment process and identified previously unrecognised technique and equipment-related problems. Further quality improvement cycles will focus on standardising inhaler technique assessment and education and addressing equipment-related barriers.

ABS 7

NEONATAL HYPOTONIA IN THE GENOMIC ERA

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Neonatal hypotonia is a generalized reduction in muscle tone, in which the newborn lacks the usual flexor tone and has excessive joint mobility [1]. Absent antigravity movements, at rest and on crying, are sensitive and specific for peripheral involvement, in which tendon reflexes are usually reduced or absent; retained or brisk reflexes suggest a central origin, usually accompanied by altered alertness and often seizures. Contractures, when present, often follow reduced fetal movements, and polyhydramnios, a sign of prenatal swallowing weakness, is associated with neuromuscular disease, especially when antigravity movements are also reduced. Ptosis and a weak cry may point to bulbar or neuromuscular junction involvement [1]. Individually these signs are rarely diagnostic; together they guide which test is ordered first and how urgently. Several common genetic causes are identified by targeted assays. Spinal muscular atrophy (SMA) is diagnosed by *SMN1* testing and Prader-Willi syndrome by methylation analysis at 15q11-q13 [1], a defect that exome sequencing (ES) does not detect [2]. The *DMPK* triplet expansion of myotonic dystrophy type 1 (DM1) escapes ES [2], is not reliably captured by genome sequencing (GS) [1], and requires dedicated testing. The American College of Medical Genetics and Genomics (ACMG) guideline for congenital anomalies or intellectual disability recommends targeted testing first when the presentation points strongly to a single genetic diagnosis, including one that sequencing may miss [2]. In SMA and DM1, the signs prompt these assays without neurophysiology or biopsy, reserved for inconclusive cases [1]. For most other floppy neonates, ES or GS is the next step. Rapid first-tier ES exceeds 40% in neonatal hypotonia cohorts [1]. In critically ill neonates and infants unselected for hypotonia, a 2026 meta-analysis pooled 39.4% across 26 ES cohorts (n = 2,205; 95% CI 32.8-46.3) and 39.4% across 22 GS cohorts (n = 2,101; 95% CI 34.7-44.1) [3]. Comparable yields keep ES a valid option where cost or GS capacity is limiting [3]. Where disease-modifying therapies exist, diagnostic timing directly affects outcomes [1]. In the ACMG evidence assessment, ES or GS reached 38% versus 21% for standard testing [2]. The economic model projected more diagnoses at lower cost with ES first or second [2]. Turnaround is 6-15 days (rapid) and 1-3 days (ultrarapid) [2]. ES or GS therefore belongs early in the pathway, not after an extensive work-up [2]. Newborn screening can identify SMA, infantile-onset Pompe disease, and other treatable disorders before hypotonia appears, though not every program includes them [1]. Rapid sequencing does not replace examination but depends on it: a well-defined phenotype raises the pretest probability and guides variant interpretation [1]. For a floppy neonate, the question is no longer whether to seek a genetic diagnosis but which test to request first, and the answer is needed within days.

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ABS 8

MATERNAL PERIODONTITIS AND ADVERSE PREGNANCY OUTCOMES: FROM ORAL INFLAMMATION TO FETAL AND NEONATAL HEALTH

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Adverse pregnancy outcomes, including preterm birth (PTB, defined as any live birth before 37 completed weeks of gestation), low birth weight (LBW, birth weight < 2,500 g), and preeclampsia, are major contributors to neonatal morbidity and mortality and remain leading causes of adverse health outcomes among fetuses and newborn infants [1-3]. Newborn infants affected by any of these conditions during pregnancy can develop severe long-term disabilities and may need long-term neonatal intensive care [3]. Periodontitis is a chronic inflammatory condition caused by Gram-negative periodontal pathogens, which leads to loss of connective tissue attachment, bone, and tooth. Periodontitis results in local as well as systemic increase of inflammatory mediators and cytokines [2]. Two major biological mechanism pathways linking periodontitis to PTB or LBW have been proposed [2]. One pathway is the hematogenic translocation of the oral microbes and their byproducts and inflammatory mediators via

systemic circulation stemming from oral pathogens causing periodontal infection that may serve as a reservoir of endotoxins, bacteria, and inflammatory mediators, thereby exposing placenta and the fetus [2]. The second proposed pathway is more direct where the oral microorganisms and their components directly reach the fetus through the placenta [2]. The exact mechanism is still unclear. Some studies have shown that during pregnancy and the postpartum period, periodontitis is more commonly seen in mothers with PTB or LBW, and these patients experience more gingival bleeding, gingival inflammation, recession, and worse pocket depth and attachment loss. Several epidemiological studies have shown a significant association between chronic periodontitis and LBW [2]. Evidence in literature suggests a link between pregnancy and exacerbation of the periodontitis in the pregnant woman and consequently, potential health consequences for mother and fetus [1, 2]. Periodontal treatments during the second trimester have shown to improve maternal oral health but fail to reduce the risk of PTB [2]. This may suggest the need for earlier intervention and improve care to enhance overall health outcomes for both mother and the fetus. Documenting oral health history, prevention oral health care, patient education, and timely referral to the dentist as part of general health examinations earlier during pregnancy while monitoring the same in the postpartum period can be integrated in perinatal care models to advance health outcomes for the mother and fetus.

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ABS 9

ATYPICAL PRESENTATION OF ACUTE LYMPHOBLASTIC LEUKEMIA IN A 15-YEAR-OLD BOY: A DIAGNOSTIC ODYSSEY

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Acute lymphoblastic leukemia (ALL) is the most common pediatric hematological malignancy, typically presenting with fever, cytopenias, lymphadenopathy, hepatosplenomegaly, bone pain, and bleeding. The peripheral blood smear usually shows cytopenias with or without circulating blasts. However, ALL poses a significant diagnostic challenge when presenting atypically, frequently masquerading as musculoskeletal or rheumatologic disorders. In adolescents, recurrent fever and bone pain can closely mimic autoinflammatory conditions like chronic non-bacterial osteomyelitis (CNO), leading to misdiagnosis and delayed treatment.

We report a 15-year-old boy hospitalized 4 times over 3 months before receiving a diagnosis of ALL.

A 15-year-old boy with a history of generalized epilepsy and a previous negative workup for rectal bleeding presented with fever and migratory pain initially affecting the chest, neck, shoulder, and clavicle. Suspected costochondritis was treated with NSAIDs and a 7-day course of betamethasone, leading to temporary resolution. Blood tests showed a normal complete blood count (CBC) and a slight increase in inflammatory markers. One month later, recurrent fever and thoracic pain prompted a first pediatric admission. Inflammatory markers were elevated, but CBC and peripheral blood smear were completely normal without atypical cells. Extensive infectious, autoimmune screenings (including HLA-B27), and initial imaging (ECG, echocardiogram, chest/clavicle X-rays) were unremarkable. Two weeks after discharge, a second admission was required for low-grade fever and bilateral knee pain. Inflammatory indices remained elevated, while the CBC and smear remained persistently normal. Whole-body bone scintigraphy showed increased uptake in the right clavicle, ribs, and right elbow. Due to the presentation of recurrent fever, bone pain, and persistent inflammation, the patient was admitted to Pediatric Rheumatology (third admission). Tests confirmed systemic inflammation, with a normal peripheral blood smear. Suspecting CNO, an MRI was performed and revealed widespread bone spongiosa alterations. Finally, a severe flare-up

of knee pain causing impaired ambulation poorly responsive to NSAIDs led to a fourth admission. Repeated peripheral smears unexpectedly revealed rare, immature elements. A bone marrow aspirate and biopsy were performed, confirming ALL.

ALL can present atypically, mimicking autoinflammatory rheumatic diseases. Clinicians should be aware that recurrent fever and migratory bone pain can mask hematological malignancies even with a persistently normal CBC and peripheral blood smear. When systemic inflammation and bone pain persist, a bone marrow biopsy remains mandatory to prevent critical diagnostic delays, regardless of reassuring peripheral blood findings.

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ABS 10

EARLY DETECTION OF CELIAC DISEASE IN PEDIATRIC PATIENTS: THE ROLE OF THE PEDIATRIC DENTIST

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Celiac disease (CD) is a gluten-related autoimmune enteropathy occurring in genetically predisposed individuals. Gluten is the trigger to produce specific autoantibodies causing a massive inflammatory response against enterocytes. CD usually develops during childhood and presents typical gastrointestinal symptoms such as malabsorption, diarrhea, constipation and can lead to growth retardation. Diagnosis is based on the serum detection of tissue-transglutaminase antibodies (tTG-IgA), anti-endomysial antibodies (EMA) and antibodies against deamidated-gliadin peptides (DGP-IgA and DGP-IgG) and confirmed by intestinal biopsy and genetic tests demonstrating positivity for HLA-DQ2 and/or DQ8. More complex cases may occur, particularly those which refer to nonspecific or extraintestinal symptoms. Among the latter, oral lesions show strong association with CD even in the absence of classic symptoms. Recurrent aphthous stomatitis (RAS) is an autoimmune condition of unknown etiology affecting the non-keratinized oral mucosa in genetically predisposed individuals. It is characterized by circular painful ulcerations from few mm up to 1 cm in diameter that extend into deeper layers of the mucosa. RAS appears to be the most strongly associated oral lesion with CD, with a reported prevalence up to 50%. Early childhood caries (ECC) is a condition affecting the hard tissues of primary teeth characterized by enamel hypoplasia, making teeth less resistant to acids. ECC has an extremely high global prevalence: according to metanalysis, up to 63% of children under 5 years of age present this condition, following the DMFT criteria established by WHO. This hypomineralization may bring premature tooth loss and so malnutrition, abnormal development of the maxillary bones – leading to malocclusion and orthopedic problems of the craniofacial complex – as well as psychological consequences. Causes of the prisms' malformation are unknown but studies suggest autoimmune mechanisms, perinatal infections and trauma. Autoimmune etiology may explain the high incidence of these lesions among CD patients, where enamel defects are present in approximately 27% of children. Another relatively common finding is benign migratory glossitis. This lesion typically migrates across the dorsum and lateral borders of the tongue and causes atrophy of the lingual papillae. It is usually asymptomatic, but, if associated with pain and nonspecific symptoms such as pallor and fatigue, further investigation can be suggested to detect iron deficiency. Its association with CD is less well established than that of RAS, but it should be considered in diagnostically uncertain cases. In conclusion, oral lesions should warn the clinician of the possibility of CD, since they may have early onset or may even be the only clinical manifestations of CD. Therefore, the role of the pediatric dentist is of primary importance in the early identification and diagnostic evaluation of CD.

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ABS 11

WHEN PUBARCHE CONCEALS A RARE DIAGNOSIS

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INTRODUCTION

Pubarche in pediatric age requires an accurate diagnostic assessment. Adrenocortical neoplasms represent a rare cause of precocious pubarche, but they should be suspected in the presence of rapidly progressive signs of virilization and marked hyperandrogenism.

Accurate diagnostic assessment of precocious pubarche is essential to enable timely treatment. An appropriate diagnostic workup includes medical history, physical examination, endocrinological evaluation and imaging. Pituitary, adrenal and thyroid function and calcium-phosphorus metabolism should also be assessed to exclude the main causes of hyperandrogenism in children.

CASE REPORT

S. was referred to our department at the age of 1.1 years for pubarche that had developed several months earlier. Endocrinological investigations showed increased DHEAS > 3,000 µg/dl (reference range 2-85 µg/dl), testosterone 3.51 ng/ml (reference range 0.01-0.52 ng/ml), D4-androstenedione > 10 ng/ml (reference range < 0.2 ng/ml) and 17-hydroxyprogesterone 5.81 ng/ml (reference range 0.13-2.51 ng/ml) with the remaining endocrine investigations within the normal range. Abdominal ultrasound and MRI revealed a right adrenal mass measuring approximately 5.5 cm, with hemorrhagic components and calcifications within the lesion and mild heterogeneous enhancement. Following multidisciplinary evaluation, right adrenalectomy with lymph node sampling was performed. Histopathological examination revealed an adrenocortical neoplasm with benign biological behavior. During the postoperative period, complete normalization of the hormonal abnormalities was observed but she developed transient adrenal insufficiency. This condition was compatible with delayed functional recovery of the contralateral adrenal gland and required initiation of hydrocortisone replacement therapy, which was subsequently discontinued following progressive functional recovery. Periodic endocrinological follow-up was maintained.

CONCLUSIONS

Although rare, androgen-secreting adrenal neoplasms should be considered in the differential diagnosis of precocious pubarche, particularly in the setting of persistent and progressive biochemical hyperandrogenism. A systematic diagnostic approach integrating clinical, endocrinological and imaging data enables timely diagnosis and potentially curative surgical treatment.

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ABS 12

LUNG ULTRASOUND IN BRONCHOPULMONARY DYSPLASIA: CURRENT APPLICATIONS AND FUTURE PERSPECTIVES

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BACKGROUND

Bronchopulmonary dysplasia (BPD) is a major complication of prematurity and may result in structural and functional respiratory abnormalities persisting beyond the neonatal period. The assessment of children with BPD requires a multimodal approach integrating clinical findings, imaging, and, whenever feasible, pulmonary function testing. In this context, lung ultrasound (LUS) is emerging as a non-invasive, repeatable, radiation-free imaging modality.

OBJECTIVE

To provide an overview of the main applications of LUS in BPD, with particular emphasis on its potential role in characterizing pulmonary involvement and its integration with conventional imaging and respiratory functional assessment.

CURRENT EVIDENCE

LUS allows bedside assessment of major sonographic pulmonary abnormalities and offers particularly favorable characteristics in pediatric patients, in whom reducing cumulative exposure to ionizing radiation is an important objective. Available evidence suggests that ultrasound scores may contribute to the assessment of the severity of pulmonary involvement in BPD and provide information complementary to chest radiography. Computed tomography remains more detailed for the characterization of structural abnormalities, although its serial use is limited by radiation exposure.

In a recent study from our group involving children with BPD, LUS showed good agreement with computed tomography and significant associations with pulmonary function and nocturnal respiratory monitoring parameters, supporting its potential use as part of a multiparametric patient assessment [1]. Overall, these findings, together with emerging evidence from the literature, suggest that integrating imaging, LUS, and functional measures may contribute to a more comprehensive characterization of the respiratory phenotype of BPD.

CONCLUSIONS

LUS represents a promising tool for the assessment and follow-up of children with BPD because of its safety, repeatability, and bedside applicability. Its role should not be considered a replacement for computed tomography or functional assessment, but rather as a complementary component of a multimodal approach. Prospective, multicenter, and longitudinal studies are needed to standardize acquisition and scoring protocols, establish reference values, and determine the prognostic value of LUS during long-term BPD follow-up.

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ABS 13

FROM AUTOIMMUNE HEMOLYTIC ANEMIA TO AUTOIMMUNE LYMPHOPROLIFERATIVE IMMUNODEFICIENCY (ALPID): A PEDIATRIC CASE REPORT

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Lymphoproliferation (splenomegaly, hepatomegaly and benign lymphadenopathy) and autoimmune cytopenias are common manifestations of immunohematological diseases in childhood. When these occur in combination or with other signs of inborn errors of immunity, they should raise suspicion of autoimmune lymphoproliferative immunodeficiency (ALPID). According to international consensus guidelines, a clinical diagnosis requires both primary criteria (chronic lymphoproliferation > 6 months and double-negative T cells [DNTs] ≥ 1.5%) and ≥ 1 secondary criterion (autoimmune cytopenias, elevated biomarkers, positive family history). ALPID management aims to control immune dysregulation through targeted immunosuppressive therapies, such as the mTOR inhibitor sirolimus. Prognosis depends on preventing long-term complications like hypersplenism and malignant transformation.

We report the clinical course, laboratory workup and therapeutic management of a pediatric case of undetermined ALPID (ALPID-U).

A 12-year-old boy was referred to our Pediatric Hematology Clinic for follow-up after a recent diagnosis of autoimmune hemolytic anemia (AIHA) and splenomegaly. He had been initially treated with oral prednisone for 1 month, followed by a progressive decrease over 15 months obtaining stable hemoglobin but persistent mild cytopenias (Hb 10.3 g/dL, leukocytes 2,770/μL, platelets 123,000/μL). History revealed parental consanguinity, full vaccination, and no recurrent infections. Physical examination showed mild pallor and a palpable spleen (4 cm below the costal margin), without lymphadenopathy.

Due to persistent cytopenias, splenomegaly and history of AIHA, the patient was admitted for extensive investigation. Blood tests, including a complete immunological screening, confirmed mild cytopenias and revealed an underlying immunodeficiency (B and T lymphocytopenia, hypogammaglobulinemia, poor post-vaccination antibody response, and a significantly increased percentage of DNTs [6.7%]). Abdominal ultrasound and total-body PET/CT revealed upper- and subdiaphragmatic lymphadenopathy and marked splenomegaly. ALPID was suspected, and the patient was referred to a Tertiary Care Center where known monogenic defects were ruled out via Next-Generation Sequencing. A diagnosis of

ALPID-U was established and sirolimus therapy was successfully initiated.

ALPID diagnosis in children remains a significant clinical challenge. In our case, the co-occurrence of the primary clinical criteria and AIHA enabled a solid clinical diagnosis of ALPID despite the negative genetic panel.

The evaluation of chronic cytopenias in children should always prompt a complete immunological screening, including serum immunoglobulin quantification and lymphocyte immunophenotyping (specifically DNTs). These early baseline tests are essential to differentiate primary autoimmune cytopenias from complex immune dysregulation syndromes like ALPID, ensuring timely targeted treatment and limiting prolonged steroid exposure.

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ABS 14

ILLNESS-TRIGGERED REGRESSION AS A POTENTIALLY MODIFIABLE TARGET IN *DARS2* LEUKOENCEPHALOPATHY

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INTRODUCTION

DARS2 encodes the mitochondrial aspartyl-tRNA synthetase, and biallelic variants cause a leukoencephalopathy classically defined on magnetic resonance imaging (MRI) by brainstem and spinal cord tract abnormalities with lactate elevation [1]. The same gene produces phenotypes from a prenatal form in which few milestones are acquired [1] to childhood-onset axonal neuropathy without leukoencephalopathy [2], a spectrum that may reflect allelic combinations [2]. In a series selected for unusually severe and atypical MRI, onset was preceded by fever or trauma in all 5 assessable patients, whose disease began between 2 months and 2 years of age [1].

CASE REPORT

A previously well girl lost motor skills at 12 months, with hypotonia and difficulty standing.

Brain MRI showed a leukodystrophic pattern with a lactate peak on spectroscopy; neurophysiology and echocardiography were normal. Muscle biopsy showed a biochemical complex I defect; genetic testing identified a *DARS2* defect. Mitochondrial cofactor therapy with coenzyme Q10, idebenone and riboflavin was started; standing and babbling were regained within a month. Nine months after onset, a mild febrile illness preceded regression of acquired skills, with tremor and gait difficulty; 2 months later, her gait was spasticataxic, and the cofactor regimen was intensified. Follow-up MRI showed attenuation of cerebellar lesions but increased white matter involvement, restricted diffusion, an abnormal lipid and lactate peak, and a reduced N-acetylaspartate/creatine (NAA/Cr) ratio. Gait, balance, tremor and speech improved. At 6 years, the neurological examination was essentially normal, with independent gait, adequate tandem gait and no dysmetria, while brisk reflexes, unsustained ankle clonus, fatigability and sporadic falls persisted. The Hammersmith Functional Motor Scale Expanded score was 64/66 and forced vital capacity 88% of predicted. She ran 10 m in 5.4 s and was attending mainstream preschool.

CONCLUSIONS

Onset in early-onset disease has been preceded by fever or trauma [1], and DARS2-deficient fibroblasts show reduced basal and ATP-coupled respiration with a glycolytic shift [3]. An intercurrent illness may therefore precipitate acute decompensation rather than mark disease progression. Our patient recovered after both episodes and has remained stable despite a reduced NAA/Cr ratio, whereas in the series by Stellingwerff et al., 2 of 6 improved slowly and 4 worsened [1]. A single case provides limited evidence on cofactor therapy, and no treatment of proven disease-modifying efficacy exists [3]. The trigger, however, might be addressed through anticipatory management of intercurrent illness (early antipyresis, maintenance of caloric and fluid intake, avoidance of prolonged fasting and a cautious threshold for intravenous glucose), set out in a written emergency plan at diagnosis.

Regression may then represent a reversible decompensation rather than an irreversible loss.

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ABS 15

DYSLEXIA AND ARTIFICIAL INTELLIGENCE: EVERY CHALLENGE IS UNIQUE

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Developmental dyslexia is a common and heterogeneous neurodevelopmental learning disorder characterized by persistent difficulties in accurate and fluent word reading and spelling. Despite major advances in understanding its genetic, neurobiological and cognitive bases, dyslexia is still frequently identified only after children have experienced persistent reading failure. This discrepancy represents the "dyslexia paradox": neurodevelopmental alterations associated with dyslexia may arise very early in life, whereas diagnosis generally occurs only after formal reading instruction has begun [1].

Dyslexia is increasingly viewed as a dynamic, multifactorial condition characterized by marked interindividual variability across phonological, linguistic, cognitive, executive, visuospatial and socio-emotional domains. Such heterogeneity, together with compensatory mechanisms and frequent comorbidities, may contribute to delayed or missed diagnosis. Late identification may expose children to years of unexplained academic difficulties, negatively affecting self-esteem, emotional well-being, educational achievement and quality of life.

The identification of early cognitive, behavioral and neurobiological markers offers the opportunity to move from a reactive diagnostic model toward risk prediction and early identification. Phonological awareness, letter knowledge and rapid automatized naming, integrated with familial, developmental and potentially neuroimaging information, may help identify children at increased risk before persistent reading failure becomes evident.

Artificial intelligence (AI) may accelerate this transition. Machine-learning and multimodal approaches could integrate heterogeneous longitudinal data and identify complex patterns that escape conventional assessment, potentially distinguishing transient reading difficulties from a high risk of persistent dyslexia [2]. AI may also contribute to management and prognosis by predicting individual responsiveness to educational interventions. AI should be regarded not as a replacement for clinicians and educators, but as a decision-support tool for earlier identification and referral. Prospective, longitudinal and externally validated studies are needed to establish whether AI-assisted models can safely reduce diagnostic delay across diverse linguistic, cognitive and educational settings. Ultimately, AI may help shift the paradigm from waiting for reading failure to identifying children at risk before its educational and psychosocial consequences become established [3].

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ABS 16

PEDIATRIC OTOMASTOIDITIS: AN UNUSUAL TEMPORAL CLUSTER OF TWELVE CASES IN A THREE-MONTH PERIOD

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BACKGROUND

Acute mastoiditis is a serious bacterial infection of the mastoid bone that occurs as a consequence of acute otitis media [1]. It's a rare but potentially serious complication of acute otitis media in children, with reported incidence rates generally ranging from 1.2 to 4.8 cases per 100,000 children per year [1, 2]. We report an unusual temporal cluster of pediatric otomastoiditis observed in our Pediatric Department over a 3-month period.

METHODS

We retrospectively reviewed the clinical data from 12 children diagnosed with otomastoiditis and admitted to our Pediatric Department over a 3-month period. Demographic characteristics, presenting symptoms, microbiological results, laboratory findings, imaging studies, treatment and clinical outcome were analyzed.

RESULTS

Twelve children (5 females and 7 males), aged 1-12 years, were hospitalized for otomastoiditis in a 3-month period. Clinical presentation included otalgia in all patients, with otorrhea, fever, auricular protrusion and/or retroauricular inflammatory signs occurring in most cases. Inflammatory markers were elevated at admission in all patients for whom data were available. Temporal bone CT was performed only in selected cases and demonstrated inflammatory involvement of the middle ear and mastoid air cells. Microbiological cultures identified *Pseudomonas aeruginosa* in 6 patients (50%). All children were treated with intravenous ceftriaxone; amikacin was subsequently added in patients with documented or suspected *P. aeruginosa* infection. Otolaryngology and infectious disease consultations were performed in all cases. Clinical improvement was achieved with conservative medical management, without the need for mastoidectomy or other surgical intervention.

CONCLUSIONS

The occurrence of 12 pediatric otomastoiditis cases within only 3 months represents an unusual temporal cluster considering the rarity of this condition. The detection of *P. aeruginosa* in 50% of our cases is noteworthy and is in line with previous Italian data reporting frequent isolation of this pathogen in pediatric acute mastoiditis [3]. This cluster highlights the importance of early recognition of clinical signs, prompt otolaryngology evaluation and targeted microbiological investigation. Further epidemiological surveillance may help clarify whether this represents a local temporal variation or an emerging epidemiological pattern.

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ABS 17

A THREE-YEAR STUDY ON OUTSOURCING PEDIATRICS IN ITALIAN HOSPITALS

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Healthcare outsourcing has evolved over the years, from non-clinical services (e.g., laundry, food, patient transportation, medical billing), to support services (e.g., microbiology, pathology, radiology, laboratory), and eventually to patient-facing clinical services (e.g., emergency, obstetrics/gynaecology, pediatrics), the core of the healthcare system. The first Italian application of hospital pediatrics outsourcing (HPO) occurred 25 years ago in Piedmont (northern Italy), remaining an isolated case for a long time. In 2018, the "Great Resignation" phenomenon determined acute lack of Italian hospital pediatricians and HPO became a structural need to face pediatrician shortage. However, the impact of HPO remains poorly documented in the scientific literature.

Our objective was to analyze the prevalence and impact of HPO in Italy and evaluate the effectiveness of Decree-Law 34/2023 (DL2023) in curbing this phenomenon, while identifying consequences for clinical quality and staff management.

Mainly, we studied the nationwide prevalence of cooperatives in HPO and their clinical and organizational impact; cooperatives were the most

common business organizations which provided HPO in Italy. Mapping was conducted via a telephone survey of directors of Italian Pediatric Divisions, due to the current lack of a unified national platform for collecting and monitoring outsourcing related data. The evaluated parameters included the prevalence of partial or total HPO during the 2023-2026 period (to estimate the effectiveness of DL2023) and pediatrician turnover analysis based on data from the Veneto Region HR.

Since 2018, 77% of hospital pediatrician resignations have been unplanned, with 90% of them moving into primary care. An analysis of total personnel expenses reveals that HPO entails hourly costs 3 to 4 times higher than the hourly wages of permanent staff, even though overall costs remain comparable. Despite the 12-month limit imposed by DL2023, there was no significant decrease in HPO after 1 year, with the number of HPO events dropping from 78 divisions in 2023 to 67 divisions in 2024. This reduction of just 11 divisions (either in a partial or total way) demonstrates a negligible regulatory impact of DL2023. A more significant reduction was in year 2026, with 38 outsourced Pediatric Divisions. Furthermore, the adoption of outsourcing models in pediatrics was not associated with clinical quality indicators or with regular audit procedures.

In conclusion, HPO compromises team cohesion and control over the quality of care. It is imperative that any recourse to outsourcing be subject to rigorous supervision based on clear indicators. Healthcare management must prioritize investment in collaborative models between facilities and the integration of medical residents. Only by addressing the deep causes of the "Great Resignation" in Italian hospital pediatrics, will it be possible to encourage the permanent hiring of pediatricians, and drastically reduce HPO.

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ABS 18

TOPICAL PROBIOTICS AS AN ADJUNCTIVE APPROACH IN REACTIVE GINGIVAL LESIONS: A PEDIATRIC CASE REPORT

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AIM

To assess the role of topical probiotics as an adjunctive treatment in reactive gingival lesions in pediatric patients.

METHODS

A 7-year-old male patient presented with exophytic, normochromic, painless gingival lesions involving the attached gingiva in the anterior maxillary and mandibular regions, both on the palatal and vestibular sites. The lesions extended over a substantial portion of the affected teeth. Based on the clinical findings, a reactive gingival lesion was suspected. A biopsy was indicated to exclude both a neoplastic process and dysplasia. Considering the patient's young age and the aim of minimizing surgical discomfort, a preliminary course of topical probiotics was initiated.

The patient was prescribed tablets containing a mixture of *Lactobacillus reuteri* and *Lactobacillus salivarius*, with the prescription to administer directly to the gingival tissues twice a day for 30 days, after oral hygiene. At 1-month follow-up, a marked reduction in gingival swelling was observed, allowing the biopsy to be scheduled. Histopathological examination revealed a peripheral ossifying fibroma (POF) arising from the gingiva with no dysplasia. The patient is currently undergoing regular follow-up because of the high risk of recurrence associated with this lesion.

DISCUSSION

POF is a benign reactive gingival lesion, generally asymptomatic, typically affecting young patients (median age: 11.5 years). It appears to originate from periodontal ligament cells and commonly presents as a well-defined exophytic gingival mass that is painless, non-bleeding, tender in consistency and similar in color to the surrounding gingiva. Traumatic or irritative factors, including dental eruption or orthodontic appliances, may contribute to its development.

Histopathologically, POF is characterized by a fibroblastic proliferation associated with mineralized components and an overlying epithelium generally devoid of dysplastic changes. Surgical excision remains the gold standard treatment; however, recurrence has been reported in the range from 8%

to 20%. In pediatric patients, surgical management may be challenging because of limited cooperation and the persistence of local etiological factors.
Topical probiotics may represent a useful adjunctive approach by contributing to the modulation of the oral microbiota and inflammatory response. Probiotic strains such as *L. reuteri* and *L. salivarius* have been associated with increased levels of anti-inflammatory mediators, including IL-10, and reduced levels of pro-inflammatory cytokines such as IL-2, IL-6, and IL-8. Here, topical probiotic therapy was associated with a substantial reduction in gingival swelling and an improvement in overall gingival health, thereby facilitating subsequent surgical management.

CONCLUSIONS

Several studies have demonstrated the efficacy of probiotics in reactive gingival lesions, suggesting that they can be safely used in pediatric patients.

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ABS 19

DECLINING BIRTH RATE, EFFECTS AND MISSING EFFECTS ON ITALIAN NATIONAL HEALTH SERVICE. IS OUR NATIONAL HEALTH SYSTEM SUSTAINABILITY AT RISK?

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Declining birth rate in Italy has reached the lowest number in 2025, when we had the lowest number of newborns (353,240). In 2025 CEDAP (National Registry of Deliveries) we have also: a total of 382 Birth Centers (BCs); 58.8% of births take place in BCs with more than 1,000 births/year; these BCs are 30.6% of the total BCs in Italy. Only 10.13% of births take place in BCs with < 500 deliveries/year, which are 30% of the total BCs. A total of 117 Neonatal Intensive Care Units (NICUs) are counted, 94 of them in BCs with more than 1,000 deliveries/year, 12 in BCs with less than 800 deliveries/year. In 2015 we had 500 BCs counted, 126 BCs had less than 500 deliveries/year, and 6.7% of births occurred in BCs with less than 500 births/year. The population aged 0-5 is also declining, counting 2.42 million in 2025; it was 2.85 million in 2019. In Italy, pediatrician workforce is mainly divided in hospital and primary care pediatricians (PCP). About hospital pediatricians, in the last few years we had the well-known phenomenon of the "Great Resignation" process, with tens of Pediatric Divisions largely managed by outsourcing or free contracts. About PCP we must consider the following data: we will have 1,547 planned retirements (2025-2029), and some PCP positions may remain unfilled due to low numbers of children (and low salary), creating potential coverage gaps. This uneven territorial distribution severely hinders timely access to primary care, driving families toward Emergency Departments also for minor conditions. The declining pediatric population (mainly newborns, and admissions to NICU) has also direct effects on National Health Service costs. In Trento province the Court of Accounts applied the DRG system and noted that the cost of a single birth was 20,980 euros in BCs with less than 500 births/year and 5,200 euros in a BC with NICU and more than 2,700 births/year. The cost of a single newborn transport is about 1,650 euros for a transportation center with a 250-transport yearly volume, but it is about 5,000 euros with a 50-100 yearly volume of transports. Moreover, costs are increasing for single NICU admissions, if the number of admitted preterm infants is declining. Despite a shrinking pediatric population, in order to give more pediatricians to the hospitals and pediatric primary care, we had recently the highest increase in pediatric residents: now 850 yearly positions are available and all assigned, creating a hazard for the future employment of new pediatricians.
In conclusion, the collapse of the birth rate in Italy needs a robust review of the Italian pediatric healthcare network. Geographic proximity, primary care availability and sustainable healthcare costs are the pillars of a needed National Health System reform, in order to avoid inequality in health status and availability. Demographic decline and financial sustainability make closing (when possible) low-volume facilities imperative which, in turn, would concentrate resources in safer hubs, with improved clinical quality.

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ABS 20

PARTIAL BIOTINIDASE DEFICIENCY REQUIRING HIGH-DOSE THERAPY: A CASE REPORT

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INTRODUCTION

Biotinidase deficiency (BTD) is a rare autosomal recessive metabolic disorder caused by biallelic pathogenic variants in the *BTD* gene, leading to impaired biotinidase activity, an essential enzyme for biotin metabolism.

Diagnosis is based on biotinidase enzyme activity. Deficiency is classified as profound when it is < 10% and partial when it ranges from 10-30%. To confirm the diagnosis, molecular analysis of the *BTD* gene is suggested but not mandatory. Clinical manifestations include hypotonia, seizures, developmental delay, skin abnormalities, respiratory problems, and visual or hearing impairment. The partial form may be asymptomatic or show variable symptoms, particularly during metabolic stress. The recommended treatment for both forms is oral biotin supplementation, although for partial BTD it is still debated. The availability of a therapeutic option enhances the importance of the neonatal screening, which enables an early diagnosis and prompt treatment, significantly improving the prognosis.

CASE REPORT

A female newborn presented abnormal neonatal screening for biotinidase enzyme activity (51.8 U/dl; normal value > 80 U/dl). The analysis was repeated at 16 days of life (64.29 U/dl), confirming the diagnosis of partial BTD. Genetic analysis identified two heterozygous variants: c.1330G>C (p.Asp444His), inherited from the mother, and c.1368A>C (p.Gln456His), inherited from the father. The patient's follow-up included auxological, neurodevelopmental, ophthalmological, and audiological assessments. From the first months of her life, the little girl had suffered from atopic dermatitis, poorly responsive to topical treatment. Around the age of 1, the persistent skin dryness suggested a link to BTD, so oral biotin therapy (5 mg/day) was started. After a year she presented with muscle pain, occasional limping, hair and nail fragility, and persistent skin dryness. Considering these symptoms possibly related to biotin deficiency, biotin dose was increased to 10 mg/day. At the annual medical check-up, muscle pain improved, but skin symptoms and hair fragility persisted. Consequently, biotin dose was increased to 20 mg per day, with clinical improvement.

CONCLUSION

This case report highlights the importance of starting treatment even in patients with partial BTD, underscoring the benefits of treating this partial form. Symptomatic partial BTD may require high therapeutic dose, generally used in profound biotin deficiency, and the management of the patients should be individualized on a case-by-case basis.

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ABS 21

MIGRATING WHEALS, ACRAL EDEMA AND JOINT PAIN AFTER AMOXICILLIN-CLAVULANATE: A SERUM SICKNESS-LIKE REACTION

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INTRODUCTION

Serum sickness-like reaction (SSLR) begins 1 to 2 weeks into a course of a drug, or after an infectious agent or a vaccine, with rash, arthralgia or arthritis

and, less often, fever [1]. In 488 patients, amoxicillin was the agent most often implicated, in 72.1%; joint symptoms occurred in 78.7% and corticosteroids were usually effective [1]. It is mistaken for urticaria multiforme (UM) and erythema multiforme (EM) [1].

CASE REPORT

A previously well 5-year-old boy with no drug allergy was given amoxicillin-clavulanate for otalgia. On day 7 of the course nuchal pruritus and urticarial lesions appeared, and on day 8 migrating wheals, with swelling of both feet and inability to bear weight. He was admitted afebrile, with lip edema but intact mucosae, confluent erythematous plaques on the face and trunk, acral edema of the hands and feet, and periarticular swelling of the phalangeal joints with faint ecchymotic discoloration. Fever to 38.2°C followed. The eruption migrated rather than resolved, fading at one site while fresh wheals appeared at another. On the second hospital day both knees were swollen, yet joint examination showed no signs of synovitis and revealed painless mobilization. On the third hospital day, annular, arcuate and polycyclic wheals with central clearing appeared on the trunk. CRP was 13.3 mg/L; there was no eosinophilia, and ESR, complement, ANA, ENA, ASO, rheumatoid factor and urine analysis were normal. *Borrelia* and *Rickettsia* serology and throat culture were negative; *Mycoplasma pneumoniae* IgM and IgG were both positive on a single sample, with positive *Chlamydia pneumoniae* IgG. He received cetirizine and ibuprofen, without corticosteroid. Acral edema and joint pain settled on the third hospital day and the rash on the fourth, and he was discharged on the fifth, with clarithromycin added for the *Mycoplasma* serology.

CONCLUSIONS

The annular wheals read as targets, suggesting EM or UM [2], but both imply fixed lesions [3], and these moved. The swollen hands, feet and knees suggested the onset of arthritis, and their migration recalled acute rheumatic fever, but the swelling was periarticular and nonsynovial, mobilization was painless, ASO was normal and throat culture negative. Normal complement argues against true serum sickness, and urticarial vasculitis persists beyond 7 to 10 days [3]. IgA vasculitis, another cause of rash with joint pain, spares face and trunk [3]. Mucosal sparing and the absence of respiratory symptoms make *Mycoplasma*-induced rash and mucositis unlikely, but they do not exclude the organism as a trigger, since SSLR follows infection as well as drugs [1]; IgM can persist for months, and resolution preceded clarithromycin. Onset on day 7 makes an IgE-mediated reaction less likely [3]; drug and infection cannot be separated. Movement, not morphology, is the discriminating sign, and getting it right spares the child a wrong diagnosis and an unverified allergy label [3].

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ABS 22

RECURRENT RHABDOMYOLYSIS IN CHILDREN: SIX FEATURES THAT POINT TO A METABOLIC MYOPATHY

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Rhabdomyolysis is the acute lysis of striated muscle, releasing creatine kinase (CK) and myoglobin into the bloodstream. The 276th ENMC Workshop defined it as compatible symptoms, namely muscle pain, weakness or swelling, with CK above 10,000 IU/L after exercise and above 5,000 IU/L otherwise, a peak on days 1-4 and normalization within 1-2 weeks; myoglobinuria alone also establishes the diagnosis, and in established neuromuscular disease the threshold is 5-10 times the patient's own baseline [1]. That pathway was framed for adults. The classical triad appears in under 10% of patients [2] and pediatric series report dark urine in only 3-6% of cases [1]; recognition rests on serial CK. Viral myositis, most often influenza, is the leading trigger below 12 years of age; trauma, unaccustomed exercise and drugs dominate later [1, 2]. Inherited metabolic disorders account for 3-5% of pediatric cases [1, 2]. Acute kidney injury occurs in 7-45% of pediatric cases [2]. The trigger often points to the underlying defect. Symptoms appearing minutes

into brief static or sustained dynamic effort, with myalgia relieved by rest, suggest a glycogenolytic or glycolytic defect, as in McArdle disease [1, 2]. Episodes precipitated by endurance activity, fasting or fever suggest a fatty acid oxidation defect such as carnitine palmitoyltransferase II or very-longchain acyl-CoA dehydrogenase (VLCAD) deficiency, though endurance also triggers mitochondrial myopathy [1]. Heat, volatile anesthetics and a family history of malignant hyperthermia point to *RYR1* [1, 2]. Six features spell the acronym RHABDO and help decide who needs genetic testing: Recurrent episodes; HyperCKemia persisting 8 weeks after the episode; Accustomed exercise, too ordinary to account for it; Blood CK above 50 times the upper limit of normal; Drugs and other precipitants insufficient to explain the severity; and Other affected relatives or exertional symptoms [1, 3]. Testing 122 referred patients was diagnostic in 13 (11%), across 10 genes including *ACADVL*, *CPT2*, *PYGM* and *RYR1*. All 13 had at least 2 features; 2 or more predicted a variant in 14% of cases, and none of the 30 patients with fewer carried one [3]. The median age at the first event, however, was 29 years, and prospective validation is awaited. Counting the features requires a CK repeated 8 weeks after the episode. Genetic testing should therefore be considered a first-tier test, using panels that span muscle energy metabolism, the dystrophies and excitation-contraction coupling, with reanalysis as the panels are updated and muscle biopsy after 8 weeks [1]. Prevention is then defect-specific, with medium-chain triglycerides plus long-chain fat restriction in VLCAD deficiency, carbohydrate before exercise in McArdle disease, avoidance of fasting and heat, and genetic counseling [1]. In children, recognizing these 6 features turns a recurring emergency into a diagnosable condition.

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ABS 23

ATYPICAL KAWASAKI DISEASE MASKED BY INFECTIOUS TRIGGER AND CORTICOSTEROID MISUSE: A CASE REPORT

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Kawasaki disease (KD) is an acute febrile vasculitis primarily affecting children and is associated with systemic inflammation, particularly in the coronary arteries. Timely diagnosis and treatment are crucial to prevent long-term cardiovascular complications, but atypical presentations can delay recognition. Atypical (or incomplete) KD represents a distinct clinical entity in which a prolonged fever (lasting more than 5 days) is associated with non-classical features that are insufficient to fulfill the criteria for classic KD and can mimic gastrointestinal, respiratory, or systemic infections. Therefore, maintaining a high index of suspicion for atypical KD is essential in children with persistent unexplained fever.

A 21-month-old boy with a history of nephrovascular hypertension secondary to renal ectopia, was admitted to the hospital with fatigue and dehydration following an upper respiratory tract infection empirically treated with a 2-day course of betamethasone, leading to a transient defervescence. Initial blood tests showed neutrophilic leukocytosis, elevated inflammatory markers, and increased transaminases/GGT. A respiratory multiplex PCR panel was positive for Rhinovirus, and EBV serology initially suggested a recent infection. After fever recurrence, a transient, widespread maculopapular rash developed. Due to the suspected infectious etiology, the patient was initially discharged despite the persistence of the fever.

Three days later, he was readmitted for hyporeactivity and persistent fever (6 consecutive days; 10 total days including the steroid-induced defervescence). Laboratory findings revealed newly developed thrombocytosis and negative EBV serology. Atypical KD was suspected, and the patient was promptly treated with intravenous immunoglobulin (IVIG) and anti-inflammatory doses

of acetylsalicylic acid. A baseline echocardiogram revealed a right coronary artery aneurysm (Z-score +3.6, small aneurysm) and diffuse dilatation of the left anterior descending artery. Recurrent fever 36 hours post-IVIG prompted a second IVIG dose, intravenous methylprednisolone, and antiplatelet aspirin, achieving sustained defervescence and normalization of inflammatory markers. During follow-up at a Tertiary Care Center, the IL-1 receptor antagonist Anakinra was added to the treatment regimen due to subsequent aneurysm progression (Z-score +6.7; medium aneurysm), which successfully led to the progressive regression of the coronary artery abnormalities.

In conclusion, atypical KD remains a diagnostic challenge. In this patient, a Rhinovirus co-infection, a false-positive EBV serology to which the hepatic involvement was erroneously misattributed, and empirical corticosteroid misuse obscured the true timeline of the fever, delaying diagnosis and driving IVIG resistance. Crucially, a transient therapeutic response to steroids must always be seen as a critical clinical red flag for underlying systemic or autoinflammatory conditions.

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ABS 24

CHILDHOOD-ONSET DEMYELINATING NEUROPATHY WITH PRESERVED AMBULATION EXPANDS THE *ADCY6* PHENOTYPE

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INTRODUCTION

ADCY6 encodes adenylyl cyclase type 6, which generates cyclic adenosine monophosphate (cAMP), a signal considered essential for Schwann cell myelination [1]. Biallelic variants cause lethal congenital contracture syndrome 8, which presents at birth with distal contractures, severe hypotonia, respiratory failure, impaired swallowing and areflexia [1, 2]. Five patients from three families have been reported, with deaths from within 80 days to 36 months and motor conduction velocities not exceeding 24 m/s [2]. No *ADCY6* phenotype compatible with independent walking has ever been described.

CASE REPORT

A male child born at term, with an uneventful perinatal course and milestones on time, walked independently at about 12 months. A motor disturbance with varus deformity of the right foot appeared at around 6 years. Nerve conduction studies at 8 years showed a primarily demyelinating motor and sensory polyneuropathy, with velocities of 32.1-38.9 m/s in the lower limb and 43.2 m/s in the upper limb. Right Achilles tendon lengthening at 10 years was followed by reported cessation of falls, although contractures persisted, dorsiflexion reaching neutral on the right and 10 degrees on the left. Examination at 11 years showed anterolateral leg weakness, absent lower limb and reduced upper limb reflexes, thenar hypotrophy and hand interossei graded MRC 3.5, with reported paresthesias, and reduced sensation over the lateral right foot at one of two examinations. The Hammersmith Functional Motor Scale Expanded score was 66/66 and the 6-minute walk distance 550 m, down from 577 m, with mild right steppage on exertion. A distal spinal muscular atrophy panel had returned a single maternally inherited allele of the recessive gene *SH3TC2*. Exome sequencing then identified two truncating *ADCY6* variants in compound heterozygosity, c.436del (p.Gln146ArgfsTer2) and c.862C>T (p.Gln288Ter).

CONCLUSIONS

In the family described by Laquérriere et al., the peripheral nerve lacked myelin, and *ADCY6* is essential for peripheral myelination, likely through cAMP [1]. Such a mechanism predicts a congenital failure to myelinate, yet early development here was normal. Conduction velocities of 32-43 m/s lie above the highest so far reported, 24 m/s [2], and ventilatory support has never been required, so nerve function is better preserved than in published cases. The genotype sharpens the contrast. Two truncating variants would be expected to abolish the protein, yet the phenotype is milder than in a patient whose second allele may retain residual activity [2]. Residual expression or modifying factors, rather than variant class alone, likely account for the difference. In undiagnosed arthrogryposis, targeted

sequencing identified the cause in 68 of 210 index patients, first-line exome sequencing in 55 of 90 [3]. A nondiagnostic panel, as here, does not close the workup. The mild end of *ADCY6*-related disease is compatible with walking at school age.

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ABS 25

SCHWARTZ-JAMPEL SYNDROME WITHOUT SKELETAL DYSPLASIA: A NOVEL *HSPG2* VARIANT

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INTRODUCTION

Schwartz-Jampel syndrome type 1 (SJS1), a rare recessive myotonic disorder, results from partial loss-of-function variants in *HSPG2*, which encodes perlecan, a proteoglycan that maintains cartilage integrity and regulates muscle excitability [1]. Permanent myotonic myopathy, skeletal dysplasia and a distinctive facial appearance define the phenotype [1]. Diagnosis relies mainly on the clinical picture, with molecular confirmation; management is symptomatic [1].

CASE REPORT

A 6-year-old boy was referred for muscle stiffness and dysmorphic facial features. He was born by elective cesarean section, with normal birth measurements, and walked independently at 15 months. Examination revealed blepharophimosis, a pursed mouth and the fixed, mask-like facies typical of the syndrome. Neurological assessment showed rigidity of all four limbs due to muscle hypercontraction and a stiff, jerky but independent gait. Postural transitions were preserved: he rose from the floor with one hand on the thigh, climbed stairs with alternating steps and squatted unassisted. Stature was normal; joint contractures and tendon retractions were absent, and no orthoses were required. Creatine kinase was mildly elevated (530 U/L; reference range 39-308 U/L), as was lactate dehydrogenase. Global developmental delay was documented at 4 years, with a Griffiths (GMDS-3) general development quotient below 50 and a Vineland-II adaptive behavior composite of 57. Treatment with mexiletine and carbamazepine was started. Videofluoroscopy performed for feeding difficulties incidentally showed marked tonsillar hypertrophy, and respiratory polygraphy documented numerous obstructive and mixed apneas and hypopneas, reported as severe. Imaging performed for severe constipation complicated by syncope showed fecal loading and sluggish peristalsis; feeding was restricted to a semiliquid diet. Genetic testing identified a novel homozygous splice-site variant in *HSPG2*, NM_005529.7:c.4740+5G>T.

CONCLUSIONS

SJS1 should be suspected when a mask-like facies with blepharophimosis and pursed lips is associated with muscle stiffness and impaired relaxation [1]. In the present patient, stature and joint mobility were clinically preserved, an uncommon but not unprecedented finding, since skeletal involvement is reported in almost all genetically confirmed cases [1], while the clinical burden was driven by

sleep-disordered breathing, intestinal dysmotility, feeding difficulties and developmental delay. Multidisciplinary follow-up is warranted and should include sleep studies [2] and cardiological assessment [1]; gastroenterological, nutritional and neuropsychological surveillance is suggested by the present case, as well as anesthetic evaluation for difficult airway management before any procedure [3]. Recognition of the facial phenotype opens the diagnostic pathway, genetic testing confirms the diagnosis, and coordinated management addresses these systemic complications.

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ABS 26

SIGUE NADANDO: A NICU ON THE OTHER SIDE OF THE MEDITERRANEAN SEA

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BACKGROUND

"*Sigue nadando*" – keep swimming – a well-known expression of perseverance that came to represent both my 6-month experience abroad and the experience of every parent walking through the doors of a Neonatal Intensive Care Unit (NICU). Leaving Sardinia, Italy, to train at Hospital Universitario 12 de Octubre in Madrid, Spain, meant crossing not only a sea, but also clinical, organizational and cultural boundaries.

DISCUSSION

As a fourth-year pediatric resident, I spent 6 months in the NICU of Hospital 12 de Octubre, a Spanish reference center for family-centered neonatal care. The unit is entirely based on family rooms, with individual and twin rooms designed to allow parents to remain continuously at their infant's bedside. The experience required me to rapidly adapt to new protocols, a broader case mix, a different healthcare system and, importantly, a new way of understanding the relationship between healthcare professionals, babies and families. What struck me most was that the single-family room model was not simply an architectural choice. Through the hospital's "*Cero Separación*" (i.e., zero separation) program, parents are considered active members of the care team, with the aim of avoiding separation between newborns and their families throughout hospitalization. Skin-to-skin contact, breastfeeding and progressive parental autonomy are actively promoted. Kangaroo Care may begin shortly after birth, even in very preterm or critically ill infants. Parents receive training in multiple aspects of neonatal care, while additional caregiver support is available when they cannot be present.

These practices are consistent with evidence suggesting that family-integrated care and skin-to-skin contact may improve breastfeeding, parental mental health, length of stay and selected neonatal outcomes. Studies of single-family rooms have also reported benefits in neurodevelopment and infection control. However, this model is not without challenges: physical separation may complicate staff communication and increase workload, while some families may experience isolation and reduced peer support. Its success therefore depends not only on room design, but on adequate staffing, communication systems and structured family-integration pathways.

CONCLUSION

Crossing the Mediterranean from Cagliari to Madrid taught me that neonatal care can look very different while pursuing the same goal: giving fragile newborns the best possible start. In the beginning, I was the one learning to "keep swimming" through an unfamiliar clinical and cultural environment. By the end, I realized that the same principle applied to neonatal care: progress requires moving beyond familiar models and embracing change. The experience showed me that "zero separation" is not merely about keeping families physically close, but about making them genuine partners in care. For me, this journey was therefore not only a training experience, but a lesson in adaptation, teamwork and a more human approach to neonatal medicine.

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ABS 27

BETWEEN *PAINS AU CHOCOLAT* & CRYING BABIES: MY EXPERIENCE IN PARIS

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INTRODUCTION

Training abroad during pediatric residency offers the opportunity not only to encounter different diseases and clinical settings, but also to experience different models of care and progressively develop professional autonomy. During my extra-network training in Paris, I worked at Necker-Enfants Malades and Robert-Debré in Pediatric Emergency Medicine and in the Neonatal Intensive Care Unit (NICU).

EXPERIENCE

One of the first striking differences compared with my previous training experience was the scale of activity. In 2025, Robert-Debré Pediatric Emergency Department recorded more than 91,000 visits, approximately 250 children per day. Such volumes require well-defined clinical pathways, efficient communication and standardization. The two hospitals offered different case-mixes: at Robert-Debré, sickle cell disease, infectious and tropical diseases, neurological emergencies, trauma and unaccompanied minors were frequent; at Necker, rare and genetic diseases and complex cardiac and pediatric surgical conditions predominated.

A central aspect of training was continuous discussion and reassessment through staff meetings, specialist input and debriefings. Learning by doing under supervision allowed me to assume progressive responsibility for patient assessment, the care of critically ill children, and practical procedures. In the Emergency Department, this meant performing lumbar punctures, sutures, immobilizations and regional analgesic procedures. In NICU, this included resuscitation, ventilation, intubation, surfactant administration and ultrasound.

Another lesson concerned standardization without losing personalized medicine. Clinical protocols, computerized prescribing and patient-specific dosing can reduce variability and errors while preserving clinical judgment. Dedicated pathways integrate medical and social assessment, safeguarding and follow-up.

Neonatal care further demonstrated how organization can influence outcomes. For instance, in transposition of the great arteries, prenatal diagnosis, planned delivery and immediate access to cardiology and urgent atrial septostomy show that sometimes expertise is not enough: proximity matters. Multidisciplinary care was equally essential in ethically complex situations, where collegial discussions on proportionality of care emphasized that major decisions should not be made in isolation.

This experience also led to my residency thesis on vaso-occlusive crises in sickle cell disease, showing how real-world data can help evaluate and improve clinical protocols.

CONCLUSION

Training abroad should teach us not to reproduce the same model everywhere, but to adapt evidence and acquired skills to available resources and local networks.

Ultimately, the three elements I bring home are autonomy, method and adaptability: taking responsibility, making care safer and more reproducible, and applying what has been learned in different clinical environments. The goal is not to reproduce the place where we trained, but to bring home what we learned.

ABS 28

TWO CONTINENTS, ONE JOURNEY: REFLECTIONS FROM SPAIN AND AFRICA

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BACKGROUND

International clinical experiences provide pediatric residents with unique opportunities to encounter different disease patterns, healthcare systems, and approaches to patient care. During my residency, I completed two training experiences in markedly different settings: the Therapeutic Nutrition Center of Ospedale Divina Providência in Luanda, Angola, and the Pediatric Emergency Department of Hospital Universitario Gregorio Marañón in Madrid, Spain.

EXPERIENCE

In Luanda, I worked in a center dedicated to the management of children with severe acute malnutrition. Clinical activities included nutritional assessment, recognition and treatment of complications, management of infectious diseases uncommon in high-income countries, and nutritional rehabilitation according to local and WHO protocols. Working in a resource-limited environment required careful clinical observation, prioritization of interventions, and adaptation to the resources available. More importantly, it provided direct insight into the impact of poverty, limited healthcare access, infectious diseases, and food insecurity on child health.

In Madrid, I joined the Pediatric Emergency Department and Observation Unit of a tertiary-level university hospital managing acute pediatric conditions from the neonatal period through adolescence. Under specialist supervision, I progressively gained autonomy in patient assessment, diagnostic reasoning, and emergency management. A distinctive aspect of this experience was the strong educational culture integrated into daily clinical practice. Regular simulation sessions, case discussions, and structured supervision promoted active learning and progressive responsibility. I also developed procedural skills, including wound suturing, casting techniques, and bedside lung ultrasound. The experience highlighted the value of multidisciplinary teamwork and a training model based on guided autonomy.

Despite their profound differences, both experiences shared a common principle: placing the child at the center of care and adapting clinical decisions to the context. In Angola, limited resources emphasized the importance of clinical skills, essential interventions, and prevention; in Spain, I experienced a system with advanced resources focused on structured pathways, constant training and progressive autonomy of the resident.

CONCLUSIONS

Experiencing pediatric medicine across two continents represented more than a professional opportunity: it was a journey in understanding how context shapes clinical practice. The combination of global child health and pediatric emergency medicine broadened my clinical perspective, strengthened my adaptability and clinical reasoning, and increased my awareness of healthcare inequalities. International experiences should be considered an important component of pediatric training, fostering clinical competence, cultural awareness, flexibility, and a broader vision of child health.

ABS 29

NUTRIACÇÃO EM ANGOLA: MY VOLUNTEER EXPERIENCE AGAINST MALNUTRITION

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BACKGROUND

Child malnutrition remains one of the main global health priorities of the WHO, with high prevalence rates in low resource settings. Within this context, the European Union-funded *NutriAcção* project, developed in collaboration with the Angolan Ministry of Health, aims to strengthen the diagnosis, management, and prevention of malnutrition.

OBJECTIVES

To describe the clinical and educational experience gained during a training period in Angola as a pediatric resident involved in the *NutriAcção* project, highlighting epidemiological, diagnostic, therapeutic, and preventive aspects of malnutrition in a resource-limited environment.

METHODS

Clinical activity was carried out in hospital and outpatient settings with limited diagnostic and therapeutic resources. Cases of acute and chronic malnutrition, infectious and metabolic complications were managed, alongside participation in community based prevention programs. Direct observation of technical, managerial, human, and systemic challenges provided insight into the complexity of malnutrition control in fragile contexts.

RESULTS

The experience allowed for a deeper understanding of the local epidemiology of malnutrition, improved clinical skills in assessment and management, and a clearer view of operational barriers affecting prevention strategies. Exposure to severe cases – often requiring intensive care that was not available – highlighted the impact of structural limitations on patient outcomes.

CONCLUSIONS

The period in Angola represented a significant professional and personal growth opportunity. Immersion in the local socio-economic reality revealed malnutrition not only as a clinical condition but as a phenomenon deeply rooted in social, cultural, and systemic dynamics. This experience reinforced the need for integrated and sustainable interventions to effectively combat child malnutrition.

ABS 30

CTRL+ALT+TREAT: PEDIATRICS IN A BELGIAN DIGITAL HOSPITAL

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INTRODUCTION

A training experience abroad represents an important opportunity for professional and personal growth. During my pediatric residency, I spent 18 months at Saint-Luc University Hospital in Brussels, Belgium, a setting characterized by high digitalization, strong interdisciplinary integration, and considerable resident autonomy.

TECHNOLOGY AND INNOVATION

Thanks to an integrated information system linked to the regional healthcare network, clinicians can rapidly access previous consultations, laboratory tests, and past hospital admissions, gaining a fuller, more objective clinical picture that does not rely solely on family-reported information. The same platform also supports safe prescribing, suggesting dosages and calculating the packages needed for treatment. Integrated AI tools add a further layer of support, enabling quick information retrieval and assisting diagnostic and therapeutic reasoning, without replacing clinical judgment, which remains the physician's responsibility. Another advantage is fast, cross-departmental communication through internal messaging, pagers, and dedicated discussion groups for complex cases, which also draw in the laboratory and pharmacy for the prompt sharing of microbiological results.

RESIDENT TRAINING IN A MULTIDISCIPLINARY ENVIRONMENT

This digital and collaborative environment shapes training directly. Over the residency, rotations across several Pediatric Departments – General Pediatrics, Infectious Diseases, and Pulmonology – offered a wide spectrum of cases. Because specialist support is always within immediate reach, residents can take the lead on even the most complex ones, turning each into a high-quality learning opportunity. Such cases included, in General Pediatrics, meningitis with septic shock, pneumonia with empyema requiring surgical decortication, severe salmonellosis, and mastoiditis with cerebral complications. Infectious Diseases brought a rare presentation of ear tuberculosis, together with the management of transplant candidates and mother-to-child follow-up for congenital infections. Pulmonology, finally, ranged from uncontrolled asthma and allergy desensitization therapies to complex lung disease in genetic syndromes and rare conditions such as cystic fibrosis and primary ciliary dyskinesia, alongside technical skills such as non-invasive ventilation. Throughout these rotations, autonomy and responsibility grew steadily, progressively strengthening clinical confidence and decision-making skills.

CONCLUSIONS

The Saint-Luc model shows how a digital hospital can strengthen residency training. Digital and AI tools proved valuable for information access and decision support, without replacing clinical judgment. Rather than distancing the resident from senior expertise, technology brought it closer – strengthening the learning relationship at the heart of residency, enabling greater autonomy, and laying a meaningful foundation for future practice.

ABS 31

MIDNIGHT IN PARIS: A JOURNEY THROUGH FRENCH PEDIATRIC EMERGENCY CARE

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INTRODUCTION

Working in a Pediatric Emergency Department involves dealing daily with a heterogeneous and unpredictable range of clinical presentations; each new patient encounter may lead to an entirely different diagnostic and therapeutic challenge. Working in a different country further requires adaptation to a different healthcare system, a new language, and distinct professional and cultural practices. When combined with the setting of a large city, characterized by high patient volumes, a multicultural population, and a broad age range, the experience becomes a continuous challenge.

TRAINING EXPERIENCE

My experience at Hôpital Armand Trousseau, a large Parisian Pediatric Emergency Department, highlighted several distinctive features of specialist training. Among these was the high degree of autonomy granted to trainees, always supported by supervision from experienced staff. This progressive approach – observe , perform under supervision, and gradually gain autonomy – enabled me to develop practical skills in managing increasingly complex procedures and clinical cases.

Particularly noteworthy was the strong emphasis on continuous medical education, through short teaching sessions incorporated into daily morning meetings, weekly sessions specifically dedicated to trainees, and simulation sessions involving the entire staff. This structured approach promotes the consolidation of clinical skills and greater consistency in the management of emergencies.

TROUSSEAU DE POCHE

Emergency medicine requires shared and easily accessible clinical pathways to ensure consistent patient management and contribute to reducing errors. In a setting characterized by a large, multidisciplinary team – including pediatricians, emergency physicians, general practitioners, and medical students – the availability of common protocols plays a central role.

To address this need, the "*Trousseau de poche*" (i.e., pocket Trousseau) was developed: a pocket-sized collection of the main protocols used within the Emergency Department. It was subsequently expanded into a free mobile application providing a digital version of the handbook, complemented by a wide range of practical tools like instant clinical calculators, data-plotting functions and diagnostic scores. Its content is regularly updated according to the latest scientific evidence. The intuitive design and comprehensive content have facilitated its adoption in other French hospitals.

CONCLUSIONS

An international clinical experience proved to be an important opportunity for both professional and personal growth. Interaction with colleagues from different European healthcare settings provided an opportunity to exchange clinical, organizational, ethical, and cultural perspectives, while also highlighting the common challenges faced in pediatric emergency medicine. Such an experience therefore represents not only an individual professional enrichment, but also an opportunity to identify, transfer, and adapt effective procedural and educational strategies to one's own clinical setting.

ABS 32

WHAT HAPPENS IN MEXICO… ENDS UP IN PEDIATRICS

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BACKGROUND

The saying goes that what happens in Mexico stays in Mexico – for a pediatric resident, it turned out to be the opposite: some experiences don't stay behind, they come home stitched into how you practice. My 3 months at Hospital Infantil de México Federico Gómez in Mexico City, Mexico, following training at the University of Cagliari, Italy, took me to a setting where infectious dermatologic disease remains a leading driver of pediatric morbidity, and where almost everything I saw came back with me as a souvenir I never unpacked.

EXPERIENCE

As a fourth-year pediatric resident, I spent 3 months in the Infectious Dermatologic Diseases Service of Hospital Infantil de México Federico Gómez, a national reference pediatric hospital. The volume and severity of disease were unlike anything in my home training network: staphylococcal and streptococcal skin and soft-tissue infections were seen daily, often more extensive or advanced at presentation than what I was used to managing; an active measles outbreak brought children with a disease largely eliminated from my previous clinical experience; and congenital syphilis, now rare in Italy, was a recurring diagnosis, frequently linked to incomplete antenatal care.

DISCUSSION

Working in a setting with limited resources reshaped how I approached each of these diseases. Diagnosis relied more heavily on clinical judgment, since laboratory confirmation and specialist imaging were not always immediately accessible, and treatment decisions had to account for delayed presentation, interrupted follow-up and barriers to consistent antibiotic or prenatal care. This mirrors what the literature describes: Mexico has documented a marked resurgence of congenital syphilis tied to antenatal screening and treatment gaps; measles re-emergence has become a stress test for surveillance and immunization systems already stretched thin; and staphylococcal-streptococcal pyodermas, while globally common, are disproportionately frequent and severe wherever crowding, limited hygiene infrastructure and delayed care converge. Managing this heavier burden required prioritizing pragmatic, resource-conscious decisions without lowering the standard of care and taught me to distinguish which diagnostic or therapeutic steps are essential versus ideal.

CONCLUSION

What happened in Mexico did not stay there – fittingly, for a rotation built around skin, it left its mark. The 3 months at Hospital Infantil de México Federico Gómez confronted me with a scale and severity of infectious dermatologic diseases I had

not previously managed directly, and the diagnostic instincts and resource-conscious judgment I built there are now simply part of how I practice – a skill I believe is essential for any physician working in an increasingly interconnected and unequal world.

ABS 33

AN UNEXPECTED CLUSTER OF PEDIATRIC RENAL ABSCESSES: INCREASING INCIDENCE OR IMPROVED DETECTION? A THREE-YEAR SINGLE-CENTER EXPERIENCE

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INTRODUCTION

Renal and perirenal abscesses are uncommon but potentially severe complications of pediatric acute pyelonephritis, usually from ascending infection or, less commonly, hematogenous spread [1-3]. Predisposing factors include vesicoureteral reflux (VUR), urinary tract malformations, immunosuppression, and delayed/inadequate treatment of pyelonephritis [1, 3]. Presentation is often nonspecific – fever, flank pain, vomiting, and elevated inflammatory markers – hindering early diagnosis without cross-sectional imaging [1, 3]. Contrast-enhanced ultrasonography and CT are key diagnostic tools; management ranges from IV antibiotics alone to percutaneous drainage for larger collections [1, 2].

CASE SERIES

Between 2024-2026, we retrospectively reviewed 5 children (3F/2M; aged 5-18) with renal/perirenal abscesses at our Department of Pediatrics, ARNAS Brotzu Hospital (Cagliari, Italy). All presented with fever, flank pain, and elevated CRP/procalcitonin. Two had predisposing urological anomalies (previously corrected VUR and a duplex system) [3]. Blood cultures were mostly negative; urine cultures grew *Escherichia coli* and *Klebsiella pneumoniae*, consistent with the literature [1]. Ultrasound and contrast-enhanced CT showed hypovascular, wedge-shaped areas evolving into rounded, avascular collections, with occasional perirenal extension [2]. All received prolonged broad-spectrum IV antibiotics (ceftriaxone, meropenem, vancomycin/teicoplanin). Two patients with larger collections required percutaneous drainage, with progressive resolution on ultrasound. Follow-up scintigraphy showed preserved or mildly reduced renal function, without significant scarring in most cases. All achieved clinical and biochemical recovery, with no recurrences.

DISCUSSION

Our findings support the literature: pediatric renal abscesses, though rare, warrant a high index of suspicion in pyelonephritis unresponsive to standard therapy within 72 hours [1]. Early cross-sectional imaging is essential to differentiate uncomplicated pyelonephritis from abscess formation and guide treatment [2]. Prolonged antibiotic therapy is effective for small abscesses, while percutaneous drainage may be considered for larger collections or treatment failure [1]. Underlying urinary tract anomalies warrant thorough urological evaluation at diagnosis and follow-up, given their role in recurrence risk [3]. A multidisciplinary approach – integrating pediatric nephrology, radiology, and infectious disease expertise – is key to optimizing outcomes and preserving long-term renal function [1, 2].

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ABS 34

WHEN FICTION BECOMES REALITY: AN INFANT OVER THE LINE

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INTRODUCTION

Pediatric cocaine intoxication is an overlooked cause of severe symptoms in infants, frequently mimicking sepsis, heart disease, or primary respiratory illness. Prenatal exposure, breastfeeding, passive crack-smoke inhalation, or accidental ingestion in houses where narcotics are used or stored can all cause poisoning. Because clinical signs may overlap

with more common pediatric emergencies and history is typically poor or missing, this diagnosis is often overlooked. Cocaine inhibits catecholamine reuptake, resulting in sympathomimetic toxicity, which appears as tachycardia, tachypnea, tremors, mydriasis, hyperthermia, and, in severe cases, seizures, arrhythmias, and cardiopulmonary failure. Urine toxicological screening is a simple bedside option that might redirect an otherwise misleading diagnostic work-up, despite possible cross-reactivity.

CASE REPORT

After a reported respiratory arrest during bathing, a 50-day-old male child was taken alone by helicopter to ARNAS Brotzu's Pediatric Emergency Department (Cagliari, Italy). Before emergency medical personnel arrived, the grandmother, who was the only person at home, performed rescue mouth-to-mouth ventilation with resolution of cyanosis. He was tachypneic (respiratory rate 60-66/min) upon arrival, with nasal flaring, subcostal and intercostal retractions, and a normal oxygen saturation (97-98% in ambient air). He had mottled, cold skin, generalized tremors, tachycardia (170-175 bpm), irritability, and normal pupil reflex. The remaining neurological assessment was unremarkable. Blood gas showed severe metabolic acidosis (pH 6.99, lactate 12 mmol/L, base excess -18.8 mmol/L) with hyperglycemia. Chest radiograph showed bilateral interstitial-alveolar opacities, and a cardiological examination, including echocardiography, ruled out structural or functional heart disease. A quick multipanel urine immunoassay drug test was performed due to the inconsistent history, the lack of caregivers, and a clinical picture suggestive of a sympathomimetic syndrome. The test came back positive for cocaine. Following admission, the results were confirmed through blood and urine testing. After being admitted to the hospital and receiving intensive intravenous hydration to encourage the drug's renal clearance, the baby's condition gradually improved with no further issues.

CONCLUSIONS

This case highlights how infant cocaine intoxication can present as an unexplained life-threatening condition, with significant tachypnea despite deceptively reassuring oxygen saturation. When a history is incomplete or unreliable, as is often the case with infants brought in by third parties, a low threshold for toxicological screening is crucial. Sometimes reality can be more dramatic than fiction, and a drug that was never meant for a child may be the real reason for an infant who is truly over the line.

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ABS 35

JUVENILE MYASTHENIA GRAVIS: A CASE REPORT OF GENERALIZED SUBACUTE ONSET

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BACKGROUND

Juvenile myasthenia gravis (JMG) is a rare autoimmune disorder of the neuromuscular junction characterized by fluctuating muscle weakness and fatigability [1]. Its prevalence in the pediatric population is estimated at approximately 25-38 cases per million, confirming its rarity during childhood and adolescence [2]. Although uncommon in childhood, early recognition is essential to prevent progression and potentially severe complications.

CASE REPORT

We report the case of an 11-year-old girl evaluated at our department because of an approximately 1-month history of fluctuating neuromuscular symptoms. Clinical presentation included generalized fatigue, more pronounced in the evening and after physical exertion, bilateral ptosis, nasal speech, occasional dysphagia predominantly for solid foods, and intermittent proximal weakness of the upper limbs. Neurological examination confirmed fatigability of the upper limbs appearing after sustained effort. The clinical presentation was suggestive of a neuromuscular junction disorder. Serum testing showed strongly positive anti-acetylcholine receptor antibodies (13.50 nmol/L), while anti-MuSK antibodies were negative. Repetitive nerve stimulation on electromyography demonstrated a pathological decremental response, supporting the diagnosis of JMG. According to the MGFA classification, the patient was classified as class IIA. Chest CT excluded the presence of thymoma.

RESULTS

Treatment with pyridostigmine bromide (120 mg/day) led to an improvement in muscle strength and a reduction of fatigability. Although the initial response was favorable, the bulbar signs recurred, and prednisone (1 mg/kg/day) was subsequently added to the treatment. The clinical course was monitored using functional myasthenia scales. The patient has been hospitalized for a cycle of plasma exchange that determined a rapid global improvement. However the efficacy was transient and so two additional cycles of plasma exchange were performed. The biological therapy (eculizumab) will be started in the next weeks.

CONCLUSIONS

This case highlights the importance of recognizing the fluctuating nature of neuromuscular symptoms in children and adolescents. Ptosis, fatigability, dysphonia and dysphagia, particularly when worsening after exertion and later in the day, should raise suspicion of JMG. The combination of clinical assessment, specific antibody testing and neurophysiological evaluation is crucial for establishing the diagnosis and guiding treatment. Early diagnosis and a multidisciplinary approach may improve disease control and prevent progression to more severe manifestations [2, 3]. The biological therapy, approved for pediatric onset JMG, represents the actual gold standard in the disease.

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ABS 36

A VARIANT OF UNCERTAIN SIGNIFICANCE WORTH TELLING

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BACKGROUND

Transfer RNA (tRNA) charging – the covalent attachment of a specific amino acid to its cognate tRNA – depends on a dedicated enzyme family, the aminoacyl-tRNA synthetases; cytosolic and mitochondrial sets sustain the translational machineries of the human cell. Pathogenic variants in the mitochondrial isoforms cause a heterogeneous group of disorders, often multisystemic [1, 2]. *LARS2*, at 3p21.3, encodes mitochondrial leucyl-tRNA synthetase; transcript levels are highest in organs with greatest oxidative demand, namely brain, skeletal muscle and cochlea. It has been linked to Perrault syndrome type 4 [3], a neonatal presentation with lactic acidosis, anemia and early death.

CASE PRESENTATION

A female patient came to attention at 18 months of age for failure to thrive, developmental delay and dyskinesia. She was born at 37 weeks to first-cousin parents by emergency caesarean section for fetal distress; prenatal ultrasonography had already documented microcephaly. Following admission to neonatal intensive care, examination showed dysmorphic features and microcephaly; brain MRI demonstrated a simplified gyral pattern. Trios clinical exome sequencing had identified a *de novo* likely pathogenic *SLITRK1* variant (c.829C>T), a *de novo* variant of uncertain significance in *CRYGB* (c.383dup) and multiple regions of loss of heterozygosity; none of these findings accounted for the phenotype as a whole. Feeding difficulties and delayed gastro-duodenal transit led to gastrostomy at 16 months. Fever and infections usually precipitated the neurological and nutritional picture. By 24 months, psychomotor delay was severe; walking was acquired at 30 months, expressive language was absent and the dyskinesia persisted. Given the consanguinity and the lack of recessive causative variants, the genetic laboratory was addressed to update the patient clinical features. The reanalysis revealed the homozygous *LARS2* variant NM_015340:c.1813C>T (p.Arg605Cys), currently classified as of uncertain significance.

DISCUSSION

The described picture possibly extends the *LARS2* spectrum beyond the known phenotype, combining congenital microcephaly, dysmorphism, failure to thrive, gastrointestinal issues and dyskinesia. These clinical features and the episodic worsening in fever resemble other tRNAs typical phenotype. Case reports of specific aminoacid supplementation in these conditions have shown global clinical

improvement. In conclusion, the case illustrates the yield of periodic exome reanalysis in selected cases, and how difficult but needed causal attribution can be, especially in current times of new therapeutic strategies for genetic disease. Functional studies of p.Arg605Cys remain necessary before the association can be established.

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ABS 37

METABOLOMICS AND POSTPARTUM DEPRESSION: THE MATERNA STUDY

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Pregnancy and the postpartum period are characterized by significant physiological, hormonal, and metabolic changes, accompanied by substantial psychological and emotional adaptations [1]. In this context, the study of the urinary metabolome may help explore potential associations between changes in metabolic profiles and mood-related manifestations during the postpartum period [2, 3]. As part of the ongoing MATERNA metabolomics study, 80 Italian women with singleton pregnancies and no relevant concomitant medical conditions will be recruited from 36 weeks of gestation onward.

Urine samples will be collected during the prenatal period before delivery (T0) and during the first month postpartum (T1), combining metabolomic analysis with the assessment of depressive and anxiety symptoms using the Edinburgh Postnatal Depression Scale (EPDS) and the State-Trait Anxiety Inventory (STAI).

Potentially relevant clinical and medical history information will also be collected, including dietary habits, physical activity, medication, antibiotic and probiotic use, smoking habits, and alcohol consumption. The aim of the study is to explore the potential association between the maternal urinary metabolomic profile and the presence of depressive and/or anxiety symptoms during the postpartum period, contributing to a better understanding of the potential metabolic correlates of postpartum mood alterations.

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ABS 38

URINARY METABOLOMICS AND AUTISM SPECTRUM DISORDERS: AN OCEAN (OF INFORMATION) IN A DROP (OF URINE)

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INTRODUCTION

Autism spectrum disorder (ASD) has a multifactorial etiology involving both genetic and epigenetic factors. Metabolomics, which can detect both endogenous metabolites and those resulting from environmental exposures, provides a unique window into the interplay between these factors.

AIM

In the absence of validated biological biomarkers for diagnosis, severity stratification, and monitoring of interventions in ASD, the aim of this study was to conduct a narrative review of urinary metabolites found to be altered in children with ASD.

METHODS
Twenty-one primary urinary metabolomics studies comparing children with ASD and controls were selected. A total of 166 significantly altered metabolites ($p < 0.05$) were extracted and classified according to the degree of replication across studies: ≥ 3 studies, 2 studies, and 1 study, respectively.
RESULTS
The tryptophan-kynurenine pathway was the most consistently replicated metabolic pathway, showing a shift away from serotonin synthesis toward neuroexcitotoxic metabolites. Within the gut microbiota axis, p-cresol and indoxyl sulfate emerged as robust markers of dysbiosis. With regard to oxidative stress, reduced homocysteine levels suggested impaired methylation. Within catecholamine-related pathways, reduced levels of GABA, norepinephrine, and epinephrine were observed, consistent with catecholaminergic hypofunction. Purine metabolism indicated accelerated catabolism, consistent with the "cell danger response" hypothesis, while the Krebs cycle showed reductions in 10 intermediates, suggesting widespread mitochondrial dysfunction. Finally, within the urea cycle, creatine and creatinine, among the most consistently replicated metabolites, showed a marked geographical discrepancy, with increased levels in European cohorts and decreased levels in Asian cohorts.
CONCLUSIONS
The overall picture is heterogeneous but converges on several recurrent biological axes: gut dysbiosis, mitochondrial and energy dysfunction, and an imbalance in monoaminergic neurotransmission. Despite geographical and methodological variability, these patterns support the potential of urinary metabolomics as a tool for phenotypic stratification and as a future source of non-invasive biomarkers in ASD.

ABS 39

HYPOXIC-ISCHEMIC ENCEPHALOPATHY: CHECK THE ADRENALS

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INTRODUCTION
Neonatal adrenal hemorrhage (NAH) is an uncommon condition associated with perinatal asphyxia, hypoxia, hypotension, coagulopathy, and traumatic delivery. Therapeutic hypothermia (TH) is the standard neuroprotective treatment for term and near-term infants with moderate-to-severe hypoxic-ischemic encephalopathy (HIE). We aimed to describe the incidence of NAH among asphyxiated neonates undergoing TH in our NICU and compare our findings with published data [1].
CASE REPORT
We retrospectively reviewed neonates treated with TH over a 3-year period. Perinatal asphyxia was defined by evidence of hypoxic-ischemic insult, including severe metabolic acidosis (pH ≤ 7.0 or base deficit ≥ 12 mmol/L), low Apgar score or prolonged resuscitation, with moderate-to-severe HIE. Eligibility for TH required gestational age ≥ 35 weeks, initiation within 6 hours of birth, and moderate-to-severe HIE. TH was performed at 33-34°C for 72 hours. Among 124 asphyxiated neonates identified during the study period, 99 met criteria for TH. Four of these 99 infants developed NAH, corresponding to an incidence of 4.0%. All 4 hemorrhages were right-sided, consistent with the right-sided predominance reported in the literature. NAH was diagnosed by abdominal ultrasonography. All infants underwent serial ultrasound and laboratory monitoring [1, 2].
CONCLUSIONS
In our cohort, NAH occurred in 4.04% of neonates undergoing TH for HIE. This incidence exceeds that reported in general neonatal populations, where NAH is approximately 0.3-0.4%. Perinatal hypoxia may increase adrenal vulnerability through hemodynamic instability, ischemia-reperfusion injury, and vascular stress. The exclusive right-sided localization observed in our 4 cases is in line with published data, reporting a predominant involvement of the right adrenal gland. Although direct comparison with cohorts of cooled asphyxiated infants is limited by the scarcity of dedicated studies, our findings suggest that NAH may be clinically underrecognized in this population. Abdominal ultrasonography should be considered in selected neonates with HIE, particularly with unexplained anemia, hypotension or persistent jaundice [2].
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ABS 40

ALLERGY OR CELIAC DISEASE? A CASE OF SEMOLINA FPIES IN AN INFANT

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INTRODUCTION

Food protein-induced enterocolitis syndrome (FPIES) is a form of non-IgE-mediated food allergy characterized by repeated vomiting within 1-4 hours of ingestion of the trigger food, sometimes associated with pallor and lethargy, with spontaneous resolution of symptoms within a few hours [1]. The diagnosis is predominantly clinical and is based on the presence of a major criterion and at least 3 minor criteria. However, it may be delayed due to overlap with other gastrointestinal conditions common in pediatric age, such as acute gastroenteritis [2]. While cow's milk and soy are the most common triggers, cereals containing gluten may also be responsible. We present a case of semolina FPIES that arose during weaning, with initial diagnostic doubt.

PRESENTATION OF THE CASE

A 7-month-old infant was coming to our observation. Two months earlier he had presented repeated episodes of vomiting after the introduction of gluten-containing pasta, requiring hospitalization.

On this occasion, skin prick tests were negative; stool testing for adenovirus and rotavirus was also negative. Blood tests showed a questionable positivity for anti-gliadin IgG. The introduction of gluten into the diet was therefore suspended. Upon subsequent administration of semolina, at the direction of the Caregiver, numerous episodes of vomiting appeared again within a few hours, even after taking fluids, so the patient was admitted to the Emergency Room requiring hospitalization and intravenous hydration. Blood tests (including PCR), infectious diseases and allergies were also normal on that occasion. IgA and IgG anti-transglutaminase antibodies and IgG EMA were negative. After an initial clinical improvement, a new administration of semolina was attempted in the department, with the recurrence of symptoms. In light of the repetitiveness of episodes after ingestion of gluten-containing cereals, the rapid onset of symptoms, and the absence of IgE-specific sensitization, excluding infectious causes, a clinical diagnosis of FPIES was made.

At the age of 14 months, the patient performed oral provocation tests with semolina in a hospital setting, with negative results and documented acquisition of tolerance.

CONCLUSIONS

The described case meets the FPIES diagnostic clinical criteria and highlights how the clinical presentation may initially mimic other pediatric gastrointestinal conditions, delaying correct diagnostic framing. Accurate temporal history and reproducibility of symptoms after exposure to the suspected food are key elements for early diagnosis.

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ABS 41

A CASE OF TRISOMY 13 WITH A CARDIAC-NEGATIVE PHENOTYPE: DIAGNOSTIC AND MANAGEMENT IMPLICATIONS

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INTRODUCTION

Trisomy 13 (Patau syndrome) is a rare aneuploidy (1/10,000-20,000 live births), with antenatal mor-

tality exceeding 95% and a frequently poor postnatal prognosis [1-3]. The typical clinical picture includes midline craniofacial anomalies, congenital heart defects (present in 80% of cases, predominantly VSD 28%, PDA 32%, ASD 27%, tetralogy of Fallot 9%, often associated with dextrocardia) [1], postaxial polydactyly (50%), scalp aplasia cutis congenita (30-50%, with an underlying bone defect in 15-20% of cases) [3], cleft lip/palate (50-80%), and microcephaly, with consistently severe neurological impairment [1]. The 2025 AAP guidelines recommend counseling that presents the full spectrum of therapeutic options to families, as trisomy 13 is not a uniformly lethal condition [2].

CASE REPORT

We describe the case of a neonate born at 38 weeks + 4 days of gestation, in whom the morphology ultrasound performed at 25 weeks had documented cleft lip/palate and cerebellar hypoplasia. The parents had chosen not to undergo further diagnostic testing during pregnancy. At birth, the infant required cardiopulmonary resuscitation and intubation. Physical examination revealed: aplasia cutis congenita with a calvarial bone defect over the posterior fontanelle region, trigonocephaly, right-sided cleft lip/palate, bilateral arthrogryposis, a supernumerary digit, and bilateral clubfoot. Echocardiography showed no structural cardiac defects, and abdominal ultrasound was normal; in contrast to the prenatal finding, postnatal cranial ultrasound did not confirm the presence of cerebellar hypoplasia. Postnatal karyotype confirmed full trisomy 13.

CONCLUSIONS

The phenotype is fully consistent with trisomy 13, as confirmed cytogenetically. The aplasia cutis with an underlying bone defect falls within the expected range (30-50%, with bone involvement in 15-20% of cases) [3], whereas the absence of congenital heart disease – present in 80% of cases, typically VSD, PDA, or ASD – is an atypical finding [1]. The lack of confirmation of the cerebellar hypoplasia underscores the prognostic limitations of fetal ultrasound. From a management standpoint, early cytogenetic confirmation allowed for timely counseling: consistent with the 2025 AAP recommendations, a multidisciplinary pathway was activated to share with the parents the full range of options, from active treatment to comfort care, in keeping with the principle of proportionality of care, bearing in mind that 5-25% of neonates survive beyond the first year of life with active treatment [2].

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ABS 42

NOT ALL PETECHIAE ARE A PTI: A CASE OF PEDIATRIC SUPERWARFARIN POISONING

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INTRODUCTION

Superwarfarin rodenticides were developed in response to the emergence of warfarin resistance in rodents. These compounds exhibit approximately 100-fold higher anticoagulant potency than warfarin and a prolonged plasma half-life, ranging from 20 to 60 days. Specifically, superwarfarins (e.g. brodifacoum) act by inhibiting the enzyme vitamin K-epoxide reductase, blocking the recycling of vitamin K and the synthesis of factors II, VII, IX and X. While coagulopathy is the main effect, in some cases transient thrombocytopenia may be observed, often due to peripheral consumption during active bleeding or to mechanisms of cellular oxidative damage. Accidental anticoagulant rodenticide poisoning in children is a rare but potentially serious occurrence. Clinical presentation can simulate much more frequent hematological pathologies, making the diagnostic path initially misleading.

CLINICAL CASE

We present the case of a 10-year-old patient who arrived at our Emergency Department for the onset of diffuse ecchymosis, petechiae, and macrohematuria, in the absence of recent trauma. In anamnesis, the mother reported a recent episode of upper respiratory tract infection, which had

occurred about a week earlier. Blood tests at entry showed severe thrombocytopenia (PLT 5,000/mcL), associated with prolongation of PT (INR > 10) and aPTT (ratio 1.26). Therefore, admission to the Pediatrics Ward was ordered and in the suspicion of an immune thrombocytopenic purpura (PTI), intravenous immunoglobulins (Ig) were requested. Arriving in the Pediatrics Ward, awaiting the arrival of the Ig, on further history taking, the mother suddenly remembered that in the previous days rodenticide baits ("Broditop", containing brodifacoum 0.005%) had been placed in the garden for rodent control and at that point the child, questioned again, confessed to having eaten some "colored candies" found outside the house. Therefore, in agreement with the Poison Control Center, the little one initiated intravenous vitamin K treatment 3 times a day and laboratory clinical monitoring. Coagulative factor analysis confirmed a deficiency of vitamin K-dependent factors (FIX 10%, FX 5%, FII 5%, FVII < 1%), with normal FV and FVIII levels. At the subsequent 24-hour laboratory re-evaluation, an improvement in PT and aPTT values was already evident. Based on these data, the child was discharged home on the fifth day of hospitalization with a diagnosis of "anticoagulant rodenticide poisoning", with home therapy with vitamin K orally for 3 months and clinical and laboratory follow-up, achieving complete correction of the coagulopathy.

CONCLUSIONS

This case highlights the importance of considering coumarin anticoagulant poisoning as a cause of coagulopathy in patients presenting with PT prolongation. Accidental ingestion of rodenticide should always be suspected and investigated in the presence of sudden bleeding diathesis associated with coagulation alterations in pediatric age, in the absence of known liver disease. The presence of thrombocytopenia, although less common than INR prolongation, does not exclude the diagnosis and may reflect the severity of systemic bleeding. Vitamin K1 therapy should be prolonged for several weeks due to the extremely long half-life of superwarfarins.

ABS 43

SECONDARY SHOULDER RECONSTRUCTION IN BRACHIAL PLEXUS BIRTH PALSY: AN AGE- AND DEFORMITY-BASED TREATMENT ALGORITHM

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Brachial plexus birth palsy may result in progressive shoulder dysfunction despite neurological recovery. Persistent imbalance between relatively preserved internal rotators and weakened abductors/external rotators promotes internal-rotation contracture, posterior humeral head translation and progressive glenohumeral dysplasia [1]. This review aimed to synthesize the evidence on non-microsurgical secondary shoulder reconstruction and to propose a practical treatment algorithm based on age, passive motion and joint morphology.

A targeted narrative review of the orthopedic, shoulder and upper-limb reconstructive literature was performed, focusing on soft-tissue release, tendon transfers, humeral osteotomy and glenoid procedures. Treatment was stratified according to passive external rotation, active motor deficit, glenohumeral congruity and remaining remodeling potential.

In infants and young children with a concentric joint, physiotherapy and surveillance remain first-line treatment. Progressive loss of passive external rotation or posterior subluxation should prompt imaging and consideration of contracture release with concentric reduction, as early restoration of alignment may improve joint remodeling [2]. In children with a concentric or reducible shoulder but persistent loss of active external rotation and abduction, tendon transfer is indicated to restore dynamic balance; release should be added when fixed contracture is present. Transfers to the rotator cuff improve Mallet scores, abduction and external rotation, with evidence supporting latissimus dorsi/teres major transfer as the most established option [3]. In older children with advanced, fixed dysplasia and limited remodeling potential, humeral derotational osteotomy can reposition the available arc of motion, although it does not correct the underlying glenoid deformity. Glenoid osteotomy should be reserved for selected severe but reconstructible deformities.

Secondary reconstruction should therefore progress from joint preservation and remodeling in early childhood, to dynamic muscle rebalancing in reducible deformity, and finally to functional repositioning when structural changes become fixed. Chronological age alone is insufficient: passive mo-

tion, reducibility and glenohumeral morphology should guide treatment selection.

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ABS 44

WHEN EXERCISE REVEALS ALLERGY: A CASE OF PEACH sFDEIA

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INTRODUCTION

Food-dependent exercise anaphylaxis (FDEIA) is a summation anaphylaxis, which occurs following exposure to multiple triggers (exercise and ingestion of a food), which individually do not induce anaphylaxis. There are two forms: food-specific (sFDEIA), the most common, which occurs following the intake of a food to which the patient is sensitized; and non-food-specific (nsFDEIA), which can occur with any food, to which no sensitization is observed. Among the most commonly implicated foods are wheat, seafood, seeds, cereals, nuts, milk, and eggs.

The pathogenic mechanisms of FDEIA are not yet fully understood. There would likely be an exercise-induced increase in intestinal permeability to trigger foods underlying this.

PRESENTATION OF THE CASE

G., 12 years old, came to our observation. Mother with bronchial asthma. Two weeks earlier, the patient had been evaluated in our Emergency Department for an episode of diffuse urticaria and vomiting with crampy abdominal pain that appeared a few minutes after eating peach with the peel. The girl had eaten peach previously without any problems. At the time of the episode, she had just finished her artistic gymnastics training. In Emergency Room she was administered antihistamine and intramuscular steroid with complete resolution of the symptoms. Since then she had been on a peach-free diet. At the Allergy Clinic of our Clinical Unit we performed skin prick tests with positive results for *Dermatophagoides farinae* (5 mm), *Dermatophagoides pteronyssinus* (4 mm), peach (6 mm), histamine (4 mm). We then proceeded to evaluate the specific IgE for peach proteins, highlighting positivity for peach-specific IgE (16.20 kU/L), Pru p 3 (12.6 kU_A/L) and Pru p 7 (13.2 kU_A/L). In light of peach sensitization, positivity for Pru p 3 and Pru p 7, food tolerance outside of exercise, and medical history, it was concluded for sFDEIA with prescription of an adrenaline autoinjector.

CONCLUSIONS

G.'s case represents a classic example of sFDEIA in which exercise acts as a trigger in triggering anaphylaxis following ingestion of a food to which the patient is sensitized. In the absence of physical activity, ingestion of the trigger food does not cause adverse reactions. This is not pure "exercise-induced anaphylaxis", as G. had been practicing artistic gymnastics for years without ever having experienced any episodes of anaphylaxis before.

ABS 45

PALMO-PLANTAR ERYTHRODYSESTHESIA – HAND-FOOT SYNDROME

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DEFINITION

Palmo-plantar erythrodysesthesia is caused by excessive fragility of the skin of the hands and feet. This skin weakness causes the production of patches of redness, dry skin, local edema, and sometimes small blisters. This causes painful sensations, tingling and

scratching. It can manifest itself in all eras of life, spontaneously or caused by pharmacological and oncological therapies.

TYPOLOGIES

We distinguish spontaneous or idiopathic forms that, very rarely, have also been identified in children.

At present, no genetic or autoimmune predisposition has been detected.

Many cases appear to be attributable to chemotherapy drugs, oncological therapies, and pharmacological treatments, such as "targeted therapy".

CHEMOTHERAPY PLANTAR PALMIC ERYTHRODYSESTHESIA

It is generated by the accumulation of the drug in the hands and feet. It can be limited to tingling in the hands and feet, but in more severe cases it can be accompanied by severe pain, red and dry skin, hyperkeratosis and very annoying fissures.

PLANTAR PALM ERYTHRODYSESTHESIA FROM "TARGETED THERAPY"

It may appear after the second or fourth week of therapy in the form of painful lesions on a scaly skin tissue, edematous, dehydrated, itchy.

CONSIDERATIONS

It is a not very frequent pathology, not easily classified, and is often diagnosed with excessive delay. Precise diagnostic investigations are not known for easy diagnosis. Therefore, it is almost always the experience and skill of the caregiver that indicates a precise diagnostic hypothesis. It can also occur at a young age, especially in connection with possible and prolonged drug therapies. In some cases of adult subjects, a possible familiarity has been hypothesized. Therefore, a careful patient history and any genetic investigations are always essential.

WHAT REMEDIES FOR HAND AND FOOT SYNDROME?

The disease can significantly invalidate people's lives, and especially for those patients undergoing cancer therapy it can reduce the possibility of walking. In some subjects it is so disabling that it almost completely prevents efficient movement. Consequently, recommendation for patients includes avoiding prolonged walking, utilizing compressive garments and stockings, and preventing friction, pressure, or excoriation on the hands and feet. A peculiar and scrupulous skin hygiene is very important, using delicate detergents that can preserve the skin hydrolipidic film. To calm the itching, redness and burning, the patient needs to use a protective and soothing cream, spreading it very gently on the lesions. For thickening, flaking and cracking, the patient should apply an anti-flaking cream topically.

CONCLUSIONS

This is an old pathology that presents itself subtly with multifaceted aspects, where one must be careful of the diagnosis and for the treatment.

ABS 46

CURRENT TRENDS IN THE CIRCULATION AND SPREAD OF WEST NILE AND USUTU VIRUSES: ONE YEAR LATER

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INTRODUCTION

One year after our previous report on West Nile virus (WNV) and Usutu virus (USUV) [1], we provide an update on their circulation in Italy, based on data from the national surveillance system coordinated by the Italian National Institute of Health (*Istituto Superiore di Sanità*, ISS).

EPIDEMIOLOGICAL UPDATE

According to ISS Report No. 2/2026, updated to August 5, 2026, 141 confirmed human WNV infections had been reported in Italy: 64 neuroinvasive cases, 27 asymptomatic infections detected among blood donors, and 50 cases of West Nile fever. Six deaths were reported among confirmed cases. WNV circulation showed a wide geographical distribution, involving 16 Italian regions. Three human cases of USUV infection were also reported, all in Lombardy [2].

Surveillance data for other arboviral infections provide additional evidence of the importance of travel-associated infections. From January 1 to July 31, 2026, 192 confirmed dengue cases, 17 chikungunya cases, and 3 Zika virus infections were reported in Italy. At that time, all were associated with travel abroad and no deaths were recorded [3].

CONCLUSIONS

The 2026 surveillance data confirm the continuing public health relevance of arboviruses in Italy. WNV and USUV are maintained through enzootic cycles involving birds and *Culex* mosquitoes, whereas dengue, chikungunya, and Zika virus infections reported during the period considered were travel-associated. These findings highlight the importance of integrated human, veterinary, and entomological surveillance, blood-donor screening, early diag-

nosis, and appropriate information for travelers. Continuous surveillance and accurate attribution of epidemiological data to national health authorities are essential for monitoring changes in arbovirus circulation and guiding preventive measures.

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ABS 47

GRANULOMATOSIS WITH POLYANGIITIS (GPA)

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INTRODUCTION

Granulomatosis with polyangiitis (GPA), formerly known as Wegener's granulomatosis, is a systemic necrotizing vasculitis predominantly affecting small- and medium-sized vessels. It belongs to the spectrum of antineutrophil cytoplasmic antibody (ANCA)-associated vasculitides, together with microscopic polyangiitis (MPA) and eosinophilic granulomatosis with polyangiitis (EGPA) [1]. GPA is characterized by granulomatous inflammation, particularly involving the respiratory tract, and necrotizing vasculitis that may affect multiple organs.

CLINICAL FEATURES AND DIAGNOSIS

Upper respiratory tract involvement is frequent and may present with persistent rhinosinusitis, nasal crusting, epistaxis, otitis media, or, in advanced disease, cartilaginous damage. Pulmonary manifestations include nodules, infiltrates, cavitation and, less commonly, diffuse alveolar hemorrhage. Renal disease typically presents as pauci-immune necrotizing glomerulonephritis and may progress rapidly if untreated. Constitutional symptoms, cutaneous lesions, peripheral neuropathy, musculoskeletal manifestations and ocular involvement may also occur.

ANCA testing represents an important component of the diagnostic work-up. GPA is strongly associated with cytoplasmic ANCA directed against proteinase 3 (PR3-ANCA), although ANCA negativity does not exclude the disease. The 2022 ACR/EULAR classification criteria integrate clinical manifestations, ANCA specificity, imaging findings and histopathological features [2]. Laboratory evaluation should include complete blood count, renal function, inflammatory markers and urinalysis. Chest and sinonasal imaging should be guided by the clinical presentation. Whenever feasible, biopsy of an affected organ can provide important diagnostic evidence, particularly by demonstrating granulomatous inflammation or pauci-immune glomerulonephritis.

TREATMENT AND CONCLUSIONS

Early recognition is essential because untreated GPA may result in irreversible organ damage and can be life-threatening. Current management is tailored to disease severity. For organ- or life-threatening GPA, remission induction is based on glucocorticoids combined with rituximab or cyclophosphamide; subsequent maintenance therapy aims to prevent relapse while limiting treatment-related toxicity [3]. GPA should therefore be considered in patients with compatible upper and lower respiratory tract manifestations, renal abnormalities and systemic features. Diagnosis requires integration of clinical, serological, imaging and histopathological findings, together with careful exclusion of infectious, malignant and other autoimmune conditions.

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ABS 48

IMPACT OF IMMERSIVE VIRTUAL REALITY ON PEDIATRIC EXERCISE TESTING: A PILOT STUDY

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INTRODUCTION

Standardized testing is a well-established tool to assess physical capacity in children, especially those with congenital or acquired heart disease, where reduced physical performance is common. Determining maximal effort in children can be difficult: a plateau in oxygen consumption is seen in about half of the cases, while in the others it is estimated using parameters such as peak heart rate, ventilatory reserve and respiratory exchange ratio [1]. Virtual reality (VR) can improve the exercise testing experience by creating simulated environments in which users can perceive a sense of real presence [2]. VR can also help to reduce pain and anxiety in children [3]. The integration of VR during pediatric exercise testing therefore may improve motivation and the accuracy of the assessment of functional capacity.

OBJECTIVES

This study investigates the impact of a 3D VR headset on clinical performance during cycle-ergometer exercise testing in pediatric patients and assesses tolerability, satisfaction, psychomotor engagement and potential adverse events. VR technologies are increasingly explored as tools to enhance motivation and adherence in young individuals, yet evidence supporting their use during cardiopulmonary exercise testing remains limited.

METHODS

Ten patients aged 11-18 years were prospectively enrolled. Each participant underwent a complete cardiological evaluation, including an electrocardiogram and echocardiography, followed by two standardized cycle-ergometer tests. The first test was performed in the conventional modality, whereas the second, conducted approximately 3 days later, employed a 3D VR headset that immersed the user in a virtual Patagonian mountain environment. Vital signs, such as heart rate and blood pressure, were recorded and an electrocardiogram was performed during the examination. At the end of each test, patients completed a satisfaction questionnaire.

RESULTS

In 9 out of 10 patients, peak heart rate was higher during the VR-assisted test compared with the standard procedure. Peak cardiac output was identical in 60% of participants and higher in 40% when VR was used. Blood pressure values showed no significant differences between conditions, except for a slight decrease in resting systolic pressure with VR. All patients reported that VR made the activity more engaging, and 80% stated that they would be interested in repeating the experience. No complications or significant discomfort were observed.

CONCLUSIONS

The use of a 3D VR headset during cycle ergometer exercise testing appears to be feasible, well-tolerated, and potentially beneficial in enhancing psychological and physical engagement and clinical performance in pediatric patients. These findings support further exploration of immersive VR environments in pediatric cardiology and sports medicine.

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ABS 49

EBSTEIN'S ANOMALY: THE EFFECTIVENESS OF SYSTEMATIC PHYSICAL EXAMINATION IN THE DIAGNOSTIC TIMING OF THE NEWBORN

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Ebstein's anomaly is a rare congenital heart disease (0.5% of cases) characterized by the failure of the septal and posterior leaflets of the tricuspid valve to detach, resulting in the apical displacement of the valvular orifice and "atrialization" of the right ventricle. Despite advances in diagnostic imaging, prenatal diagnosis can sometimes be missed, especially in the absence of macroscopic signs such as fetal hydrops, severe cardiomegaly, or sustained tachyarrhythmias, which represent the main warning signs *in utero*.

The present work describes a case in which the pathology remained silent during the prenatal ultrasound investigation, revealing itself only at birth through a careful neonatal physical examination. We present the case of a term newborn, classified as large for gestational age at birth, born to a mother with altered inflammatory markers (positive CRP) and intrapartum low-grade fever (body temperature 37.2°C). Although the initial physical examination did not reveal clinical anomalies or signs of cardiorespiratory distress, the serial evaluation at 6 hours of life subsequently detected the appearance of a new-onset holosystolic murmur and initial signs of respiratory effort with mild subcostal retractions and mild tachypnea. The initial hemodynamic stability, followed by the rapid evolution of the clinical and auscultatory picture, raised the immediate suspicion of congenital heart disease and therefore the indication to perform an emergency echocardiography. The investigation documented a picture of severe tricuspid regurgitation associated with estimated systolic pulmonary arterial pressure (sPAP) values of 50 mmHg.

This pulmonary hypertension, together with the valvular morphology, initially required a complex differential diagnosis, as a primary dysplasia of the tricuspid valve could not be excluded. However, the detailed analysis of the anatomical relationships allowed the confirmation of the pathognomonic criteria of Ebstein's anomaly: atrial septal defect with a left-to-right shunt, Ebstein-like tricuspid valve dysplasia (apical displacement of the septal leaflet greater than 8 mm/m^2) with severe valvular regurgitation, dilated right sections, dilated inferior vena cava, left ventricle of normal size and kinesis, and it confirmed the sPAP of 50 mmHg, with a moderate left-to-right patent ductus arteriosus. The assistance resulting from this clinical-instrumental picture included respiratory support with nCPAP, the initiation of diuretic therapy with intravenous furosemide, and fluid restriction; all interventions aimed at reducing fluid overload, pulmonary congestion, and right heart filling pressures, with an indirect improvement on respiratory dynamics. The resulting clinical improvement, supported by serial echocardiographic checks, progressively allowed weaning from respiratory assistance and the subsequent transition to oral diuretic therapy.

The picture of congestive heart disease from severe tricuspid regurgitation, as in Ebstein's anomaly, can also be associated with reduced pulmonary venous return and a consequent low cardiac output with organ hypoperfusion. This consideration requires serial monitoring of cerebral blood flow using transfontanellar cerebral color Doppler ultrasound technique, which in the case in question showed a resistance index (RI) in the ACA of 0.75-0.8 with a stable flow pattern.

It is also essential to ensure appropriate cardiological follow-up in newborns with Ebstein's anomaly, who can develop cardiac arrhythmias due to right atrial dilation and abnormalities of the heart's conduction pathways, with possible electrical re-entry circuits and an increased risk of developing potentially fatal tachyarrhythmias. An association with Wolff-Parkinson-White syndrome is documented in the literature in up to 5% of cases.

The case under examination underlines how the sensitivity of prenatal ultrasound screening does not reach 100%, highlighting the need for rigorous clinical monitoring in the postpartum period. The accuracy of the neonatal evaluation (auscultation, monitoring of pulses and skin color) still represents the cornerstone of newborn safety today, allowing for timely intervention where prenatal technology has shown intrinsic limits.

In particular, when Ebstein's anomaly is suspected, the early physical examination and the neonatological

re-evaluation in the first 48-72 hours of life assume a key diagnostic role: the physiological drop in pulmonary vascular resistance after birth drastically changes the pressure gradients and the dynamics of the "atrialized" right ventricle, making signs such as the murmur from tricuspid regurgitation or cyanosis manifest, which are otherwise silent during fetal circulation.

In this context, the initial neonatal hemodynamics (with sPAP values close to 50 mmHg) can initially mask or complicate the morphological assessment; therefore, physical signs associated with a targeted echocardiography remain the decisive tools for a correct and timely diagnosis and adequate treatment, in order to reduce the comorbidities associated with the underlying pathology and the long-term sequelae that could result from a delay in the assessment.

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ABS 50

FROM GENOTYPE TO PHENOTYPE IN CYSTIC FIBROSIS: THE CONTRIBUTION OF URINARY METABOLOMICS

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INTRODUCTION

Cystic fibrosis (CF) is a chronic, progressive autosomal recessive disease caused by mutations in the *CFTR* gene, which encodes a protein involved in epithelial chloride and water transport [1]. *CFTR* dysfunction leads to thick, viscous secretions, resulting in multisystem involvement, mainly affecting the respiratory, pancreatic, intestinal, hepatobiliary and reproductive systems. Clinical expression is highly variable and depends on mutation type, genetic and environmental modifiers, and patient clinical status [2]. In Sardinia, F508del and T338I are among the most represented mutations: F508del is generally associated with a more severe phenotype, whereas T338I is more often related to mild forms.

STUDY DESCRIPTION

Urinary metabolomic analysis was performed by gas chromatography-mass spectrometry on urine samples from 35 CF patients, divided into 3 genotypic groups: 11 F508del/F508del, 12 T338I/T338I and 12 F508del/T338I. The cohort included 22 males and 13 females, aged 1-54 years. First-morning urine samples were collected and processed according to an internal protocol. Metabolomic data were analyzed using univariate and multivariate statistics to identify metabolic differences among groups and correlate urinary profiles with genotype and clinical phenotype.

DISCUSSION

Preliminary analysis showed metabolomic differences between F508del/F508del patients and those carrying the T338I mutation. PCA did not show clearly separated clusters or relevant outliers. The three-class OPLS-DA model was not statistically significant because of overlap between T338I/T338I and F508del/T338I; however, pairwise analyses discriminated F508del/F508del patients from both T338I-carrying groups. These latter groups showed highly similar metabolomic profiles, without statistically significant differences, in line with their milder phenotype. The main discriminating metabolites included glucose, 4-hydroxyphenylacetic acid, citric acid, scyllo-inositol, 3-hydroxyisovaleric acid, maltose and HPHPA. These findings support the potential of urinary metabolomics in identifying CF-related metabolic signatures [3].

CONCLUSION

This study suggests a relationship between urinary metabolomic profile, genetic background and clinical phenotype in the analyzed CF patients. In particular, metabolomics appears to distinguish F508del/F508del-associated forms from those related to the T338I mutation, supporting the role of this approach in the study of phenotypic variability in CF. Although preliminary and limited by sample size, these findings deserve confirmation in larger studies.

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ABS 51

ACUTE KIDNEY INJURY IN THE PRETERM NEONATE: A CASE REPORT

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INTRODUCTION

Acute kidney injury (AKI) is a frequent complication associated with high morbidity and mortality in preterm neonates, with a reported incidence ranging from 18% to 40% in low-birth-weight infants, rising to 48% in extremely low-birth-weight infants according to modified neonatal KDIGO criteria; mortality among VLBW infants with AKI is significantly higher than in those without AKI. Nephrogenesis, which is completed by 34-36 weeks of gestation, renders the preterm kidney particularly vulnerable to hemodynamic, septic, and pharmacological insults, especially in infants born before 29 weeks of gestation. Diagnosis remains challenging owing to the limitations of serum creatinine as a marker – its delayed rise and its dependence on maternal renal function in the first days of life – as well as the lack of validated diagnostic thresholds for each class of extreme prematurity [1-3].

CASE REPORT

We describe the case of an extremely preterm neonate (gestational age 23 weeks + 3 days, birth weight 659 g) who, at approximately the third week of life, developed severe AKI concomitant with hypovolemic shock, characterized by anuria, progressively worsening diffuse edema, and rapid weight gain. Serum creatinine rose progressively from 1.05 to 4.00 mg/dL over approximately 10 days, accompanied by metabolic acidosis (pH 7.29, HCO_3^- 16.2 mmol/L) and hyperkalemia (K^+ 6.21 mmol/L). Renal ultrasound showed increased echogenicity with preserved vascular perfusion. A large patent ductus arteriosus with left-to-right shunt was also present. Management included hemodynamic support (norepinephrine, dobutamine, dopamine), furosemide in intermittent boluses and continuous infusion, albumin infusion, correction of acidosis with sodium bicarbonate, and careful fluid management. Because of persistent difficulty in controlling fluid overload and persistent anuria, ethacrynic acid was introduced as a bolus and continuous infusion. Pharmacological and antibiotic therapy was reassessed in light of the risk of nephrotoxicity and the decline in renal function. Despite intensive treatment, renal function did not normalize, with only minimal recovery of diuresis.

CONCLUSIONS

This case illustrates AKI of multifactorial etiology, comprising a pre-renal component secondary to a large patent ductus arteriosus causing impaired systemic and renal perfusion, together with an intrarenal component favored by exposure to potentially nephrotoxic drugs and by marked renal immaturity with reduced nephron reserve. Management of AKI in the extremely preterm neonate is complex and requires early involvement of a pediatric nephrologist and, in more severe cases, transfer to a Tertiary-Level Center with access to nephrology support and renal replacement therapy.

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ABS 52

PRIMARY CONGENITAL GLAUCOMA: FROM DIAGNOSIS TO EARLY MANAGEMENT

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INTRODUCTION

Primary congenital glaucoma (PCG) is a form of childhood glaucoma caused by a developmental anomaly of the aqueous humor drainage system, which obstructs its outflow, leading to an increase in intraocular pressure (IOP). The incidence of the pathology presents a marked geographical variability, ranging from 1 in 20,000 in Western countries up to 1 in 1,250 in Eastern Europe, with a 5-10 times higher risk in cases of consanguinity.

PATHOPHYSIOLOGY AND CLINICAL PRESENTATION

Persistent intraocular hypertension, in an infant eye characterized by high elasticity, leads to a progressive distension of the ocular structures. At the corneal level, edema with opacification, Haab's striae (breaks in Descemet's membrane), and an increase in corneal diameter (megalocornea) are observed. The expansion of the eyeball determines buphthalmos and an increase in axial length, with the possible development of axial myopia and irregular astigmatism. Ocular hypertension is also responsible for damage to the optic nerve, characterized by increased cupping of the optic disc, thinning of the retinal nerve fiber layer, and, in more advanced cases, disc pallor leading to irreversible optic atrophy. From a clinical perspective, corneal edema is the basis of the classic symptomatic triad consisting of epiphora, photophobia, and blepharospasm.

CASE REPORT

We present the case of a male newborn of Italian origin, born at $40^{+6/7}$ weeks of gestation from a pregnancy achieved through ICSI and an emergency cesarean section in the presence of clear amniotic fluid. There was good adaptation at birth, and auxological parameters for length and head circumference were normal, while weight was above the 90th percentile. Family history was negative for consanguinity; the mother was affected by thrombophilic mutations, had a previous HPV positivity, and underwent regular infectiological screening and prenatal ultrasound monitoring. The physical examination at birth revealed buphthalmos predominantly in the left eye, bilateral corneal opacity, and alterations of the red reflex (absent on the left, reduced on the right). Given the finding of this malformation, a specialized ophthalmological evaluation was performed, revealing megalocornea with corneal edema and an IOP of 22 mmHg (iCare) in the right eye (RE). The fundus oculi (FO) could not be examined because of media opacity. The left eye (LE) showed buphthalmos, megalocornea, corneal edema, and an IOP of 28 mmHg (iCare). The FO could not be examined because of media opacity. Therefore, a diagnosis of newborn onset PCG (LE > RE) was made, with an indication for surgery.

In consideration of the clinical urgency and the need to optimize preoperative conditions, the patient was started on a combined topical medical therapy with latanoprost, dorzolamide, and timolol within 24 hours of birth, in order to reduce corneal edema and IOP while awaiting surgery.

Upon re-evaluation at 9 days of life, the IOP was elevated (RE 41 mmHg, LE 51 mmHg), making it necessary to proceed with a 360° trabeculotomy in the LE (to correct aqueous humor drainage) and a possible SubCyclo® in the RE (to reduce aqueous humor production). Early treatment follows evidence indicating surgery as the gold standard for PCG, with success rates of angle procedures exceeding 90% in patients between 2 months and 1 year of age, although more limited in neonatal forms (< 50%).

CONCLUSIONS

The case emphasizes the importance of early diagnosis and immediate multidisciplinary care when PCG is suspected, in order to reduce the risk of irreversible visual impairment. Although the origin of PCG is predominantly genetic, recent literature highlights how medically assisted reproduction techniques, such as the ICSI used in our patient, are associated with a marginally increased risk of congenital anomalies (relative risk ~1.39). However, timely management allows for the control of disease progression. Long-term data show an absence of progression in 90.3% of cases at 1 year, 83.1% at 5 years, 70.8% at 10 years, and 58.3% at 34 years after adequate treatment, underlining the importance of follow-up. The visual prognosis remains strictly linked to post-treatment corneal transparency and the resolution of amblyopia, making a rigorous follow-up essential to ensure maximum visual potential.

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ABS 53

BREASTFEEDING AND COMPLEMENTARY FEEDING IN PRETERM INFANTS: THE ROLE OF CORRECTED AGE AND CRITICAL DEVELOPMENTAL WINDOWS IN AN OUTPATIENT COHORT

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The introduction of complementary feeding (CF) represents a fundamental stage in the development of infant nutrition and feeding skills [1]. In preterm infants, determining the optimal timing of CF can be challenging, as chronological age alone may not adequately reflect the degree of neurodevelopmental and oromotor maturation [2, 3]. This study evaluated the timing of CF introduction in a cohort of preterm infants followed at the Neonatal Intensive Care Unit (NICU) Follow-Up Clinic of the University Hospital of Monserrato (Cagliari, Italy), with particular attention to the role of corrected age and individual variability. An observational study was conducted between December 2025 and May 2026 and included 41 preterm infants. Data on perinatal history, feeding practices, and the timing of CF introduction were collected through a questionnaire administered to parents or caregivers and analyzed using descriptive and inferential statistical methods. The study population included 23 males and 18 females; 63.4% were moderately or late preterm infants, while 36.6% were very preterm infants. The mean age at CF introduction was 7.11 months of chronological age (range, 6-10 months) and 5.39 months of corrected age (range, 3.0-7.5 months). A wide interindividual variability was observed, with some infants starting CF as early as 3 months of corrected age, which represents the minimum age indicated by current recommendations [2]. The duration of NICU hospitalization showed a positive and statistically significant correlation with chronological age at CF introduction ($r = 0.4703$; $p = 0.0019$).

Overall, the findings highlight substantial variability in the timing of CF introduction among preterm infants and underscore the importance of considering corrected age together with clinical history and the infant's neurodevelopmental and oromotor skills. An individualized approach therefore appears relevant when determining the timing of complementary feeding, although further studies involving larger cohorts are needed to investigate the factors associated with different timings of CF introduction.

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ABS 54

FECAL METABOLOMIC PROFILING BY ^{1}H-NMR IN PEDIATRIC IBD: CHARACTERIZATION OF METABOLIC PROFILES AND POTENTIAL CLINICAL APPLICATIONS

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Pediatric inflammatory bowel diseases (IBD), including Crohn's disease (CD) and ulcerative colitis (UC), are characterized by a complex interplay between inflammation, the gut microbiota, and host metabolism. Fecal metabolomics may contribute to

defining the biological alterations underlying these diseases and to identifying potential biomarkers of disease and therapeutic response [1]. This study evaluated the fecal metabolomic profile of pediatric patients with IBD, comparing it with that of healthy controls (CTR) and assessing its evolution during follow-up. A total of 76 subjects were enrolled: 31 with UC, 20 with CD, and 25 CTR. Fecal samples were collected at baseline (T0), at 6 months (T1), and at 12 months (T2) and analyzed by ^{1}H-NMR spectroscopy. Multivariate analysis of metabolic profiles was performed using Principal Component Analysis (PCA), while longitudinal changes in metabolites were assessed using linear mixed-effects models (LMM), adjusted for age and sex. PCA showed substantial overlap among the groups, indicating considerable interindividual heterogeneity. At T0, patients with IBD tended to have higher levels of valine and alanine than CTR, in agreement with previous observations in pediatric IBD [2]. In patients with CD, significant remodeling of amino acid metabolism emerged during follow-up, predominantly within the first 6 months: leucine, valine, isoleucine, alanine, tyrosine, and phenylalanine decreased from T0, followed by substantial stabilization between T1 and T2. Valine showed a persistent reduction through T2 (T0-T1 $p = 0.018$; T0-T2 $p = 0.021$). These early metabolomic changes during treatment are consistent with previous findings in pediatric CD [3]. In patients with UC, the metabolic profile was comparatively more stable, with gentisate being the only metabolite showing a significant change over time. Overall, the results highlight distinct patterns of metabolic remodeling in pediatric IBD, which were broader and occurred earlier in CD and were more selective in UC. Fecal metabolomic profiling by ^{1}H-NMR may represent a complementary tool for disease characterization and for monitoring disease evolution and therapeutic response, although validation in larger cohorts is required.

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ABS 55

ATOPIC DERMATITIS IN PEDIATRIC PATIENTS: CORRELATION BETWEEN EARLY-LIFE FEEDING AND ALLERGIC MANIFESTATIONS IN A PERSONAL CASE SERIES

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BACKGROUND

Atopic dermatitis (AD) is a chronic, multifactorial inflammatory skin disease frequently occurring in childhood [1]. Its pathogenesis involves genetic predisposition, skin barrier dysfunction, immune dysregulation, and environmental factors. Early-life nutrition and exposure to food allergens may influence the development of oral tolerance, supporting the timely introduction of potentially allergenic foods rather than their preventive avoidance [2]. Conversely, unnecessary or prolonged elimination of previously tolerated foods may impair oral tolerance and promote food allergy, with potentially severe reactions, including anaphylaxis [3]. This study aimed to investigate associations between early-life factors, AD severity, food-related manifestations, and elimination diets.

METHODS

A cross-sectional observational study was conducted over 6 months at the "SOS Mami" and Neonatal Intensive Care Unit Follow-up outpatient services of the University Hospital of Cagliari, Italy. Sixteen children with clinically diagnosed AD were enrolled. Clinical, perinatal, and nutritional data were collected through a structured questionnaire administered to caregivers. Descriptive statistics, Fisher's exact test, and odds ratios were used for data analysis.

RESULTS

A positive family history of atopy was observed in 75% of children, with an earlier AD diagnosis observed in children with a positive family history. Moderate-to-severe AD was significantly more frequent among children receiving mixed or formula feeding than among those exclusively breastfed (87.5% vs 25.0%; OR = 21.0; $p = 0.041$). Moderate-to-severe forms were also more frequent among

children born by cesarean section than among those born vaginally (62.5% vs 37.5%), although the association was not statistically significant (OR = 2.78; p = 0.619). Food-related manifestations were reported in more than half of the children and were exclusively cutaneous. Overall, 37.5% followed an elimination diet. Only 5 children underwent allergological assessment, and no food allergy was confirmed among those evaluated.

CONCLUSIONS

Exclusive breastfeeding was associated with milder AD, while the observed association between cesarean delivery and greater disease severity requires confirmation in larger cohorts. Importantly, although food-related manifestations were frequently reported and elimination diets were adopted, food allergy was not confirmed in any of the children who underwent allergological assessment; food allergy cannot be excluded in children who were not evaluated. These findings emphasize that dietary exclusions should follow appropriate allergological assessment to avoid unjustified restrictions. Larger prospective studies are needed to confirm these associations and further investigate the role of early nutritional factors in pediatric AD.

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ABS 56

ACHONDROPLASIA: STATE OF THE ART AND THERAPEUTIC PERSPECTIVES

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INTRODUCTION

Achondroplasia is the most common skeletal dysplasia and is caused, in nearly all cases, by gain-of-function variants of *FGFR3*, predominantly p.Gly380Arg. Constitutive FGFR3 activation excessively drives intracellular pathways, particularly RAS-RAF-MEK-ERK, reducing chondrocyte proliferation and differentiation and impairing endochondral ossification. The clinical picture is multisystemic and includes, besides disproportionate short stature, neurological, respiratory, otorhinolaryngological and orthopedic complications, such as foramen magnum stenosis, cervicomedullary compression, obstructive and central apneas, and limb deformities [1, 2].

STATE OF THE ART

Understanding the pathogenic role of *FGFR3* has opened the way to targeted pharmacological therapies. Vosoritide, a C-type natriuretic peptide (CNP) analogue, activates NPR-B and the cGMP pathway, functionally counteracting MAPK cascade hyperactivation. Daily subcutaneous administration significantly increases annualized growth velocity, sustained on long-term follow-up, and it is currently the only targeted therapy approved in the European Union [1].

NEW THERAPEUTIC STRATEGIES

Development is moving toward longer-acting molecules and more direct intervention on the FGFR3 pathway. Navepegritide, a long-acting CNP prodrug, preserves the NPR-B/cGMP mechanism while allowing weekly dosing; in the APPROACH trial it increased growth velocity and showed favorable signals on body proportions, gaining accelerated FDA approval in February 2026 [3]. Infigratinib, an oral FGFR1-3 tyrosine kinase inhibitor, acts further upstream, reducing MAPK and STAT1 signaling and thereby promoting chondrocyte differentiation and proliferation, respectively. Clinical data show dose-dependent growth effects and promising results on body proportions. Heterogeneity across studies and endpoints, however, currently precludes establishing the superiority of any single therapeutic strategy [2].

PERSPECTIVES

Future directions include earlier initiation of targeted therapies, aimed at preventing complications such as foramen magnum stenosis, combination strategies of CNP with growth hormone, and more selective FGFR3 inhibitors, including dabogratinib (TYRA-300). Anti-FGFR3 antibodies, soluble receptors, CDK8 inhibitors and anti-FGF2 aptamers represent further approaches still at the preclinical or early stage [2].

CONCLUSIONS

The therapeutic approach to achondroplasia is rapidly evolving from predominantly symptomatic, multidisciplinary management toward strategies capable of directly targeting its pathophysiology.

Increased growth velocity is an important efficacy marker but cannot be the sole objective. Adult height, body proportions, function and autonomy, prevention of neurological and respiratory complications, long-term safety and quality of life must also be assessed. Pharmacological therapy therefore does not replace multidisciplinary surveillance, but constitutes an increasingly relevant component of an individualized, person-centered care pathway.

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ABS 57

LIMITS OF VIABILITY: WHEN RESUSCITATION BECOMES A LEGAL QUESTION

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The limits of neonatal viability represent one of the most challenging areas of medicine. Within the "grey zone" of viability, neither survival nor the degree of subsequent morbidity can be predicted with certainty. Decisions regarding whether to initiate, withhold, or discontinue intensive interventions require consideration not only of medical prognosis, but also of parental values, the potential for suffering, and the best interests of the newborn. The concepts of wrongful birth and wrongful life provide a broader ethical and legal framework for considering whether medical intervention may itself become a source of harm. Wrongful birth concerns the deprivation of parental reproductive choice following inadequate prenatal information, whereas wrongful life raises the more controversial question of whether survival with severe disability can constitute a legally recognizable harm [1, 2]. Although these concepts emerged primarily from obstetric and prenatal contexts, their underlying principles may be relevant to decision-making at the limits of viability. In extremely preterm infants, active resuscitation may result in survival accompanied by severe long-term morbidity. Conversely, withholding intervention may result in death despite a possibility of survival. The central question is not merely whether resuscitation is technically possible, but whether it is ethically and legally appropriate when prognosis is highly uncertain and the burden of treatment is substantial. Meaningful parental counselling and participation in decisionmaking become particularly important. However, the absence of universally accepted guidelines that incorporate parental preferences within the grey zone of viability may complicate this process [2, 3]. A challenging scenario arises when parents, after appropriate counselling, express a clear and informed wish to withhold resuscitation within the grey zone of viability, yet active resuscitation is initiated against their wishes. If such intervention results in prolonged intensive care, invasive procedures, significant suffering, and survival with severe morbidity, the ethical and legal implications extend beyond medical beneficence. The intervention may have deprived parents of meaningful participation in a decision concerning the course of their child's life. This raises a largely unexplored question: could the imposition of resuscitation against informed parental wishes represent a form of "wrongful birth", not because the child should not have been born, but because a life was medically prolonged despite an informed decision that such intervention should not be pursued? While such a situation cannot be equated with the established legal definition of wrongful birth, the conceptual parallel warrants further ethical and legal examination. At the limits of viability, the question may no longer be only whether survival is possible, but whether every possible survival ought necessarily to be pursued.

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ABS 58

GUILLAIN-BARRÉ SYNDROME IN A TWO-YEAR-OLD GIRL: DIAGNOSTIC AND THERAPEUTIC CHALLENGES AT A VERY YOUNG AGE

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BACKGROUND

Guillain-Barré syndrome (GBS) is an acute immune-mediated polyradiculoneuropathy, usually triggered by a preceding bacterial or viral infection. Molecular mimicry contributes to its pathogenesis by inducing an immune response against peripheral nerve components, resulting in demyelination and/or axonal injury. GBS is uncommon in children, with an estimated annual incidence of 0.34-1.34 cases per 100,000. We report a very early-onset case of GBS in a 2-year-old girl.

METHODS

Clinical, laboratory, microbiological, imaging, neurophysiological, treatment, and outcome data were retrospectively analyzed.

RESULTS

A 2-year-old girl was admitted to our Pediatric Department after approximately 10 days of progressive gait disturbance, lumbar pain, muscle weakness, and impaired balance. Three weeks before admission, she had experienced an episode of gastroenteritis. She was hospitalized for 2 weeks. Neurological examination revealed an ataxic gait with impaired postural stability, requiring support when bending down to pick up objects from the ground and when standing up. Patellar, Achilles, biceps, and triceps reflexes were bilaterally absent. There was no pain during lower limb mobilization and no cutaneous signs suggestive of joint inflammation. Respiratory function remained normal throughout the clinical course. Brain CT showed no abnormalities. Cerebrospinal fluid analysis revealed elevated protein levels of 295.7 mg/dl (normal range, 15-45 mg/dl), with otherwise normal findings. Brain and spinal cord MRI demonstrated meningeal contrast enhancement at the level of the conus medullaris and epiconus, together with enhancement of the cauda equina nerve roots, supporting the clinical suspicion of GBS. Routine blood tests were unremarkable. For the diagnostic work-up, electromyography was essential and revealed a pattern of demyelinating sensorimotor involvement, consistent with the acute inflammatory demyelinating polyradiculoneuropathy (AIDP) subtype of GBS. The diagnosis was established based on the clinical, neurophysiological, and radiological findings. Given the severity of the clinical presentation, the patient underwent 5 sessions of therapeutic plasma exchange (TPE). Due to a persistent incomplete clinical response, intravenous immunoglobulin (IVIg) was subsequently administered at a dose of 5 g/day for 5 consecutive days (0.4 g/kg/day; total dose, 2 g/kg), in accordance with the EAN/PNS guidelines. Clinical improvement was observed after immunotherapy, with progressive functional recovery during hospitalization and in the follow-up period. No respiratory failure or autonomic instability occurred, and intensive care support was not required.

CONCLUSIONS

This case highlights that GBS should be considered even in very young children presenting with gait disturbance and progressive weakness, particularly following a recent infectious episode. Careful recognition of early neurological signs, especially gait abnormalities and areflexia, is crucial for prompt diagnosis and treatment. Early intervention may improve functional recovery, even with an unusually early disease onset.

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ABS 59

STAPHYLOCOCCAL SCALDED SKIN SYNDROME IN A 3-YEAR-OLD CHILD: A CASE REPORT

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INTRODUCTION

Staphylococcal scalded skin syndrome (SSSS) is an acute bullous dermatopathy typical of pediatric age, caused by exfoliative toxins produced by *Staphylococcus aureus*, which cleave desmoglein 1 in the granular layer of the epidermis [1, 2].

CASE REPORT

A 3-year-old boy with a history of atopic dermatitis, cow's milk protein allergy and sensitization to dog allergens presented after a febrile episode treated with paracetamol, followed by defervescence; about 1 week later he developed a pruritic rash treated with cetirizine and paracetamol. Over the following 24 hours, painful cutaneous exfoliation appeared on the face and back. On admission the patient was in fair general condition, with erythematous patches and areas of denuded epidermis on the face, back and retroauricular region, positive Nikolsky sign, all associated with marked cutaneous hyperesthesia. Nasal obstruction with dense yellowish secretions and pathological otoscopic findings were also present. Blood tests showed neutrophilic leukocytosis (WBC 17,000/μL, neutrophils 74%), with the remaining values within normal limits. Dermatological consultation favored a diagnosis of SSSS, with bullous impetigo as the main differential diagnosis. Skin swab was positive for *Staphylococcus epidermidis*, while pharyngeal swab isolated *Staphylococcus aureus* and *Pseudomonas aeruginosa*; blood culture, urine culture and rectal swab were negative. Systemic antibiotic therapy with ceftriaxone and teicoplanin was started, together with intravenous rehydration, analgesia and local wound management with dressings and topical gentamicin. During hospitalization, progressive resolution of the skin lesions and gradual resumption of feeding were observed.

CONCLUSIONS

SSSS is a pediatric dermatology emergency and should be considered in children with rapidly progressive erythema, pain and cutaneous hyperesthesia, blistering or superficial exfoliation. Diagnosis is predominantly clinical and can be supported by cultures from possible primary sites of infection or colonization. Prompt systemic antibiotic treatment, combined with rehydration and local wound management, generally leads to rapid favorable evolution [1, 2].

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ABS 60

A SMALL LESION, A LONG TRAJECTORY: NEURODEVELOPMENTAL FOLLOW-UP AFTER NEONATAL THALAMIC ISCHEMIC STROKE

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Neonatal arterial ischemic stroke (NAIS) is a relevant cause of acquired brain injury and of potential neurodevelopmental impairment. Forms that selectively involve the perforating branches at the thalamic level are rare and have a multifactorial etiology, in which perinatal, placental, haemodynamic, and prothrombotic factors contribute to a variable extent [1-3].

A 20-month-old boy presented to our attention for a routine follow-up visit. He was born at 39 weeks of gestation (birth weight 3,230 g; Apgar scores 9 and 10) by cesarean section for transverse lie, after a pregnancy complicated by polyhydramnios and a prenatal ultrasound diagnosis of horseshoe kidney. Within the first 48 hours of life, he developed coarse limb tremors, both spontaneous and stimulus-evoked, together with difficulties in sucking and feeding. Brain MRI with MR angiography showed a band-like ischemic lesion of the right anterior thalamus in the territory of the perforating arteries, with patency of the main intracranial vessels. As part of the diagnostic work-up, a thrombophilia screen revealed heterozygosity for factor V Leiden, which was interpreted as a possible susceptibility factor rather than a proven isolated cause [4, 5]. Metabolic and enzymatic investigations showed no significant abnormalities; a small apical muscular ventricular septal defect resolved spontaneously.

The child was promptly enrolled in a multidisciplinary rehabilitation program comprising physiotherapy, neuropsychomotor therapy, speech therapy, and occupational therapy. At 11 months, CT documented a small calcified thalamic focus.

MRI at 18 months showed marked regression of the focal abnormality, with minimal residual signal due to susceptibility effects. The Bayley-III assessment at 16 months revealed an uneven developmental profile, with a cognitive age equivalent of approximately 14 months, a language age equivalent of 12-13 months, and gross motor skills at approximately the 10-month level. After a cycle of intensive rehabilitation, excellent progress was observed in crawling, postural transitions, lateral cruising with support, requestive pointing – the gesture by which a child points to something he or she wishes to obtain – and emerging symbolic play. This case highlights the value of early neuroimaging in neonates with unexplained neurological signs and shows that radiological regression of the lesion does not necessarily coincide with spontaneous normalization of the neurodevelopmental trajectory. Although the prognosis *quoad vitam* may be favourable, the prognosis *quoad valetudinem* – particularly in neuromotor and neurodevelopmental terms – requires a more comprehensive assessment and monitoring over time. In such cases, prolonged multidisciplinary follow-up and early, individualized rehabilitation are essential to achieve the best possible functional outcome.

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ABS 61

WIRELESS CONTINUOUS NEONATAL TELEMONITORING: A GOOD PRACTICE TO PRESERVE MOTHER-INFANT PROXIMITY WHILE ENSURING VITAL SIGN SURVEILLANCE

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BACKGROUND

Rooming-in from birth supports bonding, breastfeeding and family wellbeing, but is often interrupted when a newborn needs continuous vital-sign monitoring, since conventional wired pulse oximetry requires continuous bedside visual surveillance by an operator – rarely feasible with current staffing levels. Newborns too stable for intensive care but still requiring monitoring (moderately preterm, 34-36 weeks; ex-preterm with residual apnea risk; mild asphyxia; transient neurological disturbances, e.g. during sepsis) are therefore usually separated from their mothers for observation, or otherwise monitored only by parental vigilance, accepting a residual, low but non-negligible risk of an unwitnessed apneic event.

OBJECTIVES

To guarantee continuous monitoring of vital parameters for timely detection of clinical anomalies in this population while preserving rooming-in, and to optimize the use of nursing resources through a centralized surveillance system extended to obstetric maternity rooms and the delivery room.

METHODS

The system operates through a three-step signal chain: a sensor applied to the newborn transmits oxygen saturation and heart rate via Bluetooth to a bedside monitor, which stays with the mother in her room; this bedside monitor then relays the signal wirelessly to the hospital data network; the centralized station in the Neonatal Intensive Care Unit (NICU) receives the signal from the hospital network and displays it, together with any alarms, on its own monitor, allowing one nurse to supervise both NICU and rooming-in newborns simultaneously. Proximity between the NICU and the maternity rooms ensures a rapid clinical response to alarms. The project (design phase February 2023; operational phase from January 2025) was led by the ASL TO5 Department of Pediatrics and Neonatology directorate with nursing coordination and the ASL TO5 Clinical Risk Group, and funded from the existing departmental budget with no additional nursing staff required.

RESULTS

Preliminary experience shows increased rooming-in uptake and reduced NICU observation time for the target population, with favorable maternal perceived satisfaction, though formal quantitative confirmation is pending. A structured quali-quantitative evaluation is ongoing, covering connection rates, number and predictive value of alarms, interventions triggered, clinical outcomes, a maternal satisfaction questionnaire and a staff focus group. Reported limitations include occasional false alarms, the need for ongoing connection-reliability checks, device costs and initial cultural resistance among staff.

CONCLUSIONS

Centralized wireless telemonitoring reconciles continuous vital-sign surveillance with rooming-in for moderate-risk newborns, at no extra staffing cost, improving family bonding and resource efficiency while maintaining clinical safety. The model is transferable to other Units with comparable monitoring infrastructure and was presented as an institutional good clinical practice at the 7th Regional Patient Safety Day of the Piedmont Region (September 2025).

ABS 62

FROM HYPERGLYCEMIA TO CARDIORESPIRATORY COLLAPSE: A COMPLEX CASE OF *E. COLI* SEPSIS

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INTRODUCTION

Late-onset sepsis in extremely preterm infants may present with subtle and nonspecific signs before rapidly progressing to severe multiorgan dysfunction. *E. coli* sepsis may cause respiratory, cardiovascular, neurological, and hematological complications. We present a case of severe *E. coli* sepsis in an extremely preterm infant, characterized by rapid clinical deterioration [1].

CASE REPORT

A female infant was born at 27 weeks of gestation, appropriate for gestational age. She was managed with CPAP without oxygen requirement and showed feeding tolerance. The only complication of prematurity was a persistent patent ductus arteriosus, despite prolonged intravenous paracetamol therapy. On day 13 of life, hyperglycemia represented the only initial warning sign and was corrected by reducing glucose intake. Inflammatory markers were immediately positive, prompting blood culture sampling, which became positive within a few hours for *E. coli*. Antibiotic therapy with cefepime and amikacin was initiated. The clinical condition rapidly deteriorated, with poor perfusion, reduced responsiveness, and severe metabolic acidosis (pH 7.1). Echocardiographic evaluation documented bradycardia, prompting endotracheal intubation and initiation of SIPPV ventilation. One hour later, massive pulmonary hemorrhage occurred through the endotracheal tube. Intratracheal adrenaline and surfactant were administered. Subsequently, heart rate fell below 60 bpm and oxygen saturation reached 19%, requiring cardiopulmonary resuscitation with chest compressions. Following resuscitation, ventilation was escalated to high-frequency oscillatory ventilation (HFOV). Dobutamine was started for cardiovascular support, while furosemide was administered because of reduced urine output. The onset of clonic movements prompted cranial ultrasound, which showed clear signs of ventriculitis. A grade III intraventricular hemorrhage subsequently developed, requiring plasma and red blood cell transfusions. Clinical conditions slowly improved, although the infant remained critically ill.

CONCLUSIONS

This case highlights the rapid and severe clinical evolution of late-onset *E. coli* sepsis in a preterm infant. Hyperglycemia may represent an early isolated warning sign of infection. Prompt recognition, microbiological investigation, appropriate antimicrobial treatment, respiratory and cardiovascular support and continuous neurological monitoring are essential in managing severe neonatal sepsis and its potentially devastating complications [1, 2].

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ABS 63

INFANTILE HEMANGIOMAS. A CLINICAL CASE

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INTRODUCTION

Infantile hemangioma is a common vascular tumor affecting approximately 5-10% of infants during the first year of life [1]. It is often first observed in the initial weeks of life. It presents as a raised, hyperplastic vascular lesion, red or purple in color. Deeper lesions may not be detectable immediately after birth. Generally, lesions increase rapidly in size and vascularity during the first months of life. Deep hemangiomas may grow rapidly in diameter, and such growth can interfere with important functions [2]. Lesions that impair vision, compromise the airway, or affect other critical structures require prompt evaluation and, when indicated, urgent treatment. The appropriate treatment depends on the characteristics of the lesion and the patient.

CLASSIFICATION AND SYMPTOMATOLOGY

According to their depth, infantile hemangiomas are classified as superficial, deep, or mixed. Cutaneous lesions vary in appearance: superficial lesions are bright red, whereas deep lesions are bluish. They can interfere with normal physiological functions depending on their location. Lesions on the face or in the oropharynx may obstruct the airway or cause visual problems; those near the anus or urethral meatus may obstruct normal fecal or urinary outflow.

CLINICAL CASE

A 3-month-old girl was in good general health. The mother reports that, since the second month of her daughter's life, she has noticed a slight swelling in the left subclavicular region. She also reports that the lesion has rapidly increased in size, doubling over approximately 1 month. On examination, a prominent swelling extends from the lower margin of the left clavicle toward the left mammary areola. The swelling has an angiomatous appearance and a firm-elastic consistency, is mobile over the underlying tissues, and is not painful. Tortuous vessels are clearly visible. Visual assessment suggests that the mass measures at least 4 cm in length and approximately 2-3 cm in width. We diagnosed a hemangioma and performed an ultrasound examination of the region. The ultrasound report stated: "In the upper region of the left breast, an isoechoic solid lesion measuring 35 × 12 mm is present, with undulating but well-defined margins and a mildly heterogeneous internal echotexture due to the presence of vascular structures showing normal resistance indices. No acoustic attenuation or signs of infiltration of the underlying muscle are observed. The findings are primarily suggestive of a hemangioma."

CONCLUSIONS

Prompt evaluation is important because infantile hemangiomas undergo a peak proliferative phase during the first months of life, particularly between the first and fifth months. During this period, they may grow rapidly. High-risk infantile hemangiomas require prompt evaluation and, when indicated, early treatment to prevent complications. Furthermore, uncontrolled growth in the mammary region may damage the developing mammary gland and cause permanent asymmetry. Rapid growth of the lesion can also cause marked skin tension, potentially leading to ulceration, excoriation, bleeding, and infection.

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ABS 64

WHEN THE JAW OPENS THE EYE: THE MARCUS GUNN PHENOMENON

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The Marcus Gunn jaw-winking phenomenon is a rare congenital ophthalmological anomaly characterized, according to Orphanet, by congenital ptosis associated with a pterygoid-levator synkinesis, which produces a distinctive eyelid movement synchronous with movements of the mandible [1]. Its prevalence in the general neonatal population is unknown, but it accounts for approximately 2-6% of cases of congenital ptosis [1]. It is a trigemino-oculomotor synkinesis in which activation of the masticatory muscles produces involuntary elevation

of the upper eyelid. In addition to the most common form, which is associated with congenital ptosis, rarer variants without clinically significant ptosis have been described; in a dedicated case series these accounted for approximately 6.8% of the cases observed [2].

A 9-month-old male infant, born at term after a spontaneous vaginal delivery, was referred to our attention after his mother had noticed, during breastfeeding, a recurrent elevation of the right upper eyelid coinciding with the sucking movements of the mouth.

The infant subsequently underwent two ophthalmological evaluations, both of which concurred in the diagnosis of a right-sided Marcus Gunn phenomenon. The mother also reported rare, transient episodes of deviation of one eye, observed intermittently during fixation or while the child attempted to track and reach for an object, with an apparent momentary loss of ocular alignment.

On pediatric evaluation, the infant weighed 8.9 kg (approximately the 50th percentile), with a length of 70 cm (approximately the 25th percentile) and a head circumference of 47.5 cm, above the 97th percentile according to the WHO growth charts for age and sex, and therefore warranting clinical monitoring. The general and neurological examinations were normal, with no cranial nerve deficits, abnormalities of tone, motor asymmetries, abnormal movements, or other focal neurological signs. The Denver Developmental Screening Test documented neurodevelopment fully appropriate for age, with gross motor and fine-adaptive skills partly ahead of chronological age.

Although isolated forms without ptosis are rarer, their course appears generally benign and stable: in a series of 14 patients, no case required surgical treatment, no ptosis developed during follow-up, and the only case of amblyopia was associated with anisometropia [2]. The literature has predominantly described forms associated with ptosis, in which amblyopia has been reported in up to 59% and anisometropia in 25% of patients [3]. In a series of 48 patients, amblyopia and strabismus were present in 23% and 34% of cases, respectively [4].

The main prognostic concern relates to the evolution of visual function rather than to neurological outcome. The reported episodes of ocular deviation should not automatically be attributed to the synkinesis, and they justify pediatric ophthalmological and orthoptic follow-up, including assessment of ocular motility and alignment, evaluation of fixation, cycloplegic refraction, and exclusion of anisometropia and amblyopia. Parents can also be involved through video documentation and quantification of the eyelid excursion during sucking and mandibular movements.

Surgery is reserved for forms with functionally significant ptosis or marked synkinesis [3]. In the case described here, the normal neurological examination and neurodevelopmental profile represent favorable prognostic factors, and there is currently no clinical indication for neuroimaging. Continued pediatric and ophthalmological follow-up remains essential for the early identification of any abnormality of ocular motility, amblyopia, or binocular vision.

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ABS 65

A MULTI-OMICS APPROACH TO BRAIN INJURY IN VERY LOW BIRTH WEIGHT BABIES: SPECIFIC BIOMARKERS FOR THE TWO MOST COMMON BRAIN LESIONS

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Brain injuries, in particular intraventricular hemorrhage (IVH) and periventricular white matter lesions (PVWM), are main complications of prematurity leading to adverse long-term neurodevelopmental outcomes [1-3].

Although cerebral ultrasound (CUS) is the primary method for diagnosing and monitoring acute and

early brain lesions like IVH, cerebral magnetic resonance imaging (MRI) at the term equivalent age (TEA) is used to provide the diagnosis of PVWM lesions, lesions developing over a longer time compared to IVH.

The identification of early and non-invasive biomarkers could follow the long-term effects of the two different lesions. We know that IVH, occurring in the first 3-4 days of life, can promote long lasting effects in the brain perfusion [1] and white matter development [2]. PVWM lesions are a complex disease, able to reduce brain connectivity. Very recently the possibility to diagnose brain lesions has been corroborated by biomarkers identified in the urine, a very practical and non-invasive technique. In other words, urinary protein and metabolic profiles of very low birth weight babies below 32 weeks of gestation may vary according to the presence and severity of brain damage prior to discharge and may be followed using a multi-omic approach [3].

We wish to identify these biomarkers in the following days and weeks after neuroimaging diagnosis with CUS and MRI performed at an earlier stage than TEA, at 33 weeks corrected age, in order to follow the effects of such lesions on brain development of the premature babies. The combination of proteomic and metabolomic data with clinical and neuroradiological findings could contribute to a more comprehensive understanding of the pathophysiological mechanisms, particularly their long-term effects on the developing brain. The identification in the urine of early biomarker panels aims also at improving individual risk stratification [3].

Overall, we believe a multi-omics approach could be a valuable tool for early neuro-monitoring in preterm infants, as the combination of molecular, clinical, and neuroradiological data may provide a more comprehensive assessment of the evolving risk of complex, dynamic neurological injury. By identifying early biomarkers associated with brain injuries, we aim to improve individual risk stratification for adverse neurological outcomes. Furthermore, in the future, this will help to understand the long-lasting effects and complications of both early IVH and later PVWM lesions to promote neuroprotective strategies to mitigate these later effects. This may produce the best pathways to personalized clinical management once lesions have occurred, as neuroprotection does not only mean trying to avoid the occurrence of brain lesions but also to lessen the later effects of such diseases.

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ABS 66

METABOLIC SIGNATURES ASSOCIATED WITH BODY MASS INDEX ACROSS SIX COUNTRIES: FINDINGS FROM THE BETTER4U CONSORTIUM

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BACKGROUND

Body mass index (BMI) does not completely capture the metabolic abnormalities associated with adiposity. Within BETTER4U, we investigated the circulating metabolic signature of higher BMI and its variation across populations.

METHODS

This cross-sectional analysis included 314,767 participants from 9 cohorts in 6 countries. Metabolites were measured using Nightingale Health (7 cohorts; n = 312,757), Biocrates (n = 1,800), or Metaboneer (n = 210). Harmonized regression models assessed associations with BMI, adjusting for age and sex and accounting for familial clustering where applicable. Nightingale estimates were pooled using random-effects meta-analysis. The other platforms were analyzed separately. Platform-specific Bonferroni thresholds accounted for the effective number of independent tests.

RESULTS

Higher BMI was associated with triglyceride-rich lipoproteins, particularly large and very large very-low-density lipoprotein (VLDL) measures, as well as branched-chain amino acids, glycerol, and glycoprotein acetyls. In Nightingale meta-analyses, BMI regression coefficients were 1.24 (95% CI 0.74-1.75) for glycerol, 1.14 (0.59-1.68) for glycoprotein acetyls, and -1.62 (-2.48 to -0.76) for free cholesterol in large high-density lipoproteins (HDL). Large and very large HDL measures demonstrated inverse associations. Biocrates identified positive associations with branched-chain and aromatic amino acids and inverse associations with selected phosphatidylcholines, lysophosphatidylcholines, glycine, and glutamine. Metaboneer identified inverse associations with choline, phosphorylcholine, and glycine, with no positive associations surviving correction.

CONCLUSIONS

Higher BMI was associated with a coordinated pattern of lipoproteins and amino acids. These findings provide a basis for investigating metabolomic markers alongside BMI in personalized obesity prevention within BETTER4U.

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