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Resumo das Comunicações

PEDIATRIA

COMUNICAÇÕES ORAIS

PED_01
NEWBORN LIFE SUPPORT: RISK FACTORS FOR ADVANCED RESUSCITATION

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Introduction: Transition to extrauterine life involves significant physiological changes for every newborn. Although most neonates start breathing on their own, about 10% require assistance at birth, and approximately 1% require advanced resuscitation. This study aimed to identify antepartum, intrapartum, and newborn factors associated with the need for advanced neonatal resuscitation.

Methodology: Cross-sectional study conducted on neonates born at a Portuguese level II hospital from January 2018 to December 2020. Descriptive and inferential statistical analysis using chi-square test was performed in SPSS®27.0.1, with statistical significance set at $p<0.05$.

Results: Of 4604 neonates included, 97% received routine care and 3% (n=143) required extrauterine transitional support, of which 19% (n=27) consisted in advanced measures such as intubation, chest compressions, and/or vasoactive drugs.

For inferential analysis, newborns were divided into three groups according to gestational age. In the group of term and late preterm newborns (n=102), factors significantly associated with neonatal resuscitation comprised intrapartum bleeding (p=0.002), prolapsed cord (p=0.002), and need for urgent or emergent c-section (p<0.001). In the group of moderately preterm infants (n=26), a significant correlation was found between neonatal resuscitation and chorioamnionitis (p=0.043), intrapartum bleeding (p=0.043), vacuum-assisted delivery (p=0.043), emergent c-section (p=0.011), and low birth weight (p=0.031). In the group of extremely preterm newborns (n=15), the need for urgent or emergent c-section (p=0.003) and extremely low birth weight (p=0.038) were factors significantly correlated with advanced reanimation. These results support findings of previous studies.

Conclusion: In the delivery of effective neonatal resuscitation, being prepared is the first and most important step and crucial for optimal outcomes. With this study, the authors intended to highlight the risk factors that should be considered, optimizing the ability to predict and to prepare the need for advanced resuscitation.

PED_02
IT IS NOT AUTISM, IT IS SENSORY PROCESSING DISORDER: ABOUT A CASE REPORT

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Introduction: Autism spectrum disorder (ASD) and sensory processing disorder (SPD) are complex disorders related to brain development with negative impact on how the brain perceives and responds to external and internal stimuli. However, the differentiation between SPD and ASD can be challenging, as they may have very similar presentations.

Case report: Herein is presented the case of a 2.5-year-old boy referred to the Child Psychiatry Department for communication deficits, avoidance of eye contact, and reduced socio-emotional reciprocity. In the first consultation, the mother mentioned a language delay, diminished use of gestures with communicative intent, little interest in social interaction, and tactile defense behaviors. In clinical observation, absence of pleasure in interaction and difficulties in maintaining reciprocal play, little use of verbal communication, and poor eye contact were noticed. Hand flapping was observed when stimulated. With this clinical picture, the hypothesis of ASD was raised. The boy's mental development was assessed, scoring below normal. Assessment of his sensory profile showed a sensory search for multiple stimuli, resulting in psychomotor agitation and disorganized behavior, and weaknesses in motor planning and coordination. The child started occupational therapy with sensory integration and play strategies according to the DIR Floortime model to promote interaction, relationship, and communication between the child and adults. A very positive evolution was noted during follow-up: the boy began to seek interaction to share interests, initiated joint attention, and maintained reciprocal play for periods; improved the constancy of eye contact; started using a wide range of gestures with communicative intent; and improved the vocabulary. Rigid or repetitive play was not observed or mentioned. The final diagnosis according to the Diagnostic Classification (DC: 0-5™) was SPD and global developmental delay.
Discussion: Footage of the child before and after months of intervention allowed to evaluate his progress and to establish a differential diagnosis. In SPD, difficulties in processing sensory information can lead to behavioral issues, including difficulties in self-regulation, social participation, and inattentiveness, frequently leading to the misdiagnosis of ASD. Both SPD and ASD show decreased connectivity in parieto-occipital tracts involved in sensory perception and multisensory integration. However, in ASD, there is impaired connectivity in temporal tracts thought to subserve social-emotional processing. This case emphasizes the importance of a comprehensive assessment and appropriate referral according to the child's clinical situation and individual context.

PED_03

EVALUATION OF THE USE OF M-CHAT SCALE IN PRIMARY HEALTHCARE

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Introduction: Autism spectrum disorder (ASD) is a chronic neurodevelopmental disorder in which early intensive therapeutic intervention can significantly alter the course of the disease. The Portuguese National Programme for Child and Adolescent Health and the American Academy of Pediatrics recommends systematic screening for ASD using the Modified Checklist for Autism in Toddlers (M-CHAT-R) between the 18th and 24th months. If a moderate risk is identified, the Modified Checklist for Autism in Toddlers Follow-up (M-CHAT-R/F) should be used.

To date, there are no studies assessing M-CHAT application rates in primary healthcare in Portugal. The aim of this study was to evaluate the knowledge and use of M-CHAT-R and M-CHAT-R/F in this setting and reinforce the importance of these scales on ASD screening.

Methods: In this descriptive cross-sectional study, residents and specialists in Family Medicine working in the Northern Regional Health Administration were surveyed about their knowledge regarding M-CHAT-R and M-CHAT-R/F scales and how often they use them. Reasons for not using these tools, opinion about integrating them into the digital work platform, and the utility of receiving training on how to use them were also assessed. Data was collected through *Google Form*® questionnaires and analyzed using *SPSS*®.

Results: Of 250 participants, 76% (n=190) were acquainted with the M-CHAT-R scale, but only 21% (n=52) applied it consistently. A total of 39% (n=98) of participants used it in suspected ASD cases. The main reasons for not applying the scale were limited time, lack of experience with M-CHAT-R, and forgetfulness. Regarding M-CHAT-R/F, only 31% (n=77) knew the tool, which was the main reason for not using it. Forty-five percent (n=113) of participants admitted referring to a neurodevelopmental specialist without applying these tools when suspecting ASD. Finally, 81% (n=202) admitted using the M-CHAT-R scale more often if it was integrated into the digital work platform. Most participants surveyed (72%; n=179) considered receiving training in this subject useful.

Conclusion: Despite recommendations and documented advantages of using M-CHAT-R and M-CHAT-R/F in the primary healthcare setting, these tools are not routinely applied. However, using them only in suspected ASD cases may lead to underdiagnosis of milder cases, only identifying severe ones. It is important to increase the systematic use of these tools to promote early diagnosis, integrating them into digital work platforms and providing training to professionals potentially using them could be effective actions.

PED_04

16P11.2 DELETIONS AND DUPLICATIONS DETECTED BY MLPA IN THE CYTOGENETICS UNIT OF CGMJM/CHUPORTO: AN EIGHT-YEAR EXPERIENCE

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Introduction: The 16p11.2 chromosomal region is susceptible to deletions and duplications that have been reported to represent some of the most frequent genetic etiologies of autism spectrum disorder (ASD) and other neurodevelopmental disorders. 16p11.2 deletions and duplications consist of alterations in the number of copy number variants (CNVs) within that region, and both predispose individuals to ASD, intellectual disability (ID), epilepsy/seizures, and DF/CA with high penetrance, in addition to several other common phenotypes. Genetics Home Reference estimates that 16p11.2 deletions and duplications affect about 3/10,000 individuals each. This study presents an eight-year experience of CNV detection in the 16p11.2 region using SALSA® MLPA® Probemix P343 (MRC-Holland™).

Methods: Between 2014 and 2021, 505 individuals from different medical specialties were referred to the Cytogenetics Unit of Centro de Genética Médica Doutor Jacinto Magalhães of CHUPorto for genetic analysis, which was conducted using SALSA® MLPA® Probemix P343 (MRC-Holland™). The clinical indications for referral varied and included namely autism, ID, developmental delay, obesity, and family members with CNVs.

Results: Forty-five (8.9%) patients presented with CNVs in the 16q11.2 region, including 33 microdeletions and 12 microduplications, mostly in the BP4-BP5 region.

Conclusion: Although 16p11.2 deletions are probably the most well-known CNVs linked to autism, and deletions in this region have been detected in as many as 1% of individuals with ASDs, five different 16p11.2 CNV syndromes are reported in Orphanet. MLPA is a highly valuable and cost-effective technique for the detection of genomic CNV unbalances that are not detectable by standard conventional cytogenetics. Therefore, the study of the 16p11.2 region using MLPA may enable appropriate counseling for patients and their families, offering them clinical guidance personalized to their pathology and needs. The authors enhance the benefit of a strong collaboration between clinical professionals and laboratory geneticists in assuring the best genotype-phenotype recognition, particularly in cases of rare small genomic aberrations.

PED_05

PEDIATRIC STROKE – WHAT TO EXPECT?

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Introduction: Despite being a rare condition, stroke is one of the main causes of morbidity and mortality in neonates and children. Early recognition is crucial to improve the outcomes and quality of life of patients and their families. Herein is reported the clinical course and prognosis of patients diagnosed with stroke followed at a secondary hospital over the last 12 years.

Methodology: This was a retrospective and descriptive study based on data retrieved from the clinical files of patients with a history of stroke in the last 12 years and followed in a Pediatric Neurology consultation of a secondary hospital.

Results: Seven patients aged between 2 months and 12 years diagnosed with stroke in the perinatal period were included. Only one patient had first-degree relatives with factor V Leiden and prothrombin III mutations. Two patients had hemorrhagic stroke, and the remaining had ischemic stroke with initial seizure presentation on the second day of life. The etiological study disclosed the following prothrombotic causes: congenital heart disease, heterozygous prothrombin variant (G/A 20210), increased antithrombin III, and homozygosity for allelic variants MTHFR: 677C>T and PAI-1:(4G). The main sequelae observed corresponded to motor deficits.

Conclusion: In this small sample of pediatric patients, all strokes occurred in the neonatal period, with manifestations in the first two days of life. Ischemic stroke presented a more favorable course compared to hemorrhagic stroke. Despite the severity of this condition, proper follow-up can reduce the associated mortality. Early recognition of warning signs and thorough follow-up are key.

PED_06

BRAIN AND SPINE IMAGING FINDINGS IN PEDIATRIC PATIENTS WITH SICKLE CELL DISEASE

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Introduction: Sickle cell disease (SCD) is an autosomal recessive hemoglobinopathy that manifests as multisystem ischemia and infarction, as well as hemolytic anemia. Although its epidemiology is highly variable depending on the geography, SCD is a relatively uncommon condition in Western Europe.

The study's aim was to report brain and spine imaging findings of pediatric SCD, a condition that can have a broad and sometimes quite unusual spectrum of manifestations/complications and thus be disregarded.

Methodology: Descriptive retrospective study of brain and spine imaging findings of pediatric patients with SCD followed at a single level III pediatric hospital in Lisbon between 2010 and 2022.

Results: Due to morphological changes that occur in red blood cells in SCD, ischemia/infarction is the main multisystemic manifestation in these patients. Different patterns of ischemic brain injury and associated vasculopathy can occur, but also hemorrhagic complications, namely parenchymal, extra-axial, or even epicranial. Fat or septic brain embolization may also occur, with typical imaging patterns. Bone infarction, whether of the vault or spine, is relatively common in these patients. Increased susceptibility to infections is also observed due to functional asplenia, which justifies some cases of brain or spinal abscess and osteomyelitis observed.

Conclusion: Recognizing and being familiar with brain and spine complications of SCD, even those less commonly described in the literature, can allow prompt diagnosis and prognostic stratification and implementation of measures to control progressive vasculopathy, protecting against early and end-organ damage.

PED_07

EMERGING SKIN MANIFESTATIONS IN CHILDREN WITH COVID-19 – ABOUT FOUR CLINICAL CASES

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Background: The COVID-19 pandemic had an impact on almost all countries worldwide. The overall clinical presentation, course, and outcomes of SARS-CoV-2 infection, as well as the cutaneous manifestations of COVID-19 in childhood, differ from those of adults. Certain manifestations are more frequent in children, namely skin manifestations, with the most frequently reported in the literature being maculopapular, chilblain-like, urticarial, vesicular, livedoid, morbilliform, and petechial lesions. However, the frequency of COVID-19 cutaneous manifestations is difficult to ascertain, and the association of certain skin manifestations with disease severity is unclear.

Case description: Herein are presented the cases of four children aged between 20 months and 4 years who developed rash between days 1–7 of COVID-19 infection, with variable presentation (maculopapular, urticarial, papulovesicular, and morbilliform lesions). Two cases were associated with pruritus. Palmoplantar involvement was not observed. Most children were asymptomatic (n=3), with only one child presenting associated fever and rhinorrhea. Treatment included antihistamines in all cases, with the addition of topical corticosteroids in one case. All children showed complete rash resolution and COVID-19 infection recovery without complications.

Discussion: Rash caused by COVID-19 infection seemed the most likely diagnosis in the four cases presented, due to the temporal association between the infection diagnosis and the rash development. Most cases were asymptomatic and completely resolved without complications.

At the beginning of the COVID-19 outbreak, skin manifestations, when present, were not valued. As this virus continues to silently spread, mainly through asymptomatic carriers, accurate and rapid identification of these cutaneous manifestations is essential for early diagnosis and favorable prognosis.

PED_08

LOW-EXPRESSIVITY GONADOSOMATIC MOSAICISM IN HEALTH AND DISEASE: ILLUSTRATING THE DIFFICULTY AND IMPORTANCE OF GENETIC COUNSELLING

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Introduction: Mosaicism is a complex post-zygotic mechanism of genetic variation with particularly challenging genetic counseling. Mosaicism can be somatic, germline/gonadal, or gonadosomatic, due to highly varying expressivity in each tissue. Clinically, a disease-causing pathogenic variant in mosaic can lead to phenotypes with different degrees of severity due to the variable percentage of affected cells in each specific tissue. When a mosaic variant does not phenotypically manifest but is present in germline cells, there is a (usually non-predictable) risk of disease in the offspring. Herein are presented two cases of genetic conditions inherited from a parent with low-expressivity gonadosomatic mosaicism.

Case report: The first case refers to a 2-year-old female patient with normal growth and development who presented with prenatal and postnatal microcephaly. The patient was referred to the Ophthalmology Clinic due to strabismus, and a salt-and-pepper retinopathy pattern was observed. Genetic testing was remarkable for a pathogenic nonsense variant in the *KIF11* gene, which confirmed the diagnosis of microcephaly with or without chorioretinopathy, lymphedema, or mental retardation. Both parents were healthy and had no microcephaly. The familial variant was detected in approximately 10% of paternal blood cells, with the father being referred for ophthalmological testing at that point.

The second case refers to an 8-year-old female patient referred to the Medical Genetics Clinic due to ataxia, dysarthria, and early-onset absence seizures. Electroencephalogram detected epileptogenic activity in the right frontal lobe at rest, and genetic testing uncovered a pathogenic missense variant in the *SLC2A1* gene, establishing the diagnosis of GLUT1 deficiency syndrome. While none of the parents had a history of seizures or other neurological symptoms, the same variant was detected in approximately 9% of paternal blood and epithelial cells.

Conclusion: This study describes two cases of gonadosomatic mosaicism affecting blood cells and with transmission to offspring that illustrates the extreme importance of genetic counseling, not only for the prevention of further affected progeny but also for the detection of early/silent phenotypes that may be associated with low-expression mosaic variants, allowing proper follow-up of potential disease manifestations.

PED_09
FAILURE TO THRIVE AND LARYNGOMALACIA – SIGNS OF SEVERITY

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Introduction: Laryngomalacia (LM) is the most frequent cause of stridor in newborns. Although noisy breathing can be a source of concern and anxiety to parents, the vast majority of cases resolve within 12 to 18 months with conservative measures. However, some (few) patients experience significant consequences, such as apnea, cyanosis, feeding difficulties, and failure to thrive, and a multidisciplinary approach is required in these cases.

Case report: A full-term 1.5-month-old female infant, endoscopically diagnosed with type II LM (Olney classification) and gastroesophageal reflux since the second day of life, presented to the Pediatric consultation with labored breathing, fatigue while breastfeeding, and worsening inspiratory stridor. Objectively, she presented stridor at rest, vigorous chest wall retraction (video 1), and inspiratory stridor on cardiopulmonary auscultation. Additionally, poor weight gain was documented, with an increase of 10 grams per day since the 12th day of life. Endoscopic assessment by Otorhinolaryngology confirmed an omega-shaped epiglottis, shortened aryepiglottic folds, and edema surrounding the arytenoid region. Arterial blood gas test showed hypercapnia (pCO_2 50 mmHg) and compensatory HCO_3 elevation (28.3 mmol/L) without acidosis. Hemogram and biochemistry analysis were normal. The child was admitted for monitoring of respiratory symptoms and study of failure to thrive.

During inpatient care, irrespective of feeding modifications (supplementation with high-calorie formula, texture augmentation, acid suppression therapy) and after exclusion of other causes of failure to thrive, the child maintained a weight gain of only 5 grams per day. Since respiratory symptoms remained exuberant, surgical intervention was selected as treatment of choice. Supraglottoplasty was performed at the age of three months, without intercurrences. In Pediatric consultation 15 days after surgery, pronounced clinical evolution was observed, with an increase of 64 grams per day and complete resolution of respiratory symptoms (video 2).

Discussion: LM is a remarkably frequent cause of stridor in infants, but only a few cases have a significant clinical impact. In the present case, surgical treatment was of paramount importance to ensure normal growth in the infant with failure to thrive, emphasizing the impact of a multidisciplinary approach in these cases.

PED_10
CARDIAC DISEASE IN NOONAN SYNDROME

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Introduction: Noonan syndrome (NS) is an autosomal monogenic disorder involving the RAS-MAPK pathway. Distinctive facial features, short and webbed neck, chest deformity, and short stature are some of the characteristic findings. Cardiovascular abnormalities are present in 80-90% of patients and include congenital heart defects and hypertrophic cardiomyopathy.

Aim: To analyze the spectrum of cardiac phenotype and genotype and clinical outcomes of a series of patients with Noonan and Noonan-like syndrome.

Methods: Retrospective case series of pediatric patients with NS followed at a tertiary hospital. Clinical history and cardiac and genetic studies were assessed.

Results: A total of 15 patients (60% female) were evaluated, with a median age at diagnosis of 2.5 years (minimum 3 days; maximum 17 years). One case had prenatal diagnosis. Three patients had a family history of NS. Deleterious *SOS1* and *PTPN11* variants were the more commonly involved, followed by *BRAF* and *RAF1* variants. Hypertrophic cardiomyopathy (HCM), pulmonary stenosis (valvar, subvalvar, and supravalvar), and atrial septal defect (ASD) were the most common findings (Table 1). Five patients were under beta-blocker therapy and three patients required surgical intervention (ASD closure, pulmonary valvotomy/right ventricular outflow tract reconstruction [n=2] and left ventricular myomectomy [n=1]). One patient died at 80 days of life with cardiac and respiratory failure (severe HCM). One patient developed type B lymphoblastic leukemia. Other relevant findings included neurodevelopment delay in nine patients and abnormal lymphatics in one.

Conclusions: HCM was the most prevalent cardiac lesion in this cohort and carried the worst prognosis. Early HCM treatment in NS is under investigation, including with MEK/mTor inhibitors. Close and lifelong cardiac follow-up and multidisciplinary care are mandatory in NS patients to address complications potentially arising during adolescence and adulthood.

Table 1. Cardiac disorders in genes causing Noonan and Noonan-like syndrome in the study cohort (N=15)

Gene	HCM (N)	PVS (N)	Supravalvar PS (N)	Subvalvar PS (N)	ASD (N)	VSD (N)	MVanomaly (N)	AoV anomaly (N)
PTPN11	3	1			1			1
SOS1	2	3			1			1
RAF1	2							
LZTR1	1	1	1		1	1		
BRAF	2	2			1	1	1	
RIT1	1	1		1				
HUWE1						1		

ASD, atrial septal defect; AoV, anomaly aortic valve anomaly; HCM, hypertrophic cardiomyopathy; MV, mitral valve; N, number of patients; PS, pulmonary stenosis; PVS, pulmonary valve stenosis; VSD, ventricular septal defect

PED_11
ANTI-N-METHYL-D-ASPARTIC ACID RECEPTOR ENCEPHALITIS: CLINICAL VARIABILITY

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Introduction: Anti-N-Methyl-D-aspartic acid receptor (anti-NMDAR) encephalitis is the most common autoimmune encephalitis in children. The diagnosis is challenging as it produces a multifocal inflammation and leads to a variety of symptoms, including neuropsychiatric manifestations.

Methods: Retrospective descriptive study of patients with anti-NMDAR encephalitis admitted to the Neuropediatrics Unit of a tertiary hospital from January 2009 to March 2022.

Results: A total of 12 patients were diagnosed during the study period. The median age at diagnosis was 8.5 years (range 2-17), and there was a slight male preponderance (7/12; 58.3%). Psychiatric disorders were present in 11 cases (91.7%), with agitation/aggressiveness towards others being the most common presentation (7/12; 58.3%), followed by behavioral changes (4/12; 33.3%), such as social isolation, emotional lability, and anxiety. One patient had psychotic symptoms as single presentation. Movement disorders were present in 11 patients, namely involuntary movements of the limbs (7/12; 58.3%), followed by orofacial dystonia (4/12; 33.3%) and dystonia of other body parts (4/12; 25.0%). Consciousness fluctuations were identified in 75.0% of patients, and 50.0% had sleep disturbances, seizures, and language changes, such as mutism, interchanging of words in a sentence, or speech not understood by others. The least common sign was autonomic dysfunction, which presented as hypertension in two patients. Two patients presented with prodromal disease characterized by fever, vomiting, and odynophagia. All patients had NMDAR antibodies in liquor and blood, and half required admission to the Intensive Care Unit.

Conclusion: This patient cohort showed psychiatric symptoms and movement manifestations as the most prominent features of anti-NMDAR encephalitis. A rare case of monosymptomatic psychiatric manifestations was identified, showing the importance of considering encephalitis in the differential diagnosis of these patients. Autonomic instability was the least frequent sign. Only two patients presented with the typical prodromal phase.

With this study, the authors intend to raise attention to the clinical variability of autoimmune encephalitis and remind clinicians to consider this condition in the differential diagnosis of patients with varying presentations, such as acute behavioral changes, seizures, language dysfunction, and dyskinesias. By doing so, the authors hope to promote earlier diagnosis with an impact on prognosis, since prompt recognition and treatment generally induce significant clinical improvement.

PED_12
NEONATAL THROMBOCYTOPENIA: CASE REPORT OF A RARE ETIOLOGY

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Introduction: Thrombocytopenia is a common finding in the neonatal period. Most cases are transitory and secondary to non-hematological diseases. Herein is presented the case of a rare cause of severe congenital thrombocytopenia.

Case report: A 17-day-old full-term male newborn presented with spontaneous rectal bleeding. He was the first child of a healthy non-consanguineous couple with maternal history of male infant deaths. The prenatal and early postnatal periods were uneventful, and the boy was exclusively breastfed. The hypothesis of cow's milk protein allergy was initially presumed. On reevaluation, at one month of age, he presented anemia (Hb 8.8 g/dL) and severe thrombocytopenia (17.000/mcL). The crossmatch of maternal serum against paternal platelets was positive. Antibodies against human platelet antigen type IV and human leucocyte antigen class I were identified in the mother. The hypothesis of neonatal alloimmune thrombocytopenia was assumed, and the patient was treated with intravenous immunoglobulin (1 g/kg) and platelet transfusion, with adequate post-transfusion platelet count.

In the following weeks, the patient required periodic platelet transfusions. He was transferred to a level III hospital for further workup. Both immature platelet fraction (IPF) and mean platelet volume were normal, but the platelet histogram suggested microthrombocytopenia, subsequently confirmed by Forward Scatter obtained by flow cytometry. The severe thrombocytopenia, normal IPF, and adequate post-transfusion platelet count pointed to a central etiology. The patient's male gender, maternal history of male deaths, and microthrombocytopenia raised the hypothesis of an X-linked disease, namely associated with *WAS* gene mutations. Gene sequencing confirmed a pathogenic variant in this gene, establishing the diagnosis of Wiskott-Aldrich Syndrome (WAS). Additional studies revealed marked T-cell lymphopenia and dysfunction.

The child had multiple opportunistic infections during follow-up, including *Pneumocystis jirovecii* pneumonia and severe *Cytomegalovirus* infection, with multiorgan dysfunction. He was submitted to an allogeneic hematopoietic stem cell transplant at 11 months but died at 20 months after multiple complications.

Discussion: WAS is an X-linked disorder characterized by microthrombocytopenia, recurrent infections, and eczema. Despite its rarity, the multiple features highlighted in this case, along with prompt multidisciplinary approach, led to a swift diagnosis.

PED_13
CITRIN DEFICIENCY PRESENTING WITH FAILURE TO THRIVE AND DYSLIPIDEMIA IN INFANCY

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Background: Citrin deficiency is an autosomal recessive inborn error of metabolism with broad clinical spectrum. Due to a defect in NADH shuttle system function, both hepatic glycolysis and lipogenesis are impaired, leading to hepatic energy deficit. Three main phenotypes have been described: neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD) during infancy, failure to thrive and dyslipidemia caused by citrin deficiency (FTTDCD) intermediate type in older children, and recurrent hyperammonemia with neuropsychiatric symptoms in citrullinemia type 2 (CTLN2).

Case report: A 6-month-old female, born at term to non-consanguineous parents, was referred to the hospital after a symptomatic episode of non-ketotic hypoglycemia (36 mg/dL). She had failure to thrive since one-month-old with weight and length below the 3rd percentile, and head circumference in the 15th percentile. Food diversification began at the age of four months, with aversion to fruit. No history of jaundice was reported, and newborn screening results were normal.

The laboratory study revealed elevated alaline aminotransferase (94 U/L) and aspartate aminotransferase (91 U/L), normal bilirubin, alkaline phosphatase and —glutamyltransferase, and altered lipid profile (triglycerides 319 mg/dL, total cholesterol 114 mg/dL, HDL 19 mg/dL). Ammonia and blood galactose were normal. Abdominal ultrasound revealed diffuse liver steatosis without hepatomegaly. Plasma amino acids indicated elevated citrullinemia (128 µmol/L), methionine (199 µmol/L), and threonine (264 µmol/L), suggesting citrin deficiency, and molecular analysis showed homozygosity for the c.1231-1G>A variant in the *SLC25A13* gene.

The girl was started on a high-protein and low-carbohydrate diet with medium-chain triglyceride supplementation, with normalization of growth curves (15-50th percentile) and biochemical improvement. Currently, at the age of four years, she has no hypoglycemia episodes and presents normal hepatic and lipid profile and liver ultrasound without steatosis.

Discussion: The present case is compatible with FTTDCD phenotype, traditionally characterized by post-NICCD growth retardation, hypoglycemia, severe fatigue, and dyslipidemia presenting after the first year of life. This atypical presentation contributes to elucidating the clinical spectrum of the disease and illustrates the importance of considering this diagnosis in infants with dyslipidemia, poor weight gain, and/or steatosis.

Dietary intervention is key for treatment effectiveness and has been suggested to potentially prevent long-term CTLN2 complications.

PED_14
EARLY NEURODEVELOPMENTAL MILESTONES IN AUTISM SPECTRUM DISORDER

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Introduction: Autism spectrum disorder (ASD) is a chronic and complex neurodevelopmental disease characterized by persistent deficits in social communication and social interaction and restricted, repetitive patterns of behavior, interests, and activities. The condition is one of the most frequent neurodevelopmental disorders, with a prevalence of around 0.7–1%. The presentation usually occurs within the first years of life. As there are no known biomarkers, the diagnosis remains strictly clinical, based on history and observation. Early diagnosis and high-quality intervention are determinant in improving outcomes, which also greatly depend on the presence of comorbidities.

Methods: The aim of this study was to analyze the median age of acquisition of early neurodevelopmental milestones in children with ASD born since 2016 and followed at the Neurodevelopment Unit of Centro Materno Infantil do Norte (tertiary hospital). Data were retrieved from patients' electronic medical records. In addition to epidemiological data, early psychomotor developmental milestones, namely social smile, cephalic control, sitting, walking, first words, first phrases, and age of sphincter control, were recorded and analyzed.

Results: A total of 127 children were included in the study, 85% of which were male. The main reasons for referral to the Neurodevelopmental consultation were language disorder and global developmental delay. All children had acquired head control and sitting and walking competencies within the expected age. The median age of social smiling was 2 months. One hundred children (78.7%) said their first words at 18 months. The median age of sentence onset was 44 months, with less than half of children acquiring this milestone at the time of this study. Daytime and nighttime bladder control was also a delayed acquisition.

Conclusion: In the present study, the earliest developmental milestones were acquired within the expected ages. The acquisition of first words and especially sentence production were delayed in most children. Sphincter control was also a late acquisition. These findings underline the importance of delayed language acquisition as an early marker of possible ASD when associated with other symptoms evoking this pathology.

PED_15

LIPSCHÜTZ ULCER AS A POSSIBLE SARS-COV-2 MANIFESTATION

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Introduction: Lipschütz ulcer (LU) is a rare and self-limited condition characterized by rapid onset of painful ulcer of the vulva or lower vagina. It is more frequent in adolescent girls and has no association with sexually transmitted infections. The condition may be preceded by influenza-like symptoms, and some reports describe a link with bacterial or viral infections, mainly Epstein-Barr virus (EBV). It is not clear yet how these infections lead to the development of genital ulcerations.

Herein is described a case of LU in the setting of SARS-CoV-2 infection in an adolescent girl.

Case report: A previously healthy 11-year-old girl presented to the Emergency Department with itching and burning of the external genitalia with two days of evolution resulting in urinary retention and hypogastric pain. She had been diagnosed with SARS-CoV-2 infection five days earlier, which was confirmed by RT-PCR, with fever in the first three days of infection and no other symptoms.

Physical examination revealed hypogastric tenderness associated with vesical globus. On perineal inspection, three vulvar ulcers were observed (Figure 1). The most prominent was located on the left labia minora with 12 mm, red-violaceous border, and deep and necrotic base. The other lesions were two symmetrical smaller ulcers located on the right ("kissing lesions"). Imperforate hymen was confirmed, and exudate at vaginal introitus was detected.

The girl had not yet reached menarche and was not sexually active. She had no history of recurrent oral or genitalia ulcers or recent trauma and no family history of Behcet's disease or intestinal inflammatory disease.

Workup revealed no anemia or leukocytosis, normal C-reactive protein and erythrocyte sedimentation, negative serology for Cytomegalovirus, EBV, Parvovirus, Herpes *simplex* virus types 1 and 2, Syphilis, HIV, and HCV. The virus panel performed on the ulcer and vaginal exudate was negative.

After Gynecology consultation, the patient was discharged with topical treatment with repair cream, fusidic acid, and 2% lidocaine for pain control. Clinical improvement was reported ten days later, with only a residual scar observed on physical examination (Figure 2).

Discussion: LU is an uncommon cause of genital ulcers that should be considered in the differential diagnosis of painful vulvar ulceration, especially in non-sexually active adolescents.

Few LU cases have been reported in association with SARS-CoV-2 infection.

Complete resolution is expected, usually without scarring, with treatment and pain control crucial to avoid complications like acute urinary retention.

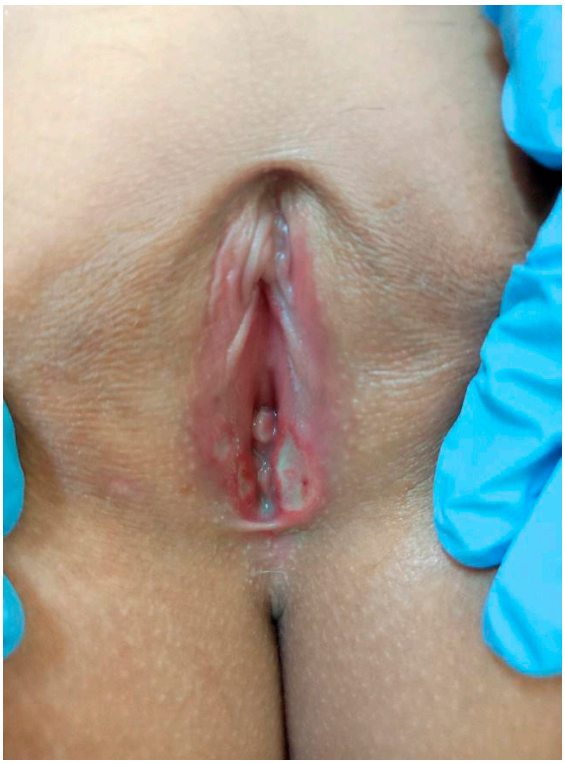

Figure 1. Physical exam on admission

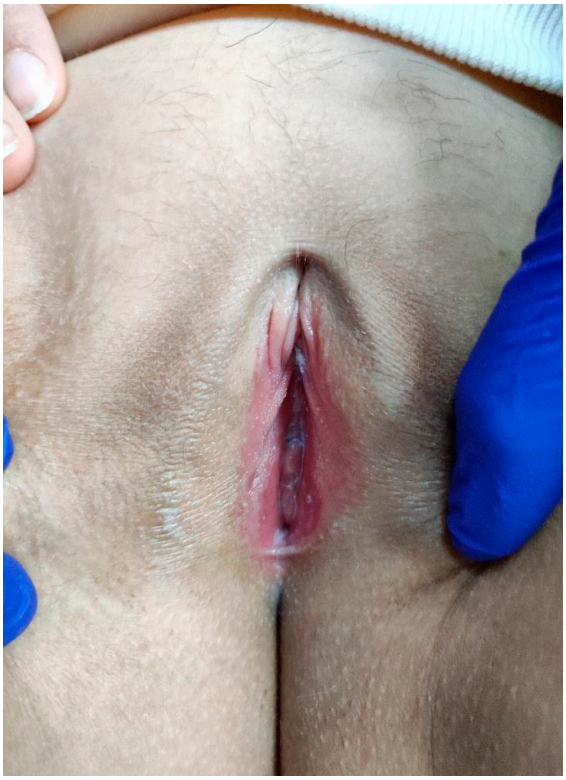

Figure 2. Physical exam on reassessment

PED_16

ECTHYMA GANGRENOSUM IN AN INFANT WITH NEUTROPENIA

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Introduction: Ecthyma gangrenosum (EG) is characterized by the presence of erythematous macules that evolve into necrotic ulcers with a central gray-black eschar. It is a rare skin condition usually occurring in immunocompromised patients, although also reported in healthy individuals. The condition might be associated with bacterial sepsis caused by *Pseudomonas aeruginosa*. Early recognition with prompt administration of antimicrobial therapy is critical for successful treatment.

Case report: Herein is described the case of a previously healthy ten-month-old female infant who presented with three ulcerated skin lesions in the anogenital region. She reported fever and diarrhea one week before admission. The physical examination revealed three necrotic ulcers with approximately 1 cm in diameter, an erythematous ring, and purulent content in the anogenital area (Figures 1 and 2). Laboratory tests showed leucocytes of 4810/uL and absolute neutrophil count of 50/uL. The patient was empirically treated with intravenous piperacillin-tazobactam and vancomycin. Wound culture was positive for *Pseudomonas aeruginosa*, and the patient was started on intravenous piperacillin-tazobactam according to drug susceptibility tests. Blood culture was negative. Immunological screening results, including serum immunoglobulins and complement (C3 and C4) levels and lymphocyte subsets, were unremarkable. Bone marrow aspirate showed normal cellularity with a reduction of segmented neutrophils. Neutrophil-specific antibodies were positive. After treatment, the patient's condition stabilized, and the necrotic area decreased. Genetic study revealed heterozygous mutations of uncertain clinical significance on *CSF3R*, *ACTB*, and *IRF8* genes. One year later, the girl still presented chronic neutropenia (120–230/μL) and monocytosis without serious infections.

Discussion: EG is characterized by fatal cutaneous infections caused by Gram-negative organisms, usually *P. aeruginosa*. Once the diagnosis is established, immunodeficiency should be ruled out. In the present case, persistent neutropenia with positive anti-neutrophil antibodies was identified. Close follow-up is required to promptly diagnose and treat possible infections.

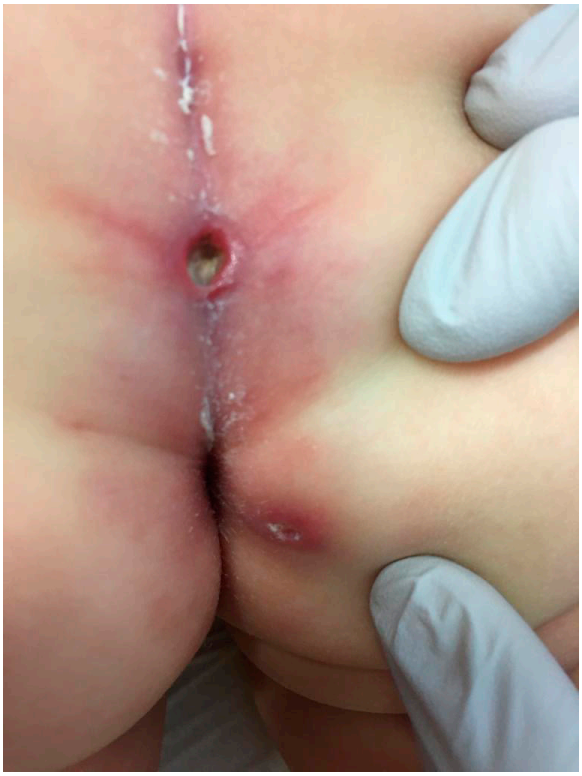

Figure 1

PEDIATRIA

POSTERS

PED_17
POLLEN-FOOD SYNDROME IN THE ADOLESCENT

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Introduction: Pollen-food syndrome (PFS) is an immunoglobulin E (IgE)-immediate allergic reaction characterized by allergic symptoms elicited upon ingestion of raw fruits, vegetables, or nuts in patients with seasonal allergic rhinoconjunctivitis. The estimated prevalence of PFS in patients with pollen allergy is 50–70%. PFS is often termed oral allergy syndrome (OAS) due to mild symptoms such as itching, urticaria, and edema mainly in the lips, mouth, and pharynx that appear after food ingestion. However, reports of systemic reactions such as anaphylaxis have been recently increasing.

Case report: The authors present the case of a 14-year-old male with grass pollen allergic rhinitis who developed PFS to fresh fruits. He reported mild oral pruritus with five years of evolution triggered by apple and pear fruit ingestion, which progressively worsened with time, manifesting as edema and urticaria when eating banana, watermelon, cantaloupe, orange, peach, and melon. The boy denied symptoms associated with other foods and had no family history of allergy.

Initial diagnostic procedures included skin prick tests, which were positive for apple 7 mm, pear 6 mm, banana 5 mm, melon 4.5 mm, orange 5 mm, peach 6 mm, and histamine 7 mm, and continued with prick-to-prick tests that were positive for apple, pear, peach skin and pulp, banana, orange, melon, and cantaloupe. Since the patient was polysensitized and had some dietary restrictions, an immuno-solid-phase allergen chip (ISAC) test was performed to clarify the specific molecular allergens involved in the disease, revealing high sensitization to grass pollen due to genuine sensitization allergens (Phl p 1, 2, 5, 6, 11) and cross-reactivity (profilins: Phl p 12; CCD-bearing proteins: Cyn d 1, Phl p 4). High sensitization to profilins was confirmed, with positive results to all profilins on microarray Bet v 2, Hev b 8, Mer a 1, and Phl p 12. Sensitization to cat and dog dander was also present (Fel d 1 and Fel d 2; Can f 5), without clinical symptoms.

After complete diagnostic workup, an open oral food challenge with cooked apple was performed (cumulative dose of 140 g), which was negative. The patient currently maintains ingestion of cooked apple, with no symptoms, and avoids ingestion of all other fresh fruits. He was treated with oral antihistamines and nasal corticosteroids during pollen season, as well as emergency medications, including oral antihistamines and systemic corticosteroids, and was prescribed automated adrenaline injector to use in case of accidental ingestion.

Conclusions: This clinical case illustrates that ISAC test allows for a better understanding of patients' sensitization profile and provides prognostic information about the severity of reactions. It also emphasizes that a high index of suspicion should be maintained in cases of pediatric patients with pollen allergy.

PED_18
SELECTIVE HYPERSENSITIVITY REACTION TO PARACETAMOL: A CASE REPORT IN PEDIATRIC AGE

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Introduction: Paracetamol is widely used as an antipyretic and analgesic in pediatric age. Hypersensitivity reactions to nonsteroidal anti-inflammatory drugs (NSAID) can be classified as immunologic and non-immunologic, depending on the underlying mechanism. Immune-mediated reactions are less common, and well-documented reactions to paracetamol are rare.

Clinical report: A 12-year-old female with adequate weight and height and updated vaccination, according to the national vaccination program, was observed at the Allergy and Clinical Immunology clinic for two episodes suggestive of NSAID hypersensitivity. She had a personal history of mild persistent rhinitis and intermittent asthma controlled with terbutaline 500 ug turbohaler as rescue medication, nasal fluticasone furoate 27.5 ug twice daily, and bilastine 20 mg as rescue medication. She was allergic to *Dermatophagoides pteronyssinus* and cat dander. The two episodes were 15 days apart, and in both, she reported urticaria, angioedema (periorbital), dyspnea, and wheezing 30 minutes after taking a 500 mg paracetamol tablet (~15 mg/kg) for fever. In both episodes, she was admitted to the Emergency Department and treated with intravenous hydrocortisone 100 mg and clemastine 2 mg, with symptom resolution within 2 hours.

The patient denied contact with other NSAID drugs and was advised to avoid them until conclusion of the diagnostic workup.

A basal lung function test with bronchodilator responsiveness was conducted, which was normal/negative, and a basophil activation test with paracetamol showed no increment of CD63 expression.

Paracetamol open-graded oral challenge with lung function spirometry monitoring was conducted according to the hospital department's protocol. Only minutes after taking 300 mg of paracetamol, the patient experienced urticaria, periorbital edema, cough, conjunctival erythema, and wheezing with a 15% fall in forced expiratory volume in one second (FEV1). A dose of 0.15 mg of intramuscular adrenaline was administered, with full recovery within 15 minutes, and paracetamol hypersensitivity reaction was confirmed.

To identify a safe therapeutic option and confirm a selective reaction, ibuprofen open-graded oral challenge was performed with recommended doses according to weight (10 mg/kg), which was well tolerated.

Discussion: This clinical case refers to a young patient with paracetamol hypersensitivity manifesting as anaphylaxis. A correct diagnostic workup is extremely important and should be conducted by experienced personnel in appropriate facilities. Besides establishing the diagnosis, information on safe therapeutic options and future guidance should be provided to the patient.

PED_19
EFFICACY AND SAFETY OF DIFFERENT PHARMACOLOGICAL TREATMENTS FOR ATTENTION DEFICIT/HYPERACTIVITY DISORDER IN CHILDREN DIAGNOSED WITH AUTISM SPECTRUM DISORDER

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Introduction: Attention-deficit/hyperactivity disorder (ADHD) and autism spectrum disorder (ASD) are neurodevelopmental disorders that commonly co-occur, with shared genetic heritability and social and executive functioning impairment. ADHD is the most common comorbidity in children with ASD. Although there is no pharmacological treatment for core ASD symptoms, pharmacological therapies are used to manage its symptoms and comorbidities. More than 80% of youth with ASD and ADHD have been prescribed medication for ADHD symptoms, and the prescription rates of psychostimulants in ASD individuals are much higher than in the general population. This study aimed to review the efficacy and safety of pharmacological treatments for comorbid ADHD in children with ASD.

Methods: Non-systematic literature review of ADHD treatment in children with ASD.

Results: Psychostimulants are recommended as front-line pharmacological therapy for ADHD in children with ASD. Methylphenidate is an effective first-line treatment, with reports of dose-dependent improvement in joint attention and self-regulation. However, compared with typically developing children with ADHD, its response rate in individuals with ASD and ADHD is lower, and side effects are more severe. Atomoxetine can be considered a good alternative to methylphenidate. Its effect size is similar to the one reported for methylphenidate in children with ADHD and ASD, with fewer intolerable side effects. Atomoxetine appears to have a more pronounced effect on hyperactivity than on inattention in patients with ASD, in contrast with its effect in children with typical ADHD. Clonidine has been reported to elicit an improvement in hyperactivity and irritability symptoms, with limited evidence of improvements in hyperarousal behaviors and social relationships. There are no known safety or efficacy data for amphetamine-based medications in children with ASD. A multicenter open-label assessment of the efficacy and safety of lisdexamfetamine in children with ADHD and comorbid ASD is currently ongoing.

Conclusion: ADHD symptoms may exacerbate ASD symptoms, with children diagnosed with both ASD and ADHD having increased treatment requirements. Treatments for ADHD symptoms in children with ASD appear to be less effective than in typically developing populations, and adverse effects are expected to be greater. Even so, pharmacological therapies play an important role in the management of these patients.

PED_20
EMOTION DYSREGULATION: THE FLIP SIDE OF AUTISM SPECTRUM DISORDER

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Introduction: Children with autism spectrum disorder (ASD) usually present with social communication and interaction deficits, restricted interests, repetitive behaviors, and atypical sensory characteristics, all of which make them more vulnerable to emotion dysregulation. Emotion dysregulation can be defined as failure to properly and effectively regulate emotions. In fact, rates of emotion dysregulation in children with ASD range from 50% to 80%. The present case highlights the preponderance of emotion dysregulation in ASD.

Case description: A 35-month-old female child was referred to the Child and Adolescent Psychiatry consultation for suspicion of ASD. The mother reported that the child had difficulties in interacting with peers and restricted interest in letters and numbers, and presented manual stereotypies. She also referred that the child presented some tantrums when contradicted, with occasional self- and hetero-aggressive behaviors. Six DIR/Floortime model sessions were hence conducted. However, the child's marked emotion dysregulation was so severe that it negatively impacted Floortime sessions. In one session, the child lost control, screamed, cried, and threw herself on the floor when the cereals she was eating in the waiting room were removed. When the doctor physically restrained her, she tried to hit her head against the doctor's body. Additionally, when the doctor removed the boxes she was playing with in the fourth session, the child broke down and had a tantrum, screaming and lying on the floor. Besides ASD with significant sensory demand in several sensory modalities, the child had an evident emotion dysregulation, with difficulties in self-regulation.

Discussion: Emotion regulation deficits in children with ASD can be both reactive and regulatory. These difficulties can be conceptualized as consequences of the core ASD symptoms. An association between poor social abilities and emotion dysregulation has been reported in the literature. A relevant study showed that restricted and repetitive behaviors were the best predictors of emotion dysregulation. This might indicate that individuals with ASD with severe repetitive and restricted symptoms are less able to regulate their emotions due to difficulties in inhibiting ongoing behaviors. All these findings raise the question of whether emotion dysregulation should be considered as part of the core features of ASD or as an associated symptom.

PED_21

A RARE CASE OF WAGRO SYNDROME ASSOCIATED WITH 11P14.2-P13 INTERSTITIAL DELETION: ADVANTAGES OF A MULTIDISCIPLINARY INTERVENTION

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Introduction: Global developmental delay/intellectual disability (GDD/ID) is a variable, heterogeneous manifestation of central nervous system dysfunction, occurring in 1-3% of the general population. The etiology is diverse, and genetic disorders have been identified in about 40% of cases. Chromosomal aberrations stand as the most prevalent genetic abnormalities associated with congenital malformation and GDD/ID. Several syndromes may be easily assumed due to their association with specific dysmorphisms, behavioral abnormalities, and multiple congenital anomalies.

Case report: A 2-year-old male patient was evaluated at the medical center due to aniridia, GDD, cryptorchidism, and obesity. He was the only son of a non-consanguineous couple, with the father presenting Axenfeld-Rieger syndrome. Due to clinical suspicion of WAGRO syndrome —a contiguous gene deletion presenting with (W)ilms tumor risk, (A)niridia, (G)enitourinary abnormalities, Mental (R)etardation, and (O)besity —, the patient underwent multiplex-ligation dependent probe amplification (MLPA) for the 11p14.1-p13 region and CGH array, which confirmed the diagnosis associated with heterozygous deletion of WT1, PAX6, and BDNF genes, arr(GRCh37) 11p14.2p13 (26759936-_34150886 7.39Mb deletion). Karyotype was performed to exclude chromosomal rearrangements, revealing a 11p14.2-p13 deletion and no rearrangements. Wilms tumor screening was initiated, with abdominal ultrasound monitoring every three months. The boy is currently attending an Early Intervention Program.

Discussion: WAGRO syndrome is a rare genomic disorder caused by chromosome 11p13 contiguous gene deletions of variable size (OMIM: #612469). Behavioral problems and GDD/ID affect most of these patients, with 20-52% of individuals presenting with autism spectrum disorder, as in the present case. Cytogenetic study plays a relevant role in the evaluation of a variety of human disorders by allowing the assessment of diagnostic and prognostic outcomes and the evaluation of family risk. In this patient, the 11p13 genotype-phenotype correlation was established and compared with previous 11p13 deletion cases, and the presence of significant genes associated with clinical features in WAGRO syndrome was discussed. Early diagnosis and multidisciplinary intervention are paramount to improving patients' quality of life.

PED_22

DDX3X SYNDROME – ABOUT A CLINICAL CASE

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Introduction: DDX3X syndrome is an emerging cause of neurodevelopmental disorders accounting for 1-3% of cases of global developmental delay (GDD) and intellectual disability. It is mainly caused by a spontaneous heterozygous DDX3X pathogenic variant and typically affects girls due to its location on the X chromosome. The condition is characterized by developmental delay, mainly at the motor and language levels, intellectual deficit, behavioral disorders (autism spectrum disease, hyperactivity, and aggression), and facial dysmorphia and hypotonia.

Case report: A 5-year-old girl with a perinatal history of term delivery by cesarean, Apgar score 8/9, light-for-gestational-age, and with sacrococcygeal pit was referred to the Neurodevelopment consultation due to global psychomotor development delay, predominantly in language, and gross and fine motor skills. She had a personal history of hypotonia associated with feeding difficulties and poor height and weight development until the first year of life. Regarding facial dysmorphia, she presented frontal bossing, hypertelorism, dysplastic ears, small nose with high nasal bridge, small mouth with thin lips, long philtrum, upturned palpebral fissures, and chin hypoplasia. She also presented associated precocious pubarche, intention tremor, gait ataxia, hyperkinetic and anxious behaviors, and self-aggression episodes. A psychometric test was performed, revealing a global intelligence quotient of 60.1. Cranioencephalic magnetic resonance imaging, metabolic study, and genetic study with karyotype, fragile-X molecular study, and array-CGH were normal. Next-generation sequencing study for intellectual disability identified a heterozygous variant in exon 13 of the DDX3X gene. Otorhinolaryngology and Cardiology assessments were performed and shown to be normal, and ophthalmologic evaluation revealed hypermetropia and astigmatism. Psychology assessment identified difficulties mainly in expressive and understanding language and attention deficit. The girl underwent multidisciplinary pediatric follow-up by Neuropediatrics, Neurodevelopment, Pedopsychiatry, Endocrinology, Ophthalmology, and Genetics and was recommended physical, speech, and occupational therapy, hippotherapy, and music therapy, with GDD improvement.

Discussion: DDX3X syndrome should be considered in the differential diagnosis of girls with intellectual deficit and/or GDD. The evaluation and follow-up should be multidisciplinary, taking into account the multisystemic involvement.

PED_23

BEHAVIORAL CHANGES IN ADOLESCENCE: AN UNEXPECTED CAUSE

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Introduction: Behavioral changes in adolescence can have multiple causes, from acute voluntary intoxication and psychiatric pathology to neurological diseases, including infectious causes. In most cases, accompanying symptoms and complementary tests are key determinants to establish a definitive diagnosis.

Case report: A 17-year-old male with a personal history of attention deficit hyperactivity disorder medicated with methylphenidate was admitted to the Emergency Department due to voluntary drug ingestion (10 tablets of acetaminophen, two tablets of diclofenac, and diazepam). He also reported fever (maximum axillary temperature of 40ºC), productive cough, myalgia, headache, and vomiting with three days of evolution. The boy presented with skin pallor, sleepiness, and slurred speech but was oriented in time and space. The remaining physical examination was normal. During the observation period, he presented episodes of verbal aggressiveness accompanied by amnesia, which were unusual according to his mother. On admission, complete and white blood counts showed no changes, C-reactive protein level was 26.4 mg/L, and acetaminophen level was 53 ug/mL (non-toxic). The urine toxicology test was positive for benzodiazepines. Cerebral computed tomography scan was normal, and lumbar puncture showed cerebrospinal fluid (CSF) with pleocytosis (mainly mononuclear cells). The boy was diagnosed with probable encephalitis, admitted, and started on parenteral ceftriaxone, ciprofloxacin and acyclovir. Polymerase chain reaction for respiratory viruses on the second day of admission identified type A influenza. Multiplex panel and CSF culture were negative, including for *Mycoplasma pneumoniae*, and magnetic resonance imaging and electroencephalography showed no changes. During hospitalization, the boy remained apyretic and his behavior normalized, with no further periods of confusion. He was discharged home after seven days with diagnosis of encephalitis probably caused by influenza A infection.

Conclusion: Encephalitis refers to an acute infectious/inflammatory process of the central nervous system characterized by focal neurological deficits and changes in behavior or state of consciousness. Its etiology is diverse, with viruses being the main agents. Although less frequent, influenza A virus can be implicated, especially in adolescents and children presenting with clinical signs suggestive of this infection. Virus isolation in CSF is not always possible, but its identification in respiratory secretions strengthens this diagnostic hypothesis.

PED_24

EXANTHEMA SUBITUM – ABOUT A CLINICAL CASE

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Introduction: Infection with human herpesvirus 6 (HHV-6) is universal in human populations, and almost all children are infected by the age of two. The primary infection can sometimes cause exanthema subitum. HHV-6 is also an established cause of neurological disease, inducing both encephalitis and encephalopathy. One-third of patients evaluated for HHV-6 primary infection present viral DNA in both peripheral blood mononuclear cells and cerebrospinal fluid (CSF), but mainly in CSF.

Case description: A previously healthy 20-month-old child was brought to the Emergency Department (ED) with high fever with 72 hours of evolution associated with vomiting and prostration. No respiratory or other gastrointestinal symptoms were reported, and no changes in physical exam, focal neurological deficits, or seizures were observed.

The initial study showed normal blood count, slight AST/ALT elevation, C-reactive protein <5 mg/L, negative urinary sediment, and normal chest radiograph.

In the ED, the boy remained feverish and somnolent, with Glasgow coma scale (GCS) of 13. Brain computed tomography revealed a "nonspecific focal hypodensity of the left parietal white matter, without associated mass effect..." and lumbar puncture showed normal CSF cytochemical examination.

The boy started treatment with ceftriaxone and acyclovir. On the following day, he became apyretic, and a maculopapular rash emerged, with cephalocaudal progression and palmoplantar involvement.

The study of nasopharyngeal secretions for Respiratory syncytial virus, *Influenza* A and B, and *Mycoplasma pneumoniae* (MP) were negative. Stool virological examination, blood culture, CSF bacteriological analysis, polymerase chain reaction (PCR) for Enterovirus, MP, Parvovirus B19, and *Rickettsia*, and Epstein Barr serologies were also negative. Cytomegalovirus serology was suggestive of past infection. HHV-6 PCR was positive in CSF and blood, and brain magnetic resonance imaging was normal.

The boy had favorable evolution and was discharged clinically well.

Discussion: Although the microbiological study can establish the presence of replicating HHV-6, it cannot establish causality or infection, making the diagnosis challenging

PED_25

MALE *MECP2*-ASSOCIATED DISORDERS – TWO CLINICAL CASES

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Introduction: Methyl-CpG-binding protein 2 (MECP2) is encoded by the *MECP2* gene, located in the subchromosomal region Xq28, required for brain development. *MECP2* mutations are associated with Rett syndrome, more typically in females. In males, these variants have traditionally been considered incompatible with life, but recent evidence suggests that they can be compatible with life and manifest in diverse forms, and have distinct clinical presentations.

Cases: The present study characterized two cases of maternally inherited *MECP2*-associated disorders, one referring to *MECP2* duplication syndrome and the other to a hemizygous pathogenic variant.

Discussion: Mutations in the *MECP2* gene should be investigated in cases of global developmental delay/intellectual disability both in females and males.

PED_26

3Q29 MICRODUPLICATION SYNDROME IN TWO SIBLINGS

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Introduction: Intellectual disability (ID) is a neurodevelopmental disorder characterized by deficits in both intellectual and adaptive functioning and affects 3% of the general population. Of these, 10% have a genetic etiology.

3q29 microduplication syndrome is a rare genomic disorder first described in 2008 and caused by a 1.6Mb duplication in the referred chromosomal region. It has a highly variable clinical presentation, with patients mainly showing mild to moderate ID, obesity, ocular and cardiac anomalies, and mild dysmorphic features but otherwise being healthy.

Case presentation: Herein are presented the cases of two female siblings referred to a Neurodevelopmental Clinic due to global developmental delay and obesity. They were the sixth and seventh daughters of a healthy mother, and both were born after uneventful full-term pregnancies. Regarding family history, only a maternal half-brother with cerebral palsy was identified. The first girl presented with mild dysmorphic features (round and long face, bulbous nose, and syndactyly) and obesity. Wechsler Intelligence Scale for Children III Full Scale IQ scored 64. She also had a disruptive behavioral profile, with reported hetero-aggressive behavior. The second girl presented with axial hypotonia, strabismus, and obesity. Griffiths Scales of Child Development scored 79. Fragile-X molecular study was normal in both girls, but Multiplex Ligation Dependent Probe Amplification (MLPA) study confirmed the diagnosis of 3q29 microduplication syndrome.

Discussion: ID is a common disorder in pediatric age with unclear etiology in most cases, causing uncertainty in the families of affected children. Clinicians should keep a high index of suspicion of ID when in presence of suspected characteristics, perform adequate genetic study, and offer families genetic counseling.

In this study, the presence of ID, obesity, and dysmorphic features in the two siblings favored the diagnosis of a genetic syndrome, subsequently confirmed by MLPA. The phenotypic heterogeneity of 3q29 microduplication syndrome may explain why it remained undiagnosed in the previous generations.

PED_27

RARE PATHOGENIC COPY NUMBER VARIATION IN THE 16P11.2 REGION ASSOCIATED WITH AUTISM AND OTHER NEURODEVELOPMENTAL DISORDERS

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Introduction: Copy number variation (CNV) plays an important role in the genetic underpinning of neurodevelopmental disorders. The 16p11.2 chromosomal region harbors deletions and duplications associated with global developmental delay/intellectual disability (GDD/ID), autism spectrum disorders (ASDs), speech sound disorders, language disorders (LDs), and obsessive and repetitive behaviors in carriers. Both neurodevelopmental and physical phenotypes are non-specific and highly variable.

Description: This study sought to characterize eight cases of 16p11.2 rearrangements (both duplications and deletions) referred to the Neurodevelopmental Unit due to learning disability (50%), GDD/ID (25%), and language impairment (25%). The median age of referral was eight years, and half of patients were male.

Discussion: In this study, GDD/ID affected all carriers of 16p11.2 deletions and duplications, with ASDs occurring in less than half. Speech and language disorders were present in all patients with 16p11.2 deletions and in one patient with 16p11.2 duplication. It should be emphasized that these results were obtained in a reference population, and thus the proportion of affected patients is probably higher than reported.

With this study, the authors intended to highlight the importance of recognizing the full spectrum of neurodevelopmental and behavioral phenotypes associated with 16p11.2 CNVs, in order to provide early diagnosis and intervention, guidance for patient follow-up and management, and proper genetic counseling.

PED_28

IN ESSENCE, WHAT DO WE HAVE HERE? A CASE STUDY

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Introduction: The assessment and final diagnosis of global developmental delay in young children are often challenging. This is particularly relevant when more than one neurological development domain is affected, as when the child presents symptoms of language, behavior, motor coordination, and attention span impairment.

During the past decades, the need to establish a final and categorical diagnosis (whether of autism spectrum syndrome, attention-deficit/hyperactivity disorder, or other) sooner than later has been emphasized. However, maybe the focus should be less on the diagnosis and more on the actual impairments that each individual presents. As acknowledged, not every child on the spectrum presents language delay or eating problems (although these comorbidities are often associated with autism spectrum disorder), which further highlights the need for early intervention and the importance of considering each individual as unique and providing individualized management approaches.

This report describes the case of a young child with global development delay and aims to raise awareness for the coexistence of symptoms and disorders in some patients and for the importance of a multidisciplinary approach in early symptomatic syndromes eliciting neurodevelopmental clinical examinations – ESSENCE.

Case study: A previously healthy four-year-old male experienced a developmental regression during the COVID-19 pandemic, according to teachers. No family history of illness was reported, only the case of an older brother who had learning issues. The mother had gestational diabetes, which was partially controlled with insulin, and Apgar score of 5´8–10 upon delivery (c-section). On clinical examination, the boy presented motor discoordination, short attention span, poor language skills, and apparent difficulty in processing information when spoken to (for instance, he could not tell his age, sex, or address, answering his own name instead). Physically, no dysmorphisms were identified. The boy began occupational therapy and follow-up by a neurodevelopmental pediatrician, which he maintains.

Discussion: As genetic causes (such as fragile X-syndrome) are discarded, tracking and psychological tests (such as SGS-II and Griffiths scale) confirm global impairments, and the child grows older, the need to establish a final diagnosis increases. Is this an attention deficit disorder? An attention deficit hyperactivity disorder (ADHD)? Or an ESSENCE instead

PED_29

NEURODEVELOPMENTAL DISORDERS ASSOCIATED WITH ABNORMAL GENE DOSAGE: SMITH-MAGENIS AND POTOCKI-LUPSKI SYNDROMES

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Introduction: Smith-Magenis syndrome (SMS) and Potocki-Lupski syndrome (PTLS) are reciprocal contiguous gene syndromes associated with deletions and duplications in the 17p11.2 chromosomal region, respectively. Changes in dosage-sensitive genes, mainly *RAI1*, are responsible for these diseases, and although SMS and PTLS share the same genomic regions, they are clinically distinct entities.

More than 100 cases of SMS have been reported worldwide. The main clinical manifestations include mild-to-moderate intellectual disability, self-injurious behaviors, sleep disorders, dysmorphic facial features, congenital heart defects, and structural renal abnormalities. Conversely, PTLS has only been globally reported in less than 50 patients and manifests through mild-to-moderate intellectual disability, hyperactivity and attention deficit, autism spectrum disorder (ASD), short stature, nonspecific facial features, and congenital heart defects.

Description: Herein are reported three cases of SMS and two cases of PTLS identified at the Neurodevelopment consultation of a tertiary hospital. Cases of SMS referred to one male and two female patients aged between three and 18 years old. All presented global development delay, hyperactivity and impulsive behavior, sleep disorders, and facial dysmorphism. Cases of PTSL referred to one male and one female patient aged between 10 and 18 years old, presenting with mild facial dysmorphism. Both had intellectual disability, in one case also with ASD with no language.

Discussion: In this study, all patients presented with the neurodevelopmental and behavioral phenotype described in the literature for these disorders. By presenting the cases seen in consultation, the authors intended to raise awareness for gene dosage-sensitive gene syndromes, mainly those involving the 17p11.2 region. Awareness of these conditions will potentially allow earlier diagnosis and improved management by a multidisciplinary team. Parents should be counseled about these disorders and supported on how to deal with them, also being informed of recurrence risks and reproductive options.

PED_30

CLARK-BARAITSER SYNDROME - A RARE CAUSE OF GENETIC GLOBAL DEVELOPMENTAL DELAY

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Introduction: Global developmental delay (GDD) is a frequent reason for referral to the Pediatric Neurodevelopment consultation. This term is used to describe failure to meet the expected developmental milestones in multiple areas of functioning under the age of five years. GDD affects 1–3% of the world population, with several etiologies acknowledged. Among these, genetic alterations are a relevant cause and should be investigated.

Case presentation: Herein is presented the case of a 7-month-old female infant referred to the Pediatric Neurodevelopment consultation due to suspected GDD. She was the second daughter of healthy nonconsanguineous parents, born at 36 weeks + 6 days after an uneventful pregnancy. History of epilepsy was reported in the mother's brother and father.

The girl presented with divergent strabismus, usual posture with closed hands, little playtime in the midline, left-hand preference in object manipulation, absence of object transfer from one hand to the other, and inability to sit with support. On physical examination, she showed mild facial dysmorphism. Cerebral magnetic resonance imaging evidenced mild enlargement of frontal lobe subarachnoid spaces bilaterally. Array-based comparative genomic hybridization (aCGH) was normal, and the genetic panel for intellectual disability (ID) revealed a pathogenic missense variant in the TRIP12 gene, compatible with Clark-Baraitser syndrome.

Discussion: Clark-Baraitser syndrome is a genetic cause of GDD and ID, with presumed autosomal dominant inheritance. It presents with multisystemic clinical manifestations with an impact on the child's physical, psychological, and social health. As in other GDD and ID etiologies, the approach should be multidisciplinary and personalized to each child and family.

PED_31

SMITH-KINGSMORE SYNDROME – TWO CLINICAL CASES

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Introduction: Smith-Kingsmore syndrome (SKS) is a rare genetic disorder with autosomal dominant inheritance caused by germline or mosaic mutations in the *mTOR* gene on chromosome 1p36. Despite clinical variability, the most consistent findings are macrocephaly, overgrowth, global developmental delay/intellectual disability (ID), and seizures. Unspecific facial dysmorphic features are not always present.

Description: Herein are reported the cases of two girls aged 12 and 14 years with SKS. One girl was born prematurely at 25 gestational weeks, and the other was born after an uneventful pregnancy. Both had progressive congenital macrocephaly with delayed closure of the anterior fontanelle and current height above the 97th percentile. In one girl, transfontanellar ultrasound revealed mildly dilated ventricles, and magnetic resonance imaging later showed a white matter reduction in the left parietal region.

None of the patients showed major dysmorphisms. Attention deficit and hyperactivity disorder (ADHD) and ID were diagnosed in both, with concomitant Fabry disease also accidentally identified in one patient during ID etiological study.

Metabolic screen, karyotype, and array comparative genomic hybridization were normal. *mTOR* gene mutation was identified in one patient at the age of 11 years by exome sequencing and in the other by overgrowth syndrome panel.

Discussion: SKS is a rare genetic disorder, with about 150 cases reported worldwide. It has a high phenotypic similarity with other syndromes associated with the *mTOR* pathway, which can lead to underdiagnosis. The two cases presented showed macrocephaly, overgrowth, and ID, which is in agreement with what is reported in the literature. Although seizures are reported in more than 75% of patients, none of the present patients had epilepsy. Noteworthy, although ADHD is reported in only around 11% of cases, it was present in both patients in this study.

With this report, the authors intend to alert for this syndrome in the hope of contributing to earlier diagnoses, individual therapeutic interventions, and genetic counseling for patients and families.

PED_32

RENAL PAPILLARY NECROSIS - AN UNDERRECOGNIZED COMPLICATION OF SICKLE CELL DISEASE

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Introduction: Sickle cell disease (SCD) is a group of hemoglobinopathies that can lead to sickling and vaso-occlusion. Renal papillary necrosis (RPN) and renal infarction are underrecognized complications of SCD and not commonly seen in the daily practice, but with potentially severe implications when diagnosed late.

Case description: A 13-year-old male with SCD with SS homozygosity was referred and admitted to the Emergency Department for painless, macroscopic hematuria over the course of that day. He denied fever, dysuria or pollakiuria, and abdominal pain. Three weeks before hematuria onset, he was hospitalized for a vaso-occlusive crisis and medicated with non-steroidal anti-inflammatory drugs (NSAIDs) for seven days. On physical examination, the boy was normotensive and had normal heart rate, presented slightly discolored conjunctivae, and the abdomen was soft, non-tender, and with no organomegaly. Blood work showed hemoglobin of 10.3 g/dL, urea of 25 mg/dL, creatinine of 0.55 mg/dL, and normal coagulation tests. Urinalysis showed 70 mg/dL of protein, >50 red blood cells, and 2-5 leucocytes per high-power field. Urine culture was negative. Renal ultrasound followed by computerized tomography urogram identified a hypovascular nodulariform area in the right kidney upper pole and a slight papillae indentation of some caliceal groups, supporting the diagnosis of renal infarction and papillary necrosis. Hyperhydration with intravenous fluids, urine alkalinization, and bed rest were maintained until hematuria cleared up four days later. Two weeks after discharge, at the follow-up visit, the boy was asymptomatic and with no proteinuria or hematuria on urinalysis.

Conclusions: Sickle cell nephropathy is a common condition, but RPN and infarction are rare complications, especially in children and with right kidney involvement. Clinicians should be aware of this rare condition in patients with SCD presenting with macroscopic hematuria, since prompt recognition and treatment are of high importance to reducing long-term kidney sequelae.

PED_33
A RARE CASE OF TONGUE BITING DURING SLEEP IN CHILDHOOD

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Introduction: Tongue biting during sleep is a rare cause of sleep disturbance. Although highly suggestive of epilepsy, the differential diagnosis with other entities, such as bruxism, facio-mandibular myoclonus, hypnic myoclonus, and geniospasm, should be pursued.-

Case report: A 2-year-old boy was referred to a Pediatric Sleep Center due to recurrent tongue biting during sleep since the age of 12 months. He had normal neurodevelopment and unremarkable past medical history**.** No family history of movement disorders or epilepsy was reported. Parents described movements like myoclonus followed by tongue biting, which caused swelling and bleeding lacerations on the lateral sides of the tongue during sleep, with very frequent awakenings and crying episodes. The patient's physical (including neurological) examination was normal, except for the tongue lesions (lateral lacerations).

As part of the diagnostic workup, the patient underwent two electroencephalograms (EEG), which were normal, and level 1 polysomnography (PSG) with expanded EEG and masseters electromyography (EMG). PSG reported a sleep efficiency of 76.3%, 15.9 arousals per hour, 35 awakenings coincident with increased masseter and chin EMG tone, and intermittent elevations of chin EMG tone during NREM and REM sleep, without visible anomalous movements or epileptiform activity.

Tongue lesions improved with time, but awakenings were still very frequent, and a low dose of clonazepam was started, with excellent clinical response. After two months of treatment, the patient started sleeping thirteen hours a day without awakening.

Discussion: Geniospasm is a rare genetic disorder characterized by paroxysmal rhythmic or irregular movements of the chin and lower lip due to repetitive contractions of the mentalis muscle. It can be associated with tongue biting during sleep, usually with onset between 10 and 18 months of age. Given the child´s age and intermittent elevations of chin EMG tone seen in PSG, geniospasm was the most likely diagnosis. Chin movements are described as benign conditions and usually decrease during early childhood. However, when causing sleep disturbance, benzodiazepines such as clonazepam are recommended. In the present case, the definitive diagnosis and proper treatment was crucial for improvement of sleep and quality of life of the child and family.

PED_34
GENDER IDENTITY: A SCHOOL APPROACH – INTERVENTION PROJECT

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Introduction: The individual's gender identity reflects a deep and experienced sense of one's own gender. In recent years, there has been a significant increase in reports of non-acceptance of gender assigned at birth, a condition termed gender dysfunction.

The goal of this study was to expand adolescents' knowledge of gender identity with the purpose of identifying gender dysphoria.

Methods: An on-site training session with dynamic content (powerpoint presentation, games) was held for students of the 11th class of São João da Madeira school. Students—knowledge was assessed before and after training through an 8-question survey with dual answers (True/False) using Google Forms®. Paired sample T-tests were used to compare the number of correct answers obtained before and after the session. Microsoft Office Excel® and Statistical Package for the Social Sciences® were used for statistical analysis, and the Shapiro-Wilk test was used to investigate data normality. $P < 0.05$ was considered statistically significant.

Results: A total of 24 students participated in the session, and 23 questionnaires were retrieved (pre-session rating: 67.4%; post-session rating: 92.4%). A statistically significant difference was found in the number of correct answers before and after the intervention (15.50 ±3.964 vs. 21.25 ±1.389, p=0.004).

Conclusion: Gender identity derives from one's individual experience, and its construction and experience are influenced by the psychosocial environment. Since this identity can have a variable impact on mental health, it is crucial to raise awareness of young population about this matter. The results obtained suggest that training sessions allowed students to acquire new knowledge about gender identity and gender dysphoria. The family doctor has a relevant role in disease prevention and health promotion, which includes children and adolescents' personal and social development.

PED_35
NURTURING A FRAGILE BOY'S HEART

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Introduction: Avoidant/restrictive food intake disorder in early childhood is a frequent and complex nosological condition that often involves dysfunction of sensory regulation and of the emotional and cognitive response.

Clinical case: A 30-month-old boy with congenital heart disease, hypothyroidism, and failure to thrive was referred to the Child and Adolescent Psychiatric consultation for reduced food intake. He was overprotected by adult caregivers who, centered on the organic disease, had difficulties in losing focus on food and nurturing the boy's emotional needs. A brief psychotherapeutic approach was carried out, focusing on early maternal experiences, impact of the disease on the family, sister's birth, ghosts of the disease, and food control. Additionally, vestibular hyporesponsiveness, tactile hyperresponsiveness (namely intraoral), and fragility in the masticatory muscles were identified, and the boy underwent occupational therapy with sensory integration and speech therapy. During follow-up, an increase in food intake and diversity, greater autonomy, and improvement in the quality of parental relationship were reported.

Discussion: A clear association has been reported between congenital heart disease and eating disorders. Indeed, malnutrition and failure to thrive are common in these children. Parents of children with congenital heart disease have significantly higher stress levels than parents of children with other types of chronic conditions. Differences in the attachment process have been described, with a lower proportion of secure attachment occurring in these children.

PED_36
HELSMOORTEL VAN DER AA SYNDROME – SMALL CASE SERIES AND LITERATURE REVIEW

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Introduction: Helsmoortel van der Aa syndrome (HVdAs) is an autosomal dominant condition. It is estimated to represent 0.17% of all etiologies of autism spectrum disorder (ASD) and is frequently associated with intellectual disability (ID). HVdAs is caused by heterozygous deleterious variants in ADNP and presents multisystemic involvement. It typically occurs *de novo* and has variable expression. Herein are reported the cases of three patients diagnosed with this condition, together with a literature review.

Methods: Patients with molecular diagnosis of HvdAs were identified, and their clinical features were compared with those reported in the literature.

Result: This report refers to three male patients aged 9, 11, and 17 years old, respectively, with heterozygous pathogenic or likely pathogenic ADNP variants identified by WES-based gene panel. All patients presented with ID of variable severity. Dysmorphic features and behavioral problems were present in all three. Epilepsy, endocrinological features, abnormal teeth development, and overweight were present in two patients. One patient presented anomalous fat deposition and skin dimples.

Discussion: Anomalous fat deposition and skin dimples have not been previously reported in HvdAs. In this study, these features were found in the patient most severely affected by the condition, presenting with the most dissimilar dysmorphic features.

The absence of a recognizable gestalt in HVdAs makes a high throughput sequencing-based approach necessary for diagnosis. The present small cohort highlights the variability of HvdAs and the challenge it represents for genetic counseling and prognosis determination. Treatment may be an option in the future, reinforcing the importance of accurate and timely diagnosis. Efforts to increase the size of this cohort through collaboration with other national centers are underway and are important to gradually clarify the impact of HVdAs in patients with ASD/ID.

PED_37

PMM2-CDG: CLINICAL CHARACTERIZATION OF TWELVE PATIENTS

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Background: Congenital disorders of glycosylation (CDG) are genetic defects in the synthesis and attachment of glycoprotein and glycolipid glycans. PMM2-CDG is the most common CDG and is characterized by severe multisystemic disease in infancy, neurologic disease and developmental delay in childhood, and/or stable intellectual disability in adulthood.

Methods: Phenotypic and genotypic characterization of patients with PMM2-CDG followed at the Metabolic Unit at Centro Hospitalar Universitário do Porto.

Result: A total of 12 patients (5 females and 7 males) with ages between seven years and adulthood were assessed. The age of diagnosis varied from one month to 13 years.

Five patients had infantile multisystem form, with inverted nipples, abnormal subcutaneous fat distribution, facial dysmorphism, severe psychomotor retardation, and cerebellar hypoplasia. The five patients had pericardial effusion, strabismus, and severe feeding problems in the first year of life, with the need for nasogastric tube, and four patients had renal involvement. Three of these patients died in the first years of life.

Seven patients had a less severe form of presentation, with psychomotor delay and cognitive impairment, and all showed progressive balance problems and mild-to-severe cerebellar ataxia. Among these cases, six had strabismus and one had mild proteinuria.

Magnetic resonance imaging was performed in all patients, showing mild-to-severe cerebellar atrophy in all. Based on the transferring electrophoresis pattern, the 12 patients were classified as CDG type I. Molecular analysis was also performed in all, revealing *PMM2* gene mutations causing phosphomannomutase 2 enzyme activity deficiency in the 12 patients. The c.193G>T; p.D65Y mutation was the most frequently found. No correlation could be established between genotype and phenotype.

Conclusion: The present study corroborates PMM2-CDG clinical variability. As the phenotypic spectrum of PMM2-CDG is highly variable and affects multiple systems, no clear correlation could be established between genotype and phenotype. Although severe cases frequently suggest this diagnosis, milder presentations can be easily underdiagnosed, despite being more frequent. The authors suggest that transferrin electrophoresis should be performed in children with development delay (with or without mild dysmorphic features or ataxia).

PED_38

DOMPERIDONE AND ELECTROCARDIOGRAPHIC CHANGES IN PEDIATRIC AGE? — AN EVIDENCE-BASED REVIEW

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Introduction: Domperidone is a dopamine D2 receptor antagonist that acts peripherally, not crossing the blood-brain barrier. The drug promotes relaxation of the esophageal gastric tract, facilitates gastric emptying, and promotes intestinal motility, functioning as an antiemetic. Domperidone is widely used for the symptomatic relief of vomiting and gastroesophageal reflux disease (GERD). However, its use has been disputed in recent years.

This study aimed to retrieve evidence of the effects of oral administration of domperidone on electrocardiographic tracing in pediatric age.

Methods: A search was conducted in the following databases using MeSH terms “Domperidone” AND “Infant”: National Guideline Clearinghouse, Guidelines Finder, Canadian Medical Association Practice Guidelines Infobase, The Cochrane Library, and PubMed. Studies published in the last 20 years (from 2002 to 2022), written in Portuguese, English, and Spanish, were retrieved. The Strength of Recommendation Taxonomy scale of the American Family Physician (SORT) was used to evaluate the quality of studies and strength of recommendations.

Results: A total of 102 articles were retrieved from the initial search, of which 12 were selected after reading the titles and respective abstracts. Of these, only three met the predefined inclusion criteria and were selected: two systematic reviews and one prospective study. Retrieved evidence did not allow to establish an association between domperidone oral administration and electrocardiographic changes in pediatric age.

Conclusion: Domperidone should not be used as first-line treatment of GERD. Electrocardiographic assessment should be considered prior to its administration.

GINECOLOGIA - OBSTETRÍCIA

COMUNICAÇÕES ORAIS

GO_01

HEREDITARY ANGIOEDEMA WITH NORMAL COMPLEMENT: OUTCOMES DURING PREGNANCY

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Introduction

Hereditary angioedema (HAE) is a rare and underdiagnosed disease characterized by recurrent episodes of mucocutaneous swelling. HAE with normal C1 inhibitor (C1-INH) activity in plasma (HAE-N) results from different genetic causes with a similar phenotype. During pregnancy, the rise of estrogens may trigger a parallel rise of acute episodes. We report three cases of pregnant women with HAE-N and how their condition was managed during pregnancy.

Case report

Patient A: A 24-year-old pregnant female with Behçet's disease (BD) was first observed during an acute episode of mucocutaneous angioedema with no identifiable triggers and no response to antihistamines. She recalled similar episodes during adolescence while taking estrogen-containing contraceptives and during her first pregnancy. No family history of angioedema was reported. The diagnosis of HAE-N was established after the identification of factor XII gene (FXII) mutation. After a subsequent episode with airway involvement, long-term prophylaxis was started. Due to the prothrombotic risk associated with BD and since no subcutaneous formulation was available, administration of intravenous (IV) C1-INH concentrate was decided (20 UI/kg every 3-4 days until delivery). A few minor episodes of angioedema were subsequently reported, and vaginal delivery was uneventful.

Patient B: A 31-year-old pregnant female with a history of multiple episodes of facial angioedema unresponsive to antihistamines starting while she was on estrogen-based oral contraception presented with a positive family history. HAE-N due to FXII mutation was subsequently diagnosed. During pregnancy, she had several minor cutaneous episodes and three major episodes (one with uvula edema), which reverted without specific treatment. She delivered by cesarean section after IV administration of 20 UI/kg of C1-INH concentrate without complications.

Patient C: A FXII mutation was identified in a 31-year-old pregnant female related to patient B (first-degree cousin) during familial screening. She recalled a few previous episodes of labial angioedema while on combined oral contraception, the last occurring three years ago. No episodes were reported during pregnancy, and the patient had a successful vaginal delivery without short-term prophylaxis.

Conclusion

HAE-N course during pregnancy is unpredictable and may result in adverse outcomes for the mother and child. Therefore, timely diagnosis and proper management are crucial.

GO_02

FETAL AND MATERNAL OUTCOMES IN PATIENTS WITH LUPUS NEPHRITIS

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Introduction

Lupus nephritis (LN) frequently affects female patients of childbearing age and is considered a major contributor to morbimortality associated with systemic lupus erythematosus (SLE). As renal flares or preexisting kidney disease can be associated with adverse maternal and fetal outcomes, patients with LN are currently advised to achieve stable remission of their renal disease for at least six months before conceiving.

The aim of this study was to evaluate maternal characteristics of women with active or prior history of LN, as well as disease impact on perinatal outcomes.

Methods

This was a retrospective cohort study of all singleton pregnancies of women with SLE followed at a tertiary center between January 2010 and December 2019. Two groups were considered: women with active or prior history of LP (LN group) and women with SLE without renal impairment (control group). The primary objective was the assessment of maternal characteristics, as well as antenatal (hypertensive disease, fetal growth restriction [FGR], miscarriage/fetal death, preterm birth) and postnatal (birthweight, Apgar index, neonatal SLE, neonatal death, congenital heart block, Neonatal Intensive Care Unit [NICU] admission) obstetric outcomes.

Results

A total of 123 pregnancies in women with SLE were considered, with a mean maternal age and gestational age at birth of 32 years and 37 weeks + 5 days, respectively. Seventeen pregnant women were considered in the LN group, corresponding to 14% of cases. Regarding maternal characteristics, a statistically significant difference was found in the prevalence of chronic hypertension in the two groups (50% vs. 4%, p<0.01; odds ratio [OR] of 25.8 in the LN group). Regarding laboratory findings, the LN group showed a higher prevalence of anti-dsDNA (69% vs. 36%, p=0.01) and anti-SSA/Ro (50% vs. 25%, p=0.04) autoimmune antibodies compared to the control group. Concerning antenatal outcomes, preeclampsia (OR 4.3), gestational hypertension (OR 6.3), and FGR (OR 8.6) were significantly higher in the LN group. In the postnatal period, newborns were lighter (2573 gr vs 2991 gr, p=0.04) and neonatal SLE (31% vs 9%, p=0.02) and NICU admissions (25% vs 6%, p=0.02) were more frequent in the LN group.

Conclusion

According to study findings, women with LN are at increased risk for adverse maternal and fetal outcomes, including hypertensive disease of pregnancy and FGR. This emphasizes the need for multidisciplinary follow-up during the periconceptional period and adequate disease control before conception as a way to promote better obstetrical outcomes.

GO_03

ABSENCE OF FETAL HEART RATE CYCLING ON INTRAPARTUM CARDIOTOCOGRAPH ASSOCIATED WITH PROLONGED INTRAPARTUM DECELERATIONS

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Introduction: Cycling is an overlooked feature in most international cardiotocography (CTG) guidelines. It consists of alternating periods of reduced and normal fetal heart variability, reflecting changes in fetal behavioral state. The occurrence of active and quiet sleep cycles is considered a hallmark of fetal autonomic nervous system integrity, demonstrating a healthy interaction between the parasympathetic and sympathetic nervous systems. This study sought to investigate whether fetuses with no cycling in early labor have a higher frequency of prolonged deceleration during labor.

Methods: A database of sequentially acquired intrapartum CTG traces from a high-risk population followed at a single center was retrospectively assessed. Prolonged decelerations on CTG —also called acute hypoxic events —were defined as decelerations with more than 3 minutes of duration. Two groups were compared: fetuses with and without cycling.

Results: One hundred cases of singleton pregnancies over 36 weeks of gestation and with cephalic presentation with epidural analgesia were analyzed. Absence of cycling from the beginning of intrapartum CTG recording was noted in 30% of cases. No differences were found between the two groups regarding gestational age, induction of labor, type of delivery, birth weight, and Apgar index at 1 and 5 minutes. Cases without cycling were more likely to have prolonged decelerations ($p<0.05$) and STAN events ($p<0.05$). No statistically significant differences were found between the two groups regarding average baseline fetal heart rate or presence of accelerations at the beginning of CTG recording or occurrence of repetitive decelerations, sinusoidal patterns, or overshoots.

Conclusion: The results retrieved showed an association between the absence of cycling and prolonged decelerations and ST events but no significant difference in perinatal outcomes. Cycling is a CTG feature that can be potentially useful for identifying fetuses at higher risk of intrapartum fetal compromise.

GO_04

CTG FEATURES ASSOCIATED WITH HISTOLOGICALLY CONFIRMED CHORIOAMNIONITIS

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Introduction

Chorioamnionitis is an inflammation of the chorion and amnion associated with adverse perinatal outcomes, including a five-fold increased risk of cerebral palsy. Clinical chorioamnionitis refers to the presence of fever and at least one of the following: maternal and/or fetal tachycardia, uterine tenderness, maternal leucocytosis, and purulent or foul amniotic fluid. However, cases of subclinical chorioamnionitis, in which histologically confirmed acute chorioamnionitis develops before women show any signs or symptoms, have been reported.

This study aimed to recognize cardiotocography (CTG) patterns in women with histopathologically confirmed chorioamnionitis, independently of intrapartum clinical signs.

Materials and methods

Retrospective study of placentas with histopathological assessment over a 12-month period (between January and December 2021). Placentas from singleton pregnancy live births, with gestational age ≥36 weeks, apyretic on admission, delivered at Kingston Hospital NHS Foundation Trust, London, were included. Placentas were evaluated by experienced perinatal pathologists using the Amsterdam consensus framework. All CTG traces were systematically assessed by two investigators blinded to antenatal circumstances and neonatal outcomes, who agreed on the definitions of CTG features.

Results

Of 242 placentas assessed, 58 were diagnosed with acute chorioamnionitis and/or associated funisitis. In the chorioamnionitis group, 17 women had pre-labor rupture of membranes for more than 24 hours. The rate of cesarean section and Apgar index <7 at 5 minutes was 52.1% and 8.7%, respectively. One case of neonatal death was reported. The presence of repetitive decelerations was observed in 71.9% of cases. Ninety-two percent of CTGs had a >10% increase in baseline fetal heart rate (FHR), of which 58.5% had an increase in baseline FHR before decelerations. Loss of cycling was noted in 86.4%. Periods of sinusoidal pattern were present in 21.1%, a ZigZag pattern in 28.1%, and overshoots in 25.1% of cases. Uterine tachysystole was identified in 8.9% of cases.

Conclusions

An increase in baseline FHR (>10%) and loss of cycling were the most common CTG features in cases of histologically confirmed chorioamnionitis. In most cases, fetal heart rate increase occurred before repetitive decelerations.

GO_05
DIAGNOSTIC ACCURACY OF HYSTEROSCOPY IN POSTMENOPAUSAL WOMEN

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Introduction

Hysteroscopic-guided endometrial biopsy is currently the gold standard in the diagnosis of endometrial pathology. The aim of this study was to assess the accuracy of hysteroscopy in the assessment of postmenopausal women, correlating hysteroscopic findings (clinical assessment) with histologic results (laboratory assessment).

Methodology

A retrospective study was conducted on postmenopausal women submitted to hysteroscopy at Centro Materno-Infantil do Norte during 2021.

Results

Of 296 postmenopausal women submitted to hysteroscopy, 272 underwent endometrial biopsy/lesion excision. Of these, 13 were excluded due to insufficient/low-quality sample, resulting in a final sample of 259 women included in the analysis. Indications for hysteroscopy included focal or diffuse endometrial thickening (n=148; 57.1%), abnormal uterine bleeding (AUB; n=14; 5.4%), AUB plus endometrial thickening (n=92; 35.5%), and other reasons (n=5; 1.9%). The sensitivity (S), specificity (E), positive predictive value (PPV), and negative predictive value (NPV) of the procedure for endometrial cancer diagnosis were 50.0%, 99.2%, 66.7%, and 98.4% (kappa 0.560), and for endometrial hyperplasia (with or without atypia) were 33.3%, 90.2%, 17.2%, and 95.7% (kappa 0.163), respectively. The S, E, PPV, and NPV of hysteroscopy for malignant or premalignant lesions were 39.1%, 89.0%, 25.7%, and 93.8%, respectively (kappa 0.228). Among patients with benign hysteroscopic findings and endometrial carcinoma on histological assessment (1.5%), two had endometrial atrophy, one endometrial polyp, and one uterine fibroid histology. Among patients with hysteroscopic findings compatible with malignancy presenting benign histology (0.8%), one had mucinous tubal metaplasia and one endometrial polyp histology.

Conclusion

The results of this study reinforce the importance of performing endometrial biopsy/lesion excision even when hysteroscopic findings are suggestive of benignity, since the S of hysteroscopy in the detection of malignant and premalignant lesions is overall low (39.1%). Of note, in 5.4% of cases (or 1.5%, considering only malignant lesions), a malignant or premalignant lesion presented as a benign finding in hysteroscopic assessment, suggesting that one in every 20 patients may be misdiagnosed if endometrial biopsy/lesion excision is not performed.

GO_06
OBSTETRIC AND PEDIATRIC OUTCOMES IN PREGNANT WOMEN WITH ANXIETY DISORDERS

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Introduction: Perinatal anxiety disorders in pregnant women increase the risk of adverse birth outcomes and can have an impact on long-term neurodevelopmental outcomes of the offspring.

Methods: This was a retrospective review of pregnant women with anxiety disorders followed at Centro Materno-Infantil do Norte between 2016 and 2021. Data were retrieved from clinical records and included medical history and obstetric, neonatal, and pediatric outcomes. Exclusion criteria comprised multifetal gestations and comorbidity with depression.

Results: A total of 66 pregnant women with anxiety disorders were identified. Generalized anxiety disorder was the most prevalent condition (88%), followed by panic disorder (9%) and agoraphobia. The mean maternal age was 30 years, and the mean length of disease at conception was 10 years. Nine percent of women had a previous induced abortion, and 45% were multiparous, four with preterm labor history. Half of women were under psychiatric drugs (antidepressants and anxiolytics), and 61% were without medication before pregnancy. A total of 42% of women used antidepressants, most commonly sertraline. No dose adjustments were required, and no disease flare was registered. Obstetric complications (gestational diabetes, hypertensive disorders, placental abruption, threatened preterm labor) occurred in 15% of pregnancies, with threatened preterm labor being the most frequent. The mean weight gain during pregnancy was 10 kg. Labor was spontaneous in 81.5% of pregnancies, and the mean age at delivery was 39 weeks, but preterm labor occurred in 9% of cases. Cesarean section (C-section) occurred in 26% of cases. Although the commonest indication for C-section was fetal malpresentation, two women had planned surgery due to extreme maternal anxiety. The mean birth weight and APGAR score at 5 minutes were 3.111 kg and 10, respectively. No neonatal complications were registered. Postpartum depression was diagnosed in three women. A total of 72% of newborns were exclusively breastfed during the first six months. With a median follow-up of five years, 11% of these children displayed behavioral abnormalities requiring Pedopsychiatry support. No significant correlation was found between the incidence of behavioral disorders and the type of maternal disorder, duration of disease, or therapy during pregnancy.

Conclusion: The role of anxiety in obstetric and pediatric outcomes was neglectable in this study. Although this might be attributed to disease stability under adequate treatment during pregnancy, more evidence is required regarding long-term outcomes in pregnant women suffering from these disorders.

GO_07

POLYMALFORMATIVE SYNDROME AND CYTOMEGALOVIRUS REINFECTION IN PREGNANCY – A CASE REPORT

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Reinfection by cytomegalovirus (CMV) in pregnancy is rare, and the risk of fetal infection is estimated to be around 0.15-2%.

A 37-year-old pregnant woman, 5G2P (2 miscarriages, 2 vaginal deliveries), was referred to the Fetal Medicine consultation due to abnormal second-trimester ultrasound. Personal history included ankylosing spondylitis, and family history was irrelevant, with healthy offspring. The first-trimester ultrasound was normal, and the first-trimester combined screening showed a low risk of major aneuploidies. Prenatal cell-free fetal DNA corroborated the previous result (fetal fraction of 4.7%). First-trimester bloodwork was normal, showing immunity to rubeola and toxoplasmosis and probable immunity to CMV (IgM 0.157 UI/mL, IgG 1003 UI/mL). The second-trimester ultrasound at 20 weeks showed hypertelorism, bilateral ventriculomegaly, cerebellar vermis hypoplasia, intestinal hyperechogenicity, ascites, shortened fetal long bones (<p5), single umbilical artery, severe fetal growth restriction (estimated weight of 208 gr), oligohydramnios (single deepest vertical pocket of 1.2 cm), placentomegaly, and intermittent absent diastolic umbilical blood flow. The situation was explained to the couple at the Fetal Medicine consultation, as well as their options. The couple chose to perform an invasive diagnostic procedure and required medical interruption of pregnancy. A chorionic villus biopsy was performed at 21 weeks due to anhydramnios. Karyotype was 46,XY, and the array showed maternal contamination. The request for medical interruption of pregnancy was approved by the Ethics Committee and occurred uneventfully.

No growth was observed in fetal skin biopsy. Placental anatomopathological examination showed a three-vessel cord, villous edema, and chronic villitis lesions. Immunohistochemistry staining for parvovirus and CMV showed positive cells for the latter. Fetal autopsy revealed subcutaneous edema, bilateral ventriculomegaly, cerebellar vermix hypoplasia, hypertelorism, retrognathia, hypoplastic thymus, bilateral pleural effusion and peritoneal effusion, unilateral clubfoot, and hepatic necrosis. Immunohistochemistry staining for CMV in the brain, kidneys, and liver was positive.

Although the incidence of fetal abnormalities in cases of CMV reinfection is lower than in cases of primary infection, this study documents a rare case of CMV reinfection leading to a polymalformative syndrome.

GO_08

DOUBLE Z–PLASTY WITH V–Y ADVANCEMENT (JUMPING MAN FLAP) FOR THE TREATMENT OF POSTERIOR FOURCHETTE FISSURE

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Introduction

Dyspareunia is a common but poorly understood condition affecting around 7.5% of sexually active women aged 16-74 years. Isolated, persistent, posterior fourchette fissure is a cause of dyspareunia, and many patients with this complaint are referred to vulvar specialists and sexual therapy services, but the incidence of the condition and clinical pathway followed by these patients remain largely unknown. The aim of this study was to highlight a common plastic surgery method not usually applied in the treatment of posterior fourchette fissure with associated dyspareunia.

Case report

Herein are reported two cases of women with posterior fourchette fissure with associated dyspareunia.

The first case refers to a 44-year-old female, 4G1P (c-section), with a previous subarachnoid hemorrhage due to brain aneurism rupture and ischemic stroke, who was referred to the vulva pathology consultation for complaints of vulvar pruritus, posterior fourchette chronic fissure, and apareunia. On physical examination, lesions compatible with lichen sclerosus were noted in addition to the posterior fourchette fissure. The patient had been previously submitted to fissure excision under local anesthesia and received topical treatment (topical corticoid), without symptom improvement. A double Z–plasty with V–Y advancement was performed under general anesthesia without complications (Figures 1-3). Four months after surgery, dyspareunia had improved, and the fissure had not recurred.

The second case refers to a healthy 20-year-old female, 0G0P, who was referred to the vulva pathology consultation for dyspareunia since the beginning of sexual activity. On physical examination, a posterior fourchette fissure was noted. Lesion excision under local anesthesia was unsuccessful, and the patient was submitted to double Z-plasty with V-Y advancement under general anesthesia a few months later. The procedure went without complications, with the patient reporting painless penetration four months later.

Discussion

Evidence for the management of fourchette fissures is overall scarce. This study describes two cases of successful posterior fourchette fissure treatment in women presenting with dyspareunia. In both cases, surgical refashioning of the introit may help to release the associated contracture, thus avoiding fissure recurrence. Double Z–plasty with V–Y advancement may be considered in selected patients with isolated, persistent, posterior fourchette fissuring.

GO_09
MANAGING MONOCHORIONIC TWIN PREGNANCIES – FOUR-YEAR EXPERIENCE

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Introduction

Chorionicity is a major factor in twin pregnancies when it comes to perinatal outcomes. Monochorionic (MC) twin pregnancies have a higher risk of perinatal morbidity and mortality than dichorionic pregnancies, with a higher risk of preterm birth, growth discordance, and MC-specific complications, such as twin-twin transfusion syndrome (TTTS), twin reversed arterial perfusion (TRAP) sequence, and selective intrauterine growth restriction (IUGR). Monoamnionicity carries an increased risk of adverse pregnancy outcomes.

The primary aim of this study was to investigate the management and maternal and fetal outcomes of MC pregnancies at a tertiary center.

Materials and methods

This was a retrospective cohort study of MC pregnancies delivered at Centro Materno-Infantil do Norte (CMIN) between January 2018 and December 2021. Cases of MC monoamniotic pregnancies were excluded. General clinical data and data regarding prenatal examination and pregnancy outcomes were retrieved from clinical records and analyzed.

Results

Among 271 twin pregnancies, 58 MC diamniotic twin pregnancies were identified in the considered time period. Three cases of MC monoamniotic pregnancies were identified and excluded, making a final sample of 55 women with a mean age of 32.4 years. Of these, 57.4% were primipara. The mean gestational age (GA) at birth was 34.7 weeks. Fifteen (27%) fetal deaths were reported, with a mean GA of 18.3 weeks. Pregnancy complications included selective IUGR (27.3%), non-selective IUGR (12.7%), TTTS (12.7%), and TRAP (1.8%). Preeclampsia was diagnosed in 3.6% of pregnancies, and cholestasis in 1.8%. Hospital admission was required in 71% of cases (mean hospital stay of 11.1 days), and 62.5% completed antenatal corticosteroid therapy. Induced labor occurred in 28.6% of cases. Uncomplicated vaginal delivery was documented in 32.7% of cases, and operative vaginal delivery in 10.2%. Regarding the second twin, 34.7% of cases referred to uncomplicated vaginal delivery, and 4.0% to operative vaginal delivery. Cesarean was performed in 57.1% of cases (48.4% of which elective). Around 50% of newborns were admitted to the Neonatal Intensive Care Unit, with a mean stay of nine days and a mean Apgar score at 1 minute of 8.

Conclusions

MC twin pregnancy carries a high incidence of pregnancy complications and perinatal mortality. Accurate early diagnosis and optimal management, monitoring, and individualized treatment are crucial for avoiding potentially adverse outcomes.

GO_10
UTERINE ARTERY PSEUDOANEURYSM: A RARE BUT THREATENING CAUSE OF POSTPARTUM HEMORRHAGE

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Introduction

Postpartum hemorrhage (PPH) remains one of the leading causes of maternal mortality globally. Most PPH cases are caused by uterine atony, laceration, and bleeding disorders. Pseudoaneurysm of a uterine vessel, arteriovenous malformation, and choriocarcinoma represent rarer causes of the condition. Pseudoaneurysm can arise following a cesarean section, dilation and curettage, hysterectomy, myomectomy, oocyte collection for intrauterine fertilization, and even uncomplicated vaginal delivery. It has a characteristic sonographic appearance, comprising a well-defined anechoic or hypoechoic pulsatile cystic structure with or without associated pelvic hematoma. Computed tomography (CT) with angiography is useful in the diagnosis. Once a hemorrhagic pseudoaneurysm is diagnosed, conservative treatment is often unsuccessful. Uterine arterial embolization is a safe treatment, but surgical interventions such as hysterectomy or ligation of the internal iliac artery are preferable in hemodynamically unstable women.

Clinical case

A healthy 33-year-old woman, primiparous at 39 weeks of gestational age post-*in vitro* fertilization, arrived at the Emergency Department with premature rupture of membranes. Because of insufficient descent of fetal presentation, a cesarean section was decided. The patient had uterine hemorrhage at immediate postpartum, requiring placement of a Bakri balloon and transfusion support. After removing the balloon, blood loss restarted. During reassessment in the operating room, a conservative approach with tamponade was chosen. The patient remained stable at the hospital, being discharged on the fifth day. Two days later, she returned to the hospital with heavy vaginal bleeding. Pelvic CT revealed a pseudoaneurysm with risk of rupture. Vascular embolization at another hospital was proposed, which the patient refused, and a total abdominal hysterectomy with adnexal conservation was performed.

Conclusions

Uterine pseudoaneurysm can have severe clinical presentation, but treatment can potentially avoid hysterectomy. This diagnosis should be considered in cases of severe postpartum hemorrhage, with embolization representing a safe and effective treatment, with few reports of complications.

Keywords: arterial embolization, hysterectomy, postpartum complication, postpartum hemorrhage, uterine pseudoaneurysm

GO_11

PREDICTION OF PREMALIGNANT AND MALIGNANT ENDOMETRIAL DISEASE IN POSTMENOPAUSAL WOMEN

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Introduction

Endometrial cancer (EC) occurs mainly in the postmenopausal period and is preceded by at least one episode of abnormal uterine bleeding (AUB) in around 80-95% of cases. Although the relationship between endometrial thickness (ET) and risk of malignancy in patients with postmenopausal AUB is established, and diagnostic protocols are well defined, there is no consensus regarding the approach to abnormal ultrasonographic findings in asymptomatic women. The aim of this study was to evaluate the influence of ET and AUB on the risk of premalignant and malignant findings (PMMF) in postmenopausal women undergoing hysteroscopy and endometrial sampling.

Methods

A retrospective observational study of postmenopausal women with hysteroscopic exams with histopathological findings followed during 2021 at a tertiary center was conducted. Patient records were reviewed, and two groups were evaluated: asymptomatic women (G1) and women with AUB (G2). G2 was split into two subgroups according to ET: G2.1 with ET <5mm and G2.2 with ET ≥ 5mm. PMMF was defined by the presence of endometrial cancer and hyperplasia. Statistical analysis was performed on SPSS, with a p-value <0.05 considered statistically significant.

Results

A total of 219 women met the study's inclusion criteria, 118 of whom were asymptomatic (G1) and 101 had AUB (G2). At least one risk factor for EC was present in 72.0% of G1 and 71.3% of G2 (p>0.05). The number of years after menopause when this assessment took place and the prevalence of PMMF differed significantly between groups (G1: Md 17y vs. G2: Md 12y, p=0.02 and G1: 5.1% [n=6] vs. G2: 12.9% [n=13]; p=0.04). In G1, the ET cut-off that provided the best sensitivity (83.8%) and specificity (67.9%) for PMMF was 10.5 mm (AUC 0.79, p=0.01). The lowest ET associated with PMMF in this group was 8 mm, and all cases presented risk factors for EC. The prevalence of PMMF in G2.2 was 15.6% (n=13; 5 of which represent EC), with zero cases of PMMF identified in G2.1.

Conclusions

According to this study's results, the risk of EC in asymptomatic women is increased when ET is ≥10.5 mm. However, considering the limited number of PMMF evaluated, this finding should be confirmed by more powerful studies. Some important questions remain: what is the meaning of ET in asymptomatic women? What is the impact of individual risk factors for EC? Can EC be accurately excluded in women with AUB and ET <5mm? This uncertainty emphasizes the need for further research to help inform the management of postmenopausal women with sonographic findings with/without AUB, allowing timely and accurate EC diagnosis.

GO_12

EFFECT OF VAGINISMUS ON QUALITY OF LIFE, SEXUAL AND REPRODUCTIVE FUNCTION: RETROSPECTIVE STUDY IN A PORTUGUESE WOMEN POPULATION

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Introduction Vaginismus is defined as genitopelvic pain/penetration disorder and consists of persistent difficulties with vaginal penetration, vulvovaginal/pelvic pain, anxiety, or fear of pain in anticipation, during or after penetration and increased pelvic muscle tension. This study aims to characterize the disease, vulnerability factors and therapeutic outcomes and to describe the impact on quality of life, sexual and reproductive function.

Methods A retrospective study using data from clinical records and telephone interviews from women treated for vaginismus in a tertiary center (CMIN-CHUPorto) from 2011 to 2021. Self-answered questionnaires Female Sexual Function Index and WHOQOL-BREF(PT) were used to evaluate sexual function and quality of life. A descriptive analysis was performed. Absolute and relative differences in pain and anxiety levels before treatment and currently were compared using Wilcoxon test; Mann-Whitney test was used to identify outcome differences. p value <0.05 was considered statistically significant.

Results 31 women were included in the final sample. The most prevalent vulnerability factors were conservative values (48.4%), depression (25.8%) and anxiety (22.6%). Worsening cause by emotional triggers (41.9%) was identified. Treatment delay was greater in primary vaginismus (4.0 (1.7-5.2) vs 0.5 (0-2.5); p=0.017). The majority underwent sex therapy with dilators, guided by a gynecologist. Differences in pain levels before treatment and nowadays were significant, either absolute (9,0 (8-10) vs 4,0 (1-6); p<0,001) or relative (60 (40-90), p<0.001), as well as relative differences in anxiety levels (57,1 (38,7-100); p=0,006). More than one-third had an unsatisfactory relational and psychological quality of life. Almost 57.1% had criteria for sexual dysfunction and they had worse pain (p=0.033 and p=0.004) and anxiety (p=0.043) reduction. Considerable difficulties with desire (25%) and orgasm (17.9%) were identified. Almost all were nullipara. Of the four recorded deliveries, three were by caesarean section and one by vaginal delivery with episiotomy.

Conclusion Conservative values and psychiatric comorbidity may be factors of vulnerability. Single therapeutic modalities also show positive outcomes. Women with sexual dysfunction have a worse pain reduction. Vaginismus can have a negative impact on quality of life, sexual and reproductive function, and therefore, early recognition and treatment is essential.

GINECOLOGIA - OBSTETRÍCIA

POSTERS

GO_13
PLACENTAL SYPHILIS AND INTRAUTERINE FETAL DEATH: CASE REPORT

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Introduction

Syphilis is a sexually transmitted disease caused by *Treponema pallidum* bacteria. All syphilis stages may cause fetal infection during pregnancy, but the risk is higher in early stages of the disease. Fetal infection may occur via transplacental invasion or less frequently through contact with active skin lesions during vaginal delivery. The aim of this study was to report a rare case of fetal death observed in the clinical practice and discuss antenatal screening.

Case report

A 35-week-and-3-day pregnant 26-year-old woman was admitted to a tertiary hospital for absence of fetal movements with three days of evolution. The patient was 2G0P, with a previous non-surgical voluntary abortion, and had grade 1 obesity and smoking history. She had no surgical background. The pregnancy had been followed in Primary Care. First- and second-trimester screenings were performed at the appropriate timing, and all serologies were negative, including for syphilis, but third-trimester ultrasound and standard blood tests had not yet been performed on admission. Absence of fetal vitality was confirmed via ultrasound, and labor was induced, with vaginal delivery of a male stillborn weighing 2220 g. Standard intrauterine fetal death studies were conducted, showing positivity for both treponemal and non-treponemal tests, with positive venereal disease titer for 128 dilutions. After this result, the woman reported a painless vulvar ulcer during the second pregnancy trimester, which she did not value. Fetal autopsy showed no internal or external malformations, and death was presumed to have occurred at 35 weeks. Pathology study of the placenta revealed higher-than-expected weight, acute chorioamnionitis lesions, high-degree villitis, intervillositis, and chronic deciduitis. The immunohistochemical study for anti-treponema pallidum antibodies revealed numerous spirochetes, confirming the diagnosis of placental syphilis.

Discussion:

Syphilis during pregnancy is the second leading cause of intrauterine fetal death globally. All pregnant women should be screened during the first pregnancy trimester or at the first pregnancy appointment. In Portugal, all women are also tested during the third trimester. According to the Centers for Disease Control and Prevention, women with a high risk of infection should be screened two times during the third trimester: at 28 weeks and close to delivery. Antenatal screening and adequate treatment have reduced the incidence of congenital syphilis and should be performed in all countries.

GO_14
PRIMARY AMENORRHEA - A MÜLLERIAN AGENESIS CASE REPORT

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Introduction

Primary amenorrhea represents failure to reach menarche. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is the second most common cause of primary amenorrhea (reported in 1 in 5000 females) after gonadal dysgenesis. Herein is reported a rare case of MRKH syndrome.

Case report

A 17-year-old nulliparous woman presented to the Gynecology Clinic with primary amenorrhea. A review of her pubertal development revealed onset of thelarche at the age of 11 and pubarche at the age of 12 years. She had an unremarkable medical, surgical, and social history and was not on any medications. Additionally, there was no history of congenital anomalies among family members. The mother's obstetric history was unremarkable, with no exposure to hormonal therapy or radiation while pregnant and full-term vaginal delivery.

The patient was of average height and weight and had normal arm span. Breasts were well developed, and distribution of axillary and pubic hair was normal (Tanner Stage 5), with grossly normal external female genitalia. She had an imperforate hymen. Complete blood count was normal, and laboratory test results for follicular stimulating hormone, luteinizing hormone, thyroid-stimulating hormone, estradiol, progesterone, testosterone, and prolactin were within the reference range, confirming normal reproductive female hormones. The karyotype was 46, XX.

On transabdominal ultrasound, only one of the ovaries could be visualized. Magnetic resonance imaging of the pelvis subsequently performed revealed the presence of two bilateral ovaries but absence of uterus and vagina. No urinary tract anomalies were observed. Based on these findings, type I Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome diagnosis was established.

The patient was offered extensive counseling regarding this diagnosis and its fertility implications, being proposed surgery with neovagina creation.

Discussion

Müllerian agenesis, eponymously referred to as Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a spectrum of congenital anomalies of unknown etiology characterized by a variable degree of uterovaginal agenesis in women with normal secondary sexual characteristics and 46, XX karyotype. The differential diagnosis includes androgen insensitivity, transverse vaginal septum, and imperforate hymen, and management of the condition includes screening for associated renal and skeletal anomalies.

GO_15

UPDATE ON PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS AND ITS RELATIONSHIP WITH INTRAHEPATIC CHOLESTASIS OF PREGNANCY –LITERATURE REVIEW

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Introduction

Progressive familial intrahepatic cholestasis (PFIC) is a heterogeneous group of inherited autosomal recessive diseases that begin in the neonatal period. It has an estimated incidence of 1:50,000 to 1:100,000 births. Three main clinical forms are acknowledged: PFIC1, PFIC2, and PFIC3. Intrahepatic cholestasis of pregnancy (ICP) is the most common liver disease in pregnancy and is characterized by transient cholestasis and pruritus.

The present Master's Degree thesis intends to perform a literature review of the three main clinical forms of PFIC and investigate the association of PFIC with intrahepatic cholestasis of pregnancy, thus contributing to the early diagnosis of both entities.

Methodology

Literature search on PubMed database of articles about the three main clinical forms of PFIC published in the last five years and articles about ICP published in the last two years.

Results

Three main clinical forms of PFIC were reported –PFIC1, PFIC2, and PFIC3 –, associated with three distinct genes: ATP8B1 (encoding for the FIC1 protein) in PFIC1; ABCB11 (encoding for the BSEP protein) in PFIC2; and ABCB4 (encoding for the MDR3 protein) in PFIC3. PFIC1 and 2 account for two-thirds of cases. A transient form of cholestasis manifesting in the third pregnancy trimester triggered by hormonal and environmental factors, such as ICP, can be observed in pregnant women with PFIC gene mutations.

Conclusion

In the absence of liver transplantation, PFIC results in high morbidity and mortality. On the other hand, despite resolving spontaneously after delivery, ICP is associated with severe fetal complications. Therefore, these two entities should be considered in cases with cholestatic presentation, allowing early and effective intervention. The same genes are implicated in ICP and the three main clinical forms of PFIC (most often the *ABCD4* gene), with ICP representing a continuum of the same disease.

GO_16

PITUITARY APOPLEXY IN POSTPARTUM: A CLINICAL CASE

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Introduction

Headache is a common puerperal complaint that is consistent with multiple etiologies. Pituitary apoplexy is a rare condition characterized by sudden-onset migraines, hormonal dysfunction, and visual disturbances. As the diagnosis during pregnancy or postpartum can be challenging, the authors report a clinical case of postpartum pituitary apoplexy managed conservatively, with favorable outcomes.

Clinical case

A 37-year-old woman, gravida 2 para 1, presented with a prior history of migraines that resolved after delivering her first child. Antenatal care was uneventful, and at 38 weeks of gestation, she was admitted to a tertiary center for premature rupture of membranes. Cesarean section (CS) was decided due to arrest of labor, and the woman gave birth to a male newborn with 3290 g and Apgar Index of 9/10 at 1st and 5th minutes. The anesthetic procedure consisted of epidural blockade, and the surgical procedure was uneventful, with an estimated blood loss of 200 mL. No complications were reported in the immediate postpartum, and the woman was discharged four days after delivery. On the 13th day postpartum, she presented fever, nausea, and persistent occipital throbbing headaches, partially responding to analgesics, which limited her daily activities and sleep quality. On admission, the patient was hemodynamically stable, with normal gynecological examination and without meningeal signs or neurological deficits. CT scan showed an enlarged pituitary gland with hemorrhagic lesions in the right and median portions. These findings were confirmed by cranioencephalic magnetic resonance imaging (MRI), evoking the diagnosis of pituitary apoplexy with no compression of chiasma optic nerves. Endocrinological tests were carried out, showing a slight thyrotropic deficit with normal cortisol. During hospitalization, a triple antibiotic scheme and analgesia were administered, and the patient was discharged four days later, requiring no surgical intervention. She currently maintains Endocrinology follow-up and experienced migraine resolution and thyroid function normalization. MRI pituitary images are also normal.

Discussion

Pituitary apoplexy is a potentially life-threatening endocrine disorder resulting from pituitary ischemia or hemorrhage. Only a small proportion of cases are correctly diagnosed in postpartum, as symptoms may overlap with those of normal postpartum state or following anesthetic procedure or CS. This emphasizes the importance of being alert to warning signs and securing a multidisciplinary approach in cases with a high index of suspicious, ensuring optimal clinical management.

GO_17
SPONTANEOUS BACTERIAL PERITONITIS DURING PREGNANCY – CASE REPORT

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Introduction

Spontaneous bacterial peritonitis (SBP) is a well-known complication of nephrosis, cirrhosis, and systemic lupus erythematosus, but its occurrence in an otherwise healthy adult is rare, whereby only a few cases of SBP complicating pregnancy in the absence of such morbidities have been reported. Herein is described a case of SBP during pregnancy from presentation to diagnosis, management, and follow-up.

Case report

A 37-year-old woman (gravida 1, para 0) presented at 22+2 weeks of gestation due to acute abdominal symptoms. She had Roux and Y gastric bypass (RYGB) surgery ten years earlier and laparoscopic cholecystectomy soon after. Her body mass index had been increasing over the last few years, reaching 43 kg/m^2 at preconception. Chronic hypertension was the patient's only known comorbidity. Gestation was the result of *in vitro* fertilization due to male infertility, and pregnancy was supervised by a dedicated multidisciplinary team specializing in obesity and bariatric surgery in pregnancy.

On admission, the woman mentioned intermittent abdominal pain with vomiting, nausea, and obstipation with three days of evolution. Vital signs were normal. The abdominal examination showed diffuse tenderness with positive Blumberg's sign, and the obstetric examination was unremarkable. Laboratory tests revealed high C-reactive protein and leucocytosis. Intra-abdominal free fluid and dilated small intestine were evident in magnetic resonance imaging. Due to suspicion of ischemic bowel syndrome, an emergency midline laparotomy was performed. An abundant exudate was removed, but no etiology could be identified. The patient was on intravenous antibiotics and was discharged home eight days later. *Escherichia coli* was isolated in the exudate.

Pregnancy followed a normal course thereafter until it was complicated by superimposed preeclampsia at term. Cesarean section was performed at 37+4 days due to non-reassuring fetal status. Although fibrin-rich intra-abdominal free fluid was observed, the woman and the newborn were discharged home four days later.

Discussion

Although rare, SBP can be a life-threatening condition to both the mother and fetus. Although surgical exploration did not identify an internal hernia, intestinal sub-occlusion after RYGB has been reported. This case highlights the importance of considering every clinical presentation and feature of the maternal history.

GO_18
MULTIPLE SCLEROSIS AND PREGNANCY – A MULTIDIMENSIONAL CHALLENGE

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Introduction

Multiple sclerosis (MS) is an immune-mediated demyelinating disease of the central nervous system characterized by relapses and remissions of neurologic deficits. The relapse rate decreases during pregnancy, but some symptoms may worsen, particularly in women with very active disease. In childbearing women, the decision to initiate, stop, or maintain treatment with disease-modifying drugs should take into account the risk of relapse, probability of pregnancy, and teratogenic potential of the specific drug.

Clinical case

Herein is reported the case of a 42-year-old woman, G1P1, diagnosed with MS at 32 years old and with several relapses since, with permanent sequelae. MS control was achieved with fingolimod. In the pre-conceptional consultation, information about pregnancy outcomes in MS setting was provided, risks of stopping fingolimod were discussed, and the patient was advised to undertake a washout period of 2 months if pregnancy was a possibility.

The woman stopped fingolimod but experienced two relapses shortly after, and as pregnancy did not occur, she resumed the drug after a negative pregnancy test and instructions to start contraception immediately. She returned to consultation 10-week pregnant, having taken fingolimod up to 4 weeks post-conception. After being informed of the probability of relapse and risks associated with fingolimod teratogenicity, the woman decided to go through with the pregnancy.

At 21 weeks, she experienced a new steroid-refractory relapse requiring hospitalization and was started on natalizumab.

Vaginal delivery occurred at 37 weeks after spontaneous rupture of membranes. Although asymptomatic, the woman arrived at the Emergency Department with a fully dilated cervix. Six months postpartum and still medicated with natalizumab, no new relapses had been reported. Concerning the newborn, a small ventricular septal defect and mild anemia and thrombocytopenia were diagnosed after birth.

Discussion

Rebound of MS activity after fingolimod cessation has been reported and can be severe. Although pregnancy is believed to be protective, it has been shown to be unable to prevent MS relapses in these cases, as illustrated in this report. When considering therapy introduction, both benefits and risks (including fetal adverse side effects emerging in this case) should be weighted. This case highlights the value of a multidisciplinary approach for women with MS and the importance of counseling and therapy management starting in preconception.

GO_19

TREATMENT AND MANAGEMENT OF IMMUNE THROMBOCYTOPENIA DURING PREGNANCY: A 10-YEAR RETROSPECTIVE STUDY AT A TERTIARY CENTER

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Introduction

Immune thrombocytopenic purpura (ITP) complicates 0.1-1/1000 pregnancies worldwide. Maternal autoantibodies responsible for platelet destruction and reduced production may cross the placental barrier and cause neonatal thrombocytopenia. The management of pregnant women with ITP requires multidisciplinary collaboration to reduce maternal and neonatal risks.

Methods

The aim of this study was to retrospectively assess pregnancies in women with ITP delivering at a tertiary hospital with a mean of 3175 deliveries/year during a 10-year period (Jan 2012-Jan 2022). Medical files with ICD-9 and ICD-10 ITP classification were reviewed for medical, obstetrical, and neonatal outcomes. A literature review was conducted using the terms "immune thrombocytopenia", "pregnancy outcomes", and "neonatal outcomes". Statistical analysis was performed using SPSS Statistics v.27 and comprised the application of Chi-square, Pearson correlation, and T-student tests.

Results

Eighteen pregnancies were identified in 16 women (0.57/1000 deliveries) over the considered 10-year period. The mean (+-standard deviation [SD]) interval between disease diagnosis and pregnancy was 6.67 (+-5.40) years, with 88.9% of women being diagnosed before pregnancy. The mean (+-SD) minimum platelet value during pregnancy was 32.40+-27.12x10^3/uL. A total of 27.8% of women had hemorrhagic symptoms, and 27.8% required hospitalization. Half (50%) required treatment during pregnancy, which consisted of corticoids in 38.9%, intravenous immunoglobulin in 38.9%, and splenectomy in 11.1%. Regarding neonates, 33.3% had thrombocytopenia, and 22.2% required medical treatment. A moderate to strong correlation was found between maternal hemorrhagic symptoms (p=0.009), platelet count <50 x10^3/uL (p=0.025), and maternal need for therapy during pregnancy (p=0.003) and neonatal alloimmune thrombocytopenia.

Conclusion: Although successful pregnancy is possible in women with ITP, hemorrhagic complications, disease progression, and need for maternal treatment may be complicating factors, correlating with neonatal risk for alloimmune thrombocytopenia.

GO_20

TWIN-TWIN TRANSFUSION SYNDROME: CASE SERIES OF PERINATAL OUTCOMES AND MANAGEMENT AT A TERTIARY CENTER

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Introduction

Twin-twin transfusion syndrome (TTTS) is a complication reported in 10-15% of monochorionic diamniotic (MCDA) pregnancies. Diagnostic criteria require monochorionic placentation and presence of oligohydramnios in one sac and polyhydramnios in the other. Bimonthly serial sonographic evaluation should be considered as a screening tool for the condition, starting at around 16 weeks until delivery. A high rate of perinatal loss is reported in cases of advanced TTTS. Laser-treated TTTS has a reported perinatal mortality rate of 30-50% and up to 20% chance of long-term neurologic handicap. The aim of this study was to report a series of TTTS cases and respective perinatal outcomes.

Methods: All cases of twins followed at the Maternal-Fetal Medicine Unit of the considered tertiary center during a 4-year period (January 1, 2018 to December 31, 2021) were retrospectively identified. Among these, cases of MCDA pregnancies were selected, and cases of TTTS subsequently identified. Exclusion criteria comprised multiple gestations of >2 fetuses, vanishing twins, loss of follow-up, and insufficient data. The Quintero staging system was used to describe the severity of TTTS.

Results

A total of 335 twin cases were identified, 64 of which were excluded. Among the 271 cases selected, 55 corresponded to MCDA pregnancies. Seven cases of TTTS were identified, corresponding to 12.7% of MCDA pregnancies, all of which were spontaneous. The mean gestational age at diagnosis was 19.7 weeks. Two pregnancies were treated with fetoscopic laser ablation and one with serial amniodrainage. The two cases treated with laser had at least one live fetus at birth: in one case, the fetus was born at 30 weeks after treatment at 22 weeks, and fetal death of the donor twin at 24 weeks; in the other case, both fetuses were born at 34 weeks after treatment at 18 weeks. In the latter, both children presented global developmental delay at three years old. The pregnancy treated with serial amniodrainage had two live births at 31 weeks, but the follow-up time is still short to allow drawing conclusions regarding neurodevelopment. All four pregnancies with no treatment were complicated with fetal or perinatal death.

Conclusion

TTTS is an unpredictable complication with relevant morbimortality. Early identification of chorionicity and serial sonographic assessment can help diagnose and classify this condition as early as possible. Improvements in the treatment and management of TTTS can improve the associated outcomes, with evidence that fetoscopic laser ablation improves survival rates. However, long-term neurodevelopment impairment after laser surgery still occurs in about 10-20% of children.

GO_21

ACUTE DYSPNEA IN POSTPARTUM – CASE REPORT

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Introduction

Acute pulmonary edema in postpartum is an uncommon but life-threatening event. Contributing factors include previous administration of tocolytics, cardiac disease, preeclampsia, and iatrogenic fluid overload. The condition typically presents with acute dyspnea, and the differential diagnosis includes pulmonary thromboembolism (PTE) and peripartum cardiomyopathy (PPCM). The mother should be admitted to a specialized unit, and medical management and etiologic assessment should be performed.

Report

Herein is reported a case of postpartum acute pulmonary edema presenting as acute dyspnea after cesarean delivery of a twin pregnancy.

A 36-year-old primipara with BC twin pregnancy after *in vitro* fertilization had induced labor at 36 weeks due to fetal growth restriction of one of the fetuses. Cesarean delivery was performed due to arrest in labor, which was complicated by postpartum hemorrhage due to uterine atony. The patient was given tocolytics and fluid therapy and submitted to a blood transfusion. Two days after labor, she presented with fatigue and acute-onset dyspnea. Computed tomography angiography was performed to exclude PTE, which was normal. The Cardiology Unit was contacted to evaluate the possibility of PPCM, with transthoracic echocardiogram revealing normal ventricular systolic function. The diagnosis of acute pulmonary lung edema due to fluid overload was assumed, and the patient was put on diuretic therapy. She responded to therapy with remarkable clinical evolution and was discharged on day 7 after delivery.

Conclusions

Although uncommon, acute pulmonary lung edema can occur in postpartum, representing a life-threatening event. Timely identification of the condition and correct implementation of measures are crucial to improving the outcomes. Acute dyspnea can present in the setting of multiple pathologic conditions, with the etiologic study required to direct and individualize therapeutic measures.

GO_22

AUTOIMMUNE HEPATITIS AND PREGNANCY: A CASE-CONTROL STUDY

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Introduction

Autoimmune hepatitis (AIH) is a chronic inflammatory liver condition that manifests in women of childbearing age. It is a rare disease with a prevalence of 11 to 25 per 100 000 individuals. Data on AIH and pregnancy are scarce, with available studies suggesting an increased risk of adverse maternal and fetal outcomes. This study aimed to evaluate obstetric outcomes in women with AIH and compare them with those of a group of healthy pregnant women.

Methods

This was a retrospective case-control study including all women diagnosed with AIH and monitored during pregnancy and postpartum at the authors' institution. AIH diagnosis was established based on the International Autoimmune Hepatitis Group (IAIHG) criteria. Pregnancy outcomes were compared with those of a control group of healthy pregnant women. Two controls per case were randomly selected. Statistical analysis was performed in SPSS 28.0.

Results

Twenty-two pregnancies in 17 patients with AIH were included in the study. The disease was diagnosed at a mean (±standard deviation [SD]) age of 26.0 (±7.6) years and the median time between AIH diagnosis and pregnancy was 4.0 (9.3) years. Prednisolone and azathioprine were administered in 90.9% and 68.2% of pregnant women, respectively. No significant differences were found between the two groups regarding age (p=0.443), body mass index at conception (p=0.331), and parity (p=0.209). Obstetric complications reported in AIH patients included four miscarriages (18.2%), seven preterm deliveries (38.9%), two gestational diabetes (GD; 11.1%), one preeclampsia (PE; 5.6%), and one fetal growth restriction (FGR; 5.6%). The median gestational age at birth and birth weight in the AIH group were 37.5 (5.0) weeks and 2975.0 (879.0) grams, respectively. Low birth weight (LBW) was observed in six (33.3%) newborns. AIH flares complicating the pregnancy and/or postpartum were reported in four (18.2%) cases. Cesarian delivery occurred in 12 (66.7%) women with AIH. A significant increase in the rate of preterm birth (p=0.037), LBW (p=0.009), and cesarian section (p=0.003) was observed in AIH patients. No significant differences were found between groups in the rate of miscarriage (p=0.425), GD (p=0.646), PE (p=0.536), FGR (p=0.947), or Apgar score at 1 (p=0.174) and 5 (p=0.472) minutes.

Conclusion

The results of this study corroborate previous findings of an increased risk of preterm birth, LBW, and cesarian delivery in women with AIH. However, most cases presented experienced no obstetric complications or AIH flares. With proper management, AIH patients are expected to achieve favorable maternal and fetal outcomes.

GO_23

IS THERE A CORRELATION BETWEEN FETAL POSTERIOR VENTRICLE WIDTH AND BIOMETRIC PARAMETERS?

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Introduction

Measurement of fetal ventricle size is part of routine second- and third-trimester ultrasound (US) scans. Ventriculomegaly (VMG) is diagnosed when the width of at least one of the ventricles exceeds 10 mm. Although isolated mild VMG has been previously associated with a higher incidence of fetal anomalies, recent studies suggest that it does not necessarily represents a pathological condition and may instead be related to other large fetal biometric measurements.

The aim of this study was to assess the correlation between fetal posterior ventricle measurement (PVM) in the third trimester and weight and head circumference (HC) in US and at birth.

Methods

A retrospective observational study was conducted through search of third-trimester USs of singleton pregnancies followed at this study's hospital and registered on Astraia® between October 1 and December 31, 2021. Preference was given to cases of PVM available on record (28 to 38 weeks of gestational age [GA]). If no measurement was accessible, a new was obtained (if available images allowed to). Data on birth weight (BW) and HC were collected from patients' medical records (Obscare® and Medtrix®). Statistical analysis was performed on SPSS® 28.0, with statistical significance set at p-value <0.05.

Results

A total of 285 pregnant women with third-trimester US registry on Astraia® were identified. Of these, 92 were excluded due to PVM impossibility (n=66), twin pregnancy (n=12), availability of fetal echocardiography only (n=9), suspicion of cytomegalovirus or herpes zoster infection (n=4), and fetal demise (n=1). The mean GA was 31 weeks. Two cases of VMG were identified. PVM showed a very weak positive correlation with HC on US (rho=0.197; p=0.006). No correlation was found between PVM and GA in US, BW centile, or HC at birth.

Conclusions

In disagreement with recent findings, no significant correlation was found between PVM and biometric parameters at birth. However, this study has limitations that should be acknowledged. Firstly, it mainly included women with high-risk pregnancies (pregnant women with diabetes mellitus, gestational diabetes, previous bariatric surgery, thyroid disease, hypertensive disorder, autoimmune disease, and fetuses with growth restriction), as those with low-risk pregnancies usually do not have their third-trimester US at this study's hospital. On the other hand, only two cases of VMG were included, and PVM was performed by several different operators.

Larger studies should be conducted to further investigate the correlation between PVM and biometric parameters at birth, especially in cases of isolated fetal VMG.

GO_24

UNEXPECTED FINDINGS: A UNIQUE PRENATAL DIAGNOSIS OF TEMPLE SYNDROME

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Introduction

Temple syndrome (TS, MIM #616222) is a genomic imprinting condition caused by disturbances in the imprinted region 14q32. Imprinting disorders arise from deleterious variants affecting imprinting control regions, which exhibit parental-specific DNA methylation acquired during germline development.

TS is an ultra-rare condition with unknown prevalence and is likely underdiagnosed. This study reports the first case of prenatal diagnosis of TS.

Case report

A 29-year-old woman was referred to a Fetal Medicine Consultation at 21w+6d after findings of unilateral clubfoot and growth below the 5th centile on routine morphological ultrasound. The couple was healthy, non-consanguineous, and had no relevant family history.

In the context of etiological diagnosis, amniocentesis was performed at 22w+5d, as well as karyotype (result: 46,XX) and rapid aneuploidy (result: negative) tests. At 24w, an MS-MLPA for UPD7-UPD14 was performed using the fetal DNA sample as control, and hypomethylation of the 14q32.3 region was detected. Due to the relevance of this finding, the sample was un-pseudonymized and the couple was referred for Medical Genetics assessment. After receiving genetic counseling and information on TS, the couple opted to proceed with the pregnancy. They agreed to pursue the study, and SNP-array was performed on fetal and maternal samples. No uniparental disomy or deletions were found, asserting epimutation as the most likely disease-causing mechanism.

A female baby was delivered by C-section at 33w+6d due to fetal growth restriction and breech presentation, with APGAR score of 4/7/9. She was small, floppy, and had right clubfoot, as well as mild dysmorphic facial features. After 14 days at the Neonatal Intensive Care Unit, the girl was transferred to the local hospital. She is currently 18 months old, has proportionate short stature, adequate psychomotor development, and undergoes regular physiotherapy after clubfoot repair. She also underwent successful treatment of four angiomas with propranolol.

Discussion

Albeit unexpected, early diagnosis of TS in this case was invaluable in informing the parents and medical team on prognosis, allowing for specific pregnancy and labor preparation, as well as early postnatal referral for multidisciplinary care.

TS prenatal diagnosis was well accepted by the couple, and additional studies established a low recurrence risk for future pregnancies.

GO_25

DIAGNOSIS OF FETAL ABDOMINAL CYST AT 11-13-WEEK ANTENATAL SCAN: FIRST SIGN OF ANORECTAL MALFORMATION?

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Introduction

Intra-abdominal cysts in the first trimester are rare, usually representing isolated findings and being associated with favorable outcomes. However, abdominal cysts of uncertain origin remain a complex challenge in prenatal surveillance. Hereby is described the case of a newborn with anorectal malformation (ARM), in which an abdominal cyst at 13 weeks was possibly the first prenatal ultrasound sign of the condition.

Case report

A 28-year-old primigravida with a medical history of asthma was admitted for routine first-trimester fetal ultrasound scan. A cystic image was detected in the right lower abdomen. On the 17-week scan, the cystic image disappeared but a single umbilical artery was confirmed. An echocardiography was performed five weeks later, with no defects identified. Pregnancy was uneventful until 34 weeks, when fetal growth restriction was diagnosed (estimated fetal weight under the second centile), with umbilical artery pulsatility index above the 95th centile. In addition, a 15-mm hypoechogenic structure was identified in the fetal right iliac fossa. A single course of corticosteroid therapy for fetal lung maturation was given before labor induction, and a male newborn of 1660 grams and Apgar score 7/8/9 was delivered by vaginal birth. On physical examination, he was diagnosed with ARM with perineal fistula. On the first day of life, distended sigmoid colon lying 4 cm from the anal verge was also identified, and the newborn underwent a sigmoid colostomy, without major complications. Array comparative genomic hybridization was normal. He is currently three months old, thriving, and with good weight gain.

Discussion/Conclusion

The literature regarding fetal abdominal cysts associated with ARM in the first trimester is scarce. This case highlights the need for close prenatal follow-up of these cases since they can be related to serious gastrointestinal conditions and require expeditious interventions after birth.

GO_26

LONG-TERM OUTCOMES OF MILD FETAL VENTRICULOMEGALY

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Introduction

Ventriculomegaly is the most common intracranial anomaly diagnosed prenatally. Mild (atrial width 10-12 mm) and isolated (when not associated with other sonographic abnormalities) ventriculomegaly usually has a favorable prognosis, with about 90% of fetuses presenting normal neurodevelopment. The aim of this study was to assess the postnatal outcomes of fetuses with mild ventriculomegaly and identify features associated with poor neurodevelopmental outcomes.

Methods

Retrospective study of mild ventriculomegaly cases diagnosed during prenatal care at Maternidade Alfredo da Costa Prenatal Center between January 2016 and December 2017. The search was conducted on ASTRAIA software. Data were retrieved from electronic medical records and comprised complementary studies (TORCH test for fetal infection, detailed ultrasound examination, and invasive testing for karyotyping), obstetric outcomes, and postnatal neurodevelopmental status.

Results

Thirty-six cases of mild ventriculomegaly were identified, with an average gestational age of 23 weeks. An association with sonographic abnormalities was found in 20 cases, with brain, spine, and urinary tract anomalies being the most frequent findings. One case of aneuploidy and one case of infection were also identified. Spontaneous in utero resolution occurred in 21 cases, and ventriculomegaly progressed in three patients and remained stable in five. Six cases resulted in pregnancy termination, one of trisomy 21 and five of polymalformative syndrome (with one case of congenital infection by toxoplasmosis). The live born rate was 83% (n=30). Neurodevelopmental status was evaluated in 21 infants and considered normal in 67% (n=14). Seven infants were diagnosed with neurodevelopment delay. Six of these were boys with associated prenatal brain abnormalities, including corpus callosum anomalies, choroid plexus cysts, delayed sulcation, and ventriculomegaly progression. Of the 16 cases with isolated ventriculomegaly (12 of which unilateral), spontaneous resolution was observed in 81% (n=13), with all infants submitted to postnatal evaluation showing normal neurodevelopment.

Conclusions

Despite the high rate of data lost to follow-up, most fetuses with mild ventriculomegaly had a favorable course and near-normal developmental status in this study. Association with other brain abnormalities was the most evident feature among infants with neurodevelopmental delay, potentially contributing to unfavorable outcomes.

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COMUNICAÇÕES ORAIS

ENF_01
SAFETY CULTURE IN OBSTETRIC CARE

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Introduction: Patient safety, as a health and safety quality component, has assumed a great relevance in health politics. Error notifications should be viewed as an opportunity for improvement.

Objective: To describe the perception of health professionals (doctors and midwives) of the safety culture for patients.

Methods: Quantitative cross-sectional and descriptive correlational study based on the Hospital Survey on Patient Safety Culture (AHRQ, 2004) and socio-demographic and professional questions applied to a sample of 121 health professionals.

Results: The sample mainly consisted of female professionals (90.9%) with nurse professional category (89.3%) and graduation (86.0%). A total of 66.9% did not notify any event or occurrence in the last 12 months, and 50.4% considered patient safety as acceptable and 45.5% as very good. The "teamwork" dimension achieved the highest positive value, although that does not make it a strong argument. The dimensions classified as problematic and hence requiring priority intervention were "Nonpunitive response to error", "Staffing", "Frequency of events reported" and "Management support for patient safety".

Conclusion: Patient safety culture in the Obstetrics Department was perceived by participants in this study as a culture of fear of punitive response to error. The safety of any organization should be a central concern and regarded as an evolutionary process, open to the implementation of corrective actions and influenced by sociodemographic and health professional-related variables.

Key words: culture; maternal health; medical error; patient safety; quality of health care

ENF_02
TAKING CARE OF TWINS: WHEN ONE HAS DOWN SYNDROME

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Background: Down syndrome (DS) is the most common chromosomal disorder affecting newborns (NBs). It is usually diagnosed prenatally and rarely found in twins, occurring in 14 to 15 per million non-identical twins. For parents, the emotional implications of having premature babies in a Neonatal Intensive Care Unit (NICU) are heavy and aggravate when one is diagnosed with DS. The authors expect that the description and analysis of this case prompt a reflection on Nursing practices, diagnoses, and interventions.

Case report: Maria (pseudonym) was a 34-year-old mom working at a Radiology Unit. With infertility issues and one previous abortion, she had to go through *in vitro* fertilization. She became pregnant with twins but had gestational diabetes and pre-eclampsia, with the second fetus presenting intrauterine growth restriction. None of the prenatal exams suggested the possibility of DS. The situation deteriorated, and she had an emergency cesarean delivery at 32 weeks and one day. Babies had Apgar scores of 8 and 9, respectively. They initially presented a similar evolution, requiring continuous positive airway pressure for 12 hours, treatment with antibiotics (sepsis), and phototherapy. Vanda was ready for discharge at D32, but Pedro experienced several problems, including motor discoordination, hypotonia, difficulty eating (starting speech therapy on D35), and physical issues, including slanting palpebral fissures, flattened nasal bridge, protruding tongue, low set ears, and short neck. By D17, he was diagnosed with DS (confirmed through karyotyping) and congenital heart disease. He started diuretics on D39. On D55, the boy was able to eat through a bottle. At discharge (D59), he maintained major development and growth differences from his sister. The mother experienced a difficult time, with the need for professional and personal support. She cared for both children but was concerned with the future.

Discussion: Although the case of twins in which one has DS is rare, comprehensive support should be available for these parents during their children's stay at NICU. It is important that nurses understand the impact of DS on these families since they are responsible for guiding and teaching parents how to care for both NBs and assessing their needs. In the present study, personalized care could be provided due to the in-depth knowledge of NBs and family and understanding of their specific needs at a time of uncertainty. Nursing support and adequate interventions aiming at rehabilitation and child development were crucial.

ENF_03
IMPACT OF THE COVID-19 PANDEMIC ON THE MENTAL HEALTH OF CHILDREN AND TEENAGERS AND THE IMPORTANCE OF NURSE INTERVENTION

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Introduction: The COVID-19 pandemic caused a sudden change in family routines due to the adoption of social distancing protocols and changes in the social life of children and adolescents related to school closure and home lockdown.

Objective: To understand the impact of the COVID-19 pandemic on the population of children and adolescents of Unidade Local de Saúde de Matosinhos (ULSM) and which nursing interventions are relevant to attend to their needs and promote their mental health.

Methods: Integrative literature review (2018-2022) using the descriptors "child", "adolescent", "mental health", "covid 19", and "nursing interventions" with combination of Boolean operators "AND" and "OR". Data sources included CINAHL, MEDLINE, and COCHRANE databases. Full-text articles published in Portuguese, Spanish, and English in the five-year study period were included.

Results: A total of 260 articles describing the impact of the COVID-19 pandemic on the mental health of children and adolescents and four articles reporting Nursing interventions directed at mental health promotion were included in the analysis. Depression, anxiety, and stress were the most commonly reported symptoms.

A 37.9% increase in Child Psychology and Psychiatric appointments in ULSM was observed in the last year compared to the 2018 pre-pandemic period. The first trimester of 2022 had already registered a 4% increase compared to the same period in 2021.

Parental ability and therapeutic partnership are crucial for successful interventions.

Conclusion: The COVID-19 pandemic had a substantial impact on the mental health of children and adolescents, bringing new challenges to healthcare professionals. According to the Portuguese Standard 011/2020 of the Directorate-General of Health (DGS), mental health responses should be adequate to the specific needs brought by the pandemic, from general information dimensions to prevention and healthcare interventions. Nurses have an important role both in promoting mental health recovery in children and adolescents and in the adoption of family support strategies. It is crucial that pediatric specialist nurses have a more active role in all child health dimensions.

Reference: Direção Geral da Saúde, 2020, –COVID-19: FASE DE MITIGAÇÃO—nº 011/2020.

ENF_04
NEUROLEPTIC MALIGNANT SYNDROME: CARING FOR TEENAGERS AND FAMILIES ON INTENSIVE CARE UNIT ADMISSION – CLINICAL CASE

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Introduction: Neuroleptic malignant syndrome is a neurological emergency caused by dysregulation of dopaminergic neurotransmission, usually by exposure to a dopamine antagonist. It is characterized by hyperthermia, generalized muscle stiffness, changes in mental status, and hyperactivity of the autonomic nervous system. The diagnosis is clinical, and treatment includes interruption of the liable drug and application of supportive measures.

The condition has a reported incidence of 0.02 to 3% in patients medicated with neuroleptics and is prevalent in young adults, although potentially affecting all ages. It can resolve within two weeks or last for months in rare cases, being fatal in 10 to 20% of patients.

The increasing use of psychopharmaceuticals, including neuroleptics, in pediatric patients reinforces the importance of addressing this topic in a way that spurs critical reflective analysis. Timely training of nurses who may come into contact with such cases in the intensive care setting is crucial.

Clinical case: Herein is presented the case of a 16-year-old boy with a history of ulcerative colitis and hyperactivity/attention deficit disorder undergoing Psychiatry follow-up appointments for anxiety, insomnia, depressive mood, and cannabinoid consumption, with two previous hospitalizations for voluntary drug intoxication.

The boy was attended to due to altered level of consciousness and tachycardia after ingesting 14 olanzapine tablets (280 mg). Endotracheal intubation was performed at home, and the patient was later transferred to the Pediatric Intensive Care Unit for continuity of care. On admission, he presented with global hypertonia, diaphoresis, tachycardia, hypertension, and hyperthermia. Complementary diagnostic tests excluded other toxicological, infectious, metabolic, or neurological causes.

Discussion: Systematization of the concept of autonomous and interdependent Nursing care for teenagers with neuroleptic malignant syndrome admitted to intensive care and their families is reflected in better performance and quality of care and emphasizes the need for investment in Nursing training and education in accordance with scientific evidence.

The following areas of attention were identified in the elaboration of the Nursing process: self-care: hygiene; self-care: use of the toilet; pressure ulcer; fall; pain; nutritional intake; parental role; airway clearance; ventilation; tissue perfusion; skin; ulcer; and hyperthermia.

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POSTERS

ENF_05
CARE FOR WOMEN WITH NDOMETRIOSIS: EESMO SKILLS!

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Introduction: Endometriosis is a chronic inflammatory gynecological condition defined by the implantation of endometrial tissue outside the uterine cavity, affecting women of reproductive age by impairing their daily life and activities, personal relationships, and reproductive capacity. The greater the severity of symptoms, the lower patients' quality of life. The nurse specialist in Maternal and Obstetric Health Nursing (EESMO) plays an important role in this setting by promoting patients' autonomy, knowledge, and empowerment, hence alleviating their suffering and promoting their quality of life. The present working group aimed to highlight the emotions of women with endometriosis and the importance of EESMO in their care.

Methodology: This was an exploratory and descriptive study of data collected in the EBSCO database through a retrospective literature search.

Results: Since women have conquered their space in society, their profile has changed from passive to active, and they have started to have control over their periods of life (climacteric, menopause, and old age). However, they also had to learn to deal with new diseases, especially endometriosis, which affects more than 6 million women worldwide and has a prevalence of 10–15%. In the setting of Nursing consultation, anamnesis is extremely important to outline the various interventions, contributing to the quality of treatments undertaken. Assistance provided by nurses should be comprehensive and focus on emotional, social, and psychological symptoms to obtain the greatest benefits, making the therapeutic process more assertive in women suffering from depression and anxiety. In health education actions, an individualized and multidisciplinary approach is required to reduce symptoms.

Conclusion: Endometriosis compromises women's quality of life, with nurses having a crucial role in ensuring the provision of knowledge and empowerment of these patients. Only EESMO are able to refer women to adequate treatment and help them avoid the potentially devastating psychological problems triggered by the condition, providing more security for women to face the difficulties and possible consequences of the disease.

ENF_06
ANALYSIS OF THE CONCEPT OF OBSTETRIC VIOLENCE: SCOPING REVIEW

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Introduction: Labor and delivery are natural processes of human reproduction and are unique to each woman due to the numerous challenges they entail. The environment in which women experience childbirth can decisively influence the childbirth experience and hence women's overall well-being. Cases of dehumanized assistance, medicalization abuse, and pathogenesis of reproductive physiological processes are examples that may fall under the concept of obstetric violence. Obstetric violence has been highlighted in the political and social agenda of several countries. Efforts have been made to establish policies that allow to humanize obstetric care, guarantee parturients' rights, and respond to this form of violence. The lack of consensus on the proper terminology to name and define behaviors that constitute obstetric violence makes this process difficult. The aim of this study is to retrieve evidence of the characteristics and dimensions of the concept of obstetric violence in the care of women in labor.

Methodology: This will be a scoping review conducted according to the Joanna Briggs Institute method. The revision question was defined by adopting the PCC acronym (Population, Concept, Context): How do you characterize the concept of obstetric violence (C) while assisting the woman (P) in the context of parturition (C)? The search will be conducted on EBSCOhost, PubMed, Virtual Health Library, and Scopus databases using the descriptors "obstetric violence", "woman", and "parturition". The Open Scientific Repository of Portugal will also be considered. Inclusion criteria comprise all types of studies published since 2017 in English, Spanish, and Portuguese. Studies of women with labor experience in the hospital environment addressing dimensions of the concept of obstetric violence will be reviewed.

Results: Extracted data will be depicted in table format. A narrative approach will also be carried out to organize and categorize the main conclusions regarding the characteristics and dimensions of the concept of obstetric violence.

Conclusion: The main terminologies associated with the concept of obstetric violence, as well as its typologies, predisposing factors, and implications in the childbirth experience will be summarized, and results retrieved will be used as a basis for conducting research on this issue.

Keywords: obstetric violence; parturition; woman

ENF_07
NURSE TRAINING: A QUALITY INDICATOR

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Introduction: Single national parameterization challenged nurses to understand the new information system. Parenthood data provides a relevant indicator for assessing Nursing data, as it requires the assessment of nurses' knowledge regarding the parental role, quality standards, and Nursing indicators, as well as documentation created through clinical practice, carrying out training sessions and evaluating the impact of this training on the knowledge and documentation produced.

Objectives: To quantify the records of focus parental role as a diagnostic tool to identify barriers encountered for elaborating the diagnosis and identify the knowledge of nurses regarding the parental role, quality standards, and nursing indicators before and after training.

Methodology: This was an action research study of data collected through the application of a questionnaire to nurses before and after training. The clinical files of children admitted to the Pediatric Ward of a hospital in the center region of Portugal were analyzed after Ethics Committee approval.

Results: Before the training session, most nurses pointed out the need for training (93%; p=0.001), many admitted not updating the Nursing process adequately (87%; p=0.007), and 87% admitted finding it difficult to carry out the parental role diagnosis. The analysis of Nursing data regarding the focus on parental role is limited. After training, there was an increase in knowledge regarding quality standards (p=0.005) and quality indicators (p=0.006), and a decrease in difficulties/barriers to documenting the clinical practice (p=0.016). There was also an increase in Nursing records of focus parental role.

Conclusion: Training sessions contributed to the acquisition of knowledge and better practices by nurses, but the most important factor is the existence of a training and research policy.

ENF_08
ADHERENCE TO PEDIATRIC ROUTINE VACCINATION IN THE COVID-19 PANDEMIC

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Introduction: Difficulty in adherence to routine immunization was observed with the SARS-CoV-2 pandemic. The World Health Organization recommended maintaining routine vaccination services but offered each country the freedom to decide whether to do so according to the rate of SARS-CoV-2 contamination. This study sought to investigate adherence to routine vaccination in times of pandemic by assessing the availability of health services to maintain vaccination coverage and addressing immunization rates in each continent.

Methodology: An integrative literature review was conducted on the PubMed database, and a narrative synthesis was subsequently performed to answer the following question: What is the adherence to routine vaccination in Pediatrics in times of COVID-19 pandemic?

As inclusion criteria, the language of studies (Portuguese, Spanish, or English), time period (between 2019 and 2022), and study subjects' age (under 18 years) were considered. The following search phrase was used: "((immunization OR vaccination)) AND (routine)".

Results: Of 1299 articles retrieved, 40 met inclusion criteria and were selected. Their analysis revealed that approximately 70% of studies explicitly and unequivocally reported a reduction of routine vaccination in children related to the COVID-19 pandemic. Globally, although childhood vaccination was prioritized, communication with families was intensified, flexibility in all organizational levels of child healthcare services was increased, health centers remained open, and vaccine supply was kept, a decrease in compliance with routine vaccination occurred.

Conclusion: This study shows that adherence to routine pediatric vaccination in times of COVID-19 pandemic was greatly affected. The authors emphasize the need for healthcare services to continue supporting immunization programs, not only in pandemic setting but also afterward, since neglecting this factor may result in the re-emergence of previously eradicated conditions, with compromise of children and adolescents' health.

Keywords: children, pandemic; routine vaccination; vaccination rate

ENF_09
IMPACT IN CHILDREN AND ADOLESCENTS OF RESTRICTIONS IN ORAL HYGIENE HABITS DUE TO COVID-19

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Introduction: The impact of the COVID-19 pandemic on pediatric oral health and limited access to dental care should be addressed. Good oral health is crucial for health, well-being, and quality of life. This study sought to investigate the correlation between the oral health of children and adolescents and the actions of health professionals during the pandemic to understand the impact of the restrictions associated with it.

Methodology: This study consisted of a literature review in CINAHL and BVS libraries followed by narrative synthesis and conceptual and thematic analysis focusing on the following main question: What is the impact of COVID-19-associated restrictions on oral hygiene habits of children and adolescents? The search was conducted using the following search strategy: ("oral care" OR "mouth care" OR "oral hygiene" OR "dental care") AND ("children" OR "adolescents" OR "youth" OR "child" OR "teenager") AND ("pandemic" OR "epidemic" OR "outbreak" OR "covid-19" OR "coronavirus"). Inclusion criteria comprised articles published in the period between 2019 and 2021, written in Spanish, Portuguese, or English, and focusing on the age range between 2 and 18 years old.

Results: From 265 articles initially retrieved (34 from CINAHL and 231 from BVS), 10 were selected for analysis. Retrieved data showed a clear difficulty of parents in maintaining good oral hygiene in their children during the pandemic, as a result of the increase in eating habits detrimental to oral health, such as ingestion of snacks and sugars, and lower demand for dental health care. The fear of cross-infections was reflected in families' access to dental care, either for treatment or preventive purposes. This study highlights the need to raise awareness of children/adolescents and their families about the importance of effective oral hygiene, especially in the pediatric age group.

Conclusion: It is important to inform and alert families about the risks associated with neglecting pediatric dental health and raise their awareness regarding lifestyle habits that influence oral hygiene habits. Oral health is an integral part of general health, warranting interventions across multiple health areas, with emphasis on programs directed at children/youth and families.

Keywords: adolescent; children; COVID-19; oral health

ENF_10
IMPORTANCE OF RELAXATION TECHNIQUES IN THE ADOLESCENCE PSYCHIATRY INPATIENT DEPARTMENT

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Introduction: The excessive use of social networks and practice of online games and the invalidation and little knowledge of associated diseases are factors contributing to the impairment of mental and hence brain health. In this context, young people may require help from mental health specialists for guidance, sometimes even resulting in hospitalization.

In Adolescence Psychiatry, patients are frequently hospitalized for the following conditions: anorexia, first psychotic episode, anxiety disorder, depressive disorder, conversion disorder, manic-depressive disorder, obsessive-compulsive disorder, and behavioral changes.

Adolescence is characterized by physical and emotional changes that are of great importance for identity construction. It is at this stage that strategies for managing stressful situations are consolidated and conditional.

Muscle relaxation technique is an intervention conducted by specialist nurses in mental health and Psychiatry that is useful to encourage and enable relaxation aiming to reduce undesirable signs and symptoms, such as pain, muscle tension, and anxiety.

The objectives of this study were to:

- report the most frequent conditions in the Adolescence Psychiatry Inpatient Department;
- demonstrate the importance and effectiveness of muscle relaxation technique in Adolescence Psychiatry Inpatient Department users;
- demonstrate the feasibility of the criteria used in the application of this technique;
- document some of the skills in mental health and Psychiatry of specialist nurses.

Methodology: Qualitative and quantitative assessment of the effects of relaxation. Quantitative assessment consisted of the evaluation of vital signs before and after relaxation, and qualitative assessment consisted of the evaluation of patients' reports about their emotions and feelings before and after relaxation.

Results: The analysis showed that patients were less anxious, the evolution of the disease was favorable, hospitalization was better accepted, and there was greater availability for psychotherapeutic activities when using the muscle relaxation technique.

Conclusion: The benefits of the application of relaxation techniques by specialist nurses in mental health and Psychiatry in Adolescence Psychiatry patients are clear. This benefit is observed through a decrease in the administration of rescue medication, favorable disease evolution, and greater acceptance of hospitalization.

Keywords: adolescence; brain; mental health, relaxation

ENF_11
NURSING INTERVENTIONS FOR CHILDREN WITH DOWN SYNDROME AND FAMILIES: SURGICAL CORRECTION OF CLEFT LIP AND PALATE

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Introduction: An estimated 9.8 million children worldwide are born with congenital disabilities. Around 91% of these correspond to Down syndrome, with approximately one in every 700 presenting congenital craniofacial anomaly (cleft lip and palate), with associated complex medical and nursing care. Herein is presented a family-centered case of a child with Down syndrome who underwent surgical correction of cleft palate, highlighting the specialized Child and Pediatric Health Nursing care required in these cases.

Clinical case: A four-month-old female child diagnosed with complete unilateral cleft lip and palate was proposed for unilateral cheiloplasty and McComb rhinoplasty. Regarding personal history, she had trisomy 21, interatrial communication of predominantly cardiac chambers, obstructive sleep apnea syndrome, hypoventilation, poor weight progression, and hypotonia. As family history, trisomy 21 and cleft lip and palate were reported. The patient required interventions at the neuromuscular, cardiorespiratory, gastrointestinal, tegumentary, regulatory, and infant developmental levels. Parental need for intervention was mainly in domains related to the transition of complex parenthood.

Discussion: Specialist nurses in Child and Pediatric Health Nursing should have the infant as a priority focus of attention while simultaneously attending to the needs and training of families throughout the perioperative period. Thus, in addition to caring for children in perioperative setting, these specialists should also focus their interventions on parental care centered on the meanings attributed to illness, knowledge, self-efficacy, and abilities concerning developmental needs and special and complex health needs, aiming to empower families and ensure the quality and safety of care after hospital discharge and recognizing the priorities, expectations, and resources of each family, towards a healthy complex parenthood transition.

Keywords: complex parenthood; infant; nursing; perioperative; Trisomy 21

ENF_12
PEDIATRIC BRAIN DEATH – IMPLICATIONS FOR NURSING PRACTICE

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The concept of brain death in Pediatrics is always highly controversial. The criteria for defining brain death are applied from the first year of life, albeit with some limitations. The Portuguese legislation provides support to these cases in the twelfth article of Law no. 12/93, April 22.

In children younger than one year, clinical criteria are difficult to apply. Therefore, between two months and one year, the minimum gap between observations is 24 hours, and two isoelectric electroencephalograms are performed with the same gap. Between the ages of seven days and two months, the gap between exams and electroencephalograms is less than 48 hours. In children younger than 7 days or with less than 38 days of gestation, the criteria for brain death do not apply.

The diagnosis of brain death requires demonstration of the cessation of brainstem functions and irreversibility. If confirmation of these premises is assured, diagnostic criteria should be applied, and the necessary conditions for execution of each exam should be provided. The current presentation will discuss this last topic, as it refers to the setting where the Nursing team has the greatest involvement. The discussion will focus on ethical and legal considerations regarding the diagnosis of brain death and will be based on a literature review and critical reflection.

ENF_13

QUALITY STANDARDS AND INDICATORS SENSITIVE TO PERIOPERATIVE NURSING CARE

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Introduction: The continuous improvement of health care is the gold standard of Nursing practice. OE has defined quality standards for the definition of a strategy aimed at improving the quality of professional practice. Its implementation involves the management of the care provided and the consequent planning of what is necessary for its operationalization through the construction of indicators sensitive to Nursing care. These should ensure that the quality of Nursing care is based on a coherent system of values that includes the client, the institution, and the nurse.

Objectives: To define and document quality, monitoring, and Nursing care-sensitive indicators in the perioperative setting of the clinical practice.

Methodology: Literature review in reference databases.

Results/Discussion: A total of 102 articles were initially identified, of which six were included in the analysis after application of the predefined inclusion criteria.

The following indicators related to Nursing practice were extracted from the analysis: patient satisfaction with the care provided, surgery cancellation rate, effectiveness of the preoperative Nursing consultation, adverse event reporting, pain reduction, anxiety reduction, non-conformity reporting by healthcare professionals, hand washing rate, healthcare-associated infection rate, morbidity/mortality rate, operational efficiency, and financial effectiveness.

Conclusion: Quality indicators offer the possibility of a quick and simple overview of the quality of a service or product. Quality in health should be perceived as the achievement of the greatest benefits with the lowest risks and costs for users.

This study found that, despite the growing commitment and concern of perioperative nurses, documentation of quality standards and indicators in this specific area remains scarce. Reorganization of resources and development of structures to ensure a quality professional Nursing practice are required, as well as the implementation of systematic care assessment.

Ensuring the quality of Nursing care and the safety of patients should be the main concern of the nurse manager.

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ENF_14
DROWNING IN PEDIATRIC AGE

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Abstract: According to the World Health Organization, drowning is defined as the secondary respiratory difficulty during the process of aspiration of liquid by submersion or immersion. It is one of the leading causes of death in children and young people globally and the second most common cause of brain damage accidents in individuals under the age of 15 years. Drowning is most common in the summer months.

The aim of this work is to remind that drowning can have several causes and be classified in degrees according to its severity and that earlier implementation of rescue measures and acknowledgment of drowning circumstances allow more effective assistance to the victim. Additionally, it intends to clarify some outdated concepts.

The research was carried out using scientific literature. The main results obtained indicate that drowning is due to inadequate supervision in swimming pools, bathtubs, and other liquids in children under the age of five years. In adolescents, the incidence is higher in males and is usually associated with risky behaviors.

Efforts to significantly reduce morbidity and mortality should therefore focus on prevention, through the dissemination of information and awareness campaigns among families, children, adolescents, and the community overall.

ENF_15
NURSING CARE OF CHILDREN ADMITTED TO PEDIATRIC INTENSIVE CARE WITH MULTISYSTEM INFLAMMATORY SYNDROME IN CHILDREN (MIS-C): A SINGLE-CENTRE RETROSPECTIVE ANALYSIS

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Introduction: The new coronavirus pandemic brought global challenges to the provision of healthcare, affecting adults and elderly people with need for intensive care admission and causing high mortality. The same was not observed in children. However, an inflammatory syndrome associated with coronavirus exposure affected children and prompted the need for intensive care admission. The aim of this study was to assess the characteristics, treatments, outcomes, and nursing interventions in children with coronavirus-associated multisystem inflammatory syndrome in children (MIS-C) admitted to a level III Pediatric Intensive Care Unit (PICU).

Methodology: Retrospective review of episodes of children admitted to PICU diagnosed with MIS-C between February 2020 and February 2022. Demographic and clinical data were collected.

Results: During the period in analysis, 23 admissions of children with MIS-C diagnosis were reported, 17 of whom were male (74%) and six female (26%), with a median (interquartile range [IQR]) age of 12 (8-15) years and average body mass index of 20.4 Kg. Most children had been transferred from other hospitals (66.7%), with the remaining having been transferred from the Pediatric Emergency Department and Pediatric Ward.

The main comorbidities reported in this cohort were allergic rhinitis and excess weight or obesity. Fifteen children (65%) required inotrope support, with a median (IQR) duration of 3 (3-3.8) days, and one child required extracorporeal membrane oxygenation. Ten children (43%) required oxygen therapy, with only one invasively ventilated. Electrocardiographic changes were reported in 43% of cases. The median (IQR) PICU length of stay (LOS) was 3 (2-4) days and the median (IQR) hospital LOS was 6 (5-8.5) days. No patient death was reported.

Nursing interventions included COVID-19 precaution measures, hemodynamic monitoring with special attention to dysrhythmias, medication administration and fluid management, and support to patients and families with a special focus on uncertainty-related stress due to the new syndrome. All children discharged had follow-up by Pediatric Cardiology and Pediatric Infectious Diseases specialties.

Conclusion: Coronavirus-associated MIS-C was found to have complex presentations requiring PICU admission. The novelty of this syndrome required prompt response to the presenting ill child, with great uncertainty in treatment efficacy and outcomes, challenging the healthcare team to be updated on advances in the care of children with MIS-C and nurses to pay extra care to hemodynamic monitoring of cardiac disfunction and manage the expectations and anxiety of children and parents facing this new condition.

ENF_16
THE IMPORTANCE OF BABY MOVEMENTS

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Introduction: Counting baby movements during pregnancy is a simple way for parents to access the baby's well-being at home. As the method used for this counting varies according to country, this study intends to present different methods of counting baby movements during pregnancy.

Methodology: A systematic literature review of pregnancy studies over the last 15 years and public national guidelines from different countries was conducted on PubMed.

Results: Fetal movement counting is an accepted and simple method for parents to assess the baby's well-being at home. The Portuguese guideline for low-risk pregnancy assessment recommends counting 10 baby movements in 12 hours.

Conclusion: Midwives have a privileged role during pregnancy in educating parents about the importance of counting baby movements and how to do it.

ENF_17
SUBCUTANEOUS ROUTE AND HYPODERMOCLYSIS: A CHALLENGE IN PALLIATIVE CARE

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Introduction: Symptomatic control is a cornerstone of pediatric palliative care. Children with complex chronic conditions experience several hospitalizations during life, and the need to receive therapy is a constant reality. The oral route is preferred for therapy administration, but when not available, the subcutaneous route is assumed as a safe, fast, and effective alternative that provides suffering relief and significant quality of life improvement, with the advantage that it can be used at home, provided there is a trained caregiver.

Objectives: This study aims to propose a pathway for implementation of subcutaneous therapy and hypodermoclysis in Pediatrics and define a protocol for implementation of the subcutaneous route and hypodermoclysis in Pediatrics.

Methodology: Bibliographic search in Cinhal, PubMed, and Scielo databases; analysis of adult palliative care procedures; development of a training partnership between the adult and pediatric palliative care teams of Centro Hospitalar Universitário do Porto; reflection and development of materials; dissemination and training of professionals.

Results: The intended results include the development of protocols for hypodermoclysis and subcutaneous therapy in Pediatrics and for dilution of drugs in palliative care; theoretical training of medical and nursing teams; and sensitization and motivation initiatives for this intervention.

When caring for children and families with complex chronic conditions, some situations cannot be changed, and accepting this helps to envisage an ocean of opportunities where health professionals can make a difference. In a university hospital center, teamwork allows the creation of specialized palliative care teams supporting each other to respond to situations of great complexity from birth to adulthood.

ENF_18
ANTENATAL EDUCATION AND DEPRESSION

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Introduction: Pregnancy is one of the most vulnerable events in the life of a couple, marked by profound physical and psychological changes and thus representing a critical period for women's mental health. The aim of this study was to investigate the association between attendance of prenatal classes and depression.

Methodology: A systematic literature review was performed on PubMed using the terms "prenatal education"[Mesh] and "depressive disorder"[Mesh].

Results: The results retrieved suggested that physical activity is linked to low depression symptoms and that mindfulness training and antenatal classes may lead to benefits in maternal mental health.

Conclusion: Further studies are required to understand the relationship between antenatal classes and depression.

ENF_19
VIRTUAL REALITY AND PAIN CONTROL IN CHILDREN

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Introduction: Children who experience pain are afraid of procedures involving needles, resisting these approaches. Non-pharmacological methods are key alternatives for pain relief, as they are secure, cheap, and non-invasive (Usterner & Ayyilodiz, 2021). One of the methods used in pediatric age is distraction, of which virtual reality (VR) is an example (Gerçeker et al., 2018). The aim of this study was to understand if the use of VR in procedures involving needles was effective in controlling pain in children aged four to twelve years old.

Methodology: Through an evidence-based approach, pain control in children in the setting of procedures involving needles was identified as a clinical problem. The following starting question was thus elaborated based on PICO methodology: "Is it appropriate to use VR to control pain in children during needle procedures?" This was followed by the identification of MeSH descriptors and inclusion criteria for study search. The search was conducted in Medline (PubMed), CINHAL (EBSCO), and Scielo (EBSCO) databases during April 13, 2022, and inclusion criteria comprised studies published in the last five years, written in English, and with children aged four to twelve years old as target population. The presence of pathological conditions was an exclusion criterion.

Results: After eliminating the 10 duplicate studies retrieved in the initial literature search, a total of eight studies were considered in the analysis. From these eight studies, six had positive results concerning the use of VR in pain and anxiety control in children aged 4 to 12 years. The study by Erdogan & Ozdemir (2021) compared VR with other methods of distraction, showing similar effectiveness among them. In addition, all studies showed the benefits of implementing VR versus not using this technique by reporting lower pain scores with the first.

Conclusion: This study showed that VR represents an effective non-pharmacological strategy for pain control in needle procedures in children aged 4 to 12 years old.

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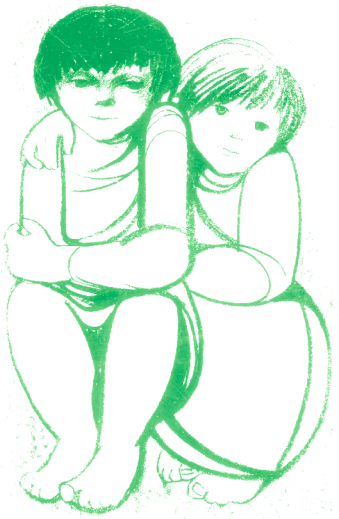

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CÉREBRO E SAÚDE: PROTEGER O CÉREBRO E CUIDAR DA SAÚDE

BRAIN HEALTH PROTECTION: HOW TO IMPROVE IT?

Chair
Alberto Caldas Afonso

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