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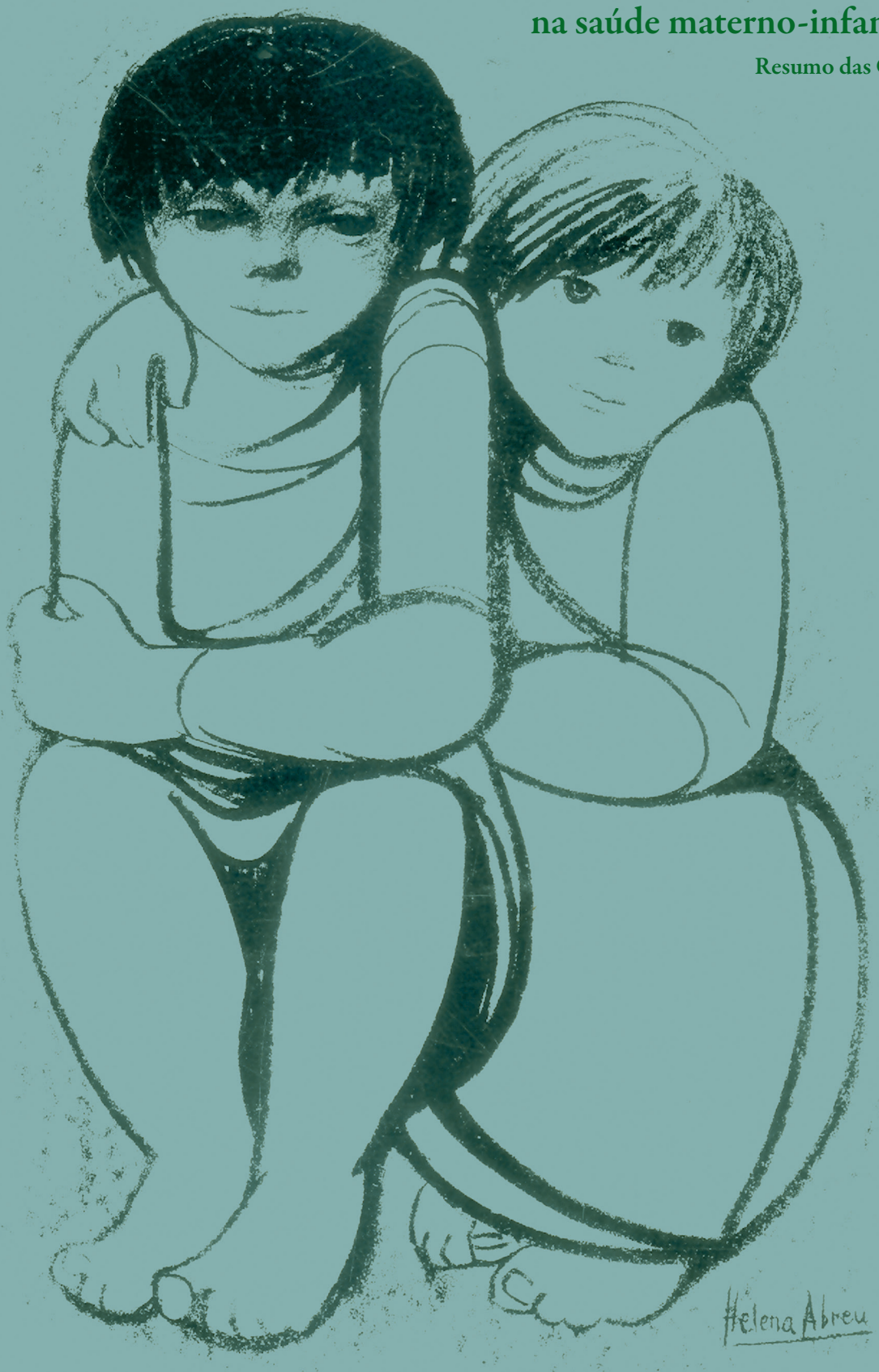
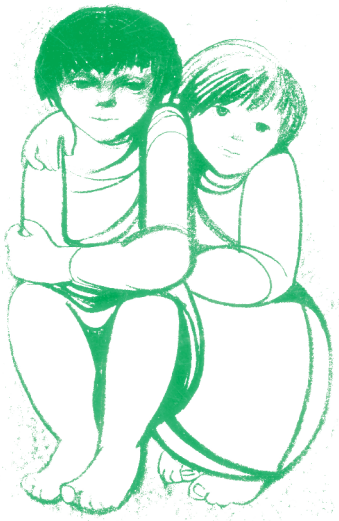
Resumo das Comunicações





PEDIATRIA

COMUNICAÇÕES ORAIS

(24562)

NARCOLEPSY IN PEDIATRIC AGE – A CROSS-SECTIONAL STUDY

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Introduction: Narcolepsy is a rare sleep disorder characterized by excessive daytime sleepiness (EDS) that manifests before the age of 18 in more than half of cases. Cataplexy (sudden loss of muscle tone usually precipitated by emotions), the key symptom of type 1 narcolepsy, may be absent at the beginning. Diagnosis delay is common in pediatric age due to lack of awareness, comorbidities contributing to EDS and atypical presentations.

Methods: A cross-sectional study involving narcolepsy patients followed in a Pediatric Sleep Center between January/2018 and December/2023–aimed to characterize this disorder in a pediatric population and analyze its clinical and diagnosis challenges.

Results: Thirty patients were included (53% females), 29 with narcolepsy type 1 and 1 type 2. Median age of symptoms onset was 8 years (3-13). Referral to specialized consultation and diagnosis happened on average 3 (1-10) and 4 years (1-12) after the first symptom, respectively. EDS was the first symptom in 28 (93%), with "sleep attacks" in 19. Cataplexy was present from the beginning in 13 (43%), without EDS in 2. In 11 (37%) it occurred on average 3 years (1-10) after EDS. In 4 cases, muscle tone loss wasn't triggered by emotions (2 with sudden falls) and there was facial involvement in 11 (1 with ptosis). Sleep paralysis was reported by 6 (20%) and vivid dreams/hypnagogic or hypnopompic hallucinations by 18 (60%). Decrease in school performance was present in 17 (57%), behavioral changes in 23 (77%), and increased appetite/weight gain in 19 (63%). Psychiatric conditions were present in 8 (27%) (4 under psychotropic medication), 8 reported restless sleep (2 with restless leg syndrome), and 15 snoring. Polysomnography revealed periodic limb movements (PLMS) ≥5/hour in 2 and obstructive sleep apnea (OSA) in 6.

Conclusions: Median age of presentation was lower than reported (peak at 15 years old). Typical symptoms like "sleep attacks" and cataplexy were absent in one-third and over half of the cases. Cataplexy not triggered by emotions or involving facial muscles can occur, as well as comorbidities like OSA, PLMS and psychiatric conditions, which can delay the diagnosis. Yet, the diagnostic process was shorter than previously reported (6 to 10 years). It's crucial to thoroughly investigate symptoms suggestive of narcolepsy in any patient with EDS or atypical cataplexy such as sudden falls or ptosis.

Palavras-chave : narcolepsy

(24578)

NAVIGATING PARENTHOOD IN THE AGE OF SOCIAL MEDIA: UNDERSTANDING THE ROLE OF ONLINE PLATFORMS IN HEALTH INFORMATION SEEKING

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Introduction: In contemporary society, the dynamic landscape of social media has profoundly influenced how parents seek and engage with health information. The exponential growth of online platforms has transformed the way individuals access, share, and interpret health- related content. This phenomenon is particularly salient within the realm of parenting, where caregivers often turn to social media for guidance on various health-related matters concerning their children. This work endeavors to delve into the multifaceted relationship between parents and social media as a conduit for health information.

Methodology: An anonymous and individual questionnaire was prepared, consisting of 23 questions, aimed at parents of children observed in the context of child and youth health and open consultation at a Health Center from March to April 2024. The data were subsequently recorded in an anonymized database using Microsoft Office Excel® and statistical analysis was carried out using SPSS version 28.0 and Microsoft Office Excel ®.

Results: We received 72 responses, with the majority of participants being female (77.8%), married, holding a degree and 50% aged between 31-40. We also obtained an equal number of responses from residents in urban and rural areas. Regarding the use of social networks/websites to search for information, 58.3% of respondents answered affirmatively to this question, with Internet pages being the most used source, less than once a week by the majority, but 26.2% use this feature several times a week. The areas that parents most seek information from in these places are nutrition, general pediatrics, development and sleep. When there is a discrepancy between the information provided by the family doctor or pediatrician and the information from these sources, 95.2% report following the instructions of their family doctor or pediatrician. 42.9% of parents who use these sources say it is because they feel the need for more information and 35.7% say they need to use these resources because they do not feel comfortable contacting their family doctor/pediatrician to clarify any doubts.

Discussion: While the use of social media to gather information about children can seem convenient and reassuring for parents, it carries significant risks that cannot be overlooked. Therefore, it is imperative for parents to approach online health information cautiously and always verify findings with reputable medical sources or healthcare professionals.

Palavras-chave: social media, parents

(24587)

IS THE ARTIFICIAL INTELLIGENCE THE FUTURE OF CYTOGENETICS?

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Cytogenetics enables the identification of chromosomal alterations associated with genetic disorders. Most common alterations in pre/postnatal diagnosis are numerical and may be detected using G-banded karyotyping or high-resolution chromosomal microarrays. This study aims to highlight potential advantages/disadvantages of artificial intelligence (AI) in detecting chromosomal rearrangements and the elaboration of cytogenetic reports.

An english literature review from 2020 to 2024 was performed to identify studies concerning the state-of-art of AI in cytogenetics. Studies that demonstrate the application of AI in cytogenetic analysis using search key-words “artificial intelligence”, “machine learning”, “cytogenetics” and “karyotype” in PubMed, Google Scholar and Scopus, were included.

Analysis of databases revealed some key ideas. AI-generated methods offer remarkable advantages, including significant time reduction, possibly up to 40%, compared to traditional karyotype analysis. AI methods are diverse, such as software able of transforming poor quality karyograms into high quality digital images, or an object detection based system with 99.32% chromosome detection probability and about 75.71% chromosome abnormality accuracy. However, challenges remain, particularly in improving diagnostic accuracy, sensitivity and specificity. There is also a clear need for larger and standardized chromosomal databases. Limitations also include the potential for systematic error and bias in the interpretation of results as well as ethical considerations regarding privacy and security of patient data.

Integrating AI into cytogenetics presents both opportunities and challenges. In prenatal and postnatal diagnostics, AI appears to have the potential to speed up chromosome counting and abnormality detection, which could lead to faster responses. Despite progress, challenges remain in AI, particularly in recognizing distorted, overlapped or blurred chromosomes, in a metaphase.

Ensuring high quality, representative and unbiased data, the regular evaluation of AI systems is essential to train AI effectively and maintain diagnostic accuracy. Increased regulation is needed to address ethical considerations, particularly data privacy. While AI seems promising, it may never substitute manual diagnosis, due to limitations in databases, lack of validation, standardization issues and the inherent risk of systematic errors.

Palavras-chave: Artificial Intelligence, Machine Learning, Cytogenetics, Karyotype

(24597)

CHILDHOOD BILATERAL BASAL GANGLIA DISORDERS IN SUSPECTED MITOCHONDRIAL PATIENTS – MRI PATTERNS AND DIAGNOSIS

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Background: Basal ganglia involvement is a red flag for neurometabolic diseases, more commonly due to mitochondrial disorders (MD). It can be clustered by MRI pattern: T2-weighted hyperintensities in the putamen; or globus pallidus; or globus pallidus, brainstem, and cerebellum with diffusion restriction; or less commonly as T1-weighted hyperintensities.

Methods: Retrospective study of brain MRI findings of paediatric patients with suspected mitochondrial disorder with basal ganglia involvement. All patients had extensive genetic work-up: mitochondrial (mtDNA) and nuclear DNA (nDNA).

Results: A total of 21 patients (57% female) were included with a median age [IQR] of first symptoms at 9 [0-27] months. Age at first brain MRI was 6 years [1-14]. Final diagnosis was mtDNA-MD in 11 patients, nDNA-MD (EARS2 and POLG) in 2, pyruvate dehydrogenase deficiency (PDH) in 1, biotin-thiamine-responsive basal ganglia disease (BTBGD) in 1, GABA transaminase deficiency (GABA-T) in 1 and 1 with genetic encephalopathy (STXBP1). Four patients remain undiagnosed.

T2 hyperintensities were observed in: i) putamen in 5 patients (4 mtDNA-MD, 1 PDH); ii) globus pallidus in 5 (BTBGD, GABA-T, STXBP1, 2 undiagnosed); iii) globus pallidus, brainstem and cerebellum involvement in 5 (2 nDNA-MD, 2 mtDNA-MD, 1 undiagnosed). T1 hyperintensities was only observed in one patient with mtDNA-MD. Five patients could not fit any of these patterns particularly: 2 Leigh patients with T2 hyperintensities in the substantia nigra; a POLG patient with predominant white matter involvement and diffusion restriction in the lentiform nucleus.

Discussion: Recognition of neuroimaging patterns in the basal ganglia can aid diagnosis. Bilateral basal ganglia involvement is a common finding in MD, although not pathognomonic nor with a specific pattern. Isolated globus pallidus involvement appears to be more common in non-MD patients.

Other genetic causes are to be excluded and treatable (or reversible) causes are to be considered simultaneously at first approach.

Palavras-chave: Basal Ganglia, Mitochondrial Disorders

(24620)

PERFUSION INDEX AS AN EARLY PREDICTOR OF AORTIC COARCTATION

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Introduction: Perfusion index (Pi) monitoring is a non-invasive method of assessing real-time peripheral perfusion, derived from and displayed by the pulse oximeter.Pulse oximetry has moderate sensitivity for detecting obstructive lesions of the left heart, especially coarctation of the aorta (CoA), highlighting the need for improved screening methods for these conditions.

Clinical case series: We present two full-term newborns (NB) who underwent monitored pregnancies.The first NB underwent serial fetal echocardiograms due to suspected CoA, based on right heart chambers predominance,a “gothic” aortic arch with a narrow isthmus, and turbulent flow.Admitted to the Neonatal Intensive Care Unit (NICU) since birth for surveillance, he remained asymptomatic in the first days, with echocardiograms showing turbulence after subclavian emergence (20mmHg gradient), without CoA flow, and ductal closure on day 3.At this time, he presented a post-ductal PI <0.7 and a pre and post-ductal PI differential >50%.On day 5, a systolic murmur was detected, along with decreased femoral pulse amplitude and pressure gradient between upper and lower limbs, without a peripheral oxygen saturation (SpO2) differential.An echocardiogram confirmed CoA on this date.The second NB had a prenatal diagnosis of subaortic and muscular ventricular septal defect (VSD) and ductus venosus agenesis.Admitted to the NICU after birth due to grunting and respiratory distress, he presented a systolic murmur on day 1, and an echocardiogram confirmed VSD, showing a “gothic” aortic arch with flow acceleration in the isthmus region (15mmHg gradient). On day 2, he had an increase in oxygen needs and started noninvasive ventilation, presenting a post-ductal PI <0.7 and a pre and post-ductal PI differential >50%.At this time, no pressure gradient between upper and lower limbs or SpO2 differential existed, but a 2D echocardiogram confirmed aortic coarctation.Both NB started prostaglandin E1 infusion and underwent corrective surgery.

Conclusion: In both cases,the low post-ductal PI value with a pre and post-ductal differential >50% emerged at an earlier stage compared to clinical indicators such as decreased femoral pulse amplitude or pressure gradient between upper and lower extremities. Although further studies are needed and it is not yet recommended as a screening parameter for congenital heart defects,monitoring the microcirculation through PI may hold potential for earlier diagnosis in cases of suspected CoA.

Palavras-chave: Aortic coarctation, Capillary perfusion index, Congenital heart defects screening

(24617)

PERIVENTRICULAR-INTRAVENTRICULAR HEMORRHAGE AND PERIVENTRICULAR LEUKOMALACIA IN VERY LOW BIRTH WEIGHT NEWBORNS

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Introduction: Peri-intraventricular hemorrhage (PIVH) and periventricular leukomalacia (PVL) are among the main neurological injuries affecting premature newborns, potentially leading to developmental sequelae. The aim of this study was to identify the frequency of PIVH and PVL and associated protective factors in neonates with gestational age <32 weeks and very low birth weight (VLBW).

Methodology: We conducted a retrospective observational study of all newborns with gestational age less than 32 weeks and very low birth weight born between January 2018 and December 2023, who were admitted to a neonatal intensive care unit. Descriptive and inferential analysis ($p<0.05$) was performed using SPSS (v.27).

Results: 176 newborns were included, 56.8% male, with a mean gestational age of 28.6 ± 1.70 weeks and a birth weight of 1080 ± 246g. PIVH developed in 43.2% and PVL in 30.1% of cases. The occurrence of PVL in newborns with PIVH occurred in 45.3% of cases. Magnesium sulfate (MgSO4) was administered for fetal neuroprotection in 60% and corticosteroid therapy for fetal lung maturation in 63.6% of cases. A lower incidence of PIVH was observed in the group of newborns who received MgSO4 compared to those who did not (31% vs 48% PIVH; $p=0.017$). Newborns undergoing corticosteroid therapy for fetal lung maturation had significantly fewer cases of PIVH compared to those who did not receive corticosteroid therapy ($p<0.001$). Regarding PVL, significantly fewer cases were recorded in newborns receiving corticosteroid therapy ($p=0.020$). No statistically significant differences were found regarding sex, type of delivery, twinning, or maternal age.

Discussion: This study supports that prenatal exposure to MgSO4, as well as corticosteroid therapy for fetal lung maturation, had a protective effect on the development of PIVH. Additionally, corticosteroid therapy appears to also have a protective role in the occurrence of PVL, unlike MgSO4.

Palavras-chave: Periventricular-intraventricular hemorrhage, Periventricular Leukomalacia, magnesium sulfate, corticosteroids

(24633)

NUTRITIONAL IMPACT OF MODULATOR THERAPY IN CHILDREN WITH CYSTIC FIBROSIS: A TERCIARY CENTER EXPERIENCE

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Introduction: Patients with cystic fibrosis (pwCF) are at increased risk of malnutrition due to the suboptimal or absent function of the CFTR protein. This happens due to exocrine pancreatic insufficiency (EPI), intestinal inflammation and increased energy expenditure from chronic pulmonary inflammation. Modulator therapies, such as lumacaftor/ivacaftor (LUM/IVA) and elexacaftor/ivacaftor/tezacaftor (ELX/TEZ/IVA), have shown promise in improving disease outcomes. This study aims to explore the impact of modulator therapy on nutritional outcomes in pediatric pwCF: improvement of BMI and EPI; reduction in pulmonary exacerbations (Pex) and intestinal inflammation.

Methods: Observational and retrospective study analyzing clinical data from all pediatric pwCF undergoing treatment with LUM/IVA and/or ELX/TEZ/IVA over a period of 5 years. Demographic and clinical data (including genotype and PEx rates, BMI, fecal elastase and calprotectin pre/post treatment) were collected. Descriptive statistical analysis was used.

Results: The final sample comprised 19 patients, ages 8 to 17 years old (M 15; IQR 12-16), 53% female (n=10), 15 f508del homozygotes, 3 f508del heterozygotes, and 1 N1303K homozygote. 10 patients initially received LUM/IVA, with subsequent transition of 9 patients to ELX/TEZ/IVA, resulting in a total of 18 patients under that therapy.

For those on LUM/IVA, a median increase of 17,7 kg/m^2; IQR (15,8 kg -18,0 kg) to 18,45 kg; IQR (17,2 kg -20,3 kg) and a 71.0% (IQR 62.5) reduction in PEx were observed over the first year. Transition to ELX/TEZ/IVA showed a median BMI increase of 19,1 kg/m^2; IQR (18,0 kg - 21,0 kg) to 21,1 kg/m^2; IQR (18,3 kg - 22,0 kg) and a 100% (IQR 20.8) reduction in the frequency of PEx over the first year. None of the patients had improvement in fecal elastase. A median decrease in calprotectin of 422,0 ug/g (IQR 158,0 ug/g -1349,5 ug/g) to 89.0 ug/g (IQR 70,5 ug/g -161,0 ug/g) was observed with the overall therapy.

Most patients sustained this profile throughout the subsequent treatment years (up to a maximum of 3 years of follow-up). Side effects were residual and non relevant in all patients.

Discussion: Along with lung disease, malnutrition is one of the main burdens of disease in CF. Modulator therapies have effectively improved nutritional outcomes and reduced inflammation. However, EPI is irreversible. By 12 months of life, approximately 85% of the pwCF have EPI, so future studies should examine the effects of CFTR modulators when started early in life.

Palavras-chave: Modulator therapies, cystic fibrosis, IMC, nutricional

(24631)

CARDIOVASCULAR MANIFESTATIONS IN RASOPATHIES

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BACKGROUND AND AIM: RASopathies are a group of multisystemic genetic disorders affecting the Ras/MAPK signaling pathway including frequently cardiac phenotype. Our aim is to present the spectrum of cardiovascular manifestations, molecular genetic testing and management of a pediatric population in a single tertiary centre.

METHODS: Retrospective review of clinical charts of patients diagnosed with RASopathies followed in Pediatric Cardiology and Genetic Department for a 10 year-period (2014-2024). Data analysis included: demographics, prenatal diagnosis, molecular genetics, cardiovascular presentation and management.

RESULTS: The study included 25 patients (60% females) with RASopathies, medium age at diagnosis 4,5 years (ranging from pre-natal diagnosis to 13 years), medium follow-up 48 months. Noonan syndrome (NS) represented 88% (22/25) of the subjects, with heterozygous variants in six different genes: *PTPN11* (9), *SOS1*(5), *RAF1*(3), *LZTR1*(1), *MAPK1* (1) and *RIT1*(1); 2 patients with clinical diagnosis of NS; three patients presented Cardiofaciocutaneous (CFC) Syndrome (heterozygous pathogenic variant in *BRAF).* Pre-natal ultrasound findings were polyhydramnios (3), increased nuchal translucency (2), Dandy-Walker malformation (1). The latter had NS family history and had prenatal diagnosis. In 5 cases NS was confirmed in one of the parents after the child's diagnosis.

Major cardiac manifestations included pulmonary valve (PV) stenosis (41%) followed by hypertrophic cardiomyopathy (HCM) (27%), obstructive in 5 cases, atrial septal defect (ASD) (17%) and left ventricle hypertrophy (17%). HCM was associated with variants in *RAF1*(3), *PTPN11*(2) and *LZTR1*(1). During follow-up, 3 patients underwent cardiac surgery (one had multiple surgeries), namely pulmonary valvuloplasty, Morrow myomectomy and ASD closure, and one ICD implantation (age: 13 years). One patient diagnosed by day 7 of life with a deleterious *RAF1* variant and severe obstructive HCM died at 80 days.

CONCLUSIONS: RASopathies have an important prevalence of cardiac disease with a spectrum that extends from mild to severe and potentially fatal outcome. In pediatric population the finding of HCM associated with additional cardiac lesions (PS, ASD, mitral valve dysplasia) or particular cardiac phenotypes (e.g. biventricular hypertrophy) should raise suspicion of NS. Further investigation is needed to a more accurate picture of the cardiologic landscape in RASopathies and the development of new therapeutics for these debilitating disorders.

Palavras-chave : RASopathy, Genetics, Cardiology

(24634)

MAYER-ROKITANSKY-KÜSTER-HAUSER SYNDROME: THE EXPERIENCE OF A TERTIARY CENTRE

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Backgroud: Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, also known as Müllerian aplasia, is a congenital disorder characterized by agenesis of the uterus and upper part of the vagina, in females with normal ovarian function. It has an estimated prevalence of 1 in 5000 female live births and represents an important cause of primary amenorrhea, which frequently prompts referral to Paediatric Endocrinology. In recent years, much progress has been made in investigating genetic causes and the possibility of uterus transplantation. The goal of this paper was to characterize the cases of MRKH syndrome in a tertiary centre.

Methods: We describe five cases of MRKH syndrome with follow-up at our hospital centre, over a period of 17 years (2007-2024).

Results: The median age at diagnosis was 15 years old (min. 10 – max. 16) and the most common presentation was primary amenorrhea, in four of the cases (80%). One patient (20%) was diagnosed earlier, at 10 years old, in the context of agenesis of the vaginal opening. All had appropriate pubertal development, with normal FSH, LH and estradiol levels. Karyotype analysis was performed in two patients (40%), both with 46,XX. Pelvic imaging exams (ultrasound and magnetic resonance) showed complete uterine agenesis in two patients (40%) and, in the remaining three (60%), the presence of a residual uterus. Regarding vaginal evaluation, three patients (60%) had vaginal agenesis, and one (20%), the most recently diagnosed, still awaits gynecological evaluation. Of the three patients with vaginal agenesis, two underwent surgical vaginoplasty with bowel graft (at ages 16 and 17) and one chose to postpone this decision. All patients were studied for concurrent extragenital malformations, of which two (40%) had nephro-urological anomalies (one patient with unilateral renal agenesis and one with duplicated collecting system). In the remaining three (60%), no extragenital anomalies were identified. Four of the five cases described (80%) have already reached adulthood and, so far, none of them have embarked on motherhood through adoption.

Discussion and conclusion: MRKH syndrome is a complex entity with great physical and psychological impact, hence requiring a multidisciplinary approach. It is essential to consider this differential diagnosis in cases of primary amenorrhea with normal pubertal development. Uterus transplantation has been successfully performed in some countries and presents itself as a promising option for treating infertility in these women.

Palavras-chave: Mayer-Rokitansky-Küster-Hauser syndrome, Müllerian aplasia, Primary amenorrhea, Uterus transplantation

(24593)

MOHR-TRANEBJAERG SYNDROME TIMES THREE: "ADDING UP" ON A RARE GENETIC NEURODEVELOPMENTAL DISORDER

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Background: Mohr-Tranebjaerg syndrome (MTS) is a rare X-linked neurodegenerative disorder caused by mutations in the TIMM8A gene. It typically manifests in childhood with hearing loss, followed by dystonia and optic atrophy. Intellectual disability may range from mild to severe. The diagnosis is based on clinical evaluation and confirmed by genetic testing. Management primarily focuses on symptomatic treatment.

Clinical case: We present three siblings diagnosed with MTS, following the maternal uncle's diagnosis and subsequent familial genetic counselling. The eldest, aged 13, was referred to neurodevelopment consultation due to generalized learning difficulties and sensorineural hearing loss diagnosed by the age of 9. His physical examination was unremarkable but hyperkinetic behavior was evident. He had moderate intellectual developmental disorder. His 9-year-old twin brothers were referred due to attention deficit hyperactivity disorder (ADHD) suspicion and behavioral problems. Both had unilateral strabismus, macrocephaly and moderate IDD. No significant dysmorphisms were observed but hyperkinesia and language and speech impairments were present. After an ADHD diagnosis, all three brothers started methylphenidate with noticeable improvements in behavior, attention and school performance. In the twins, risperidone was also effective in reducing symptoms of aggression and impulsivity. Currently, all of them maintain multidisciplinary follow-up and benefit from speech and occupational therapies as well as special education.

Discussion: This report highlights the wide spectrum of the disease, concerning neurodevelopmental, physical and sensory impairments, which emphasizes the need of a comprehensive multidisciplinary approach of these patients.

Conclusion: We underscore the importance of always considering familial history and genetic counselling for families of affected individuals. Early identification and appropriate management strategies are crucial for optimizing outcomes.

Palavras-chave: genetics, neurodevelopment, neurodegenerative, ADHD, multidisciplinary

(24615)

THE EXPANDING ROLE OF COMPOUNDED MEDICATIONS IN A PEDIATRIC HOSPITAL (2021-2024)

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The pharmaceutical industry often fails to provide tailored medicines for pediatric patients, including appropriate dosages, compositions and pharmaceutical forms. This forces caregivers and nursing staff to manipulate oral solid medications, such as crushing tablets or opening capsules or sachets and mixing their contents with food or liquids to administer the required dose. However, this recurrent practice results in undocumented administration and uncertain dose accuracy. Research evidence significant dose deviations depending on medication characteristics and manipulation methods. Pharmacists preparing compounded medications (CM) with precise qualitative and quantitative compositions emerge as a more suitable practice. Therefore, expanding the availability of CM tailored for the pediatric population becomes imperative and Pharmaceutical Services of Santo Antonio Hospital have made efforts to expand this area.

Applying descriptive statistical analysis, quantify CM specifically designed for pediatric use added to Santo António Hospital therapeutic formulary since 2021 and identify the main reasons for their preparation.

Since 2021, 73 new CM have been introduced, with 19 in 2021, 29 in 2022, 20 in 2023 and 5 in the first few months of 2024. Before 2021 the number of new CM per year was residual. The new CM were mainly introduced as liquid preparations forms, such as suspensions (76%) and solutions (10%).Personalised dosage (78%) was the main reason for preparing CM, followed by personalised composition (11%). Tailored dosage forms and filling a therapeutic gap each accounted for 5%.

The recent focus on CM development has led to a significant increase in the number of CM available. Most of these medicines have been introduced in liquid form, which is the gold standard for pediatric use. The necessity to personalize the dosage is the main reason for preparing these medicines at Santo António Hospital, which is in line with what is described in the literature. Althoug less frequent, the customization of the pharmaceutical form (useful, for example, for patients with feeding tubes and PEG), the customization of the composition (beneficial, for example, for those on a ketogenic diet) and the resolution of therapeutic gaps, such as market shortages, are also important needs met by these medicines. Technology is expected to have a major impact on CM in the coming years. Artificial intelligence has the potential to assist in galenic development, enabling formulations with optimized composition, greater stability and longer expiration date. 3D printing of CM also has potential in pediatrics, for example, to print tablets, chewing gums and orodispersible films. In summary, CM plays a central role in pediatric therapy, ensuring quality and safety through customization of dosage, form and composition. Multidisciplinary teamwork is essential for optimal pediatric care.

Palavras-chave: compounded medications, personalized medicine, Pharmaceutical Technology, compounding Pharmacy

(24627)

MULTIPLE EPIPHYSEAL DYSPLASIA: WHEN TO CONSIDER.

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Introduction: Multiple epiphyseal dysplasia (MED) is a skeletal disorder affecting bone and cartilage in the epiphyses. Unlike many other skeletal disorders, it may not have a significant effect on the final adult stature. Both dominant and recessive types display mild symptoms such as joint pain and early-onset arthritis, with most individuals reaching normal height in adulthood. Here, we discuss four cases from three families, illustrating the diverse phenotypic expression of MED.

Clinical cases: The first case concerns an 8-year-old boy with lower limb dysmetria and suspected short lower limbs, with normal height. He also reported experiencing joint pain in his knees. Radiographic assessment revealed bilateral shortening of the proximal femoral epiphyses and bilateral bilaminar patella. Due to strong clinical suspicion, Sanger sequencing for a recurrent variant in SLC26A2 was requested, which was confirmed to be present in homozygosity, thus confirming the diagnosis of MED type 4.

The second case concerns another 8-year-old boy displaying gross motor abnormalities, coordination difficulties, and muscle pain following exertion. He has pes planus, hip hypermobility, and underwent surgery at six months for interventricular communication and pulmonary stenosis. His father also had motor issues in infancy and now experiences ongoing joint pain. Following inconclusive genetic testing for congenital myopathic syndromes, recent x-rays unveiled metatarsal and epimetaphyseal tarsal dysplasia with segmental shortening. Furthermore, an elbow x-ray performed in a post-traumatic setting revealed irregularities in the proximal epiphysis of the radius and distal humerus. An NGS panel for skeletal dysplasia was requested, revealing a paternally inherited heterozygous splicing variant in COL9A3.

The variant was classified as likely pathogenic, confirming the diagnosis of autosomal dominant MED type 3.

The final case concerns a kindred, with two brothers presenting epiphyseal dysplasia in infancy and sideroblastic anemia in adolescence. Only one of them developed short stature and iron overload. Both brothers were genetically studied in adulthood, at different times, revealing the same two heterozygous variants in the HSPA9 gene. A diagnosis of a mild form of EVEN-plus syndrome was made within this family.

Discussion/Conclusions: Together, these cases underscore the diverse clinical presentations associated with MED. It also reinforces that short stature is not a recurrent feature on this group of congenital skeletal disorders, and that syndromic forms of this disorder need to be considered.

Palavras-chave: Multiple epiphyseal dysplasia, Skeletal dysplasia, Paediatric presentations, Phenotypical variability, Genetics

(24607)

ACUTE RESPIRATORY SYNCYTIAL VIRUS BRONCHIOLITIS IN NEONATES

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Introduction: Respiratory Syncytial Virus (RSV) is a major cause of acute bronchiolitis, with significant associated morbidity, especially in the neonatal period. This study aims to determine protective factors and characterize hospitalized newborns regarding the treatment, complications, and disease progression.

Methodology: Observational, retrospective study of newborns with acute RSV bronchiolitis hospitalized in a neonatal intensive care unit between January 2019 and December 2023. Demographic data, severity measures, and clinical outcomes were analyzed, including non-invasive ventilation and invasive mechanical ventilation (IMV).

Results: 64 cases were identified, 57% male with a median age of 18 days (IQR 14-25). The majority (92%) were born at term and with direct contact with other infected infants (87%). Clinical presentation with apnea was observed in 4.6% of cases, and 7.6% presented signs of severe respiratory distress. High-flow nasal cannula therapy was initiated in 65% of cases, while 33% required non-invasive positive pressure ventilation. IMV was used in 4.7% of patients. The most frequent complication was atelectasis (14%), followed by superinfection bacterial pneumonia (4.6%). The prevalence of exclusive breastfeeding at admission was 64.6%. The mean length of hospital stay for newborns exclusively breastfed was shorter compared to formula-fed newborns (6.5 days vs. 9.2 days; $p<0.05$). Peak admissions occurred in the months of November, December, and January.

Discussion: Acute RSV bronchiolitis continues to be an important cause of hospitalization in neonatal intensive care units. A significant proportion of newborns had no other recognized risk factors besides age. The role of breastfeeding as a protective factor is highlighted, associated with shorter hospital length of stay. In this study, RSV seasonality remained unchanged and was not modified by the COVID-19 pandemic, unlike other recent studies.

Palavras-chave: bronquiolite, VSR, ventilação, neonatal

(24655)

ARTIFICIAL INTELLIGENCE APPLIED TO TYPE 1 DIABETES – EXPERIENCE OF A TERTIARY PEDIATRIC CENTER

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Background: Automated insulin delivery (AID) systems are transforming the management of type 1 diabetes mellitus (T1D). The most recent technology consists of hybrid closed-loop systems that automatically adjust insulin delivery in response to real-time sensor glucose levels. This technology aims to optimize glycemic control and improve quality of life, presenting few limitations. This study evaluated glycemic control in children and adolescents on AID in a pediatric tertiary center.

Methods: We reviewed the cases of T1D patients on AID at our center, and characterized them in terms of demographics, glycemic control based on HbA1c and Ambulatory Glucose Profile (AGP) of the previous 28 days, and acute complications (hypoglycemia and ketoacidosis).

Results: Our sample included 22 patients using the hybrid closed-loop system MiniMed TM 780G. Mean age was 10.1 years (min.3.6-max.18.4) and 59% were males. Average duration of disease was 4.6 years. The patients had been using the AID system for an average period of 20.0 months (min.2.5-max.34.5) and, at the time of the start of the system, 8 children (36%) were under 7 years of age. Total daily insulin dose was, on average, 0.84 units/kg/day (min.0.6-max.1.2), with a mean basal insulin of 39.7% and mean bolus of 60.3%. The automated mode (SmartGuard TM) was active 95.9% of the time (min.81-max.99). Mean HbA1c was 6.6% and glucose management indicator (GMI) was 6.9%. Mean glucose value was 150 mg/dL, with a mean coefficient of variation (CV) of 35.0%. Percent time in target range (70-180mg/dL) was 73.2%; percent time with high glucose values (>180mg/dL) was 19.0% and very high values (>250mg/dL) was 6.0%. Regarding low glucose levels, the percent time at level 1 hypoglycemia (<70mg/dL) was 1.9% and at level 2 hypoglycemia (<54mg/dL) was 0.2%. No episodes of ketoacidosis or severe hypoglycemia occurred since AID system placement.

Discussion/Conclusion: Mean HbA1c achieved in our sample is within the recommended target (<7%). With regards to the continuous glucose monitoring metrics, presented in the AGP report, average values of all patients were within the international established goals - GMI<7%, CV <36% and time in range >70%. We highlight the low percentage of time below range, with only 0.2% at level 2 hypoglycemia. Our sample included 8 children with AID placement under 7 years old, an age group where these systems are not yet authorized, however they proved to be effective and safe in these younger children.

(24613)

EXPLORING REFERRAL PATTERNS AND CLINICAL APPROACHES FOR FORESKIN SYMPTOMS IN BOYS: INSIGHTS FROM PEDIATRIC SURGICAL PRACTICE

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Introduction: Foreskin-related symptoms frequently prompt referrals to pediatric surgical consultations, yet a considerable portion of these cases are discharged following initial observation. In this study, our objective is to assess the reasons behind such referrals and the corresponding initial management strategies applied by pediatric surgeons during the first consultation, identifying opportunities for improving the efficacy of referrals related to foreskin symptoms.

Methods: We performed a retrospective data collection, including boys who attended their initial pediatric surgery consultation from January to December 2023. The collected data included a variety of factors, such as demographic details, history of UTIs or balanitis, the specialty of the referring doctor, and the surgeon's initial diagnosis and management. Statistical analysis was carried out using IBM SPSS®, with a predefined level of statistical significance set at $p < 0.001$.

Results: We analyzed 556 referrals of boys with foreskin symptoms, with a median age of 6 years old. Five hundred and eight (89,8%) patients were referenced by their primary care physician and waited an average of 21.14 days for the initial consultation. Topical steroids were already prescribed to 170 (30%) boys before their first appointment. Physiological phimosis was diagnosed to 146 (25,8%) patients. Diagnosis of preputial adhesions was made to 128 (22,6%) boys, 88 (15,5%) were diagnosed with a preputial ring and 29 (5,1%) with a pathological phimosis. There were 274 (48,4%) boys discharged at the first consultation, 213 (37,6%) of which were discharged without being offered any other measures. Older boys were more likely to receive a surgery proposal ($p<0,001$). Older age at referral was also positively correlated with discharge at the first appointment ($p<0,001$). Younger boys were more likely to be prescribed with topical steroids ($p<0,001$).

Conclusion: Our study highlights the significant proportion of patients discharged during their initial consultation, with a notable subset of boys discharged without the recommendation of additional therapeutic measures. A substantial portion of consultation time is allocated to boys with normal preputial development, who may not need specialized surgical care. There should be an investment for educational initiatives targeting primary referring medical specialties to optimize the allocation of healthcare resources and improve patient outcomes.

Palavras-chave: Foreskin-symptoms, Phimosis, Circumcision

(24579)

ON THIN WHEELS: PATTERNS AND SEVERITY OF E-SCOOTER ACCIDENTS IN CHILDREN

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Introduction: Electric scooters are being used worldwide as a new mean of transportation and as no drivers' license is required are very popular amongst adolescents. We aimed to assess if e-scooters pose a risk to children by assessing and describing the patterns and severity of injuries related to their use.

Methods: We performed a retrospective review of all pediatric emergency department referrals relating to e-scooter use from January 2022 to July 2023 at a level 1 trauma and tertiary center. Data including patient demographics, mechanism of injury, trauma score, diagnosis, and treatment were collected.

Results: Fifteen patients were identified in this series, of which 67% were male. Median age was 13 (± 2 years). Eleven fell due to unbalance, 3 were low speed frontal collisions and 1 was a high-speed collision. Five reported minor head traumas with no brain injury, but 80% suffered trauma to the lower limbs. There were 8 contusions and 2 fractures, one avulsion fracture of the Chaput tubercle and one fracture of the trapezium, both treated conservatively. Also, 11 reported wounds, of which 3 required sutures. There were no hospital admissions, nor need for surgery, but 27% were followed-up on consultation.

Discussion: E-scooters pose a significant risk to children and can be associated with severe injury, as they can reach velocities higher than 20km/h without mandatory protective gear. In a 19-month period, 15 pediatric patients visited the emergency department due to e-scooter accidents. Although it does not represent a high volume, it is in line with other trauma centers. Opposing to those, in this limited cohort there were no hospitalizations, surgeries or mortality to report.

Conclusion: Nonetheless, the risk e-scooters pose to the pediatric population should not be overlooked and these findings emphasize the importance of ongoing surveillance to monitor trends to shape public policy regarding the restriction of electric scooter use in children.

Palavras-chave: e-scooter; accidents; trauma; public policy;

(24551)

CONGENITAL SUBGLOTTIC CYST IN A NEWBORN – CASE REPORT

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Background: Congenital subglottic cysts represent a rare etiology of neonatal stridor, typically associated with prematurity and a history of tracheal intubation. However, they can also occur in term neonates without prior airway intervention. The subglottic region, being the narrowest segment of the pediatric airway, renders these cysts capable of causing significant respiratory compromise. Prompt recognition and appropriate management are essential to ensure a safe and adequate airway.

Clinical Case: We present the case of a newborn male, born at 37+1 weeks of gestation via induced eutocic delivery, from a mother with type 1 diabetes mellitus, and a pregnancy complicated by pre-eclampsia. Apgar score was 8/9/9 and the birth weight was 4745g (>99th percentile). Despite unremarkable events, the neonate developed respiratory distress characterized by tachypnea, biphasic stridor, and hypoxemia on the 5th day of life. Initial management with non-invasive respiratory support, oxygen, aerossolized adrenaline and systemic steroids resulted in temporary improvement. However, persistent stridor prompted further evaluation, revealing a subglottic, adherent lesion on laryngoscopy and bronchoscopy. Subsequent laryngotracheoscopy in the OR confirmed the diagnosis of subglottic cyst, and endoscopic marsupialization was performed, with histopathological examination demonstrating rare fibrin exudate. The neonate was discharged without respiratory symptoms.

Discussion: Subglottic cysts are retention cysts of subepithelial mucosal glands, which may be congenital or acquired. The non-specific symptoms may present a diagnostic challenge, so a high level of suspicion is essential. The most common symptoms are dysphonia, dysphagia, hoarseness, stridor, and dyspnoea. This condition may be misdiagnosed as laryngomalacia, which is more common, therefore diagnostic nasofibroscopy and bronchoscopy are indicated.

Conclusion: This case highlights the importance of prompt recognition and multidisciplinary approach of stridor in neonates. Diagnosis and appropriate management are crucial for ensuring optimal outcomes in affected infants.

Palavras-chave: Subglottic cyst, Stridor

(24559)

COVID-19 AND VASCULITIS: A PEDIATRIC CLINICAL CASE

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Background: Vasculitis in pediatric age is a common condition, with IgA vasculitis being the most common systemic vasculitis. It is characterized by the deposition of Immunoglobulin A (IgA) in small vessels, with palpable purpura and involvement of at least one of these systems: joint, gastrointestinal, and renal. Vasculitis may be preceded by upper respiratory tract infections, insect bites, drugs, etc. Therefore, infection by severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) can trigger a systemic inflammatory response. This inflammatory cascade can activate specific immunoglobulins, leading to the appearance of endothelial lesions and inflammation.

Case: A 12-year-old adolescent, male, previously healthy, presented to the Emergency Department (ED) with a 5-day history of skin lesions on the lower limbs. On physical examination, he exhibited a papulovesicular rash, some with honey-colored crusts, accompanied by cough. Initially diagnosed with Bullous Impetigo, he was discharged with flucloxacillin.

He returned to the ED after 72 hours due to worsening lesions (on Day 3 of antibiotic therapy). He appeared reasonably well, hemodynamically stable, with a papulovesicular rash evolving into palpable purpura, symmetrically distributed, predominantly on the lower limbs, progressing to the thighs. He also had ankle edema and difficulty walking. He continued to have cough and rhinorrhea but remained afebrile. Laboratory tests showed leukocytosis with lymphocytic predominance, and other inflammatory parameters were within normal limits. SARS-CoV-2 swab test returned positive.

Diagnosis of vasculitis related with COVID-19 was made, leading to a 9-day hospitalization. He responded well to treatment without requiring corticosteroids. Follow-up in outpatient clinic showed resolution of lesions.

Discussion/Conclusion: This case illustrates vasculitis associated with SARS-CoV-2 coinfection, with minimal respiratory symptoms. It could be IgA vasculitis, as there is palpable purpura and joint involvement, but it is not the typical age or presentation. However, the presence of endothelial injury and inflammation is undeniable.

COVID-19 has opened a new era in the practice of pediatric rheumatology since it has been associated with inflammatory complications such as vasculitis and arthritis. This is rarely reported as a complication of COVID-19, underscoring the importance of recognizing this association for prompt diagnosis and prevention of further complications. Whether COVID-19 is the reason of the vasculitis or only the trigger remains unknown.

Palavras-chave: vasculite, COVID-19, pediatrics, rheumatology

PEDIATRIA

POSTERS

(24573)

A CHALLENGING CASE OF RESTLESS LEGS SYNDROME IN A CHILD WITH TYPE 1 DIABETES MELLITUS

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Introduction: Restless legs syndrome (RLS) is a neuromotor disorder marked by an urge to move the legs, often accompanied by discomfort, typically occurring during rest in the evening or at night and relieved by movement. It can cause severe sleep disruption with insomnia and restless sleep. Risk factors include family history, iron deficiency, and certain medical conditions. Treatment involves lifestyle adjustments and iron supplementation if ferritin is <50ng/mL.

Case report: A 6-year-old male (now 11), recently diagnosed with type 1 diabetes mellitus (DM) and managed with an insulin pump, began having trouble falling asleep, leg pains described as "spikes on his legs" at bedtime that alleviated by leg massaging or rubbing, and restless sleep with rhythmic foot movement. Since the DM diagnosis, sleep time also became a stress factor due to glycemic control and one of his parents always slept with him. His mother was diagnosed with "growing pains" as a child and his polysomnography revealed 10 periodic limb movements per hour. Oral iron and vitamin D supplementation were started due to low ferritin (23 ng/mL) and vitamin D levels, with partial improvement. Follow-up revealed persistence of symptoms (specially during higher glycemic levels) and ferritin levels <50 ng/mL (min 32, max 48), prompting the maintenance of oral iron for periods of 6 months and the addition of gabapentin, with no success. Intravenous iron administration resulted in significant clinical improvement and resolution of insomnia, which also coincided with better glycemic controls. Currently, the patient is successfully tapering gabapentin.

Conclusions: RLS is an underdiagnosed cause of insomnia and often mistaken for growing pains. While RLS is a common sleep disorder associated with diabetes in adults, this association is still poorly understood in children. Further research is needed to explore the relationship between glycemic levels and the worsening of RLS symptoms, as reported by these parents. Sleep deprivation and stress/anxiety can trigger and exacerbate symptoms, potentially contributing to treatment resistance in this case. While oral iron supplementation is usually effective, intravenous iron or alternative pharmacological treatments may be necessary in refractory cases. This case highlights the importance of recognizing RLS, as well as the challenges faced, particularly with resistance to first-line treatment. Exploring available therapeutic options is crucial to effectively manage this condition and improve sleep quality.

Palavras-chave: Restless Legs Syndrome, Type 1 Diabetes Mellitus, Sleep disorders

(24596)

A RARE CASE OF NEONATAL DERMATOPHYTOSIS CAUSED BY TRICHOPHYTON RUBRUM

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Background: Dermatophytes are fungi that cause infections of keratin rich areas, such as the skin, hair and nails. Tinea corporis is a type of dermatophytosis, specifically affecting the epidermal layer of the skin in exposed areas like the trunk; the most frequent causal agent for all ages is Tricophyton rubrum.

In the neonate, noncandidal fungal infections are uncommon, but can result in high mortality and morbidity. Incidence of neonatal fungal infections appears to be increasing worldwide, with preterm infants being the most affected age group.

Clinical case: A preterm neonate (born at 26 weeks + 6 days) with a birth weight of 970 grams, inpatient at the neonatal intensive care unit, presented at day 25 of life with annular plaques with peripheral erythema and scaling, and a central clearing, located in the face and dorsal region. She was otherwise systemically well, and had been inpatient at the neonatal intensive care unit since birth. The mother had similar lesions in her body.

After evaluation by a dermatology specialist, skin scrapings were collected for potassium hydroxide microscopy, and empirical treatment with ketoconazole was started. Tricophyton rubrum was later identified in the fungal culture, confirming the diagnosis of tinea corporis by this agent. Complete resolution of the rash was observed within 1 month of treatment.

Discussion: To date, only 5 cases of tinea corporis by Tricophyton rubrum have been reported in neonates. In this population, fungal infections can present unique challenges due to their delicate and developing immune systems and skin barrier. Moreover, Tricophyton rubrum infections can manifest differently and may require specialized care and attention.

One aspect to consider is the mode of transmission. Neonates can acquire Trichophyton rubrum infection through contact with infected individuals (like caregivers) or contaminated objects.

Diagnosing Trichopyton rubrum infections in neonates also requires a careful evaluation. Symptoms like redness, scaling or rash on the skin can be indicative, but due to similarities with other dermatological conditions, a definitive diagnosis often requires laboratory testing, such as microscopic examination of skin scrapings, or fungal cultures.

Conclusion: In summary, while Trichophytum rubrum infections are more commonly found in adults, they can also affect neonates, requiring specific considerations in terms of diagnosis, treatment and prevention of transmission.

Palavras-chave: neonatology, pediatrics, dermatophytosis, preterm, tinea corporis

(24585)

ASTHMA PREDICTIVE INDEX IN A CASE SERIES OF HOSPITALIZED RECURRENT WHEEZING EXACERBATIONS

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Introduction: The risk of a child with recurrent wheezing (RW) being diagnosed with asthma at the age of 6 is four to seven times greater in the presence of certain risk factors (RF) until 3 years old. It can be calculated using the asthma predictive index (API), widely tested and used in longitudinal studies.

Aim: Predict the diagnoses of asthma in a case series, of hospitalized RW exacerbations (RWE).

Methods: API is positive, individually, through the existence of RW, plus at least one of the two *major* criteria: parental asthma (PA) and/or atopic dermatitis (AD), or two of the three *minor* criteria: allergic rhinitis (AR) and/or wheezing apart from respiratory infections (WaRI) and/or peripheral eosinophilia (PEo) ≥4%. Retrospectively, the prevalence of these RF in RWE hospitalizations was analyzed. Only patients with at least one white blood count were included, and those over the age of 3 were excluded.

Results: From October 2022 to March 2023, there were 45 hospitalizations due to RWE, in 36 patients (78%, n=25 male). 32 met the inclusion and exclusion criteria, and their median age was 17 months [3; 47], SD 10.24. The prevalence of PA was 25% (n=8) and the presence of AD was 13% (n=4). A personal history of AR was found in 19% (n=6), WaRI in 34% (n=11) and PEo in 41% (n=13). The API was positive in 47% of cases (n=15). One-third of the patients had at least one *major* criteria, while in 13% (n=4) the API was positive exclusively with the association of *minor* criteria. One-quarter of the sample did not have any of these RF.

Discussion & Conclusions: In preschoolers asthma symptoms are variable and nonspecific, and wheezing can coexist due to other etiologies. The existence of predictive indices regarding this chronic pathology allows us to be more interventionist, particularly with maintenance therapy and even try to change the disease natural course. In this series, as in the literature, males stood out in the incidence of RW. By the API calculation, almost half of these patients are likely to suffer from asthma. For this result the most preponderant *major* criteria was PA, while the existence of WaRI and PEo were the most verified *minor* criteria.

Palavras-chave: recurrent wheezing, asthma, risk factors

(24630)

AN UNLIKELY COMPLICATION OF ACUTE OTITIS MEDIA

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Introduction: Luc's abscess is an exceedingly rare complication of acute otitis media in the pediatric population. It develops when infection spreads subperiosteally to reach the temporal fossa, deep to the temporalis muscle. Early identification of this condition is vital as it can often spare the patient an unnecessary mastoidectomy. We present the case of a patient with Luc's abscess and discuss its successful conservative management.

Case report: A 9-year-old boy presents to the Emergency Department with swelling over the temporozygomatic area for two weeks with significant increase in size the day before admission. The child also mentioned minor complaints of ear pain since the previous month. Fever, headache, focal neurological deficit or other associated symptoms were denied, as well as recent trauma. No relevant past medical history and up-to-date vaccination schedule. On physical examination, there was facial asymmetry, pain, and hard edema localized in the left temporozygomatic region, with no other local inflammatory signs. Normal cervical and temporomandibular joint mobility. Otoscopy showed bilateral effusion. No auricular protrusion, sulcus effacement or retroauricular inflammatory signs. Unremarkable laboratory tests. Soft tissue ultrasound showed a small collection with an elongated morphology with scarce septations and homogeneous anechoic internal content in the left temporal region, adjacent to the external table. Head computed tomography (CT) showed complete occupation of the mastoid cells and middle ears without mastoid involvement, thickening of the epicranial soft tissues in the left temporal region and a small epicranial abscess. The patient was admitted and treated with intravenous ceftriaxone for 7 days and methylprednisolone with favorable clinical outcome. At discharge, the temporal swelling had completely resolved.

Conclusion: Luc's abscess is a rare but manageable complication of acute otitis media. Although most published literature suggested surgical management, our case shows a successful conservative approach.

Palavras-chave: Otitis media, Luc's abscess

(24628)

BULLOUS HENOCH-SCHÖNLEIN PURPURA WITH NEPHRITIS: A CASE REPORT

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Background: Henoch-Schönlein Purpura (HSP), also known as IgA vasculitis, is one of the most common systemic vasculitis of childhood and may affect skin, joints, gastrointestinal tract, and kidneys. Skin manifestations include a non-thrombocytopenic palpable purpura on pressure-bearing sites, especially on lower extremities and buttocks. Although other cutaneous lesions may occur, in pediatric patients, hemorrhagic bullae and vesicles are rare. It has been speculated that they may reflect a more severe disease course.

Case report: A previously healthy 11-year-old girl presented with purpuric rash on her lower limbs, ankle arthralgia and abdominal pain. No previous history of infection/vaccination. Laboratory tests, including blood count, kidney function and coagulation studies as well as abdominal ultrasound were unremarkable, and a prescription of acetaminophen was made. However, the following week, due to worsening skin lesions, ongoing arthralgia and abdominal pain, further medical evaluation was warranted. Urinalysis and ankle ultrasound revealed no abnormalities. NSAID therapy was initiated and a referral to a pediatric rheumatology appointment was made. A week later, despite symptomatic improvement, cutaneous lesions evolved to vesicles and bullae in the lower third of both legs. Moreover, four weeks after initial presentation, urinalysis revealed an elevated protein/creatinine ratio, indicating renal involvement; prednisolone 1mg/kg/day was prescribed by pediatric nephrology. Despite initial worsening of proteinuria, urine sediment was normal and two weeks later both proteinuria and skin lesions started to improve; corticoid was tapered. In the follow-up visit, 4 months after presentation, urinalysis was normal and skin lesions were healing.

Discussion/Conclusion: Our patient met EULAR classification criteria of HSP. Even though HSP exhibits diverse array of manifestations, long-term prognosis is mainly determined by the extent of renal involvement. Hemorrhagic bullous and vesicular lesions are rare cutaneous manifestations in pediatric patients (<2%) having been described as worse prognosis factors. Recent literature review showed similar prevalence of abdominal and renal involvement in these cases.In IgA vasculitis nephritis, corticosteroids treatment may reduce the risk of end-stage renal disease. arly use of systemic corticosteroids has been suggested in bullous HSP patients, but their beneficial effect on the extent of lesions and/or sequelae of disease has not been proved.

Palavras-chave: Bullae, Henoch-Schönlein purpura, Nephritis, Case report

(24625)

BASAL GANGLIA DISEASE IN A CHILD WITH RECURRENT ATAXIA: IMPORTANCE OF TIMELY RECOGNITION OF TREATABLE DISEASES

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Recurring episodes of neurological deficits following mild disease raise a high suspicion of metabolic disorders, namely mitochondrial disorders presenting with Leigh syndrome. A “treatable-first” differential diagnosis approach is advisable in uncommon presentations since early recognition and intervention are crucial for prognosis.

Female child, 5 years old, first daughter of a healthy non-consanguineous Venezuelan couple, with normal growth and development except for a mild language delay. At 22 months after an acute episode of diarrhea, she had an episode of lethargy, ataxia, and dysphagia requiring admission to an intensive care unit in Venezuela and invasive mechanical ventilation for 10 days. Brain MRI showed recent FLAIR bilateral hyperintensities lesions of the basal ganglia, thalamus, and midbrain. Electromyography was normal. No metabolic study was performed. She was treated for presumed Miller Fisher Syndrome and discharged after 3 weeks, with right hemiparesis (recovered in two months).

Family migrated to Portugal. At the age of 3 years, she was admitted with ataxia, lethargy, and decreased muscle strength on the right. On admission she had normal blood and cerebrospinal fluid counts, negative CRP, and sterile blood culture., plasma lactate of 2.8 mmol/L, high plasma glycine and alanine, and normal urinary organic acids profile and acylcarnitines. Brain MRI showed T2 hyperintense foci in the basal ganglia, enlarged perivascular spaces or cavitated areas, with an atrophic appearance of the lenticular nuclei. She began coenzyme Q10, thiamine, L-carnitine, and biotin in line with a very likely mitochondrial disorder (Leigh syndrome) and discharged with normal neurological exam.

Genetic studies revealed two pathogenic variants in heterozygosity in the SLC19A3 [c.980-14A>G and novel p. Trp59 (c.177G>A)] giving a final diagnosis of thiamine-responsive basal ganglia disease. She started supplementation with high doses of biotin and thiamine, with significant clinical improvement. Currently, at 9 years old, she attends primary school with minor learning difficulties and unremarkable neurological exam.

This clinical case demonstrates the importance of including treatable diseases in the differential diagnosis of basal ganglia hyperintensities. Early recognition of thiamine-responsive basal ganglia disease is key to better outcome since not all previous lesions can be reversed with treatment. Follow up MRIs are advised.

(24618)

WHEN SHOULD PEDIATRIC KNEE PAIN SOUND THE ALARM?

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Introduction: Musculoskeletal complaints are common in Primary Care (PC) practice, including in pediatric age, representing up to one-third of the problems diagnosed at this stage of life. Knee pain is frequent in childhood and adolescence, and its etiologic factors are mostly benign and self-limited, with no significant long-term repercussion.

Case Report: 15-year-old female with personal history of leg-length discrepancy, followed by Orthopedics in 2008 and treated with an insole for the left foot. In August 2023, she presented with limping due to sporadic left gonalgia for three months, exacerbated by load and karate practice. Furthermore, she reported an increase in severity and frequency after having fallen off her skateboard two weeks earlier, with the pain now also appearing at night. Despite relief at rest, it had a low response to NSAIDs. There were no inflammatory signs or other accompanying symptoms, and her mother believed that similar pain was related to low adherence to the prescribed insole use. On physical examination (PE), she displayed a good general condition, adequate anthropometry, no deformities, and bilaterally preserved mobility, strength and sensitivity, but pain on palpation of the medial region of the left knee. Full spine and lower limb X-rays were requested, showing a 5 mm asymmetry, with shortening of the left lower limb, a lytic lesion on the left femoral distal metadiaphysis, with ill-defined limits in a sunburst pattern, and findings of a Codman's triangle, with cortical rupture, suggestive of osteosarcoma. Although no changes were apparent on an ultrasound, a targeted MRI confirmed the diagnosis. In September 2023, the patient was referred urgently to a dedicated oncology center and started neoadjuvant chemotherapy. In January 2024, an *en bloc* tumor resection with total knee replacement was performed, and the patient recovered *with no complications.*

Discussion & Conclusion: In PC practice, physicians must be able to recognize warning signs that should lead to an on-time referral to secondary care, so long-term morbidity and mortality can be prevented, such as in bone tumors, namely osteosarcoma, which has a poor survival rate. Even though a detailed clinical history, along with a complete PE, can provide vital clues to the underlying condition, it is crucial to cross this information with the child's age and changes in gait, so the correct diagnosis will not be overlooked.

Palavras-chave: knee pain, pediatric age, osteosarcoma, primary care

(24609)

STREPTOCOCCUS PYOGENES MENINGOENCEPHALITIS: A RARE CLINICAL PRESENTATION OF A COMMON BACTERIA

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Background: *Streptococcus pyogenes* (Group A *streptococcus*, GAS) is associated with a wide spectrum of diseases in pediatric age. Meningitis is a rare clinical manifestation of SGA invasive infection, presenting a high mortality rate and risk of neurological morbidity.

Case report: Previously healthy eight-year-old child, with up-to-date national immunization programme, admitted to the emergency department with a two-day history of fever, vomiting, frontal headache and prostration. He was hemodynamically stable, prostrated with doubtful meningeal signs, presenting scarlatiniform rash, perineal petechiae and strawberry tongue. Pharyngeal swab rapid antigen test for GAS was positive. Laboratory evaluation showed thrombocytopenia (112.000/uL) and elevated C-reactive protein (283.0 mg/L). Intramuscular benzathine penicillin was administered for suspected scarlet fever. In a few hours, he developed right periorbital oedema and erythema. Cerebral CT scan showed increased CSF density in the pericerebral spaces in the left frontoparietal convexity, suggesting meningitis, with subdural empyema and periorbital cellulitis. Cerebrospinal fluid analysis revealed pleocytosis (2405 cells/uL; 91.4% polymorphonuclear), hypoglycorrhachia (15 mg/dL; CSF/serum glucose ratio 10%) and hyperproteinorrachia (2.89 g/L). Antibiotic treatment (ceftriaxone and clindamycin) was initiated for presumed bacterial meningitis. Due to neurological deterioration (Glasgow coma scale 9 and focal neurological signs), he was transferred to a pediatric intensive care unit. Intravenous dexamethasone and Human Immunoglobulin were added to treatment. On the fifth day, his clinical condition worsened due to status epilepticus, needing invasive mechanical ventilation, intracranial pressure monitoring and antiepileptic treatment. A multi sensitive *Streptococcus pyogenes* was isolated on both blood and CSF cultures, switching Ceftriaxone for Penicillin. He recovered well after 14 days of clindamycin and 8 weeks of penicillin. On day 28, he was discharged to a physical rehabilitation center to proceed clinical care.

Discussion/ Conclusion: The authors highlight that GAS should be considered as potential cause of meningoencephalitis. Early recognition and adequate treatment are crucial to ensure a better outcome in cases of invasive GAS disease, like this.

Palavras-chave: Meningoencephalitis, Streptococcus pyogenes, Group A streptococcal infection, Invasive disease

(24605)

CARDIOVASCULAR ANATOMICAL VARIANTS: AN ADOLESCENT'S JOURNEY THROUGH STROKE AND DYSPHAGIA

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Background: Patent foramen ovale (PFO) and aberrant right subclavian artery (ARSA) are usually considered anatomical variants. However, PFO represents the substrate for embolic disease as cryptogenic stroke, even in young population, and ARSA can compress adjacent structures, particularly the esophagus and cause dysphagia. We report a case of an adolescent with these two entities who underwent successful surgical repair.

Case report: A 15-year-old girl, with hypermobility spectrum disorder, chronic widespread pain (CWP) and Tourette syndrome, presented to the emergency department with a right hemiparesis. CT angiography revealed a thrombus in the M1 segment of the left middle cerebral artery and the patient was submitted to mechanical thrombectomy; during the procedure an ARSA was detected. Subsequently, antiplatelet therapy was initiated, and risperidone and contraceptive pills were stopped. The following investigation revealed the presence of a tunnel shaped PFO with left-to-right shunt. There was full neurological recovery and the PFO was percutaneously closed using NobleStitch® system. During the subsequent follow-up she complained of progressive dysphagia and echocardiogram showed persistent PFO. Upper gastrointestinal endoscopy revealed a pulsatile extrinsic compression of the esophagus, consistent with dysphagia lusoria. After multidisciplinary discussion, she underwent surgical ARSA reimplantation and PFO closure. Following the surgical procedure, she reports reduction of dysphagia, and continues a multidisciplinary follow-up, with no recurrence of thromboembolic events.

Discussion/Conclusions: Recent randomized controlled trials showed that PFO closure plus antiplatelet treatment is superior to medical treatment alone for secondary stroke prevention, with low risk of embolism recurrence post-complete PFO closure. The fact that the patient only complained of severe dysphagia after the stroke represented a clinical challenge regarding the true cause of this symptom, but the significant improvement after the ARSA reimplantation corroborates that this vascular anomaly was an important contributor. This case shows the difficulties of clinical management of a patient with CWP and cardiovascular anomalies and the importance of a multidisciplinary team approach.

Palavras-chave: patent foramen ovale, aberrant right subclavian artery, stroke, dysphagia

(24550)

RASH ASSOCIATED WITH INFLUENZA – CASE REPORT

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Background: Rashes are common in children. They can be caused by many different factors, including infections, drugs, or allergies. However, its etiology can be elusive, and many times a definitive diagnosis cannot be achieved.

Clinical Case: A 7-year-old male, with a history of allergic rhinitis, medicated with bilastine and montelukast, presented in the ER for the second time with a 2-day history of fever and a pruritic rash, that appeared on the same day. Associated symptoms were rhinorrhea and cough. He had no history of other allergies and all his vaccinations were up to date. He had taken amoxicillin in the previous week for an acute otitis media. He had no reactions the previous times he took the same antibiotic. On physical examination he had slightly elevated, erythematous, well-circumscribed papules and plaques, compatible with an exuberant urticarial rash, on the face, trunk, and limbs, with no lesions on the palms nor the soles of the feet. Other findings were a runny nose and a discreetly erythematous oropharynx, with no further changes on physical examination. The presumptive diagnosis of a viral exanthem was made, later confirmed by PCR result positive for Influenza A. He was discharged with an antihistamine drug and a corticosteroid with complete resolution of symptoms.

Discussion: Rash is a rare manifestation of an Influenza infection. Nevertheless, there are some reported cases, more commonly in children. It has been described as petechial, macular, papular, maculopapular, reticular, or purpuric. It can be localized or generalized, as well as pruritic or not. Proposed mechanisms for Influenza-associated rash remain speculative.

Conclusion: This case highlights a rare presentation of a common disease, reminding that an Influenza infection should be included in the differential diagnosis of a febrile exanthem.

Palavras-chave: Influenza, Rash

(24558)

CHROMOSOME 2Q INTERSTITIAL DELETION INCLUDING SATB2 GENE IN A CHILD WITH CARDIOPATHY

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Background and Aim: Chromosomal interstitial deletions of the long arm of chromosome 2 are rare events and patients' clinical manifestations vary according to the size and location of the deleted fragment. We report a 2 years-old child with a very small interstitial deletion of the long arm of chromosome 2 (q32.1q33.3) defined at the molecular level by array comparative genomic hybridization (array CGH).

Clinical Case and Methods: This boy is the first child of a nonconsanguineous healthy couple, who presented with global developmental delay, facial dysmorphisms, neonatal hypotonia, feeding difficulties, posterior cleft palate, laryngomalacia type 1 and Obstructive Spleep Apenia Syndrome, conduction deafness, gastro-oesophageal reflux, supraventricular tachycardia with extrasystoles and small atrial septal defect. Brain MRI revealed small corpus callosum. Chromosome analysis with GTL banding on metaphases obtained from conventional lymphocyte cultures and arrayCGH Cytoscan 750K were performed.

Results: Initial conventional chromosomal analysis showed an abnormal chromosome 2 with a deletion apparently in 2q32.1q33.3 [46,XY,del(2)(q32.1q33.3)]. Since the karyotypes of both parents were normal, this alteration was considered *de novo*. Microarray-based comparative genomic hybridization confirmed the 20Mb loss in region 2q32.1q33.3, which includes the *SATB2* gene: arr[GRCh37] 2q32.1q33.3(185531376_20615736)x1.

Conclusions: *SATB2* gene is correlated with Glass syndrome (OMIM#612313, ORPHA:251028) that is a multissistemic pathology characterized by intellectual disability of variable severity and dysmorphic facial features, including micrognathia, downslanting palpebral fissures, cleft palate, and crowded teeth. The authors compare the child's phenotype with the published literature and enhance the importance of the combination of high resolution banding with appropriate molecular cytogenetic techniques in the characterization of chromosomal rearrangements, enabling better genetic counselling for patients and their families.

Palavras-chave: Cytogenetics, Deletion; Glass syndrome

(24566)

COLONIC ATRESIA: A RARE CAUSE OF NEONATAL INTESTINAL OBSTRUCTION

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Introduction: Colonic atresia (CA) is a rare cause of neonatal intestinal obstruction, usually located proximal to the splenic flexure. Typically, diagnosis is postnatal, suggested by signs of low intestinal obstruction. Notably, colonic atresia may be associated with Hirschsprung's disease (HD) in <10% of cases.

Clinical case: A full-term female neonate, born after an uneventful pregnancy, presented at 48 hours of life with food intolerance characterized by bile vomiting and absence of meconium passage since birth. Physical examination revealed a distended abdomen and a perforated and normal anus. Biliary content drainage followed nasogastric tube placement. Abdominal radiography showed significant colonic distension and absence of pelvic gas, prompting suspicion of intestinal occlusion. Given the suspicion, a contrast enema was performed which was suggestive of an ascending CA. The neonate underwent exploratory laparotomy where a type IIIa CA was identified at the level of the ascending colon/hepatic flexure, without other identifiable malformations. A colostomy was performed due to significant caliber discrepancy between colonic segments and risk of HD. Enteral feeding commenced on the 2nd post-op day, after colostomy functioning. Full feeds were achieved on the 3rd day post-op. The patient was discharged on the 6th day post-op without complications. Paralelly, rectal biopsies were conducted for HD exclusion, which results were negative. At 11 months of age, the colonic anastomosis was performed. Due to significant distension of the ascending colon and distance of 4cm between stoma-ileocecal valve, this segment was resected and an ileocolonic termino-terminal anastomosis was performed . Pathology examination showed no changes in intestinal innervation. The patient was discharged without postoperative complications and remains asymptomatic after 21 months.

Conclusion: CA is a rare clinical entity that mandates high clinical suspicion. Prompt diagnosis and surgical intervention are crucial when managing a CA as there is a high risk for precocious intestinal perforation. Long-term follow-up, including exclusion of other malformations and rectal biopsies to rule out HD, ensures optimal outcomes. This case underscores the importance of meticulous management and multidisciplinary care in addressing rare gastrointestinal anomalies in neonates.

Palavras-chave: colonic atresia, neonatal obstruction, Hirschprung's disease

(24586)

CYTOGENETIC CHARACTERIZATION OF A "PURE" 10P PARTIAL DUPLICATION: A CASE REPORT

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Introduction: Trisomy 10p (ORPHA:171929) is a rare chromosomal syndrome caused by total or partial duplication of the short arm of chromosome 10, phenotypically characterized by intellectual disability and multiple congenital malformations. Several partial 10p trisomies have been published, but most of these reports are based on patients with another chromosomal contribution as a result of missegregation of a balanced familial translocation. "Pure" 10p duplications are extremely rare events and may be the result of other mechanisms such as *de novo* duplication, pericentric inversion in one of the parents and supernumerary marker chromosomes. The presence and severity of symptoms depend on the size and location of the duplicated segment and the genes involved. The most frequent breakpoint is located in the p11 band, however, if the break is more distal it results in partial trisomy.

Clinical Case: We report the case of a 6-month-old girl referred for cytogenetic studies due to global developmental delay, axial hypotonia, and failure to thrive between the age of 15 days and two months, with diagnosed feeding difficulties. At 13 mo, length and weight centiles evolving between centiles 3 and 15, and occipitofrontal circumference in P85. She presented with mild frontal bossing, but dysmorphic evaluation was otherwise unremarkable. The proband's karyotype revealed a structurally abnormal chromosome 10 with additional material in the short arm. MLPA, FISH and array-CGH analysis were performed to identify the origin of the segment involved and to define the breakpoints. The complete karyotype was established as: 46,XX,dup(10)(p15.3p12.1). The length of the tandem duplicated segment comprises 24.8Mb. Parental samples are not available for genetic studies at this time.

Discussion: The phenotypic manifestations of cases with pure trisomy 10p described in the literature are inconsistent. To date, seven pure 10p15.3p12.1 duplications have been reported and the associated clinical features were similar to cases of full 10p duplication, but with a milder phenotype. The authors establish the correlation between the "classic" "Trisomy 10p Syndrome" and the clinical findings of our patient. The description of the present case contributes to a better characterization of pure Trisomy 10p Syndrome and delineates genomic segments with potential phenotypic impact.

Palavras-chave: Trisomy 10p, Chromosome duplication, Intellectual disability, Congenital malformations

(24603)

EYES WIDE OPEN: BEYOND APPEARANCE, TOWARDS WELL-BEING IN TREACHER COLLINS SYNDROME

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Introduction: Treacher Collins syndrome (TCS) is a rare genetic disorder that affects craniofacial development due to malformation of the first and second branchial arches. This case report emphasizes the importance of multidisciplinary follow-up, particularly focusing on ocular manifestations, in children with TCS.

Case report: This case presents a 4-year-old male diagnosed with TCS, who has been under pediatric ophthalmology follow-up since he was 14 days old. He was born of a nonconsanguinous marriage and at full-term by cesarean delivery with an Apgar 9/10. Prenatal ultrasounds were described as normal.

During examination after birth, numerous abnormalities in the craniofacial structures were found: bilateral microtia with complete absence of the external auditory canals, malar and zygomatic bones hypoplasia, retrognathia, inverted and droopy eyelid slits, hypoplasia of the lower eyelid fissure and complete cleft palate. The rest of the examination was normal. No family history was found for similar craniofacial dysmorphias. Until the 10th day of life, he was being fed orally complemented with nasogastric tube feeding due to swallowing difficulties. Breathing remained unaffected.

Multidisciplinary follow-up included: pediatrics, genetics, otolaryngology, stomatology, pediatric and plastic surgery, physical medicine and rehabilitation. He has been using a Bone Conduction Hearing Aid with Softband since he was 4 months old. At 2 years old, he underwent cleft soft palate correction. At 3 years old, mandibular distraction and bilateral cantopexy of the lower eyelids were perfomed. The child demonstrated progressive language development and achieved key developmental milestones.

The ophthalmological evaluation up to four year-old showed antimongoloid slant of the palpebral fissures and absence of eyelashes in the entire extent of the lower lids. Centered Hirchberg reflex with apparent orthotropia, normal cover test and eye movements. Cycloplegic objective refraction indicated mixed astigmatism on the right eye and simple hyperopic astigmatism on the left eye. Fundoscopy showed no abnormalities.

Conclusion: Many infants with TCS may exhibit ocular differences, which can contribute to a saddened facial appearance. Other ocular findings include eyelid coloboma, amblyopia, anisometropia, strabismus, congenital cataracts, occasional microphthalmia, dacryostenosis, among others. Vision may still be normal in TCS because the retina does not develop from the affected branchial arches. Therefore, in most cases, despite the major orbital malformations, the ophthalmological prognosis isn't compromising.

Multidisciplinary care aims not only to correct anatomical anomalies, address physical, cognitive and social challenges associated, but also to promote overall development and enhance the patient's quality of life.

Palavras-chave: Treacher Collins syndrome, Multidisciplinary, Ophthalmological, Wellness

(24567)

PEDIATRIC URACHAL REMNANTS: A CASE SERIES HIGHLIGHTING DIVERSE PRESENTATIONS AND MANAGEMENT

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Introduction: The urachus forms during embryonic development from the separation of the allantois and the ventral cloaca, corresponding to the future bladder. It gradually narrows and eventually disappears by birth, transforming into the median umbilical ligament. When failure of its obliteration happens a urachal remnant (UR) originates. This study aims to present a case series of our department showcasing the diverse forms of UR and its presentation in the pediatric age.

Methods: Retrospective study of all patients diagnosed with UR between January 2013 and February 2023. Demographic data, clinical manifestations, complementary diagnostic exams performed, therapeutic approach and postoperative complications were collected.

Results: The study encompassed 11 patients, with 72.7% (N=8) being male. The diagnosed urachal anomalies comprised urachal cyst (N=6; 54.5%), patent urachus (N=3; 27.2%), and urachal sinus (N=2; 18.2%). Median age at presentation varied across urachal anomaly types: 4.2 years (range: 0.7-15.4) for urachal cysts, 5.2 years (range: 0.1-16.3) for patent urachus, and 6.9 years (range: 0.2-13.6) for urachal sinus. Clinical presentations of urachal cysts included infection (N=2), urinary tract infection (N=1), recurrent abdominal pain (N=1), umbilical drainage (N=1), and incidental intraoperative finding (N=1). Among patients with patent urachus, two presented with umbilical drainage and one, at a later age (16.3 years), with recurrent abdominal pain. Both urachal sinuses manifested as urachal infection with palpable infraumbilical swelling. Pre-operative clinical suspicion of UR was present in all patients except one, which discovery was incidental, and another patient who was suspected to have Meckel's diverticulitis. Abdominal ultrasound was the most frequently used diagnostic imaging modality (N=8; 72.7%), followed by abdominopelvic computed tomography (N=2; 18.2%). Surgical resection was performed in all cases, with urachal sinuses addressed via umbilical exploration and the remaining UR primarily approached laparoscopically (N=6; 67%). No complications arose intra or postoperatively.

Conclusion: Our study highlights the wide range of clinical presentations of UR and emphasizes the importance of considering UR in the differential diagnosis of pediatric umbilical or abdominal complaints. Early recognition aided by diagnostic imaging is essential for optimal management. Laparoscopic surgical resection has proven to be effective with minimal complications.

Palavras-chave: urachal remnant, pediatric

(24610)

WHEN THE NECK TILTS THE DIAGNOSIS TOWARDS A MORE SERIOUS CONDITION

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Introduction: Torticollis is characterized by lateral inclination of the head to the shoulder, causing torsion of the neck and deviation of the face. It can be congenital or acquired and different types of pathophysiologic processes can produce torticollis, from benign conditions like upper airway infections to more serious conditions like retropharyngeal abscess or a tumor.

Case Report: A seventeen-month-old child presenting to the Emergency Department with complaints of fever, cough, rhinorrea, and transient head tilting to the left. After evaluation, the diagnosis of acute left-sided otitis media was made and the child was treated with amoxicillin with clavulanic acid. Due to maternal concern regarding more frequent periods of irritability, the child was reevaluated in the Emergency Department one month later, revealing persistent left cervical torticollis, reduced mobility of the right upper limb, right hand clenched, and refusal to crawl. When testing the grip movement of the right hand, it was ineffective, leading the child to preferentially use the left hand. A cervical ultrasound revealed only small reactive cervical lymph nodes, and an head CT scan showed no suggestive image of an expansive lesion. Ophthalmological examination was within normal limits. The patient was discharged with a neuro-axis MRI scheduled. However, five days later, the child returned to the Emergency Department due to worsening pain when standing up and crawling, spending more time laying down at daycare. A neck CT scan was performed, showing no cerebral hernia or any other findings regarding significant canal involvement or myeloradicular compression. Subsequently, a neuro-axis MRI was performed, revealing a large space-occupying lesion measuring 58.8 x 15.1 mm, centrally located from C4 to D3, with spinal cord enlargement and medullar edema. The patient was then transferred to a tertiary hospital and, based on the child's age and location of the lesion, it was decided to proceed with biopsy with electrophysiological monitoring. The histological diagnosis revealed a Grade I Pilocytic Astrocytoma.

Discussion and conclusions: There is a vast spectrum of pathologies that may present as torticollis. Therefore, a comprehensive evaluation is crucial to accurately diagnose the underlying cause and implement appropriate treatment strategies. This case underscores the significance of a systematic approach, including detailed medical history, thorough physical examination, and imaging studies, to ensure timely intervention and optimal patient outcomes.

Palavras-chave: Torticollis, Limb mobility impairment, Central nervous system, Astrocytoma

(24619)

HIVES FROM HEAT AND SWEAT: ARE AN ADOLESCENT IN GREATER RISK? – CASE REPORT.

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Introduction: Chronic inducible urticaria requires a detailed clinical history, especially in cholinergic urticaria, a rare subtype, especially in pediatric age. The symptoms are triggered by an increase of body temperature and the differential diagnosis is made with other non-physical or physical subtypes, like heat contact urticaria or solar one.

Case Report: a 17-year-old adolescent, followed in Allergy and Clinical Immunology since the age of 11. He was referred from primary care due to seasonal respiratory complaints, with a family history of atopy of asthma and food allergies on maternal side. He was diagnosed with pollinosis and at the age of 14, underwent on grass pollen specific immunotherapy for a year, which was prematurely interrupted for economic reasons and needed to maintain therapeutic control with topical nasal steroids for periods, and ketotifen eye drops and oral antihistamine as resource. At the age of 16, on a follow-up appointment, he started complaints of skin itching lesions, which were micropapular and with a visible erythematous halo on photographic records he took. The identified triggers were "begin to feel warm or sweaty". First episodes got solved spontaneously in less than one hour and with no residual lesions. The condition is reproducible during physical exercise, when he got in heated places or if he stayed outside on sunny days. He does not take hot baths and denies symptoms related with stress. Until then, the study performed had no significant changes and it was assumed a cholinergic urticaria. Avoidance measures were explained, and a SOS second-generation H1-antihistamine (AHs) was prescribed.

Discussion & Conclusions: The suggestive clinical history was diagnostic but could be confirmed with a specific provocation test – Exercise or Immersion Test. Avoiding triggers, in this case stimuli that increase body temperature, is essential. Currently, only AHs in standard dose (age- or weight-based) are considered as a first-line pharmacological therapy. In this case, despite the impressive picture records and no signs of remission, it seems like an easy-to-control condition that not particularly interfere with the patient's daily life that has, generally, a favorable prognosis.

Palavras-chave: Chronic inducible urticaria, Cholinergic urticaria, second-generation H1-antihistamine

(24626)

GENERALIZED BULLOUS ERUPTION IN PEDIATRICS – FROM A RARE DIAGNOSIS TO A SERIOUS COMPLICATION

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Background: Bullous diseases are uncommon in Pediatrics. Linear IgA dermatosis (LAD) is a rare bullous skin disease, that can be idiopathic, related to other diseases or induced by drugs or infections. Skin disruption and immunosuppressant drug use increase infectious risk.

Case Report: A previously healthy 3 year old female child, 2 days after an adenotonsillectomy, developed papules and blisters in her hands and face, with no response to antihistaminic and topical corticoid. Over 10 days, plaques and bullae formed, and the lesions progressed to involve multiple areas of the skin while sparing mucous membranes, associated with pruritus and anorexia. She was admitted to her local hospital and fulfilled 14 days of systemic corticoid (3 methilprednisolone 10 mg/Kg bolus followed by 10 mg/Kg/day of prednisolone) with no improvement. After that, she was transferred to a tertiary pediatric hospital to further study and treatment. Upon admission, she exhibited a symmetrical dermatosis with circular plaques with erosions, tense serous blisters and bullae, organized in "strings of pearls", mostly distributed on the trunk, perineum, and limbs. Skin biopsy was performed, and low-dose oral dapsone was initiated given the clinical suspicion of LAD. On the 31st day of disease she initiated fever with increase in acute inflammatory markers (C-reactive protein 34,7 mg/L). E. coli was identified in blood and the patient had a good outcome after 15 days of antibiotic therapy (Piperacillin/Tazobactam). The biopsy with direct immunofluorescence revealed neutrophilic subepidermic bullous dermatosis with IgA deposits at the basal membrane, confirming LAD. Autoimune and celiac diseases were excluded. Dapsone was gradually increased to 1.5 mg/kg/day. She was discharged after 41 days of hospitalization, with mostly cicatricial lesions. She is currently in her fifth month of treatment with dapsone, with minimal recurrence of bullae.

Discussion: Drug exposure (anaesthetic and opioid use) might have been the trigger behind this case, however confirmation is not possible. Short-term systemic corticosteroids may expedite initial improvement, but dapsone is more effective in medium-long term disease control. Skin disruption and immunosuppressant drug use increase infectious risk, hence the low threshold to initiate antibiotic treatment in the presence of fever. Recurrence of lesions may persist for months to years before total resolution.

Conclusion: LAD is a rare cause of bullae in Pediatrics. Systemic corticoids must be used with caution, in a short period of time and signs of complications must be closely monitored. Dapsone at higher doses can provide disease control. Case reports of rare diseases are important to disclose treatment outcomes and to generate further inquiry.

Palavras-chave: Bullae, Linear IgA dermatosis, Infectious risk, Pediatrics, Dapsone

(24629)

AN UNCOMMON PRESENTATION OF INFLUENZA A INFECTION: A CASE REPORT

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Background: Influenza virus is one of the main causes of acute upper respiratory tract infections in pediatric patients. Although rare, it is a recognized etiological agent of viral encephalitis, with a prevalence similar to that of Herpes Simplex Virus.

Influenza-associated acute encephalitis (IAE) affects mainly children under 5 years of age, and its diagnosis is difficult because clinical presentation varies significantly and the influenza virus is rarely detected in cerebrospinal fluid (CSF). IAE can result in neurological sequelae or even death

Case report: A 12-year-old male teenager, with a history of asthma and allergic rhinitis and an updated national immunization programme, presented with a 2 day long fever, headaches and abdominal pain followed by confusion and 2 episodes of tonic-clonic seizures.

After hospital admission his neurologic state quickly worsened, requiring admission to an intensive care unit (ICU). Mechanical ventilation, vasopressors and inotropes support and antiepileptic drugs were initiated. Initial blood and CSF analysis and cerebral MRI were unremarkable. He tested positive for influenza A in nasal swab and oseltamivir was added. During ICU stay, he presented rhabdomyolysis (CK 4468 U/L) , acute renal injury (creatinine 0,91 mg/dl and urea 58 mg/dL) and hepatitis (TGO 52 U/L, TGP 123 U/L) . He was gradually improving, extubated on D4 and discharged from ICU after 6 days. During the stay in the pediatric ward, he recovered under physical rehabilitation. Additionally, he was diagnosed with femoral thrombosis at the site of the central venous catheter. He was discharged after 20 days in hospital.

Discussion: Authors point the challenges in the management of IAE in pediatric patients. As reported in this case, its non-specific presentation and no detection in CSF are challenging. Its possible muti-organ involvement highlights the multidisciplinary approach and needed surveillance for potential sequelae.

Palavras-chave: Influenza-associated acute encephalitis (IAE)

(24632)

LUDWIG'S ANGINA IN CHILDREN: THE IMPORTANCE OF A RAPID DIAGNOSIS AND AN APPROPRIATE MANAGEMENT

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Ludwig's angina (LA) is an acute inflammatory condition characterized by cellulitis of mouth floor that can lead to edema, distortion, and airway obstruction and, consequently, asphyxiation and death. Typically originates from an infected or recently extracted tooth, but can also arise from mouth or facial trauma, submandibular sialadenitis, and other underlying diseases. In children, deep neck space infections subsequent to upper respiratory tract infections represent the majority of the cases. Predominantly seen in middle-aged individuals, has been rarely reported in children. A 7-year-old child, presented to the hospital due to the sudden growth of a sublingual mass that had been present for 5 days. Additionally, reported fever that started on that day and sialorrhea, halitosis and odynophagia. The patient presented a significant midline sublingual swelling, causing an upward and left deviation of the tongue, which prevented speech but did not cause labored breathing or stridor. The oropharynx was not visualized due to interposition of structures and asphyxiation risk, but a dental cavity was reported. Due to impending risk of airway obstruction, anesthesiology and otorhinolaryngology were promptly involved. Blood work revealed leukopenia (2000/µL) with neutrophilia (85%) and C-reactive protein (CRP) of 27.82mg/L. A cervical CT scan revealed a densification of adipose tissue of the neck, as well as a suppurative collection within the floor of the mouth, measuring 64x27x37mm and slightly enlarged cervical lymph nodes. Antibiotic therapy was initiated with clindamycin (40mg/kg/day), amoxicillin and clavulanic acid (100mg/kg/day), as well as methylprednisolone (3mg/kg/day). The patient underwent abscess drainage under general anesthesia and orotracheal intubation and was transferred to the intensive care unit. CT scan repeated on the third day, revealed a significant reduction in the collection size. Repeated blood work showed 7200 leukocytes/µL, with 80% neutrophils and CRP 20.2mg/L. Pus collected during surgery isolated a Streptococcus mitis, described as a common contamination agent . The child was extubated on the third day and transferred to the pediatric ward, being discharged from the hospital after 15 days of intravenous antibiotic, with a substantial reduction of the sublingual mass. The incidence of LA in children has steadily decreased in the last decades due to modern dental care and the surge of the antibiotic era. Albeit rare, it is a life-threatening condition, highlighting the need for a high index of suspicion and a rapid intervention due to the potential progression to severe respiratory and airway compromise. Timely initiation of antibiotic therapy assumes a pivotal importance. Despite the considerable morbidity and mortality, early recognition, empirical antibiotic administration, and prompt surgical interventions offer favorable prognosis.

Palavras-chave: Ludwig's angina, Deep neck infection, airway compromise, sublingual mass

GINECOLOGIA - OBSTETRÍCIA

COMUNICAÇÕES ORAIS

(24611)

ARTIFICIAL INTELLIGENCE AND COLPOSCOPY: AUTOMATIC IDENTIFICATION OF VAGINAL AND VULVAR SQUAMOUS CELL CARCINOMA PRECURSORS

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Introduction: Despite Human Papillomavirus (HPV) established role in cervical cancer, its impact extends to vaginal and vulvar cancers. While colposcopy offers a comprehensive examination female genital tract, its diagnostic accuracy is suboptimal. Artificial intelligence (AI) integration holds promise for improving the cost-effectiveness of colposcopy. However, there is no dedicated AI models specifically designed to differentiate low-grade (LSIL) from high-grade (HSIL) squamous intraepithelial lesions in the vagina and vulva. This study aims to develop and validate an AI model for differentiation of HPV-associated dysplastic lesions in these regions.

Methods: Two independent convolutional neural network (CNN) models were developed to differentiate HSIL from LSIL in colposcopy images: one for vaginal lesions and another for vulvar lesions. The vaginal model was developed on a dataset of 13,372 frames (90% training, 10% testing) obtained from 32 colposcopies. The vulvar model, comprising a total of 837 frames from 5 colposcopies, was also divided in a training and testing set. The model was evaluated through its sensitivity, specificity, accuracy and area under the receiver-operating curve (AUROC).

Results: For LSIL/HSIL differentiation in vagina, the CNN demonstrated a sensitivity of 94.4%, a specificity of 98.2%, and an accuracy of 95.0%. For LSIL/HSIL differentiation in vulva, detection the CNN demonstrated a sensitivity of 99.7%, a specificity of 100.0%, and an accuracy of 99.7%. The AUROC were respectively 94.4 and 1.00.

Conclusion: While potential selection bias and overfitting warrant further investigation to definitively assess generalizability, these results are promising. They represent the initial step towards a comprehensive, AI-powered colposcopic assessment encompassing the entire female genital tract.

Palavras-chave: Cervical Squamous Cell Carcinoma, HSIL, LSIL, Colposcopy, Artificial Intelligence

(24595)

OVARIAN HYPERSTIMULATION SYNDROME AND SPONTANEOUS PREGNANCY: A CASE REPORT

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1 CMIN

Introduction: Nowadays, ovarian hyperstimulation syndrome (OHSS) is a rare complication that occurs during assisted reproductive treatments.

Clinical Case: A 36-year-old woman with three years of primary infertility was submitted to controlled ovarian stimulation with antagonist protocol, using recombinant human luteinizing (rhLH) and recombinant human follicle-stimulating hormones (rhFSH) (225 UI/day) for 11 days with contraceptive primmer. Triggering was performed with recombinant human chorionic gonadotropin (r-hCG) and 15 oocytes were retrieved. On the trigger day estradiol measure was 3.619 pg/ml and there was a premature elevation of progesterone (10.19 ng/mL). Due to the risk of OHSS it was decided to do a freeze all cycle.

On day seven post oocyte retrieval she presented at the emergency department with progressive abdominal pain and distension. She had mild dyspnea. She did not have fever, urinary or intestinal symptoms. She had minimal relief while taking paracetamol. Pelvic ultrasound revealed bilaterally enlarged ovaries (maximum 85 and 94 mm) and moderate ascites. Blood analysis revealed: hemoglobin 15,0 g/dL, hematocrit 44%, AST 75 U/L; ALT 63 U/L, renal function and ionogram were normal.

The patient was hospitalized. A culdocentesis was performed with aspiration of 1500 mL of citrine liquid and IV (intravenous infusion) albumin reposition, with immediate relief of symptoms. She continued treatment at home with enoxaparin, analgesic medication, and cabergoline.

Six days later she had persistent nausea, vomiting, and abdominal distension. Transvaginal ultrasound revealed persistent liquid in the vaginal sac and it was visualized two intrauterine gestational sacs. Serum Beta-hCG was 11821,0 U/L with mild elevation of AST and ALT.

Two weeks later, she reported persistent nausea. Transvaginal ultrasound confirmed a dichorionic gestation, with two embryos compatible with 6 weeks + 5 days and 6 weeks+1 day with positive cardiac activity. The patient maintains pregnancy follow-up.

Discussion: In this case, OHSS was prevented by the decision to do a freeze all cycle. Pregnancy during ovarian stimulation increases placental hCG release, which stimulates ovarian VEGF output and exacerbates symptoms.

Conclusion: This is a rare case of spontaneous pregnancy during ovarian stimulation. Early detection and adequate supportive therapy are recommended to guarantee a positive outcome.

Palavras-chave: ovarian hyperstimulation syndrome, assisted reproductive treatment

(24616)

COMPARISON OF PREGNANCY OUTCOMES IN WOMEN UNDERGOING FRESH VERSUS FROZEN EMBRYO TRANSFER IN IN VITRO FERTILIZATION CYCLES

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Introduction: Fresh and frozen embryo transfer (ET) are commonly used techniques in *in vitro* fertilization cycles to increase the chances of a successful pregnancy in infertile couples. However, there is still debate about which method offers better pregnancy outcomes. This study aims to compare pregnancy outcomes between women undergoing fresh and frozen ET.

Methods: We conducted a single-center retrospective cohort study involving 358 patients who achieved pregnancy post fresh or frozen ET at our Reproductive Medicine Center between 2021-2022. Frozen ET were performed 3-5 days post-ovulation with a programmed menstrual cycle. Our primary outcome aimed to compare the live birth rates, defined as viable newborn delivery ≥ 24 weeks of gestation, between women who underwent fresh and frozen ET. Secondary outcomes included miscarriage and biochemical pregnancy rates, as well as obstetric outcomes, assessed across both groups. Obstetrics outcomes were compared in pregnant women with singleton pregnancies following fresh (n=92) and frozen (n=97) ET who delivered live births.

Results: Patients age (fresh ET: 34.6±3.7 vs. frozen ET: 34.3±4.0 years, p=0.673) and body mass index (fresh ET: 25.3±4.8 vs. frozen ET: 24.9±4.8 kg/m^2, p=0.583) showed no significant differences between groups. Live birth rates were comparable between fresh ET (62.2%) and frozen ET (69.1%) groups (p=0.171), as were miscarriage rates (fresh ET: 14.4% vs. frozen ET: 17.4%, p=0.442). However, the fresh ET group had a higher rate of biochemical pregnancy (23.3% vs. 13.4%, p=0.016).

Pregnant women of the frozen ET group had a lower rate of preterm delivery (16.3% vs. 6.2%, p=0.027). Incidence of hypertensive disorders of pregnancy (fresh ET: 9.8% vs. frozen ET: 8.2%, p=0.487), intrauterine growth restriction (fresh ET: 15.2% vs. frozen ET: 10.3%, p=0.384), gestational diabetes (fresh ET: 5.4% vs. frozen ET: 6.7%, p=0.445), placenta previa (fresh ET: 3.3% vs. frozen ET: 1.0%, p=0.358) and cesarian section (fresh ET: 56.5% vs. frozen ET: 59.8%, p=0.648) were comparable between groups. Newborns from the frozen ET group were heavier (3244.4±452.2 vs. 2990.5±616.1 gr, p=0.001) and had a reduced incidence of low birth weight (6.2% vs. 17.4%, p=0.016). Rates of macrosomia (fresh ET: 3.3% vs. frozen ET: 4.1%, p=0.754) and perinatal mortality (fresh ET: 1.1% vs. frozen ET: 0%, p=0.229) were similar between groups.

Discussion: No significant difference was observed in the live birth rate between frozen and fresh ET. Frozen ET correlated with a reduced risk of preterm delivery and low birth weight, along with higher birth weight compared to fresh ET. Other maternal and neonatal outcomes did not significantly differ between the groups. While our finding align with some prior studies, larger prospective and randomized trials are necessary to validate our results. The choice among fresh or frozen ET should be tailored to each patient's scenario.

Palavras-chave: frozen embryo transfer, fresh embryo transfer, in vitro fertilization, pregnancy outcomes

(24623)

ARTIFICIAL INTELLIGENCE AND COLPOSCOPY: AUTOMATIC IDENTIFICATION OF CERVICAL SQUAMOUS CELL CARCINOMA PRECURSORS

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Introduction: Proficient colposcopy is crucial for adequate management of cervical cancer precursor lesions, nonetheless its limitations may impact its cost-effectiveness. The development of artificial intelligence models is experiencing exponential growth, particularly in image-based specialties. The aim of this study is to develop and validate a Convolutional Neural Network (CNN) for automatic differentiation of high grade (HSIL) from low grade dysplasia (LSIL) in colposcopy.

Methods: A unicentric retrospective study was conducted based on 70 colposcopy exams, comprising a total of 22,693 frames. Among these, 8,729 were categorized as HSIL based on histopathology. Total dataset was divided into training (90%, n= 20,423) and testing set (10%, n = 2,270), the latter being used to evaluate the model's performance. Main outcome measures included sensitivity, specificity, accuracy, positive predictive value (PPV), negative predictive value (NPV) and the area under the receiving operating curve (AUC-ROC).

Results: Sensitivity was 99.7% and specificity was 98.6%. PPV and NPV were was 97.8% and 99.8%, respectively. Overall accuracy was 99.0%. The AUC-ROC was 0.98. The CNN processed 112 frames per second.

Conclusions: We developed a CNN capable of differentiating cervical cancer precursors in colposcopy frames. The high levels of accuracy for the differentiation of HSIL from LSIL may improve the diagnostic yield of this exam.

Palavras-chave: Cervical Squamous Cell Carcinoma, HSIL, LSIL, Colposcopy, Artificial Intelligence

(24624)

DEEP LEARNING AND MULTIDEVICE COLPOSCOPY: AUTOMATED DETECTION AND DIFFERENTIATION OF CERVICAL AND ANAL SQUAMOUS CANCERS PRECURSORS - A TRANSATLANTIC STUDY

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Introduction: Human papillomavirus (HPV) infection presents neoplastic risks in both the cervix and anus. With a growing interest in examining finer anatomical details, colposcopy has become vital in assessing not only the female genital tract but also the anal region. However, its complexity leads to a shortage of proficient physicians, especially in early diagnosis. This study aims to develop an artificial intelligence (AI) algorithm for identifying and differentiating HPV-related dysplastic lesions, specifically between low-grade and high grade squamous intraepithelial lesions (LSIL and HSIL, respectively) in both cervix and anus.

Methods: In this retrospective multicenter study, a Convolutional Neural Network (CNN) was developed using 295 colposcopy and anoscopy examinations, involving 3 different device types. A total of 80,167 frames were utilized, categorized as LSIL or HSIL based on pathological analysis. The dataset was split into training (90%, n= 71890) and test sets (10%, n = 8277) to assess model performance. Diagnostic metrics including sensitivity, specificity, accuracy, positive and negative predictive values (PPV and NPV, respectively) and an area under the receiving operating curve (AUC-ROC) were calculated.

Results: In the test set, the accuracy of the CNN in differentiating LSIL from HSIL is 93.9%, with 91.5% sensitivity, 96.1% of specificity, 95.6% of PPV and 92.4% of NPV. The AUC-ROC was 0.96.

Conclusions: HPV infection is ubiquitous and its impact extends beyond the cervical region. This study we developed a pioneer AI deep learning model, the first of its kind, for differentiating HPV-related dysplastic lesions, during both cervical and anal examinations. This model achieved promising results, suggesting its potential to improve detection accuracy and cost-effectiveness in clinical practice.

Palavras-chave: HSIL, LSIL, Colposcopy, Anoscopy

(24599)

MATERNAL FETAL SURVEILLANCE IN MIGRANT PREGNANT WOMEN: A PROSPECTIVE OBSERVATIONAL STUDY IN A TERTIARY CENTER IN 2023

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Introduction/Goals: The exponential growth of the immigrant population in Portugal, and the consequent increase in the number of pregnant migrants, has proved to be a real challenge for health services, particularly obstetrics.

Portugal provides maternal care to all pregnant women regardless of their nationality, but migrant pregnant women are at greater risk of insufficient follow-up, complications during labour and inadequate puerperal care, and these outcomes are often attributable to difficulties in accessing maternal care. Other limitations pointed out include the language barrier and the perception of unequal treatment by the health professionals who assist this population.

This study aims to understand the perception of migrant pregnant women followed at CMIN on their pregnancy surveillance.

Methods: Prospective observational study focused on migrant women who gave birth at CMIN in 2023. The analysis was based on the answers to an anonymous questionnaire.

Results: A total of 100 surveys were analyzed, with the majority being Brazilian (58.5%), followed by Angolan (4.9%), Spanish, Italian, Russian, Bangladeshi and Cape Verdean (3.7% each). Almost half of the women (45.1%) had lived in Portugal for less than a year.

The majority of respondents (61.7%) understood how to access healthcare in Portugal. As for the language barrier, the majority considered that they always understood the information they were given (75.3%) and felt well-informed during their pregnancy (94.9%), although 10.5% confessed to having given consent to procedures at some point without fully understanding them. Migrant mothers did not perceive unequal treatment from health professionals (80.2%). Around 76.3% felt that their requests were always taken into account, and 87.8% never felt pressured to do something they didn't want to.

Overall, the migrant mothers felt safe (92.4%) and satisfied (98.7%) with the healthcare they received, with only 10.1% considering that they would have had a better experience in their country of origin.

Conclusion: Although outcomes vary according to the country of origin and reception, there is a greater tendency for worse obstetric outcomes in migrant pregnant women. In recent years, there has been greater awareness of the racial disparity in maternal care, but still, little is known about the personal experiences of these women.

Our study shows very optimistic results, reflecting a collective effort by health professionals to create an inclusive, optimized and user-centered environment.

Palavras-chave: migrants, maternal, fetal

GINECOLOGIA - OBSTETRÍCIA

POSTERS

(24569)

OVARIAN AND FALLOPIAN TUBE TORSION IN PREGNANCY - A CASE OF SUCCESS

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Background: Adnexal torsion – the complete or partial torsion of the ovary and/or fallopian tube on its ligamentous support, often disrupting its blood supply – is one of the most common gynecological surgical emergencies. In pregnancy, its frequency seems to be higher than in non-pregnant women. Timely recognition allows for an early treatment, with benefits for fertility preservation and less associated morbidity.

Clinical case: A 26-years-old primigravida, with a spontaneous pregnancy of 8 weeks, was observed in the Emergency Department due to a 2-hour-evolution sudden left lumbar pain radiating to the pelvis, associated with nausea and an episode of vomiting. In the ultrasound (transvaginal and abdominal - in collaboration with Radiology), an in-utero pregnancy of a living embryo with a crown rump length for 8w1 d, a normal right ovary and a left ovary with 75*45mm, rounded, with edema, non-mobile, and with numerous peripherical cysts of less than 30mm, were observed. The pain was uncontrolled, despite analgesia with paracetamol, tramadol and butyl scopolamine.

Laparoscopy, due to a suspicion of left adnexal torsion, was proposed. Intraoperatively, an enlarged uterus (according to the gestational age), a normal looking right ovary and an edematous purple-colored left ovary (with a smooth-walled cystic lesion of 4 cm) and fallopian tube, twisted on their pedicle twice, were seen. The pedicle was distorted, the adnexa gradually recovered its normal color, and the cyst was punctured, with an outlet flow of transparent fluid.

The post-operatory period was uneventful, being the patient discharged home on 2 days after surgery. Due to this surgical procedure, the pregnancy was monitored in the obstetric clinic. The pregnancy was uneventful and ended with a vacuum-assisted birth at 39w2d of a male newborn with 34959 and an Apgar score of 9/10/10.

Discussion: The ultrasound multidisciplinary approach was essential for an early diagnosis of the adnexal torsion, minimizing the chances of a negative impact on the ongoing pregnancy. During the surgical procedure, if the corpus luteum was compromised or removed, then the pregnancy could have been negatively affected and eventually culminating in a first trimester loss.

Conclusion: The timely diagnosis of adnexal torsion allows for the appropriate treatment, enabling the preservation of the affected adnexa and the reduction of associated surgical morbidity, which is especially relevant in adnexal torsion during early pregnancy.

Palavras-chave: torção ovária, adnexal torsion. ovarian torsion, torão anexial, pregnancy

(24570)

RELATIONSHIP BETWEEN SOCIAL NETWORKS AND POSTPARTUM DEPRESSION

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Introduction: The puerperium is distinguished as a phase in women's life cycle of extreme importance in terms of the consolidation of the roles assumed by women, either at an individual level, as women, or at a family, social and professional level. It is distinguished by being a time of great physical, psychological, and emotional challenges, which may lead to changes in women's mental health, and there is therefore a risk of developing postpartum depression. Social networks have become platforms for the search of information and social support of high relevance to puerperal women.

Objective: To investigate the relationship between the use of social networks and postpartum depression

Methodology: A research was conducted on July 3, 2022, in the CINAHL and Pubmed databases for scientific publications of quantitative study types, which assess the use of social networks by puerperal women and postpartum depression.

Results: From a result of 536 publication, after applying the exclusion criteria, three articles were selected for analysis. Two articles relate the potential of publications on social networks to be a tool to identifying postpartum women the risk of postpartum depression, and the other study evaluates the impact of interventions carried out through social networks on depressive symptoms of postpartum women.

Conclusion: There are a few investigation with a high level of evidence that direct social networks as a resource to support postpartum women, fundamentally in postpartum depression. This review concludes the relationship between the use of social media and postpartum depression in two different ways: identifying postpartum women at risk of depression, and the effectiveness of interventions through social networks in postpartum women with depressive symptoms. The need for further experimental studies on this topic is identified.

Palavras-chave: postpartum depression; puerpera; social networks

(24612)

ADENOCARCINOMA OF THE UTERINE CERVIX: A CASE REPORT

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Introduction: Since the implementation of cervical screening, the incidence of cervical carcinoma has decreased dramatically. However, there has been a proportional increase of adenocarcinoma in relation to cervical intraepithelial neoplasia (CIN), especially at a young age, although its prevalence is significantly lower.

Established risk factors include infection with the human papilloma virus (HPV), mainly 16 and 18, and the use of oral contraceptives. Unlike CIN, adenocarcinoma is not associated with smoking, and the sensitivity of screening is lower.

The lesions of adenocarcinoma more commonly involve the glandular epithelium, unlike CIN which is almost always found in the transformation zone. Diagnosis therefore requires excisional procedures (conization) and endocervical curettage.

Clinical case: A 47-year-old non-Caucasian woman from Angola was referred to the gynecology appointment for abnormal uterine bleeding. During the gynecological examination, a globose and easily bleeding cervix was observed, which motivated HPV virus testing and cytology, which revealed endocervical adenocarcinoma and HPV 18 positivity. The woman was then referred to the Cervical Pathology Unit.

A colposcopy was performed which identified an extensive ectropion, with a lesion suggestive of invasive adenocarcinoma, with dense acetowhite areas, relief and atypical vessels. Biopsies were then taken from the lesion, the histological result of which revealed an adenocarcinoma in situ.

For diagnosis, a LASER and scissor conization was carried out, with subsequent endocervical curettage, which was uneventful.

Conclusion: Cervical adenocarcinoma is a rare disease with variable patterns on colposcopy and a tendency to extend into the endocervical canal. Colposcopic sensitivity is lower than for CIN. Therefore, definitive diagnosis requires excision of the lesion and evaluation of the margins.

Palavras-chave: Cervical adenocarcinoma, HPV, Colposcopy

(24602)

COMPARATIVE OF VERY ADVANCED MATERNAL AGE PREGNANCIES: SPONTANEOUS PREGNANCIES VS. ASSISTED REPRODUCTIVE PREGNANCIES.

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Introduction: Very advanced maternal age (≥ 45 years-old) is frequently associated with maternal and fetal complications. Despite the risks, the prevalence of pregnancies in this age group has increased in recent years, largely due to advancements in assisted reproductive technology. We aimed to compare the characteristics and outcomes between spontaneous pregnancies (ESP) and assisted reproductive pregnancies (ARP) in women with very advanced maternal age.

Methods: We performed a retrospective single-center cohort study which included pregnant women with follow up and labor in a tertiary portuguese hospital between 2019 and 2023.

Results: A total of 66 women were included (45,5% had fertility treatments) with a mean age of 46,3 ± 1,8 years. The ARP group was in average two years older, 63,3% were nulliparous (25,0% for ESP), 40% used donor oocytes and 6% were twin pregnancies. In general, 51% had comorbidities, mostly chronic hypertension (16,7% for ESP vs. 23,3% for ART). During pregnancy, 22,7% (22,2% for ESP vs. 23,3% for ARP) were diagnosed with gestational diabetes, 7,6% had preeclampsia (8,3% for ESP vs. 6,7% for ART) and 21,2% were hospitalized (11,1% for ESP vs. 26,7% for ART). The mean labor gestational age was 37,2 weeks (37,7 for ESP vs. 36,7 for ARP) and the percentage of cesarean deliveries was higher in the ART group (41,7% for ESP vs. 76,7% for ARP). The mean newborns' weight was 2,827 Kg (2,951 Kg for ESP vs. 2,677 Kg for ARP).

Conclusion: Very advanced maternal age pregnancies due to assisted reproductive technology are more likely to be older, primiparous, hospitalized in a maternal fetal unit and have a cesarean delivery. Knowing these outcomes may influence a better prenatal counseling and posterior pregnancy surveillance.

Palavras-chave: Assisted Reproductive Technology, Pregnancy, Very advanced maternal age

(24604)

GESTATIONAL WEIGHT GAIN AFTER POST-COVID-19 PANDEMIC

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Introduction: Maternal prepregnancy body mass index (BMI) and gestational weight gain (GWG) have an important role in maternal and neonatal outcomes. During the COVID-19 pandemic strategies were developed to fight sedentarism and recent studies show this period was not associated with greater GWG. We aim to compare the GWG during and after the COVID-19 pandemic.

Methods: GWG of pregnant women admitted in a tertiary portuguese hospital for delivery during January 2021 and 2023 were compared. Missing data, pre-term and twin pregnancies were excluded.

Results: 314 women (N = 155 COVID and N = 159 Post-COVID) with a mean age of 31,1 years-old (30,5 ± 5,6 COVID and 31,6 ± 5,8 Post-COVID) were included. The mean BMI was 25,5 Kg/m 2 (26,0 ± 6,0 Kg/m 2 COVID and 25,1 ± 4,9 Kg/m 2 Post-COVID) and the mean GWG was 13,6 Kg (13,6 ± 5,3 Kg COVID and 13,7 ± 5,9 Kg Post-COVID). There were no statistically significant differences in GWG between groups (p>0,05) and the excessive weight gain was similar (41,3% COVID and 41,1% Post-COVID).

Conclusion: The GWG was not affected by the COVID-19 pandemic and the percentage of women with excessive BMI and GWG is still very high. Preconception counseling and encouragement for exercise and diet during pregnancy surveillance are needed. In a digital era, better strategies such as technological tools must be developed for weight management.

Palavras-chave: COVID-19; Gestational Weight Gain; Pregnancy

(24606)

CONTRACEPTIVE CHOICES AFTER VOLUNTARY TERMINATION OF PREGNANCY

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Introduction: In Portugal, there is a promotion of universal access to family planning consultation, as well as free distribution of contraceptive methods with the intention to reduce unwanted pregnancy. However, an important number of voluntary terminations of pregnancy (TOP) occur a result of non-use of contraception or irregular use of the chosen method. The contraception counseling should be addressed since the beginning of the process and an efficient method should be provided as soon as possible, in order to prevent new unwanted pregnancies.

Objective: To compare the contraception use of women before and after a TOP in tertiary hospital.

Methods: Retrospective study between January 2017 and June 2023. All cases of TOP were recorded, and data retrieved. Cases with information about contraceptive use were included in the analysis. Statistical and descriptive analysis was performed using SPSS version 29 and a value of $p<0.05$ was considered statistically significant.

Results: A total of 4649 women underwent voluntary TOP during the described period. A total of 3276 cases fulfilled the inclusion criteria. Median age of women was 28.60 ±6.89 years [13, 47], with a median parity of 0.78 ± 0.98 [0,7] and an average gravidity of 2.21 ± 1,40 [1,14]. The TOP was performed between 4 weeks + 0 days and 10 weeks and 6 days, with a mean of 8 weeks ± 1.26. Women had an average of 0.29±1.26 [0,5] previous interruptions of pregnancy.

The most chosen method before and after the process was the contraceptive pill (45.8 and 42.8%, respectively). The most frequent motive of contraception failure reported by women was its irregular use (25.3%).

There was a significant reduction ($p< 0.001$) of women without contraceptive method (from 24.8% to 1.2%) or use of condom alone (from 23.8% to 2.1%) and a significant increase ($p<0.001$) of women who opted for long-acting reversible contraception (1.2% to 37.6%), being the IUD second method of choice after the TOP.

Conclusion: In this study, there was a significant modification of contraception use in women who underwent TOP, with an increase of very efficient methods like subcutaneous implant and intra-uterine devices, as reported previously in literature. These methods have the benefits of being user independent and not being subject to irregular use. An important number of women chose contraceptive pill as the contraceptive method after TOP, in which the counseling of correct use is central to prevent another unwanted pregnancy and the pills should be provided in the family planning consultation.

Palavras-chave: Contraceptive, family planning, voluntary terminations of pregnancy, long-acting reversible contraception**Obstetrícia | Casos Clínicos**

(24635)

LATE POSTPARTUM HEMORRHAGE IN THE CONTEXT OF SUSPECTED PLACENTA ACCRETA SPECTRUM: A CASE REPORT

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Introduction: Placenta accreta spectrum (PAS) is a generalized term that encompasses placenta accreta, increta, and percreta, resulting from abnormal trophoblast growth, which can adhere to the myometrium, invade the myometrium, and even penetrate the serosa, respectively. The surge PAS prevalence is primarily attributed to increasing cesarean section rates, especially significant in cases of prior cesarean section combined with placenta previa (risk of 3%). Even though its prenatal diagnosis remains difficult, PAS is associated with significant morbidity and mortality, particularly maternal.

Clinical Case: 39-year-old healthy women, G2P1 (with a previous cesarean section due to breech presentation) underwent her second pregnancy, resulting in a vaginal delivery. Antenatal care was provided by her private gynecologist. Her pregnancy appeared uneventful, except for the diagnosis of posterior placenta previa with multiple placental lakes in the second trimester ultrasound. Later transitioning to inferior inserted placenta. The patient had a spontaneous vaginal delivery complicated by an early postpartum hemorrhage that required manual uterine revision with suspected PAS, resulting in severe anemia requiring packed red blood cell transfusions. She was discharged 48h after the delivery, stable and with normal loquiae. However, on the 5th postpartum day, she presented with profuse vaginal bleeding, leading to hospitalization. Transvaginal ultrasound revealed a hyperechoic vascularized image, occupying the entire thickness of the posterior wall. Magnetic resonance imaging identified a posterior nodular lesion measuring 61 x 50 x 48 mm, involving the entire posterior myometrium, with intact serosa, suggesting a possibility of placenta increta. After discussion of treatment options, the patient opted for surgical intervention.

Conclusion: Early recognition and management of PAS are critical to reduce maternal morbidity and mortality associated with postpartum hemorrhage. Nonetheless, its prenatal diagnosis remains difficult. Healthcare providers must be aware of the risk factors and be vigilant in diagnosing PAS to improve patient outcomes.

(24614)

CONDYLOMA OF THE CERVIX: A CASE REPORT

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Introduction: Cervical condyloma is a presentation of genital condyloma, a sexually transmitted infection caused by the low-risk human papilloma virus (HPV) types 6 and 11.

It is estimated that 80% of cases occur between the ages of 17 and 33, however around 90% exposed to the virus will not develop condylomas and the majority, 80%, will eliminate HPV within 18 to 24 months.

Genital condylomas are mainly diagnosed clinically, making biopsy generally unnecessary, except in cases of immunosuppressed patients or when a malignant lesion is suspected. Typically exophytic, they can range from small lesions smaller than 5 mm to large masses in both the genital and anal regions.

Although often asymptomatic, cervical condylomas can cause itching, pain or bleeding. The preferred treatment for genital condylomas is destructive, with LASER vaporization indicated preferentially for extensive lesions**.**

Clinical case: A 25-year-old non-Caucasian woman, from Brazil, was referred to the Cervical Pathology Unit for vulvar lesions suggestive of condylomas and a large white lesion on the cervix. A cytology was performed and revealed Atypical squamous cells of undetermined significance (ASC-US) and high risk HPV.

A colposcopy was carried out at the Unit, which identified the cervix with an exuberant mass resulting from a group of condylomas affecting all quadrants.

A biopsy was carried out, due to the size and relief of the lesion, which confirmed the clinical suspicion.

Treatment with LASER vaporization was proposed, which went without complications, and the patient was advised was suggested to be vaccinated against HPV and to change her lifestyle habits in order to reduce risk factors.

Conclusion: Although the incidence of genital condylomas is declining, largely due to vaccination, there are still cases of exuberant lesions that require destructive treatment in the Unit. This case highlights the importance of early detection and appropriate treatment to prevent complications.

Palavras-chave: Condyloma, HPV, Colposcopy

(24669)

NEOPLASIA INTRAEPITELIAL DA VAGINA (VAIN) – UMA NOVA REALIDADE

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Introdução: O diagnóstico de Neoplasia Intraepitelial da Vagina (ValN) tem vindo a aumentar, como resultado do uso sistematizado da colposcopia e da implementação do rastreio para a presença de Vírus do Papiloma Humano (HPV). A ValN é um diagnóstico raro, com uma incidência estimada de 0,2 casos / 100 000 mulheres. A idade média ao diagnóstico encontra-se entre os 43-60 anos. Entre os fatores de risco encontra-se o antecedente de histerectomia por neoplasia intraepitelial cervical (CIN) de alto grau, pelo que doentes que desenvolvem ValN pouco tempo após cirurgia por CIN podem apresentar extensão vaginal da doença cervical que não foi detetada e tratada. As mulheres encontram-se, normalmente, assintomáticas, e a maioria dos diagnósticos é feita em exames de rotina.

Devido à sua raridade, a história natural da doença não foi ainda completamente caracterizada. No entanto, estudos retrospetivos sugerem que 2-8% dos casos vêm a progredir para carcinoma invasor da vagina.

Apresentamos um caso de mulher com antecedentes de histerectomia por patologia benigna, que manteve vigilância em rastreio, ainda que sem critérios para vigilância pós-operatória, com posterior achado de ValN e com seguimento ao longo de 10 anos.

Descrição Do Caso: Apresentamos o caso de uma mulher de 62 anos, com antecedentes de histerectomia total aos 30 anos por hemorragia uterina anómala em contexto de fibromiomas uterinas. Foi mantida em programa de rastreio (com realização de citologia e teste HPV), tendo sido inicialmente referenciada à consulta de Patologia do Colo da nossa instituição por persistência histológica de ValN2 em biópsia vaginal em 2019 (previamente com ValN1 desde 2011, até surgimento de ValN2 em 2016), tendo sido submetida a exérese da lesão com LASER, tendo o estudo anatomopatológico da peça revelado presença de ValN2/3.

A doente manteve vigilância em consulta, tendo realizado citologia, teste HPV e vaginoscopia em timing apropriado (com citologia NILM e posteriormente ASCUS, e HPV AR positivo para outros, que negativou em 2022). Voltou a realizar biópsia vaginal com achados de ValN1 em 2022.

Dados os achados e história clínica, a utente foi orientada para manutenção de seguimento no hospital da área de residência.

Conclusão: Mulheres com antecedentes de histerectomia por patologia benigna (não-CIN) não têm critérios para manter vigilância em rastreio, no entanto, são cada vez mais frequentes os casos de neoplasia intraepitelial da vagina diagnosticados neste contexto.

Devido à sua raridade, a história natural da doença não foi ainda completamente caracterizada, no entanto, devido à evolução científica e tecnológica na área da Patologia do Colo e do Trato Genital Inferior, tem sido cada vez maior a taxa diagnóstica. O follow-up é realizado normalmente ao longo de vários anos, devido ao risco de recorrência das lesões, e o risco de progressão para carcinoma invasor é ~2-8%.

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COMUNICAÇÕES ORAIS

(24560)

PUMPING BREAST MILK DURING HOSPITALIZATION IN PEDIATRIC INTENSIVE CARE UNIT- NURSING INTERVENTIONS TO LACTATING MOTHERS

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When a child is admitted to hospital, specially to pediatric intensive care unit, it is very common to occur interruption of breastfeeding because these infants/children are too sick to be breastfed and/or too sick to receive any type of enteral nutrition.

Breast milk production can be compromised in a few days due to the sudden interruption of breastfeeding so there is a need to establish an exclusive pumping of breast milk if the mother wants to continue breastfeeding after discharge.

There are not many studies about exclusive pumping and the information available comes from specialists in breastfeeding. So, is it important to have guidelines to support and encourage these mothers to pumping breast milk to maintain milk production.

As pediatric nurses working on a pediatric intensive care unit, nursing interventions should be divided in three areas: pumping equipment, pumping routine and *mindset*.

The selection of appropriate pumping equipment as well as establish a pumping routine are crucial to maximize milk production and to help maintain breastfeeding when the child is able to be breastfed again. As pediatric nurses, it's our responsibility to support these mothers as well as their child.

These mothers also need emotional support because sometimes they can be too overwhelmed with the health of the infant/child to think about breastfeeding. Therefore, it is paramount that the lactating mother is fully encouraged and supported by the health care professionals, especially nurses due to close care to the patients (child and family).

The benefits of breast milk are undeniable, specially to a critical ill child. It is important to support the lactating mother while her child can't be breastfed in order to retake breastfeeding when possible. Nurses should be able to guide mothers as they wish, to maintain or not breastfeeding afterwards.

Training pediatric intensive care nurses on this theme is essential to help mothers maintain breastfeeding during and after hospitalization.

Palavras-chave: exclusivepumping

(24581)

VULVAR CANCER – IMPACT ON WOMEN'S LIVES

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Introduction: Vulvar cancer is a rare gynecological neoplasm of the lower genital tract, representing 4% of all gynecological cancers. It mainly affects postmenopausal women, with a higher incidence in the 6th and 7th decade of life. However, there is currently an increase in the number of cases in young women, with invasive tumors diagnosed under the age of 40 representing 7%. This increase is attributed to the rising rates of Human Papilloma Virus infection, and is enhanced by smoking.

Non-specific symptoms, shame and embarrassment make women delay seeking help. The diagnosis of vulvar cancer can affect your physical and mental health, as well as your marital and social relationships, compromising your quality of life. The Specialist Nurse in Maternal and Obstetric Health assumes responsibility for interventions that allow you to reach your health's maximum potential.

This working group aimed to raise awareness of the impact of vulvar cancer on women's lives, and the importance of Specialist Nurse in Maternal and Obstetric Health in their care, as the lack of information/support that users with this pathology feel is notorious.

Methodology: This is an exploratory study of data collected through a retrospective literature search.

Results: In Portugal, in 2022, 196 cases of vulvar cancer were diagnosed, and projections indicate an increase in the number of new cases, according to estimates there will be 282 cases in 2050.

Estimates indicate that in Europe in 2022 there will be 17.500 cases, and in 2050 the number of cases of vulvar cancer will be 22.500.

This neoplasm can be curable when diagnosed at an early stage, with survival rates greater than 90%.

Discussion/Conclusion: Vulvar cancer is considered rare, but that does not mean it is any less important. Nursing interventions must focus on the physical, emotional and sexual aspects in order to minimize the negative impact, promoting and optimizing each woman's strategies to face the disease. Not forgetting that family is one of the fundamental pillars during this process.

The Specialist Nurse in Maternal and Obstetric Health has a determining role and can make a difference in each woman's life, empowering her with the aim of avoiding abandonment of her treatment, monitoring pre and/or post-surgery, empowering her in decision making.

Palavras-chave: vulvar cancer, nursing care, impact

(24584)

DATA MINING, MACHINE LEARNING AND ARTIFICIAL INTELLIGENCE – APPLICATIONS IN MIDWIFERY AND OBSTETRICS

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The use of artificial intelligence (AI) has experienced rapid growth over the past few years, in the field of medicine in general and in midwifery and obstetrics in particular. In parallel with the technological progress experienced worldwide, AI capabilities have largely improved, as new learning algorithms, theories and advanced computing power came into play, bridging the gap from theoretical models to practical use. Data mining techniques have been used to extract relevant data from abstract clinical data and less frequently with narrative clinical data. Machine learning encompasses methods of data analysis in order to produce models that can be used to represent and solve problems in several domains. Their main objective is user safety and evidence-based practice, being used to monitor human error, reasoning based on clinical cases and inducing rules for specialised electronic systems.

The aim of this review was to analyse research studies on data mining, machine learning, and intelligent systems applications in midwifery and obstetrics.

This review was performed in two databases (PubMed and CINAHL). Only articles that discussed data mining, machine learning or/and intelligent systems applications in midwifery and obstetrics were considered in this review. No time or language limit was applied.

Six articles were included in this review. The results suggest that data mining, machine learning and intelligent systems have produced successful applications for a broad list of midwifery and obstetrics topics, such as: gynaecology (e.g. cervical cancer diagnosis); assisted reproductive technology (e.g. implantation outcome of IVF, classification of sperm cells, embryo selection); pregnancy risk assessment (e.g. hospitalisation due to hypertensive disorders in pregnancy); labour progression and monitoring (e.g. forecast vaginal delivery in twins, fetal monitoring, predicting successful vaginal deliveries, preventing dystocia); postpartum risk assessment (e.g. incidence of postpartum haemorrhage) and neonatal/infant health (e.g. neonatal mortality prediction).

Despite some limitations, data mining and machine learning both represent a very promising area of AI applications for the development of practical and highly effective interventions that can support midwives in a variety of clinical domains. More research is needed to explore its vast potential.

Palavras-chave: data mining, machine learning, artificial intelligence, midwifery, obstetrics

(24598)

IMPORTÂNCIA DO PROCESSO FORMATIVO E COMPETÊNCIAS INSTRUMENTAIS DO ENFERMEIRO PERI-OPERATÓRIO NO CONTEXTO DA CIRURGIA ROBÓTICA

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The importance of the nurse's training process and instrumental skills in the context of robotic surgery

Authors:

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Theme of the work: The topic presented is in the field of knowledge "importance of the training process and instrumental skills of perioperative nurses in the context of robotic surgery".

The aim of the study is to identify the main challenges in the training process and the perspectives of peri-operative nurses in robotic surgery.

Introduction: According to the Mesh descriptors, robotic surgery is defined as "surgical procedures performed remotely using a computer that controls surgical attached to mechanical arms designed to perform those tasks of the surgeon." (2015).

Robotic surgery has revolutionized healthcare, providing greater precision, less invasiveness and faster invasiveness and faster recovery of the person, and the perioperative nurse is a highly trained professional who plays a crucial role in the success of the robotic procedure.

Methodology: An integrative literature review was carried out to prepare this study. The search for databases Academic Search Complete, CINAHL Compelte, Mediclatina and Medline Complete databases, using the EBSCOhost Web search engine done in April 2024. In order to understand the importance of the training process and instrumental skills in robotic robotic surgery, we administered fifteen questionnaires in the Operating Room of the Unidade Local de Saúde Santo António to peri-operative nurses who develop instrumental skills as a research tool.

Results: The analysis of the included studies and the questionnaire applied in the Operating Room of the Unidade Local de Saúde Santo António leads to the conclusion that the training process in the context of robotic surgery deepens knowledge and instrumental skills, such as: fundamentals of robotic technology, configuration and operation of the robotic system, specialized instrumentation, techniques for handling robotic surgical instruments and personal safety.

CONCLUSION:

The role of the perioperative nurse is dynamic and encompasses knowledge and mastery of new technologies, instrumentation techniques and the challenges involved in the use of robotic surgery, guaranteeing the safety of the person (Ulmer, 2010, cited by Martins Trevilato, Jost & Caregnato, 2019).

Palavras-chave: Educação, Enfermeiro Peri-Operatório, Procedimentos da Cirurgia Robótica, Competências Instrumentais

(24685)

DESAFIOS DA INTELIGÊNCIA ARTIFICIAL NA SAÚDE: A IMPORTÂNCIA DA QUALIDADE DOS DADOS NA VIGILÂNCIA EPIDEMIOLÓGICA

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Introdução: A Inteligência Artificial (IA) tem um potencial revolucionário na otimização dos cuidados de saúde e no suporte à tomada de decisão através da síntese rápida e essencial de informação. O desempenho dos modelos de machine learning e de deep learning, está fortemente relacionado com a quantidade, qualidade e diversidade dos dados que lhe são fornecidos. Portanto, para os modelos terem um bom desempenho são necessárias grandes quantidades de dados de treino diversificados e de boa qualidade. A experiência prática das autoras enquanto enfermeiras de controlo de infeção permite observar limitações significativas impostas pela qualidade dos dados no software de vigilância epidemiológica e nos registos clínicos consultados. Estas limitações estendem-se também ao potencial da IA para identificar padrões e riscos de forma precisa e eficiente e contribuir para a prevenção da transmissão cruzada da infeção.

Objetivo: Analisar os desafios e a importância da qualidade dos dados na aplicação da IA na vigilância epidemiológica.

Metodologia: Revisão da literatura dos últimos 5 anos, na base de dados Cinahl (via EBSCO), e Elsevier, considerando os descritores: “data quality”; “artificial intelligence”; “health care”; “clinical records”. Oito artigos foram selecionados pelo titulo e resumo. Análise reflexiva sobre a experiência prática das autoras, enfermeiras especialistas a exercer funções na UL-PPCIRA da ULSSA com a utilização de software de vigilância epidemiológica Hepic®, cuja base de dados é alimentada por múltiplos sistemas de informação.

Resultados: A integração da IA na gestão dos cuidados de saúde é incontornável para criar valor e promover o desenvolvimento das organizações. As análises preditivas tornaram-se ferramentas indispensáveis que permitem ao enfermeiro, através de tecnologias capazes de analisar conjuntos de dados complexos, monitorizar em tempo real, detetar precocemente alterações da condição do doente, desenvolver planos de cuidados personalizados e adaptados às necessidades individuais, prever as necessidades dos doentes e auxiliar na tomada de decisões clínicas, transformando os cuidados de saúde num sistema integrado, preditivo, preventivo e personalizado para todos. A promessa da saúde digital depende principalmente da capacidade de obter dados de qualidade. A IA combina a ciência da computação e conjuntos de dados para possibilitar a resolução de problemas. Utiliza algoritmos de machine learning e deep learning para fazer previsões ou classificações baseadas em dados de entrada, quanto mais robustos forem estes dados maior é a possibilidade de o modelo proporcionar melhores resultados. No entanto, o desempenho destes modelos está fortemente relacionado com a quantidade, qualidade e diversidade dos dados que lhe são fornecidos. Na área da saúde, a falta de dados de qualidade e representativos deve-se à fragmentação dos dados em diversas bases, falta de dados com anotações de especialistas e registos incompletos ou inexatos dos dados, o que pode levar o modelo a previsões incorretas. O Hepic® é capaz de identificar automaticamente doentes que compartilharam o mesmo espaço físico, permitindo a identificação de contactos de risco e oferecendo um potencial substancial para a modelagem preditiva de riscos de infeção. No entanto, enfrentamos desafios práticos como transferências fictícias, alocações de camas não registadas e erros administrativos que comprometem a integridade dos dados. Estes problemas não só limitam a capacidade de identificar contactos de risco, mas também restringem a aprendizagem efetiva por parte da IA.

Conclusões: A IA não só melhora a qualidade dos cuidados prestados pelos enfermeiros, como contribui para intervenções oportunas, reduzindo a probabilidade de eventos adversos e complicações. A qualidade dos dados é um fator crítico para o sucesso dos modelos de IA. É necessário um investimento em dados robustos e equipas multidisciplinares que estabeleçam as bases e adotem as melhores práticas para reduzir o viés algorítmico. O investimento nesta área, através da otimização dos registos clínicos e dos sistemas de informação e respetiva interoperabilidade resultam em melhoria imediata para os cuidados de saúde e contribuição significativa para o processo de inovação tecnológica com recurso à IA.

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Palavras-chave: Qualidade de dados, inteligência artificial, cuidados de saúde, registos clínicos

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POSTERS

(24675)

ARTIFICIAL INTELLIGENCE IN NURSING AND ITS IMPACT ON PATIENT CARE

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Introduction: Artificial intelligence (AI) is playing an increasingly important role in nursing, bringing significant innovations and improvements to patient care.

Objective: To understand how AI-powered tools can have an impact on patient care.

Methods: Integrative review of scientific literature by searching SIECLO and PubMed electronic databases. The inclusion criteria were papers published between 2019-2024, available in full text, in Portuguese and English.

Results: Nursing, a profession historically based on empathy, care and personalised attention, finds in AI a complement that can enhance the skills of nurses and at the same time help them simplify routine tasks, reduce errors and the entire workload, allowing more time for direct care of users and consequent improvement of the care provided. One of the applications of AI in nursing has been the development of decision support systems to help nurses make informed decisions by analysing data to manage complex cases where there are a variety of factors.

Discussion/Conclusion: In the literature there is strong evidence of AI in nursing in the future, with a strong impact on healthcare, not only in personalising care for users, but also in improving safety. There are, however, some constraints relating to important ethical issues to be considered that, regardless of the tool used, have to be safeguarded, namely user privacy and data security. Finally, under no circumstances can AI replace the touch, empathy and human connection that nurses provide to their patients. Finding the right balance between the use of AI and human care is key to maintaining high-quality healthcare.

Palavras-chave: Artificial intelligence, nursing, nursing care

(24676)

BURN MANAGEMENT IN THE PEDIATRICS OUTPATIENT CLINIC – ABOUT A CLINICAL CASE

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Introdução: A gestão da queimadura na criança visa o tratamento da área lesada, considerando a frequência na realização dos tratamentos, o conforto da criança e controle da dor, a minimização de sequelas, os aspetos hemodinâmicos e nutricionais e a prevenção de complicações.

Objetivos: Demonstrar a relevância da avaliação, escolha do material e execução dos tratamentos na gestão da queimadura em pediatria, para a rapidez na cicatrização e ausência de sequelas.

Relatar um caso clínico de queimadura profunda parcial, seguido na consulta de cirurgia pediátrica do CMIN.

Metodologia: Apresentamos um caso clínico, de cariz longitudinal, referente a uma queimadura, resultante de lesão acidental com líquido fervente, numa criança 2 anos, saudável, sem antecedentes relevantes. A colheita de dados realizou-se por observação direta, consulta ao SClínico e registo fotográfico ao longo de 4 meses. Após a lesão no tórax com líquido fervente, a criança foi conduzida ao serviço de urgência do ULS São João, executado o primeiro tratamento D1, e referenciado para a consulta externa de cirurgia pediátrica do CMIN. O acompanhamento do caso no CMIN iniciou a D2, manteve-se até D24 na execução dos tratamentos, com posterior vigilância na consulta. O tratamento implantado considerou o tipo de tecido danificado, gestão da dor, gestão do exsudado e pele periférica. Foi utilizada uma enzima alginogel que se mostrou eficiente nas fases de cicatrização. As áreas foram monitorizadas através de avaliação da área lesada, registo clinico e registo fotográfico.

Foram utilizadas técnicas farmacológicas e não farmacológicas para o controle da dor e colaboração da criança durante os tratamentos. O estado emocional, a aceitação do processo, o ensino para prevenção de complicações, foram alguns dos aspetos trabalhados em todos os tratamentos com os pais.

Utilizado o método expositivo e fotográfico na apresentação do caso.

Resultados: Eficácia na execução do tratamento com a utilização de enzima alginogel, melhoria rápida e significativa do tecido cutâneo e epitelização completa. Follow up aos 4 meses, cicatrização sem sequelas.

Conclusões: A utilização da enzima alginogel, a avaliação e monitorização rigorosas da área queimada com programação eficiente dos tratamentos em função da evolução da queimadura permitiu completa cicatrização.

A aplicação de técnicas não farmacológicas e a execução dos tratamentos com enfermeiros de referência permitiu total colaboração da criança, satisfação dos pais e profissionais.

Palavras-chave: Burns, Nursing, Child

(24680)

OESOPHAGEAL ATRESIA IN NEWBORNS: WHEN AND HOW TO USE THE REPLOGLE TUBE

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Introduction: Oesophageal atresia is the most common congenital malformation of the upper digestive tract. It is defined as an anomaly of embryonic development that results in the interruption of the oesophagus, associated or not with a tracheo-oesophageal fistula. The use of a replogle tube is indicated if surgical correction is impossible within 48 hours of life, due to the characteristics of the malformation, namely the existence or not of a tracheoesophageal fistula or the distance between the tops, which are sometimes very far apart, making anastomosis impossible. The Replogle tube is a double-lumen tube, inserted through the nostril to the proximal top of the esophagus and kept under continuous suction in order to avoid bronchoaspiration of saliva. This procedure requires continuous nursing intervention and care.

Objectives: To establish a nursing protocol for the use of the Replogle continuous suction tube. To standardize care for newborns diagnosed with oesophageal atresia, in relation to the use of the Replogle tube.

Methods: Integrative review of the scientific literature by searching the electronic databases SIECLO, PubMed and Uptodate. The inclusion criteria were papers published in 2017-2024, available in full text, in Portuguese, Spanish and English.

Results: The bibliographic research will allow the updating of the protocol for the use of a Replogle probe in newborns with esophageal atresia.

Discussion/Conclusion: The suspected diagnosis of esophageal atresia usually occurs in the third trimester of pregnancy, based on indirect echographic signs. However, the predictive value of these signs is low, so only fetal magnetic resonance imaging or birth will guarantee a diagnosis. In the neonatal period, the presence of persistent airy sialorrhea in the mouth and/or nose, associated with signs of respiratory difficulty and the impossibility of progressing the gastric tube are considered warning signs. The neonatal diagnosis is confirmed by the observation on the chest X-ray of the radiopaque orogastric tube wrapped around the upper esophageal pouch. The definitive treatment is surgery. However, when surgery needs to be postponed, airway patency is ensured by continuous suctioning at low pressure, using a Replogle tube and raising the incubator headboard. The implementation of an action protocol for the use of the replogle probe guarantees standardized nursing care.

Palavras-chave: Atresia Esofágica

(24681)

APPLICATION OF ARTIFICIAL INTELLIGENCE IN THE HEALTH LITERACY OF PARENTS OF PRETERM NEWBORNS

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Introduction: The use of Artificial Intelligence (AI) in healthcare is expanding rapidly, and one of its most promising applications is in improving health literacy. As a topic of growing popularity due to the relevance and impact it has on healthcare, it is considered one of the priority objectives, with specific action plans by the World Health Organization and the Directorate-General for Health. In the context of neonatal care, health literacy becomes crucial for parents to understand the need for care for their preterm children, and is one of the main challenges faced by nursing professionals.

Objectives: To map and analyze the scientific evidence in the literature on the importance of using AI to promote health literacy as a facilitating strategy in the transition to parenthood in neonatal intensive care.

Methods: A descriptive, reflective and critical approach was favored, based on scientific evidence. A search was carried out using all the keywords and indexing terms identified as relevant in the EBSCO and MEDLINE databases. The inclusion criteria were studies published in 2017-2024, available in full text, in Portuguese, Spanish and English.

Results: Nurses' clinical practice in neonatal intensive care units must ensure that communication and discharge planning take into account and promote parental health literacy. Preparing for discharge is complex and multidimensional, taking into account the need to acquire parental knowledge and skills that guarantee the security and confidence needed to transition and autonomously care for the child at home, and considering the parents' individual, family, social and cultural characteristics.

Discussion/Conclusion: Promoting health literacy in neonatology is an important determinant of newborn health. The use of AI allows parents' knowledge and skills in health matters to be strengthened, contributing to the safe and autonomous provision of differentiated care for their child at home related to prematurity.

Palavras-chave: Artificial intelligence, health literacy, parents, nursing, neonatology

(24683)

DOES ARTIFICIAL INTELLIGENCE OPTIMISE THE MANAGEMENT OF NURSES' WORK SCHEDULES IN HOSPITAL?

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Introduction: The President of the European Parliament has stated that, despite the challenges posed by artificial intelligence (AI), we should not fear the future, emphasising that any technological advance must respect fundamental rights and democratic principles. Although AI cannot digitise human emotions, will and decisions, its applicability in the healthcare context has the capacity to free clinical resources from administrative tasks related to document management, allowing for greater interaction with users. The application of AI in drawing up nursing work schedules has the potential to transform hospital management, bringing efficiency, employee satisfaction and improved patient care.

Objectives: To reflect on the applicability of AI in the management of nurses' work schedules in hospital.

Methods: Integrative review of the scientific literature by searching the SIECLO and PubMed electronic databases. The inclusion criteria were studies published after 2020 and available in full text.

Results: The application of AI in drawing up nursing work schedules offers a number of significant benefits for hospital management and for the healthcare professionals themselves. AI makes it possible to organise shift distribution more efficiently by adjusting schedules in real time. It can also take into account nurses' individual preferences and restrictions, resulting in more balanced and personalised schedules, avoiding overload and promoting a healthier working environment. In addition, automating the preparation of schedules reduces human error and ensures compliance with labour regulations and policies. Adequate provision of nurses also improves the quality of patient care. Implementing AI in nursing scheduling faces challenges, such as integration with existing scheduling systems, ethical and privacy issues, and the need for ongoing maintenance and updating to reflect changes in policies and legislation.

Conclusion: AI has the potential to reinvent experiences and applications, creating new opportunities and improving operational efficiency and productivity in hospitals. However, the implementation of AI faces challenges such as a lack of specialised skills, ethical issues and the need to invest in infrastructure and data security.

Palavras-chave: Artificial Intelligence, Shift Work Schedule, Nursing

(24688)

VIRTUAL REALITY AS A NON-PHARMACOLOGICAL STRATEGY FOR CHILDREN PAIN

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Virtual reality as a non-pharmacological strategy for children pain relief in a hospital context undergoing invasive and painful procedures: scoping review

Background: Distraction is often used as a strategy for managing pain in children associated with health procedures. Virtual Reality (VR), as an advanced digital model, allows children to be immersed in a controlled virtual environment, with three-dimensional vision and real-time movement. The child is distracted from their nociceptive sense, and there may be a reduction in the perception of pain during painful procedures.

Goal: Identify the scope of available evidence on the effectiveness of using VR as a non-pharmacological strategy for children pain relief in a hospital context undergoing invasive and painful procedures.

Methods: This "scoping" review followed the guidelines released by the Joanna Briggs Institute. We used the databases through the EBSCOhost Research Databases platform.

As a research strategy, were used a Boolean phrase "virtual reality an pain control" limited to the title and abstract. The search was only restricted to studies that included children (<18 years old).

23 articles were identified that were subject to analysis by title and abstract, 12 articles were excluded and 11 articles were included for content analysis in this scoping review.

Results: The majority of articles (10) focus on randomized controlled clinical trials. Only one of the articles addresses a systematic review with meta-analysis.

There is a growing use of VR as a children pain relief strategy in hospital settings undergoing invasive and painful procedures. VR is a multi-sensory technology that allows children to immerse themselves in a virtual world. Studies highlight that VR creates a non-pharmacological form of analgesia, altering the activity of the pain modulation system. In this way, the person pays less attention to painful procedures, reducing the input of negative and painful stimuli.

Studies show that using VR during blood collection is a method that reduces the duration of crying and increases adaptation to the procedure. The studies allow identifying barriers/facilities in the use of VR, particularly those associated with the use of the equipment, the physical and emotional condition of the child, the physical and human environment (health professionals and family members).

Conclusions: The use of VR reduces pain and anxiety during invasive procedures in hospitalized children. Knowledge of the barriers and ease of using VR as a children pain relief strategy allowed us to find strategies that guarantee the successful implementation of VR in pediatric services.

Palavras-chave: Virtual Reality; Pain; Children

(24689)

ARTIFICIAL INTELLIGENCE: REFLECTION ON ITS APPLICABILITY IN PALLIATIVE CARE

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Artificial Intelligence: Reflection on its Applicability in Palliative Care

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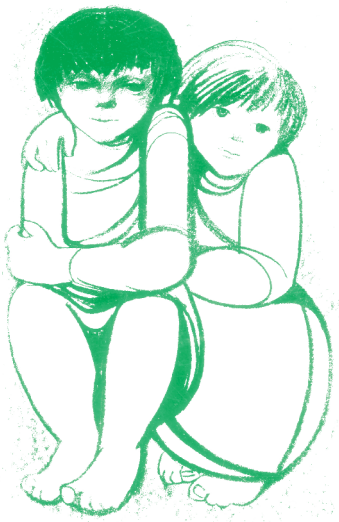
Objective: To reflect on the use of artificial intelligence in Palliative Care, analyze the present and envision the future.

Methodology: Theoretical reflection elaborated through bibliographic research carried out in the databases via EBSCO through the website of the Order of Nurses. Reading, analysis and interpretation of scientific articles on the use of Artificial Intelligence in Palliative Care were carried out. Results: Artificial intelligence should be seen as an emerging potential in palliative care, enhancing the four fundamental pillars: symptom control, organization of care and teamwork, communication and family support, improving accessibility for citizens, creating referral and decision algorithms and overcoming barriers of geographical dispersion.

Palliative Care Professionals should assume the roles of technology coordinators, in a philosophy of continuous improvement of the quality of care for people with palliative care needs.

Descriptors: Palliative Care; Artificial intelligence.

Palavras-chave: Palliative Care; Artificial intelligence.



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