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GENDER DIFFERENCES IN E-CIGARETTE USE AMONG YOUTH: A COMPREHENSIVE SURVEY ANALYSIS

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Introduction. Electronic cigarettes, considered as safer than combustible cigarettes, simulate smoking by heating nicotine or tobacco into vapours. Their growing popularity among teens and young adults raises health concerns, with studies linking them to cardiovascular, respiratory, immune issues, lung injuries, and a potential gateway to combustible smoking and substance addiction.

Objective. The aim of this study is to explore the underlying motivations among young individuals to use e-cigarettes and to uncover gender differences in usage patterns.

Methods. The quantitative study sample included individuals up to 25 years old, without differentiation based on gender or specific location. A comprehensive online survey was conducted in November and December of 2022 using a questionnaire designed by the author. Following the preparation of the online survey, it was shared on youth-oriented social platforms. The statistical analysis was performed using "Microsoft Excel" and "R Studio" software. To analyse the data, the Chi-square test was conducted.

Results. The survey, which included 1,335 respondents up to 25 years old, revealed significant gender difference in initiation patterns, with a higher percentage of females (84.2%) than males (80.1%) trying electronic cigarettes, often starting in adolescence. Recent use was reported by 80.3% of respondents, indicating widespread consumption. The survey also found a preference for higher nicotine concentrations and different acquisition patterns between genders, with males preferring specialized shops and online sources, and females more often obtaining electronic cigarettes through social connections. The main reasons for starting included social influence, stress relief, and flavour variety. Reported symptoms post-use varied by gender, indicating distinct physiological impacts.

Conclusions. The findings highlight a gender variance in electronic cigarette usage among the youth, with a higher inclination observed in females. The trend underscores the need for gender-targeted prevention strategies, especially for females, to limit early exposure. The significant recent electronic cigarette use among youths, coupled with a preference for high nicotine content, signalling a concerning trend towards addiction and emphasizing the need for heightened awareness of nicotine risks. Moreover, gender-specific adverse reactions following electronic cigarette necessitate personalized health education and interventions to address these distinct physiological reactions effectively.

Keywords: electronic cigarettes; e-cigarettes; smoking; carcinogens; nicotine; adolescent; youth; young adults.

BURDEN OF RSV INFECTION AMONG PEDIATRIC PATIENTS IN THE POST-LOCKDOWN PERIOD: A SINGLE CENTER EXPERIENCE

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Introduction: Respiratory syncytial virus (RSV), a leading cause of acute lower respiratory tract infections in infants and young children, puts a high pressure on the health care system. Implementation of COVID-19 quarantine interventions in many countries, including Lithuania, resulted in a strong reduction of RSV activity in 2020-2021, followed by a rapid return of RSV cases to pre-pandemic levels in the post-lockdown period.

Objective: This study aimed to evaluate the burden of RSV infection among pediatric patients in the post-lockdown period (in 2021-2022 and in 2022-2023 cold seasons).

Methods: A single-center retrospective cross-sectional study was performed at the pediatric emergency department in Vilnius during two cold seasons – from October 1, 2021, to April 30, 2022 (Season I) and in the same period in 2022-2023 (Season II). Patients under 18 years, who had been tested for RSV, were enrolled in the study. Nasopharyngeal swabs were tested for RSV using real-time reverse-transcriptase polymerase chain reaction (RT-PCR) tests.

Results: RSV PCR tests were positive for 14.5% of tested children (1021/7058). The infection predominated among patients under the age of 3 years (67.8%), especially infants (31.1%). Positive tests increased from 8.8% (430/4875) in Season I to 27.1% (591/2183) in Season II. More than a third of patients (38.3%, n=391) were hospitalized, 7.2% of them (n=28) required treatment in pediatric intensive care unit (PICU). The proportion of RSV hospitalizations was higher in Season II (33.7% vs. 41.6%, p=0.01). Patient rates in PICU did not differ between seasons (p=0.450). More cases of X-ray confirmed pneumonias were diagnosed in Season II compared to Season I (23.6% vs. 14.2%, p=0.010). RSV types were specified in 65 patients. Type A prevailed in Season I (56.7%, 17/30), and type B – in Season II (88.6%, 31/35). Type B tend to be more associated with pneumonia (33.3% vs. 28.6%) and treatment in PICU (25.0% vs. 9.5%) compared to type A, but the differences were insignificant ($p>0.05$).

Conclusions: Burden of RSV infection was high among pediatric patients in the post lockdown period, especially in 2022-2023 (Season II), as there was higher number of cases, more RSV infected patients with pneumonia and higher hospitalization rates, compared to 2021-2022 (Season I). Children under three years of age, particularly infants, were the most vulnerable group. Type B dominated in Season II and there was a trend for type B to be associated with pneumonias and hospitalizations in PICU.

Keywords: RSV; children

HEAVY METALS IN SCHOOL ENVIRONMENT AND ATOPIC DERMATITIS

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Introduction: One of the most common skin diseases in childhood is atopic dermatitis (AD). AD is caused by multiple factors including air pollutants (like heavy metals), especially in house dust. Our hypothesis is that AD is associated with higher concentrations of heavy metals in environment.

Objectives: To compare the incidence of AD and eczema in children between schools with higher concentrations of vanadium and lead and schools with lower concentrations.

Methods: The results from a study of aerosol pollution and concentration of heavy metals in dust in Vilnius schools were extrapolated. Concentrations of microelements from 11 schools were collected in 2017-2018. Atopic dermatitis morbidity data of years 2017-2020 was provided by the National Institute of Hygiene. The pediatric population of 6 -11 years was analysed. Chi Square test of independence was used to calculate the statistical significance.

Results: The highest concentration of vanadium was found in Zaros gymnasium 52.09 ppm., the lowest was in Baltupiu school, 12.69 ppm. The highest concentration of lead was found in Nemunelio school, 504 ppm, the lowest was in Grigiskiu gymnasium, 14 ppm. In Zaros gymnasium (population 280) the relative AD morbidity in 2018 was 6,79, while in Baltupiu school (population 463) it was 3,67 ($p=0,35$; $\alpha=0,05$). In Nemunelio school (population 306) the relative AD morbidity in 2018 was 6,21 compared to Grigiskiu gymnasiums 4,62 (population 130), ($p=0,76$; $\alpha=0,05$).

Conclusions: Relative morbidity in Baltupiu school is lower than in Zaros gymnasium in 2018, as is it is lower in Nemunelio when compared to Grigiskiu school, however, it is not statistically significant. The results may have been impacted by locations, both Nemunelio and Zaros schools are within 1 kilometer from railways, and Zaros is nearby various industries, while Grigiskiu school is in outskirts of Vilnius, outside the city main. Other pollutants and heavy metals also may have affected the results. There is a need of a larger scope studies to assess the association between AD and heavy metals in environment.

Keywords: atopic dermatits, air pollution, heavy metals, children.

PARENTAL NONADHERENCE TO THEIR CHILDREN'S EPILEPSY TREATMENT PLAN

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Introduction: Parental nonadherence to their child's epilepsy treatment plan may result in reoccurrence of seizures. It may be unintentional, such as healthcare inaccessibility, or intentional, parents not following the treatment plan in fear of side effects, poly-therapy and multiple dosing being too much of a burden.

Objectives: To examine adherence to antiepileptic drugs (AEDs) in a cohort of parents, whose children have epilepsy, and assess the barriers to treatment from their perspective.

Methods: A cross sectional study was conducted using a questionnaire. 46 parents of children, being treated in a tertiary centre for epilepsy, answered questions assessing demographics, usage of various medications and alternative medicine methods, individual barriers to adherence and their beliefs surrounding epilepsy treatment.

Results: 91.3% of respondents were female (mean age 38.63 (SD±5.86) years). The children's mean age was 7.84 (SD±3.57) years and 54.3% of them were female. 47.8% of children had focal seizures, 37% – generalized seizures and 15.2% of parents did not know what type of seizures their child experienced. On average, the children took 2.33 (SD±1.48) AEDs and 2.13 (SD±0.911) food supplements. 91% of respondents experienced difficulties adhering to their children's epilepsy treatment plan. The most common problems were (1) AEDs having an unpleasant taste (45.65%) (2) or being too difficult to swallow (30.44%), (3) some experienced obstacles when buying AEDs (23.9%). 20% had difficulty getting the prescription from their doctor and 11% had missed a dose because they ran out of AEDs. 2% believed AEDs were not necessary to control their child's epilepsy, 22% thought the prescribed medications were not effective, and 39% believed food supplements help keep epilepsy under control. The amount of AEDs and food supplements combined negatively correlated with parent's age ($p=0.021$), while the amount of food supplements used positively correlated with their education ($p=0.021$). The amount of AEDs depended on the frequency of seizures ($p=0.002$). Children with focal seizures took significantly less food supplements than those with generalized seizures ($p=0.007$).

Conclusions: Most parents experience difficulties adhering to their children's epilepsy treatment plan. Education, age and seizure type plays a role in the amount of medications and food supplements children with epilepsy receive, however only the amount of seizures makes a difference in how many AEDs they take.

Keywords: Epilepsy, antiepileptic drugs, adherence to treatment.

UNDERSTANDING DYSLIPIDEMIAS IN PEDIATRIC POPULATION: PROGRESS REPORT FROM AN OBSERVANT PREVALENCE STUDY

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Introduction. The main etiological factor of early morbidity and mortality from cardiovascular diseases is atherosclerosis, which develops already in childhood. Atherosclerosis at a young age is caused by dyslipidemia.

Objectives. The aim of the study is to determine the prevalence of dyslipidemia in the population of children aged between 5 and 10 years, determine the causes, characteristics of dyslipidemias, and the relationship with risk factors.

Methods. Children aged 5-10 years were included in the biomedical study "Prevalence study of dyslipidemias in children" conducted at Vilnius University Hospital, Santaros Clinics. A questionnaire inquiring about sociodemographic characteristics medical histories, detailed histories of diet, physical activities was completed. Body composition analysis data was evaluated using the machine TANITA MC-780MA-N: weight, fat mass (kg; %), trunk fat content (kg; %), muscle mass (kg), BMI. Standard lipid panels were also evaluated. The study protocol was approved by the Vilnius Regional Biomedical Research Ethics Committee of Lithuania (No. 2023/1-1497-955). Written informed consent to participate in this research was obtained from their parents or legal guardians.

Results. A total of 54 participants were included in the analysis, with a median age of 7.21 (5; 10) years, and boys accounting for 50%. Girls were non-significantly younger (7.1 y. vs. 7.3 y., p=0.637) with a higher BMI (16.5 (14.35; 17.05) kg/m2 vs. 16.2 (14.25; 17.65) kg/m2, p = 0.965). Increased total cholesterol was measured for 9 (17.3%), LDL-Ch for 10 (19.2%), TG for 0 patients, decreased HDL-Ch for 6 (11.5%) patients. There was a total of 17 patients with dyslipidemia. 9 (52.9%) of those had a family history of cardiovascular diseases. Body composition analyzer of children with dyslipidemia (N =17) revealed the following factors: a median weight of 27.5 (23.5; 48.7); fat mass 5.8 (4.5; 14.4) kg; trunk fat content 2.5 (1.8; 5.9) kg; BMI 16.4 (14.8; 22.0) kg/m^2; waistline 54,5 (53.0; 70.5) cm. Median body weight was significantly higher by 2.8 kilograms in children with dyslipidemia (p=0.046). Comparing these two groups, significant differences were also found between the medians of fat masses (5.8 (4.5; 14.35) kg vs. 4.7 (3.8; 6.3) kg, p=0.033) and trunk fat content (2.5 (1.8; 5.9) kg vs. 1.9 (1.6; 2.7) kg, p=0.035). Increased BMI (>85thpercentile) is statistically significantly different between children with and without dyslipidemia (58.3% vs. 41.7%) p=0.031. Our questionnaire revealed that there were significant differences between diets. Children with dyslipidemia consumed less vegetables (17.6% vs. 57.1%; p=0.007) and fruits (0% vs. 22.6%; p=0.032) than children without dyslipidemia.

Conclusions. Our study highlights the concerning prevalence of dyslipidemia among children, with approximately one-third of participants exhibiting abnormal lipid profiles. Children with dyslipidemia displayed different characteristics such as higher body weight, increased fat mass, and lower consumption of vegetables and fruits compared to healthy children.

Keywords. Dyslipidemia, children, causes, risk factors.

DIAGNOSIS AND OUTCOMES OF CRITICAL CONGENITAL HEART DISEASES FROM ANTENATAL TO NEONATAL PERIOD IN LATVIA FROM 2018 TO 2022

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Introduction. Critical congenital heart disease (CCHD) is a severe condition that requires surgical intervention in the first year of a child's life. Approximately one in four infants with congenital heart defects are diagnosed with a critical form, which can lead to cardiogenic shock and death if left untreated. Fetal echocardiography is the main diagnostic tool for detecting
CCHD, followed by postnatal pulse oximetry screening within 24 hours of birth.

Objective: Determine the prevalence of CCHD and investigate cases identified during antenatal and pulse oximetry screening, and subsequently, the long-term outcomes of the disease.

Materials and Methods: A retrospective study using the Children's Clinical University Hospital (CCUH) electronic system "Andromeda", including medical histories and outpatient records of patients observed or/and treated at CCUH from 2018 to 2022.

Results: A total of 167 patients were analysed, of whom 62.3% (n = 104) were boys, 35.3% (n = 59) were girls and in 2.4% (n = 4) the sex was not specified. Continued pregnancies were present in 65.3% (n = 109) and terminated pregnancies or foetus mortus in 34.7% (n = 58). 70.1% (n = 117) of CCHDs were detected during prenatal screening, with the highest proportion of 29.3% (n = 49) between 18 and 22+6 weeks gestation. Pulse oximetry screening was positive in 60% of 25 known cases. Most common were duct dependent systemic blood flow lesions 41.3% (n = 69) and duct dependent pulmonary blood flow lesions 38.3% (n = 64). Prostoglandin E1 infusion was administered in 39.4% (n = 43) of neonates and 50.5% (n = 55) of patients underwent surgery in the first month. The percentage of *exitus letalis* was 9.5% (n = 9), with boys predominating at 55.5% (n = 5) and newborns with hypoplastic left heart syndrome predominating at 33.3% (n = 3).

Conclusions: Newborns with CCHD who are diagnosed early and treated appropriately have a better chance of a favourable prognosis and long – term outcomes.

Key words: CCHD, pulse oximetry screening.

GENETIC SPECTRUM AND CLINICAL PRESENTATION OF AUTOINFLAMMATORY DISEASES: A SINGLE CENTER STUDY

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Introduction. Autoinflammatory diseases (AIDs) are a group of diseases caused by dysregulation in the innate immunity. Since clinical manifestations are varied and the disease can often be polygenic or multifactorial, the diagnosis delay may occur, which leads to severe complications like amyloidosis.

Objectives. To determine the frequency of autoinflammatory disease genes among children with suspected inborn errors of immunity (IEI) and to evaluate clinical manifestations in children with the presence of genetic variants of AIDs.

Methods. Sequence analysis and deletion/duplication testing of the 429 genes in the Primary Immunodeficiency Panel were conducted in 33 patients with suspicion of IEI, among whom 7 had suspected AIDs. The genetic testing was carried out in the laboratories of Invitae (USA) and Veritas (Spain) with the support of the Jeffrey Modell Foundation.

Results. Among the 33 children who underwent genetic testing, autoinflammatory gene variants were identified in 8 (24.2%) children, including 4 with suspicion of AID. The age of the children ranged from 3 to 16 years. Among the 3 children with suspected AID who did not have any AIDs gene variants, one had a systemic JIA, one had recurrent pericarditis, and one had PFAPA syndrome. In three children, variants in the *MEFV* gene associated with Familial Mediterranean Fever (FMF) were identified. In 2 of them, the same variant c.2084A>G (p.Lys695Arg) was found and classified as a variant of uncertain significance (VUS). In only one case, there was suspicion of AID. In one child with a pathogenic variant in *SERPING1*, another pathogenic variant (c.2082G>A(p.Met694Ile)) was identified in *MEFV*, although clinical manifestations of FMF was not observed in the boy. Variants in the *PLSG2* gene were revealed in two children. In one case, a diagnosis of systemic JIA was established, while in the other, suspicion of immunodeficiency arose due to severe meningoencephalitis caused by opportunistic infections. Variants in the *NLRC4* gene were detected in two other children with clinical features of PFAPA syndrome.

Conclusions. The variants of only four autoinflammatory genes were identified in 24.2% of children with suspected IEI and in 57.1% of children with suspected AIDs. Correspondence with clinical symptoms was observed in 50% of patients with variants of uncertain significance, indicating that they might contribute to susceptibility to autoinflammation.

Keywords: autoinflammatory diseases, inborn errors of immunity, PFAPA, systemic JIA, FMF.

PREDICTORS OF NEONATAL OUTCOMES IN PRETERM NEWBORNS WITH HYPOXIC INJURY OF THE CENTRAL NERVOUS SYSTEM

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Introduction. Neonatal hypoxic–ischemic brain injury is a leading cause of child mortality and morbidity, including cerebral palsy, epilepsy, and cognitive disabilities. Currently, the possibilities of timely diagnosis and treatment of prematurely born children with perinatal damage to the central nervous system are limited.

Objective. To clarify the possible role of malone-D-aldehyde (MDA) and sialic acids (TSA) urine concentrations in the early diagnosis and outcomes of hypoxic-ischemic CNS injury, by identifying associations with the clinical consequences of intrauterine hypoxia in preterm neonates.

Methods. The study included 100 preterm newborns: main group (n=50) – with hypoxic ischemic CNS injury, control group (n=50) – relatively healthy newborns. Maternal, anthropometric, clinical, and biochemical parameters were studied in both groups.

Results: Newborns with CNS injury had significantly higher levels of MDA (OR 1,13, 95%CI 1.01-1.26, p=0,03), and TSA (OR 2,15, 95%CI 1.02-4.5, p=0,03**)** in urine. According to the results of multiple logistic regression analysis, in preterm newborns an increase in the level of urinary MDA, also delivery via CS and the decrease of urine output on the 3rd day after birth increases the chances of having IVH (AUC- 0,7529).

Conclusion: After conducting studies on a larger sample of patients, it is possible to consider determining the levels of MDA and TSA in urine as a minimally invasive screening method for newborns with hypoxic CNS injury.

Key words: preterm newborns, hypoxic-ischemic CNS injury, malone-D-aldehyde, sialic

ROUTINE BLOOD PRESSURE MEASUREMENT IN CHILDREN IN PRIMARY CARE SETTING: ARE WE DOING IT RIGHT? SURVEY FOR ADOLESCENTS

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Introduction: A higher prevalence of sedentary behaviour and overweight contribute to increased blood pressure (BP) among children and adolescents. The European Society of Hypertension (ESH) emphasizes the importance of routine BP measurement in children and adolescents to identify youngsters at risk for cardiovascular disease.

Objectives: Study aimed to evaluate the real-life BP measurement experiences and compliance with ESH guidelines in Lithuanian adolescents in primary healthcare centers (PHC).

Methods: A cross-sectional survey study was conducted in Lithuania from October 2023 to March 2024. Answers were collected remotely using Google Forms or in-person distributing hard copies for 14-17 year old pupils at various schools and non-formal education centers. The questionnaire consisted of 15 quantitative questions about overall health of the respondents and their experience with routine BP measurement techniques. Data analysis was performed using MS Excel.

Results: There were 448 respondents with mean age of 15.45 ± 1.14 years, (59.8% females) from 26 different districts. Chronic diseases like diabetes, kidney or heart disease were found in 7.6%. 14.5% were born premature or stayed in the neonatal intensive care unit. The diagnosis of AH was reported by 5.1%. 16.6% of the adolescents stated that their BP was not measured in the PHC at all. Majority of those, who had their BP measured, claimed that BP measurement was done annually once in PHC (79.7%), whereas 11.2% stated that BP was checked less than once a year. Concerning the technique, BP was measured at rest in 59.6% of cases, 41.4% reported an inappropriately sized arm cuff and 38.2% mentioned incorrect body positioning. Most of the responders (78.3%) stated that BP was measured only once during the visit. Physicians do not provide adolescents with insights on their BP results in 40.4% of the cases.

Conclusions: Study showed that PHC physicians in Lithuania tend to be inconsistent with proper BP measurement for adolescents and often do not follow the recommended ESH guidelines in daily clinical practice. These results represent the demand of raising health professional awareness in assessing BP measurement.

Keywords: Blood pressure, measurement, adolescents.

EXPLORING THE CORRELATION BETWEEN CORTICOPHOBIA AND QUALITY OF LIFE AMONG CHILDREN AND PARENTS OF CHILDREN WITH ATOPIC DERMATITIS

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Introduction: Atopic dermatitis (AD) is a chronic inflammatory cutaneous disorder. Topical corticosteroids (TCS) combined with emollients remain the mainstay of AD treatment. Their efficacy and safety, when appropriately used, has been clearly established. Paradoxically, the fear of using TCS (usually called „topical corticophobia (TCP)“) is a frequent concern for patients and their parents. TCP may impair adherence to treatment regimen, leading to an uncontrolled disease with a negative impact on patients' and parents' quality of life.

Objectives: To asses TCP and investigate the correlation between TCP and Dermatology Life Quality Index (DLQI) scores of children and parents of children with AD.

Methods: Single-center cross-sectional study conducted between December 2020 and December 2022, of patients (0–17 years), diagnosed with AD, recruited through consecutive sampling during dermatology outpatient clinic appointments. Enrolled participants completed self-report questionnaires, including, DLQI according to age and the adapted Lithuanian version of the TOPICOP questionnaire. TOPICOP responses were scored on a four-point Likert scale, with a maximum cumulative score of 36, while FDLQI and DLQI scores measured on a scale 0 to 30. The study conducted statistical analyses using Microsoft Excel and R Commander package for R. A p-value <0.05 was considered significant. Study was approved by Biomedical Ethics Committee (Approval No. 2020/8-1251-733).

Results: We analyzed 238 TOPICOP and DLQI questionnaires. Girls represented 49% (n=119). Mean age of patients was 6,84±4,43 years. The prevalence of corticophobia in our study population was 55% (n=126). It was found that 32% (n=76) of the group had a high risk, 46% (n=109) had an intermediate risk of corticophobia. All DLQI scores of the impact of AD on the quality of family life (r=0.225), child's life (r=0.192) and infant's life (r=0.222) showed a statistically significant positive correlation with the severity of corticophobia ($p<0.00$; $p=0.022$; $p=0.03$).

Conclusions: The study revealed that corticophobia is prevalent among considerable number of AD patients and their parents that fall under high or intermediate risk. Notably, higer levels of corticophobia are associated with greater impact on both family and childs with AD quality of life. Therefore we suggest that educating patients about the use of TCS could reduce the risk of phobia and improve adherence to treatment plans, ultimately improving quality of life.

Keywords: Atopic dermatitis; corticophobia; pediatrics; topical steroids; Topicop; DLQI

USE OF CONTINUOUS GLUCOSE MONITORING IN GLYCOGEN STORAGE DISEASES

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Introduction. Fanconi-Bickel syndrome (FBS) is a rare autosomal recessive genetic disorder linked to a faulty glucose transporter 2 (GLUT2) due to SLC2A2 gene mutations. FBS manifests with metabolic challenges like acidosis, fasting hypoglycemia, postprandial hyperglycemia and growth retardation. Yet, there's a lack of data on effective nutritional treatments, underscoring the need for improved clinical assessments.

Objectives. We aimed to evaluate the efficacy of a diet with extended release waxy-maize cornstarch (ERWMC) and continuous glucose monitoring (CGM) on metabolic response, biochemical changes and growth in FBS patients over time.

Materials and methods. We report three FBS cases and their tests findings in a 2 year time period to compare them before monitoring and during it. Patients were treated in Vilnius University Hospital Santaros Klinikos with uncooked corn starch (UCCS) or ERWMC (Glycosade®), carbohydrate-restricted diet (CRD) and applied CGM system using a smartphone.

Results. **Patient 1** 19-year-old female with FBS diagnosed at 10 months. Managed with CRD from 2 years age and UCCS from 4 years age that helped maintain slow but steady growth. Glycemic data insufficient for evaluation. Cholesterol increased with fluctuations (6.41->7.4- >9.04->7.59), and triglycerides initially rose then decreased to 3.26. **Patient 2** 6-years-old female. With FBS diagnosed at 3 months age. CRD and UCCS was prescribed at 1 year 9 months of age, but parents did not comply with prescribed regime and diet. The glucose management with CGM was introduced at 4 years age, but discontinued after a few weeks. Noticeable growth retardation (height growth from 2 cm below 3rd percentile of height by age at 3 months to18 cm below 3rd percentile of height by age at 4 years 9 months). Fasting glucose before monitoring was below the lowest normal limit but later increased over the highest limit(1.9->8.4->6.6->5.6). Cholesterol, triglycerides have prominently increased over a 2 year period. (3.19->6.97->9.32 and 1.61->11.99->8.8). **Patient 3** 4-year-old female, diagnosed with FBS at 1.5 months. Monitored with CRD and UCCS from 8 months age, switched to ERWMC at 20 months age, and started CGM at 2 years 2 months. This led to adequate growth (height growth from 1 cm below 3rd percentile of height by age at 1.5 months to a 3rd percentile of height by age at 4,5 years). Glucose levels fluctuated widely (4.1->8.8->5.3->3.9), cholesterol increased slightly above normal (3.14->3.07->5.31->5.82), and triglycerides was 2.8 times lower than the lowest limit before the monitoring and it has increased during.

Conclusions. In our observation, using a CGM system can benefit FBS patients by helping determine ERWMC doses, making informed dietary choices, and maintaining stable blood sugar levels. This fosters positive metabolic outcomes and supports healthy growth.

Keywords. Continuous glucose monitoring, Fanconi-Bickel syndrome, Glycogen storage diseases, Growth retardation.

VISITING HABITS OF THE EMERGENCY MEDICINE CENTER OF THE CHILDREN'S CLINICAL UNIVERSITY HOSPITAL IN CASES OF ACUTE CHILD ILLNESSES

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Keywords. Parents; Health literacy; Acute illness

Introduction. Around 200 children per day visits the Children's Clinical University Hospital Emergency Medical center in Riga for acute illnesses, of which more than 75% are patients whose life is not in danger and whose treatment could be provided by the child's family doctor. Only 4% of patients have life threatening health condition. The large number of non-acute patients delays the help to children, who are in urgent need of emergency medical assistance.

Objectives. The aim of the study was to understand and analyze parental decision-making processes and actions in cases of acute illness of a child when visiting Children's Clinical University Hospital Emergency Medicine center.

Methods. A total of 192 patients' parents were surveyed using a questionnaire. The study included children who were triaged in two main groups: green (life is not in danger) and white (patient does not need emergency help).

Results. Most commonly, patients will seek medical assistance for symptoms of acute respiratory illness (43%) and elevated temperature (42%). Parents in 77% (n=146) of cases were concerned about their child's health, but half of all parents lacked knowledge about how to help their child. In 68% (n=95) of cases, medications used at home did not improve the child's health condition and 47% (n=73) indicated that a consultation with the family doctor was not available. Half of parents noted that one of the main reasons for a visit to the hospital was that it had more accessible diagnostic methods. Parents rated their health literacy as good in 58% (n=111) of cases. A statistically significant result ($p<0.05$) was found between parental health literacy and the frequency of a child's acute visits to the emergency department in the last 12 months.

Conclusions. Parents make decisions to seek acute care at a Children's Clinical University Hospital based on their health literacy. It's important to educate parents about children health and how to act when a child is acutely ill. If a child's primary care provider is unavailable, parents will seek other options to help their child.

SMALL FIBER POLYNEUROPATHY POTENTIAL THERAPY IN PEDIATRICS

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Introduction: Small fiber polyneuropathy is a disease that occurs because of damage to small myelinated and/or unmyelinated nerve fibers. Due to its general symptoms, small fiber polyneuropathy is often not correctly diagnosed and is labeled as an idiopathic condition. Only in recent decade has more attention been paid to the specific combination of symptoms, which include tingling and burning, stabbing pain in the extremities, often accompanied by autonomic and enteric dysfunction.

Case description: In the clinical case, a 6-year-old girl is being examined, whose clinical symptoms correspond to small fiber polyneuropathy. The clinical description of symptoms consistent with the diagnosis includes pain in the legs, which worsens when the legs are immersed in cold water, muscle hypotonia, impaired temperature sensation -when the feet are immersed in cold water, there is no sensation of icy cold water, unclear ethology tachycardia, episodes of vomiting, hypertension, sweating, psychomotor developmental delay, fatigue, dry irritating cough, fever episodes, hematuria, headaches. Lack of appetite, difficulty chewing solid food.

Based on the clinical picture and ineffective therapy so far, intravenous immunoglobulin infusion of 30 g (2g/kg) administered over 5 days is initiated, and five months after the initial IVIG infusion, the therapy plan is changed to 7.5 g (0.5g/kg) as a single dose once a month. After two and a half years, the dose is increased to 15 g (1g/kg) once a month, as the effectiveness of the therapy decreased.

Conclusions: After the administration of IVIG, the patient's overall well-being, physical endurance increased, and developmental delay decreased, leading to progress in physical development. Sweating and hypertension decreased, and the patient started eating on her own. The dry cough decreased, she was able to walk without falling, started attending dance classes, and could hold a spoon. The quality of life for both the patient and her caregivers improved.

Keywords: small fiber polyneuropathy, pediatric, rare disease, fibromyalgia pain, psychomotor development delay, intravenous immunglobilin.

FAMILIAL BETHLEM MYOPATHY IN A PEDIATRIC PATIENT: A CASE REPORT

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Introduction. Bethlem myopathy is a rare subtype of congenital muscular dystrophy characterized by progressive muscle weakness and joint contractures. It is a milder phenotype of the spectrum of collagen VI-related myopathies, which are caused by mutations in genes encoding collagen VI proteins, namely *COL6A1*, *COL6A2*, or *COL6A3*. Clinical manifestation may begin at any time from the prenatal period to late adulthood, presenting a range of symptoms from mild muscle weakness to severe disability impacting the individual's mobility and quality of life.

Case description. A 13-year-old patient presented with progressive weakness affecting both arms and legs, accompanied by an inability to stand flat-footed, walk on heels, squat and rise, and experiencing calf pain after walking longer distances. Additionally, the patient struggled with lifting and carrying various objects. Since the age of 2, an abnormal gait characterized by toe walking had been observed. At 11 years old, the patient underwent Achilles tendon surgery. Physical examination revealed scapular winging, elbow contractures, muscle atrophy in the arms and legs, and follicular hyperkeratosis in the humeral and thigh regions. Elevated levels of creatine kinase were noted, along with myopathic changes evident on electroneuromyography. At 15 years old, muscle biopsy demonstrated myopathic changes without dystrophinopathy-related immunohistochemical alterations.

At the age of 20, next generation sequencing analysis revealed a heterozygous variant c.1053+1G>A; p.(?) of *COL6A2* gene (NM_001849.4) which is reported as pathogenic.

The patient's mother, her mother, brother, and niece reported similar symptoms. Segregation analysis in affected family members revealed the same variant of *COL6A2* gene.

Conclusions. We present a rare case of familial Bethlem myopathy with symptoms evident from early childhood. Understanding the clinical presentation and progression of Bethlem myopathy in children is crucial for early detection, effective management, and genetic counseling. We highlight the importance of a multidisciplinary approach involving neurologists, orthopedist, geneticists, and rehabilitation specialists in providing care for patients with Bethlem myopathy, particularly in pediatric cases where early intervention can significantly impact quality of life.

Keywords. Bethlem myopathy, Collagen type VI-related myopathy, *COL6A2*, Joint contractures, Proximal muscle weakness.

CLINICAL CASE – DIFFERENTIAL DIAGNOSIS OF ACUTE PNEUMONIA IN A PATIENT WITH A HISTORY OF INTRAVENOUS DRUG USE AND NEWLY EXPERIENCED HIV-HCV CO-INFECTION.

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The current study presents a 16 years old patient, with a history of intravenous drug use for 1 year who suffers from severe pneumonia. Chest CT images indicated the possibility of either heroin induced lung damage or atypical pneumonia – multinodular lesions within the pulmonary interstitium, more pronounced in the right lobe. In further investigation the patient tested positive for HIV and HCV infection. Antibody tests have confirmed the presence of *Mycoplasma pneumoniae* IgM, furthermore, there was observed augmentation in the antibody titer over time.

No significant changes were detected during bronchoscopy, and in bronchoalveolar lavage, *Pneumocystis carinii antigen, Aspergillus antigen, Legionella pneumophila,* as well as *Mycobacterium tuberculosis* DNA, were not identified.

The patient received empirical antibacterial therapy with azithromycin and ceftriaxone, following wich clinical improvement was observed, additionally, repeated X-ray of the lungs showed a reduction of pneumonia. The primary diagnosis was *Mycoplasma pneumoniae* - induced complicated bacterial pneumonia.

Significance of this case:

In the context of complicated infectious diseases, the consideration of atypical pathogens and thorough review of medical and epidemiological history were crucial for the accurate differential diagnosis of heroin-induced lung damage. Determining IgM antibody titers, along with detecting HIV and HCV infections, was essential to ascertain the underlying cause of pneumonia and initiate an appropriate and effective treatment regimen.

CONCURRENT PRESENTATION OF SUPERIOR MESENTERIC ARTERY SYNDROME AND IGA VASCULITIS IN AN 11-YEAR-OLD GIRL. A CASE STUDY

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Keywords: superior mesenteric artery syndrome, IgA vasculitis.

Introduction: IgA vasculitis (IgAV) is the primary systemic vasculitis in childhood, commonly diagnosed clinically with typical rash, often accompanied by gastrointestinal, musculoskeletal, or renal symptoms. In contrast, superior mesenteric artery syndrome (SMAS) is a rare proximal small bowel obstruction caused by compression of the third portion of the duodenum. While rare, SMAS is critical to consider due to potential severe consequences such as malnutrition, dehydration, electrolyte abnormalities, gastric perforation, emphasizing the importance of prompt diagnosis and management.

Case description: We report a case involving an 11-year-old girl presenting initially with spasmodic abdominal pain, subfebrile temperature, vomiting and diarrhea. Laboratory analysis indicated leukocytosis with neutrophilia, elevated CRP, and presence of RBCs in urine. Initially admitted to the infectious disease department due to suspected bacterial gastroenteritis, on the fifth day of illness, the patient developed haemorrhagic purpura accompanied by arthralgia and arthritis. Subsequently, she was transferred to rheumatology department where she was diagnosed with IgAV. The pulse therapy with methylprednisolone was initiated; however, despite this intervention, the patient continued to experience persistent stomach-aches, vomiting, and diminished appetite, resulting in weight loss. Following consultation with the gastroenterologist, a new diagnosis of SMAS was suspected, subsequently confirmed by contrast-enhanced CT scan. Oral feeding was discontinued, and a jejunal tube was inserted. After a month of enteral feeding, the tube was removed, and the patient was discharged from the hospital after a 5-day monitoring period, during which improvement in condition was observed.

Conclusions: The case emphasizes the importance of considering alternate diagnoses in patients with evolving clinical presentations, especially when initial treatments fail to yield improvement. It is significant to have multidisciplinary approach in managing complex medical conditions, ultimately improving patient care and prognosis.

CASE REPORT: CONGENITAL HYDROCEPHALUS

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Introduction: Congenital hydrocephalus (CH) is a condition affecting 0.5-0.8/1000 newborns in Europe and USA. It is caused by the impaired cerebrospinal fluid outflow which leads to build-up and expansion of the cerebral ventricles**.**

While it is promptly diagnosed during regular prenatal ultrasound, condition can get severe - the cerebral parenchyma damaged by mass-effect or ischaemia. The child then suffers from developmental issues, distinct cosmetic features such as "sunsetting eyes" and macrocephaly, also to be prone to irritability. The characteristic changes in vital signs include Cushing's reflex - distinct bradycardia, irregular respiratory rate and abnormally increased arterial and pulse pressure.

Case description: We describe a case of Caucasian male delivered on 38th gestational week in Paul Stradins CUH with antenatally diagnosed septo-optic dysplasia, with septum pellucidum agenesis and congenital hydrocephalus due to inborn obstruction of the cerebral aqueduct of Sylvius. After C-section (Apgar score 7/8) he was transported to the Children's hospital for a ventriculoperitoneal shunt with a Codman-Hakim programmable valve (VPS) placement to be then discharged on the 13th day after surgery. During the recovery period patient had recurrent shunt dysfunctions due to the blockage of the valve which required additional surgical treatment to replace it.

Conclusions: Congenital hydrocephalus is a treatable condition which is managed by the VPS placement. The dysfunction rate is 30-40% within the first year and it poses the ethical question of quality of life when there're additional in-born defects. That is why medical professionals should be aware of the risk factors for the development of the condition to consult future parents about healthy choices antenatally.

Keywords: cerebrospinal fluid, congenital hydrocephalus, prenatal diagnostics, septo optic dysplasia, septum pellucidum agenesis, ventriculoperitoneal hunt

TWO RARE PRIMARY BRAIN TUMOURS IN A FANCONI ANEMIA PATIENT: A CASE REPORT

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Keywords. Medulloblastoma, neuroblastoma, Fanconi anemia, BRCA2, TP53

Introduction. Fanconi anemia (FA) is a rare inherited condition, characterized by multiple anomalies, progressive bone marrow failure, higher risk of developing solid malignancies, including central nervous system tumors. Molecular genetic testing is a crucial factor in diagnostics of FA, including mutations in FANCA, BRCA1, BRCA2, PALB2, BRIP1, RAD51, TP53 genes. Consecutive primary malignant tumors are rare and are often considered unfavorable prognosis factor.

Case description. We describe a 4-year-old female patient, who presented in 2021 with congenital hypothyroidism, short stature, multiple *cafe-au-lait* spots, clinodactyly, pancytopenia. Cytogenetics revealed normal karyotype 46,XX, spontaneous and DEB-induced chromosomal breakage. Next-generation sequencing showed pathogenic BRCA2 variant. FA was genetically confirmed. In June 2022 tumor in right temporal lobe was found on MRI. Histopathology, immunophenotype tests confirmed neuroblastoma. Bone marrow biopsy finding in July 2022 revealed neuroblastoma cell complexes, dissemination to bones, bone marrow. Resection of the primary tumor was performed. Proband sequencing established oncogenic mutation in TP53, MYCN amplification. The patient received induction chemotherapy according to COJEC protocol, dinutuximab. A new lesion appeared in left temporal lobe. Additional immunophenotype testing of cerebellar tumor was done in Lithuania, which verified positivity for Synaptosyn, NeuN, p53, Ki-67 40-50%. Primary medulloblastoma was confirmed according to histomorphology, flow cytometry. Medulloblastoma was resected and relapsed soon afterwards. In face of second malignancy developed on therapy, parents choose palliative care.

Conclusions. We report a patient with consecutive primary undifferentiated neuroblastoma, high-risk medulloblastoma associated with FA. Genetic testing revealed mutations of BRCA2, TP53 genes, MYCN amplification. In relation to MYCN amplification, TP53 mutation on poor outcome, novel treatments targeting MYCN, TP53 should be developed for pediatric patients with primary CNS tumors.

Summary. The current study highlights the challenges of BRCA2, TP53 mutation, MYCN amplification presentations in FA patients, diagnosis, complications, management, and surveillance.

ISOPRINOSINE THERAPY IN SUBACUTE SCLEROSING PANENCEPHALITIS

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Introduction: Subacute Sclerosing Panencephalitis (SSPE) is a rare, incurable neurological disorder, caused by persistent measles infection, primarily affecting children. It is characterized by cognitive decline, seizures, and coma. Isoprinosine (IP) is the only medication with official indications for SSPE in multiple countries. Globally, other treatments, including combination therapies, are gaining popularity, therefore understanding the efficacy of IP monotherapy is crucial. This review aims to assess the effectiveness of IP monotherapy in treating SSPE, comparing outcomes based on treatment initiation stage and the time frame of case reports.

Results: We conducted a systematic search on PubMed using keywords "SSPE", "Subacute Sclerosing Panencephalitis". Inclusion criteria: SSPE treated with IP monotherapy, specified stages before treatment, outcomes. Exclusion criteria: reviews, immunocompromised patients, animal/in vitro studies, unrelated or unfinished studies. We analysed 53 cases: 35 (66%) started treatment in stage I-II, and 18 (34%) in stage III-IV. In earlier stages, progression occurred in 27 cases (77%), with only 8 (23%) cases stable or improved. Among stages III-IV, SSPE progressed in 8 cases (44%), with 10 (56%) cases stable or improved. In studies published before 1994, 43% showed stable or improved outcomes (19% in stage I-II, 24% in stage III IV), whereas in the last 30 years, no cases in our search exhibited stability or improvement.

Discussion: In our analysis, disease progression was more prevalent in st. I-II compared to st. III-IV. The outcomes varied: 43% of cases, reported before 1994, showed stabilization or improvement compared to none after 1994. Pritha et al. (2022) examined this treatment from 1999 onwards: only 5 out of 22 patients showed stabilization/improvement. This, together with our observation, suggests worsened outcomes of IP monotherapy in recent decades. We also identified a shift towards other therapies: in our initial search, focused on all types of treatment in the last 30 years, only 10 out of 34 articles described IP monotherapy. This could be due to availability of alternative treatments or challenges in conducting research on rare diseases. We excluded articles that did not specify SSPE stages – this resulted in fewer studies in our analysis. However, a review by Sliva et al. (2019) did not classify the patients according to stage or timing: out of 149 patients treated with IP monoterapy, 62 (41.6%) improved/stabilised, while 87 (58.4%) progressed. They decided that IP was clinically useful, either alone or in combination. While IP monotherapy may offer some benefit, the variability in SSPE outcomes suggests the importance of further research of other treatment strategies.

Conclusions: Starting IP monotherapy in the early stages of SSPE does not consistently result in improved outcomes. There is also a decline in reported stabilization or improvement in recent decades, compared to cases from over 30 years ago. Going forward, researchers should investigate other treatments to improve outcomes for SSPE patients, while also emphasizing the importance of measles vaccinations to prevent the development of SSPE in the first place.

Keywords: Subacute sclerosing panencephalitis, SSPE, isoprinosine, measles infection.

THE OCCURENCE OF MANIA AND SUICIDALITY IN THE BACKGROUND OF SELECTIVE SEROTONIN REUPTAKE INHIBITORS TREATMENT IN CHILDREN

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Introduction. The use of selective serotonin reuptake inhibitors (SSRIs) in pediatric depression management has surged in recent years. Despite their perceived efficacy and safety, concerns have been raised regarding the possibility of adverse psychiatric effects, particularly the onset of mania and suicidal ideation. The purpose of this review is to examine the literature to assess the prevalence and risk factors of mania and suicidality associated with SSRI treatment in children.

Results. The literature review found 14 studies from 2019 to 2024 meeting inclusion criteria. Prevalence rates of mania and suicidality among pediatric patients receiving SSRIs are increased in the majority of studies. Notably, patients displayed a heightened risk of suicidality within the initial 6 weeks following SSRI initiation, with paroxetine and venlafaxine demonstrating the highest risk profiles. No statistically significant increase in completed suicides was observed. However, SSRI did not significantly differ from placebo in precipitating manic episodes. Factors such as age, gender, SSRI treatment duration, psychiatric comorbidities, and history of self-harm or suicide attempts influenced the occurrence of adverse events.

Discussion. The review underscores the intricate relationship between SSRI treatment and the onset of mania and suicidality in children, revealing conflicting findings and the need for individualized risk assessment across studies. Furthermore, many studies exclude individuals at risk of self-harm or suicide, limiting confidence in the medication's effects on this subgroup.

Results are conflicting, with some suggesting an increased risk, others showing no change, and some indicating reduced risk.

Conclusions. The relationship between SSRI treatment and adverse psychiatric effects in children is complex. Clinicians must consider the benefits of SSRIs against the risks of adverse psychiatric effects, like mania and suicidality, in children, while also emphasizing personalized treatment and close monitoring. Further research is needed to clarify these associations and identify risk factors.

Keywords. Adolescent; children; mania; psychosis; selective serotonin reuptake inhibitors (SSRI); suicidality.

PAEDIATRIC HERPES ZOSTER: IMPORTANCE OF TIMELY VACCINATION

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Introduction: Shingles (herpes zoster) is a painful skin rash as the result of reactivation of latent varicella-zoster virus which gained access to sensory ganglia during varicella. Herpes zoster mostly occur in those who are 50 years and older, but it also may develop in children who had chickenpox before 1 year of age, because they may not stay immune. It is important to receive the vaccine at the recommended times indicated.

Case description: A 4-year-old girl was admitted to the Department of Paediatric Infectious Diseases at Children's Clinical University Hospital due to itchy rashes on the skin, which has persisted for 3 days. The main physical examination findings on admission included herpes zoster-type lesions that are visible on the skin parallel to the course of the intercostal nerves, mainly on the back and on the front wall of the chest, forming a belt-like skin lesion, small maculovesicular rash in the hairy part of the head, in the occipital region, on the face and in some places on the lower limbs. It is known that the patient has such a rash for the first time. According to the mother, the girl got chickenpox at the age of 5 months, when her older sister also had chickenpox. The patient is partially vaccinated, she did not receive the varicella vaccine according to the immunization schedule for children aged 12-15 months. A scraping from the area of the skin lesion was taken and DNA of the Varicella zoster virus was identified proving the diagnosis of herpes zoster. The patient was treated with intravenous acyclovir and acyclovir cream, and several days later was discharged in a compensated state for further outpatient treatment.

Conclusions: This case highlights a 4-year-old girl's development of herpes zoster which is also known as shingles, despite typically occurring in older individuals, due to her early exposure to chickenpox. It is important to recognize that children who had chickenpox before 1 year of age may also be at risk due to potential waning immunity.

Keywords: herpes zoster, shingles, early exposure, vaccination.

A CASE OF TUBERCULOUS OTITIS MEDIA IN INFANCY – MANAGAMENT AND SEQUELAE

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Introduction. Tuberculous otitis media (TOM) occurs rarely, and associated symptoms are non-specific therefore making the diagnosis challenging. Delayed or inappropriate therapy, or even with timely and effective therapeutic measures, TOM can lead to serious complications and long-term sequelae, such as hearing loss.

Case description. A 3-month-old boy presented to the Childrens' Hospital in Riga, Latvia with complaints of purulent, haemorrhagic discharge from the left ear that initiated a month earlier and showed no improvement after empiric antibacterial therapy with *Amoxicillinum*. The child was born at 40 weeks gestation, the mother had been diagnosed with human immunodeficiency virus (HIV). Due to unknown HIV status, vaccination was delayed. After failed treatment with *Ceforuxime,* the symptoms persisted and computed tomography (CT) scan of the temporal bone was performed with suspected findings of congenital lithic tumour of the temporal bone. To investigate further, magnetic resonance imaging was performed with findings suggesting rhabdomyosarcoma and intracranial advancement. Biopsy was obtained from mastoid, posterior ear canal and retro-aural lymph nodes and revealed caseous necrosis masses in all samples thus leading to confirmation of diagnosis of TOM. Milliary tuberculosis of the lungs was also found. Although remission of tuberculosis was achieved, the patient had to undergo several surgical interventions because of fibromatous changes of the temporal bone directly caused by tuberculosis and lymphadenopathy and has received a hearing aid due to mixed type hearing loss in the affected ear at 9 years of age.

Conclusions. Awareness of uncommon clinical manifestations in paediatric patients at risk of tuberculosis is crucial for specialists to successfully diagnose and treat the disease and minimize the risk of complications especially in regions endemic of tuberculosis.

Keywords: Tuberculous otitis media; Extrapulmonary tuberculosis; Tuberculosis in neonate.

CHILD TO ADULT. HEALTH CARE TRANSITION FOR CHILDREN WITH COMPLEX DISORDERS

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Introduction. The main aim of this report is to discuss the importance of successful transition to adult health care for children with complex disorders. To make it easier to understand the difficulties and challenges related to medical care that arise when children with complex disorders reach the age of 18, I will review a clinical case involving Charge syndrome. Charge syndrome is a genetically inherited disease that affects multiple organ systems, including the heart, nerves, genitals, ears, and other vital organs. These patients require special care from an early age, provided by a multidisciplinary team of doctors. Difficulties arise when patients reach the age of 18, since they can no longer be treated by paediatricians. It is very important to ensure a consistent transfer of health care from children to adult physicians, ensuring quality care in adulthood. Children with Charge syndrome not only have many congenital pathologies, but compared to healthy people, their development and maturation also lag behind. For these children multidisciplinary care is especially necessary.

Objectives. The successful transition of health care to adults is becoming an increasingly relevant and concerning part of the lives of children with complex disorders. This means that we aim to emphasize the challenges of proper care for patients with complex disorders and ensuring their increased independence, not only in childhood but also as they start adult life.

Methods. Research articles were reviewed primarily to provide an impression of the successful patient transfer from paediatric care to adults' care. A patient with Charge syndrome was discussed to demonstrate the needs of children during this challenging time in their lives.

Results. Research articles have revealed that successful health care transfer is very important for children with complex disorders. It depends on the experience of the paediatric and adults' doctors who take over the needs of a child with complex pathology to properly care for the patient. Patient case demonstrates that it is difficult to find specialists willing and trained to take care of patient with multiple health problems. Successful transfer of health care ensures the patient's ability to continue to be cared for by specialists who will monitor patient's health, help the patient integrate into society, achieve independence, and live a fulfilling life.

Conclusions. Most specialists have little experience with patient transition from paediatric to adult care for children with complex disorders. As this issue raises significant concern, further research and increased cooperation is needed to maximize the success of healthcare transitions to support adult patients as they leave paediatric care.

Keywords. Transition, paediatrics, complex disorders, health care.

"LET'S TAKE A PICTURE FIRST". RADIOLOGICAL ANSWERS ON QUESTIONS IN SYNOVIAL SARCOMA CASE

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Introduction. Synovial sarcoma is a rare mesenchymal tumour with partial epithelial differentiation in adolescents and young adults, accounting for about 2.5 to 10% of soft tissue sarcomas.

Case description. A 17-year-old boy presented with initial complaints of pain in the left hip after trauma. Initial imaging was MRI, which showed a large tumour (11 x 7 x 6 cm AP LL x CC) in the interfascial space of the adductor longus and adductor magnus muscles, with femoral artery and vein passing through. The subsequent biopsy revealed a synovial sarcoma in conjunction with the t(X;18)(p11.23; q11.2) translocation. To obtain precise information about the tumour spread, a whole – body MRI was performed, which showed negative dynamics (tumour enlargement 14 x 9 x 8.7 cm AP x LL x CC), multiple small metastases in both lungs and metastatic nodes in the small pelvis. After progressive complaints from the patient about local pain and swelling, an ultrasound scan was performed to exclude deep vein thrombosis, but the true reason was iliac vein compression from small pelvis metastatic nodes. During the treatment phases, the patient struggled with various complaints and complications, including severe pain, bilateral pneumothorax, hypertension, and massive epistaxis. Various surgical and medical procedures were performed to alleviate the symptoms and control the disease, e.g., chemotherapy, pleural drainage, and analgesic therapy with paracetamol, metamizole, ketorolac and morphine. Although the therapies led to different results, the patient is still coping with the disease, and the plan is to continue the treatment and consider the possibility of radical surgery after the completion of chemotherapy. After 3 months of intensive treatment, there is a positive dynamic – the mass in the left thigh (11 x 6 x 5 cm AP x LL x CC) and the metastatic nodes in the small pelvis have decreased, the bilateral pneumothorax has been absorbed.

Conclusions. A multimodal approach should be used to confirm an accurate diagnosis, complement existing imaging results, and obtain a clear clinical picture. Even the most accurate diagnostic examination does not always allow us to recognise the consequences.

Keywords. Synovial sarcoma.

EFFECTIVENESS OF DEFIBROTIDE FOR PROPHYLAXIS AND TREATMENT OF SINUSOIDAL OBSTRUCTION SYNDROME IN CHILDREN: A SYSTEMATIC REVIEW OF CLINICAL STUDIES

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Introduction. Sinusoidal obstruction syndrome (SOS), previously known as veno-occlusive disease, is a rare life-threatening complication of chemotherapy, mostly associated with hematopoietic stem cell transplantation. SOS occurs in approximately 22% of children and 30% of infants undergoing treatment for oncological diseases, which is 2-3 times more often than in adults. Defibrotide (DF) is currently the only drug that is approved for the treatment of SOS, but due to the rarity of the disease and lack of prospective studies, the evidence of DF efficiency remains uncertain. Several studies suggest the benefit of SOS prophylaxis with DF.

Objective. Systematically assess the effectiveness of the use of DF in children for prophylaxis and treatment of SOS in clinical trials, published within the last 10 years.

Methods. Literature search in PubMed and Google Scholar was performed between the 1st of November 2023 and the 1st of December 2023. We used "defibrotide", "sinusoidal obstruction syndrome, "veno-occlusive disease", "pediatric" and "children" as keywords. The systematic review was performed in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines. Case reports, reviews, meta-analyses, studies published more than 10 years ago and studies with no children were excluded.

Results. 15 publications – 10 retrospective studies, 3 prospective studies, one randomized clinical trial, and one controlled clinical trial - met the inclusion criteria and were analyzed in this review. In total, 7655 patients were included into the studies, with 3121 (40,7%) children <18 y. Eight studies evaluated only SOS treatment with DF, four studies described only SOS prophylaxis with DF, and three studies - both prophylaxis and treatment. The DF dose range was between 12,5 and 60,0 mg/kg/day, with the mean dosage of 25 mg/kg/day. Three studies showed the benefit of SOS prophylaxis with DF while three studies concluded that the prophylaxis is not useful. The studies evaluating DF treatment reported overall survival rates ranging from 25% to 90.4%. All studies supported the benefit of DF for the treatment of SOS.

Conclusions. This systematic review suggests that the treatment of SOS with DF in children is effective and should be applied in clinical practice. However, the most recent studies do not show enough evidence that SOS prophylaxis with DF has significant benefits.

Keywords: sinusoidal obstruction syndrome, veno-occlusive disease, pediatric, defibrotide, hematopoietic stem cell transplantation

SYSTEMIC JUVENILE IDIOPATHIC ARTHRITIS WITH THREATENING MACROPHAGE ACTIVATION SYNDROME

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Introduction: Systemic juvenile idiopathic arthritis (SJIA, also called Still's disease) is a rare auto-inflammatory childhood disease with prevalence between 1 – 9 per 100.000. It accounts for 10-20% of all types of juvenile idiopathic arthritis and is the most severe subtype with significant morbidity. Onset usually occurs between 3 – 5 years of age.

Macrophage activation syndrome (MAS) is a life-threatening complication of rheumatic disease that, for unknow reasons, occurs more frequently in patients with SJIA. MAS has been reported in up to 10% of patients with SJIA, whereas subclinical MAS is assumed to be present in approximately 30-40% of patients.

Case report: A 6- year-old boy hospitalized with complaints of daily spiking fevers, pain and swelling in the right knee joint and maculo-papular rash on the body, that worsens with temperature rises. A week before, patient had cough and runny nose.

During an objective examination, peripheral lymph nodes (neck, inguinal) up to 1 cm in diameter are palpated. In the ultrasonography examination, signs of synovitis are observed in both knees. Laboratory tests showed increased erythrocyte sedimentation rate (ESR) 110 mm/h (N= 0-15); increased ferritin level 777,5 ng/mL; increased INR 1.36 (N= 0.8-1.2); decreased prothrombin 59.6 % (N-76.6 – 116.2); increased D-dimer 2.4 mg/FEU (N=0-0.55). Trepan biopsy is performed and showed: morphologic changes may match with SJIA. In dynamics, temperatures rises are more frequent, inflammatory markers (C-reactive protein, ESR), and ferritin are increasing. Possible development of MAS. Therapy with Anakinra was initiated with rapid improvement of symptoms.

Conclusion: SJIA is a rare auto-inflammatory disease childhood disease. MAS is a severe life-threatening complication of SJIA. MAS prevalence in patients with SJIA is 10%, whereas sublinical MAS is assumed to occur in 30 – 40% of patients. Early, close clinical monitoring and appropriate treatment are important to prevent the development of serious SJIA complications such as MAS.

Keywords: Systemic juvenile idiopathic arthritis; Macrophage activation syndrome; life threatening complication

CLINICAL COURSE AND MANAGEMENT OF REFRACTORY AUTOIMMUNE PROCESSES AND UNSPECIFIED IMMUNE TOLERANCE DISORDERS

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Introduction. Primary immunodeficiency diseases are a group of disorders which affect components of the innate and adaptive immune systems, are predisposing to recurrent infections and allergic reactions, are hard to diagnose and often do not respond to selected treatment (Marzieh et al, 2019) (Renzi et al, 2020).

Case description. We report a case of a 11-month-old boy diagnosed with severe autoimmune hemolytic anemia with hemoglobin (HGB) up to 43 g/L, acute kidney injury, hyperbilirubinemia, liver damage, thrombocytopenia. Infections and hemolytic uremic syndrome were excluded from differential diagnosis, anti-DNA, anti-ds DNA were within normal range. Patient received treatment with methylprednisolone, azathioprine, renal replacement therapy. Immunosuppressive therapy was gradually reduced until completely canceled (1 year and 4 months) and patient was allowed to receive vaccination according to his age (2-year-old). However, at the age of 2 years and 7 months patient was admitted to the hospital with the 2nd episode of severe acute autoimmune hemolytic anemia presented with cold antibodies and HGB 30 g/L, resistant to immunosuppressive therapy. The child developed livedo reticularis, hepatosplenomegaly, jaundice. For his further treatment patient received cyclophosphane, vincristine and fludarabine course. During the next year patient experienced two episodes with recurrent idiopathic thrombocytopenic purpura, unresponsive to immunosuppressive therapy. Patient received course of prednisolone per os, methylprednisolone and dexamethasone pulse therapies, intravenous immunoglobulins, azathioprine, rituximab, cyclophosphane, vincristine. After careful evaluation of this case by the multidisciplinary team, the patient was suspected of developing an immunodeficiency, which could not be confirmed by immunological and genetic testing. Extremely severe disease episodes with minimal response to intensive therapy courses were evaluated as indication for allogeneic hematopoietic stem cell transplantation (HSCT). Thus at the age of 3 years and 9 months patient received HSCT from his brother, who was tested as compatible donor.

Conclusions. Hematopoietic stem cell transplantation provides a curative option for children with complicated cases of immune dysregulation. Management of such cases should involve a multidisciplinary team for better diagnosis and effective treatment.

Keywords: Primary immunodeficiency disorders; Immunosuppressive therapy; HSCT; Children

USE OF BLINATUMOMAB IN PEDIATRIC PATIENT WITH RECURRENT B – CELL PRECURSOR ACUTE LYMPHOBLAST LEUKEMIA AND TRISOMY 21.

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Introduction: Recurrent B-cell precursor acute lymphoblastic leukemia (B-ALL) is a challenging diagnosis to treat. Blinatumomab is monoclonal antibody used as second-line treatment for recurrent B-ALL both in children and adults. Here, successful case of using blinatumomab in pediatric patient with late relapse B-cell precursor acute lymphoblast leukemia and Trisomy 21 is reported.

Case description: A 7-year old girl presented to the ER with strabismus, headache and dizziness. Recent complete blood count (CBC) revealed thrombocytopenia. In November 2019 patient was diagnosed with B-ALL without CNS damage. Patient received B-ALL therapy according to the ALL-BFM-2000 protocol with reduced doses due to genetic disorder - trisomy of the 21st chromosome. Treatment was with positive dynamic and after completing chemotherapy course remission has been achieved. Maintenance chemotherapy completed in December 2021. In August 2022 patient is diagnosed with late relapse of B-precursor ALL. Initial blasts are found in the bone marrow as well as in the cerebrospinal fluid. On the MRI leukemic conglomerates and infiltrations are found around cranial nerves. In August 2022 therapy according ALL REZ BMF-2002 protocol was started. Patient received prophase, F1 protocol. Minimal residual disease (MRD) 33% in bone marrow before F2 protocol. Blast cells are not detected in the cerebrospinal fluid after the first course of chemotherapy. Taking into consideration that the patient has trisomy of 21st chromosome, initial chemotherapy drugs dosage was reduced to 75%. Control bone marrow examination before the first course of R1 chemotherapy showed MRD 9,3% therefore it was decided to increase chemotherapy dosage to 100%, regularly evaluating the side effects and patient tolerance. MRD without positive dynamics after receiving F1, F2, 2 x R2 and 2 x R1 chemotherapy courses. Based on these results 2 courses of Blinatumomab were additionally administered achieving desired effect of reducing MRD. Considering the patient's co-morbidities, evaluating potential benefits and risks, patient's legal guardian refrains from bone marrow transplantation at the moment. After receiving the first course of therapy with Blinatumomab, MRD is no longer visualized either by flow cytometry or PCR. So far, the patient has received F1, F2, 4x R2, 3x R1 chemotherapy courses, as well as 2 Blinatumomab courses. Intensive chemotherapy course completed in September 2023.

Conclusions: Blinatumomab was tolerated without any significant complications and helped to achieve no visualized MRD by flow cytometry or PCR. Patient should be further observed to exclude late therapy complications.

Keywords: blinatumomab, recurrent B-cell acute lymphoblastic leukemia, pediatric

FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS SECONDARY TO UNC13D MUTATION (FHL3) COMPLICATED BY MYCOBACTERIUM BOVIS INFECTION: CASE REPORT

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Introduction: Familial Hemophagocytic Lymphohistiocytosis (FHL3) is a rare genetic disorder characterized by multiorgan damage and dysfunction, affecting organs such as the spleen, liver, bone marrow, and brain. In Sweden, the incidence of FHL is reported to be 1.2 in 1,000,000 children per year. Here, we present the first confirmed case of FHL3 in Latvia.

Case Description: A 37-day-old girl was admitted to the Children's Clinical University Hospital with persistent fever, hoarseness, haemorrhagic skin rash, and hepatosplenomegaly. Laboratory results revealed pancytopenia, hypertriglyceridemia, hypofibrinogenemia, hyperferritinemia, elevated CRP, IL-6, LDH, ALAT, ASAT, and increased D-dimers. Her medical history indicated a physiologically normal pregnancy, with normal birth height and weight, and she had also received the BCG vaccine. Ultrasound examination showed splenomegaly, gallbladder wall thickening, and ascites. CMV DNA was detected in urine and blood. Following the diagnosis on September 21st, she underwent HLH-2004 protocol-based treatment, which included intrathecal chemotherapy. She also received broad-spectrum antibiotics and antifungal therapy for severe neutropenia and fever, as well as intravenous Ganciclovir when the CMV DNA copy number in the blood was above 1000. Genetic testing confirmed FHL3 due to two pathogenic compound heterozygous germline variants in the UNC13D gene. The patient underwent a hematopoietic stem-cell transplant on December 15th, utilizing stem cells from a matched unrelated donor. During the patient's early post transplantation period, the skin around the BCG vaccination site became inflamed and hardened. Mycobacterium bovis bacilli were detected in a faecal sample, confirming Mycobacterium bovis infection. The patient is currently receiving anti-tuberculosis treatment.

Conclusions. This case report highlights the importance of early diagnosis, adherence to the HLH-2004 protocol, and subsequent hematopoietic stem-cell transplantation in managing and improving outcomes in Familial Hemophagocytic Lymphohistiocytosis. Furthermore, the occurrence of complications in early post-transplantation period emphasizes the need for monitoring and tailored management strategies.

Keywords. Familial Hemophagocytic Lymphohistiocytosis (FHL); UNC13D; Hematopoietic stem-cell transplantation; Mycobacterium bovis infection;

THE IMPORTANCE OF PULSE OXIMETRY IN THE DIAGNOSIS OF INFANTILE COARCTATION OF THE AORTA: A CASE REPORT

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Introduction: Coarctation of the aorta is a congenital narrowing of the aortic lumen causing obstruction to the blood flow through the vessel. This lesion, which is seen primarily in the infantile (preductal) form, is the 8th most common congenital heart defect and accounts for 6–8% of all congenital heart diseases. Without treatment infantile coarctation of the aorta can result in cardiogenic shock, as it is one of the ductus arteriosus dependent heart diseases. Unfortunately even with advances in fetal echocardiography, prenatal diagnosis of this condition still remains a challenge. So it is very important to make an early diagnosis after the birth if it did not occur prenatally. Using pulse oximetry as a screening method can be very helpful in these kind of cases.

Case description: a 5 days old girl is hospitalized in the neonatal clinic with possible diagnosis of congenital heart disease. Girl was born at the gestational age of 40 weeks from 3rd pregnancy, 2nd birth. APGAR score of 9/10 points. Three days after birth girl starts to eat less and breath more heavily. At the neonatal clinic general condition of the newborn is severe – child was tachycardic up to 180 times per minute, breathing was irregular, 60-70 times per minute, saturation of oxygen without breathing mask 70-74%, use of accessory muscles in the breathing process could be seen. Screening of pulse oxymetry was done at the maternity ward and it showed saturation on left hand 95%, on right hand – 99%, on left foot – 70%, on right foot – 60%. After the initial evaluation girl was intubated and artificial pulmonary ventilation was started afterwards. Intravenous prostaglandīns were administered. After the consultation of pediatric cardiologist the diagnosis of infantile coarctation of the aorta and cariogenic shock was made. Girl was prepared for surgery.

Summary: coarctation of the aorta is the 8th most common congenital heart defect. Unfortunately even with advances in fetal echocardiography, prenatal diagnosis of this condition still remains a challenge. So it is very important to make an early diagnosis after the birth if it did not occur prenatally. Pulse oximetry can be helpful in such cases.

Conclusion: without treatment infantile coarctation of the aorta can result in cardiogenic shock. So early diagnosis and treatment is the key to better prognosis.

Keywords: infantile coarctation of the aorta; pulse oximetry

OSTEOMYELITIS OF THE PATELLA IN A 9-YEAR-OLD-BOY: A CASE REPORT

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Intruduction: While osteomyelitis disease most commonly affects the long bones, involvement of the patella is rarely seen. Due to this rarity and the variable clinical presentation, diagnosis is often delayed. Essential surgical steps include drainage, radical debridement, administration of culture-directed antibiotics . We present a case report that describes a 9-year-old male patient with patella osteomyelitis.

Case description : On 26.02.2024. a 9 year old boy was administered to Childrens Clinical University Hospital with dynamically progressing pain in the left knee joint and subfebrile fever. Objectively, the knee joint was enlarged in volume and there was pain on palpation of the patella. Given the inflammatory parameters and the patient's condition, the patient was admitted to hospital. An in patient blood culture was taken which showed the growth of *Micrococus luteus*. The patient was started on *Cephalosporin* antibiotic therapy. A magnetic resonance imaging was preformed which indicated destruction of the patella, a finding consistent with osteomyelitis. Surgical management was indicated. On 27.02., the patient underwent left patellar excochleasis, biopsy, knee repair and drainage. Histological material confirmed the diagnosis of acute hematogenous osteomyelitis, and a biopsy of the bone was taken during surgery, which showed growth of *Staphylococcus aureus*. Another blood tests were carried out together with sensitivity to antibiotics on 27.02. The result of the blood smear was *Staphylococcus epidermidis* - resistant to Oxacillin and Clindamycin. The antibiotic therapy is changed to *Vancomycin* according to the sensitivity of the microorganisms. On 04.03. The patient continues to have inflammatory signs: pain, hyperemia, hyperthermia, in the left knee joint area. Arthroscopic, surgery on the knee was indicated. After the operation, control MRI was performed. Patient continued to receive AB therapy, swelling and pain decreased gradually. Dynamic blood tests, markers of inflammation were reduced, the microflora in the blood was no longer growing.

Conclusion: Osteomyelitis can cause local bone fractures, purulent arthritis, joint contractures and general sepsis. It can also cause complications of renal amyloidosis. Acute hematogenous is difficult to treat and treatment is prolonged.

Key words: Osteomyelitis, patella, cultured directed antibiotics.

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