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ABSTRACTS

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AIR POLLUTION KEY FOR THE UNDERSTANDING OF RESPIRATORY DISEASES IN CHILDREN: NEW EVIDENCE OF HARM

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Objectives

It's known that both the size and micro-elemental composition of particulate matter (PM) play an important role in the etiology of chronic noncommunicable diseases in children, while the impact on acute respiratory diseases is still less studied. The aim of our research presentation is an overview of the results of our Human Ecology Interdisciplinary Research Group (HEIRG) activities from 2020 to 2025, focusing on two studies of the evaluation of the impact of indoor aerosol pollution in primary schools and kindergartens on the respiratory health of children.

Materials and Methods

To determine seasonal aerosol particle number (PNC) and mass (PMC) concentrations in primary schools and kindergartens, a condensation particle counter (TSI model 3007, PNC 0.01 to>1.0 µm) and an optical particle sizer (TSI model 3330, PNC and PMC 0.3–10.0 µm) were used. Original indoor short-term (10 min) serial measurements are carried out. A micro elemental composition, including heavy metal (HM) concentrations, was analyzed in samples of dust as natural PM aggregates using a SPECTRO XEPOS X-ray fluorescence spectrometer. The annual morbidity data were collected from medical records.

Results

It was found that within one city, there was up to 7 times the difference in PM concentration at randomly selected primary schools and up to 30 times the difference in HM concentration in dust samples taken from kindergartens. In younger school-age children, we confirm the relationship between submicron-size PM and asthma morbidity. On the other hand, the concentrations of Vanadium and Nickel in dust samples were related to the incidence of upper respiratory infections in both school-age and preschool children, while the concentration of Tungsten was related to the seasonal flu morbidity in preschoolers only.

Conclusions

Our proposed methodology for the complex assessment of indoor aerosol pollution in schools and kindergartens allows us to evaluate the impact of air quality on the origin and course of both acute and chronic respiratory diseases in children.

Funded by project S-MIP-23-128 of the Lithuanian State Research Council.

CASE REPORT: HYPERSENSITIVITY PNEUMONITIS

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Objectives

Hypersensitivity pneumonitis (HP) in childhood is a rare, complex, and heterogeneous interstitial lung disease that occurs when susceptible individuals develop an exaggerated immune response to inhaled allergens. The differential diagnosis of HP requires a thorough exposure history, multidisciplinary discussion of clinical and radiological data and in some cases, assessment of bronchoalveolar lavage lymphocytosis.

A very cachectic 13-year-old girl (weight 27.7 kg, height 155 cm, BMI 11.6 kg/m^2) was admitted to the Department of Infectious Diseases with fever, malaise, weakness, productive cough, tachypnoea, dyspnoea and auscultatory crackles with wet rales. She had previously received amoxicillin for pharyngitis and clarithromycin for pneumonia without improvement. Her appetite was poor, and her mother had not realized how underweight she was. At home, she was exposed to multiple airborne allergens/pollution.

Investigations

Thoracic X-ray: diffusely increased bilateral pulmonary pattern with fine nodules; sarcoidosis or tuberculosis suspected. S-shaped scoliosis diagnosed.

Blood tests: neutrophilic leukocytosis, CRP <1 mg/L, LDH 334 U/L, IgG 32.7 g/L. Negative for all tested viral and bacterial infections, including pulmonary tuberculosis and psittacosis, cystic fibrosis, celiac disease. Mildly elevated total IgE (111 kU/L), allergen-specific antibodies negative, ECP 15.6 µg/L, Aspergillus fumigatus IgG 39.4 kU/L.

Thoracic CT: diffusely distributed, poorly defined “cotton swab-like” foci in both lungs; findings not typical for sarcoidosis or tuberculosis. Impression: hypersensitivity pneumonitis.

Bronchoscopy: tracheal and bronchial mucosa markedly inflamed with whitish granulations at airway bifurcation.

BAL cytology: macrophages 68%, neutrophils 3%, lymphocytes 28%, eosinophils 1%, no atypical cells. Spirometry: FVC 35%, FEV1 35% of predicted, FEV1/FVC 82%; positive bronchodilator test (11% FEV1 increase) with subjective

improvement.
Treatment
Prednisolone 1 mg/kg/day (25 mg) for 5 days, then 15 mg/day for 5 weeks with gradual tapering. Symbicort 160/4.5 mcg × 2, dietary therapy, physiotherapy.
The patient was treated in the hospital for 3 weeks with positive dynamics: CT thorax showed marked regression of findings, weight gain was 2.3 kg, and lung function improved significantly but was not normal.
Cooperation with a social worker. The girl lived in a private house with smoking parents. There were a zebra finch, quail, pigeon, rat, and 2 budgies indoors, which she fed and whose cages she cleaned. After the hospital stay, the patient was moved to another place until the birds were removed and the house cleaned.
Follow-up
The patient has been followed by a pediatric allergologist-pulmonologist for 3 years. Regular treatment with Symbicort 160/4.5 × 2. Lung function remains low: FVC 83%, FEV1 81% of predicted, FEV1/FVC 83. Weight 43 kg, height 159 cm, BMI 17 kg/m^2. CT thorax: emphysematous bulla in S1 of the right upper lobe, centrilobular emphysema mainly in the upper lobes. Scoliosis was successfully operated in 2023. Continuous physiotherapy. Advice on healthy eating.

Materials and Methods
x

Results
x

Conclusions
Fever is not always of infectious origin. The home environment can be a cause of serious illness in a child.

CHARACTERIZATION OF RESPIRATORY TRACT MICROBIOTA IN LATVIAN CYSTIC FIBROSIS PATIENTS, HIGHLIGHTING THE FIRST CASE OF M. ABSCESSUS AND M. CHIMAERA CO-INFECTION

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Objectives
The objective of this study was to determine the prevalence of colonization by major respiratory tract pathogens in cystic fibrosis patients in Latvia in 2023, and to compare these findings with data collected since 2020. Additionally, we analyzed the first documented case in Latvia of co-infection with Mycobacterium abscessus subsp. massiliense and Mycobacterium chimaera

Materials and Methods
Data were collected between 2020 and 2023 from the following sources: the ECFS Patient Registry, the Laboratory of the Centre of Tuberculosis and Lung Diseases (Latvia), and the patient data collection system of the Children's Clinical University Hospital of Latvia. The collected data were entered into Microsoft Excel and subsequently analyzed using standard descriptive statistical methods.

Results
As of the study period, there were 61 registered cystic fibrosis patients in Latvia. Data were obtained from 86.9% of these patients (N = 53). Of those, 68% (N = 36) were pediatric patients, and 32% (N = 17) were adults. In year 2023, 67,9% (N=36) of Latvian patients were colonized by Staphylococcus aureus, 28,3% (N=15) by Pseudomonas aeruginosa, 1,9% (N=1) by Burkholderia cepacia, 1,9% (N=1) by Stenotrophomonas maltophilia, 24,5% (N-13) by H.influenza, 11,3% (N=6) by Achromobacter and 3,7% (N=2) by MRSA. In 2020 percentage of bacterial colonization were 64,1% (N=34), 22,6% (N=12), 1,9% (N=1), 0% (N=0), 20,7% (N=11), 5,6% (N=3) and 1,9% (N=1), respectively. P.aeruginosa is most prevalent in the age group 25-39 years - 77,8% (N=7). The prevalence of Staphylococcus aureus colonization by age group was: 64% (2–5 years), 80% (6–11 years), 82% (12–17 years), 75% (18–24 years), 60% (25–28 years), 67% (30–34 years), and 100% (35–39 years). 42% (N=16) of patients in the age group 6-39 years, uses chronic antibacterial inhalations. Patient with M. abscessus subspecies massiliense and M. chimaera co-infection was diagnosed after five months of characteristic symptoms. Sputum were repeatedly investigated for Nontuberculous Mycobacteria (NTM), but only after several months were cultured positive. No mutation in rrl and rrs genes hence no resistance for macrolides and aminoglicozides. During diagnostic process, until the diagnosis of NTM pulmonary disease was made, according to symptomes and investigational results, acuta EBV infection, Clostridium difficilie infection, Aspergillus infection were diagnosed and treated. On Lung CT many confluent dense foci was seen. Sputum culture conversion was achieved after seven months of treatment. During the course of therapy, the patient experienced several drug-related adverse effects, the most significant being linezolid-induced cytopenia and amikacin-induced ototoxicity. The patient is currently in the continuation phase of NTM treatment.

Conclusions
Our data show, that S.aureus and P. aeruginosa should be considered an important pathogens in Latvia. 30% (N=16) of patients are colonised by both pathogens. Although the presence of nontuberculous mycobacteria (NTM) is rare,

co-infection with Mycobacterium abscessus subsp. massiliense and Mycobacterium chimaera may occur

EPIDEMIOLOGICAL TRENDS OF LOWER RESPIRATORY TRACT INFECTIONS IN CHILDREN (2018–2024): INSIGHTS FROM LUHS KAUNO KLINIKOS

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Objectives
This study aimed to analyze the epidemiology, etiology, clinical characteristics, and treatment of prevalent lower respiratory tract infections (LRTIs) in children at the Lithuanian University of Health Sciences (LSMU) from 2018 to 2024. The objectives included identifying the incidence of acute bronchitis, bronchiolitis, and community-acquired pneumonia (CAP), examining etiological trends across pandemic phases, assessing clinical outcomes, and evaluating changes in antibacterial therapy.

Materials and Methods
A retrospective analysis of 1,053 children treated for lower respiratory tract infections (acute bronchitis, bronchiolitis, pneumonia) at LSMU Kaunas Clinics from 2018 to 2024 was conducted. Approval from the Biomedical Research Ethics Committee received (No. BE-2-10). The study assessed incidence, etiology, clinical course, and changes in antibacterial therapy across pre-pandemic, pandemic, and post-pandemic periods, as well as different age groups. Statistical analysis was done using SPSS version 30.0, with significance set at $p < 0.05$.

Results
A retrospective analysis of 1,053 children's medical records revealed that bronchiolitis incidence remained stable (~22 cases/year), while bronchitis cases rose from 22 in 2018 to 42 in 2024. Pneumonia cases decreased during the pandemic (43 cases in 2021) but surged post-pandemic to 199 in 2024. Notably, older children (2–18 years) experienced higher bronchitis rates post-pandemic. The incidence of CAP caused by Mycoplasma pneumoniae significantly increased post-pandemic (26.5%) compared to pre-pandemic (11.1%) and during the pandemic (6.3%).
Hospital stays for bronchiolitis were longer during the pandemic, with increased complications post- pandemic, particularly acute respiratory insufficiency (ARI) in infants under 3 months. Similar trends were noted for bronchitis and pneumonia, with ARI incidences rising significantly post-pandemic. Treatment practices shifted; post-pandemic, macrolides and cefotaxime were used more frequently for pneumonia, while the use of ampicillin/sulbactam decreased.

Conclusions
In conclusion, the pandemic significantly influenced the incidence and etiology of LRTIs in children, leading to increased complications and changes in treatment regimens. The rise in bronchitis among older children and the resurgence of pneumonia cases highlight the need for ongoing surveillance and adaptive treatment strategies.

IDENTIFYING RISK FACTORS FOR SLEEP-RELATED BREATHING DISORDERS IN CHILDREN: EVIDENCE FROM A LITHUANIAN CROSS-SECTIONAL STUDY

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Objectives
To investigate the prevalence of sleep-related breathing disorders (SRBD) in Lithuanian children and identify the most significant associated risk factors using a binary logistic regression model.

Materials and Methods
This cross-sectional study included children aged 2–17 years old. Parents completed a comprehensive questionnaire, including demographic information, general health data, and the Pediatric Sleep Questionnaire (PSQ; Chervin et al., 2000). The translated and validated Lithuanian version of the PSQ was used in the study. Children with ≥8 positive responses on the PSQ were classified as having SRBD. Binary logistic regression analysis was performed to assess associations between potential risk factors and suspected SRBD.

Results
Out of 1,929 participants, 14.9% were classified as having SRBD based on PSQ criteria. No significant gender difference was observed between groups ($p = 0.485$). Logistic regression analysis identified mental illness (B 2.300, $p < 0.001$, OR 9.975, 95% CI 6.644-14.977), endocrine disorders (B 1.763, $p < 0.001$, OR 5.829, 95% CI 3.147-10.795), weight category: overweight (B 0.432, $p < 0.001$, OR 1.540, 95% CI 1.230-1.929), obesity (B 0.818, $p < 0.001$, OR 2.265, 95% CI 1.730-2.965), frequent respiratory tract infections (B 0.902, $p < 0.001$, OR 2.465, 95% CI 2.093-2.904), adenotonsillar hypertrophy (B 0.796, $p < 0.001$, OR 2.218, 95% CI 1.704-2.886), allergic rhinitis (B 0.401, $p < 0.001$, OR 1.493, 95% CI 1.184-1.883), and male gender (B -0.290, $p < 0.001$, OR 1.136, 95% CI 1.142-1.562) as significant risk factors for SRBD.

Conversely, a history of adeno-tonsillectomy was associated with a reduced risk of SRBD (B -0.320, p = 0.002, OR 0.726, 95% CI 0.590-0.893). Constant of logistic regression model was -0.504, p < 0.001. The AUC of logistic regression model to predict SRBD was 0.717± 0.009 (p<0.001, 95% CI 0.698–0. 735).

Conclusions
Approximately 15% of children in the Lithuanian pediatric population may be affected by suspected SRBD. The most significant risk factors identified were mental illness, endocrine disorders, obesity, recurrent respiratory tract infections, and adenotonsillar hypertrophy.

LONG-TERM PAEDIATRIC HOME MECHANICAL VENTILATION: A 20-YEAR EXPERIENCE IN LITHUANIA

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Objectives
To characterize the clinical profile and outcomes of paediatric patients receiving long-term home mechanical ventilation (LTHMV) at the Hospital of Lithuanian University of Health Sciences, Kauno Klinikos (KK) - one of the largest national centres for paediatric LTHMV, serving patients from all regions of Lithuania.

Materials and Methods
This retrospective study includes paediatric patients who were initiated on and followed with LTHMV at KK between 2005 and 2024. Follow-up began on the first day of ventilatory support and ended upon transition to adult care, discontinuation of ventilation, or death. Ethics approval (No.: BE-2-67) was obtained for the study.

Results
The first paediatric LTHMV case in Lithuania involved a patient with spinal muscular atrophy type II, who has remained on ventilatory support since 2005. Over the 20-year period, 65 children were initiated on LTHMV, including 44 boys (67.7%) and 21 girls (32.3%). Invasive ventilation via tracheostomy was used in 31 patients (47.7%), and 34 patients (52.3%) received non-invasive ventilation (NIV), including 3 patients (8.8%) managed with continuous positive airway pressure (CPAP). A notable increase in LTHMV initiations was observed after 2017, coinciding with the introduction of polysomnography and the implementation of national regulations supporting LTHMV. Since 2018, NIV has become the predominant mode of ventilatory support, accounting for 57.8% of cases compared to 42.2% receiving invasive ventilation. The median age at LTHMV initiation was 6.67 years (range: 0–17.9; 95% CI: 5.83–9.05). The most frequent indication was neuromuscular or other neurological disorders (n = 35; 53.8%), followed by obstructive sleep apnea (n = 6; 9.2%), airway malformations (n = 6; 9.2%), Prader–Willi syndrome (n = 4; 6.2%), central sleep apnea or sleep-related hypoventilation (n = 3; 4.6%), spinal cord injury (n = 2; 3.1%), brain tumors (n = 2; 3.1%), Arnold–Chiari malformation (n = 2; 3.1%), and other rare conditions (n = 5; 7.7%). Median age at LTHMV initiation varied by diagnosis: 1.3 years (95% CI: −1.12-8.43) in neuromuscular disorders, 16.04 years (95% CI: 11.78–18.58) in obstructive sleep apnea, and 0.95 years (95% CI: −1.60 to 6.38) in airway malformations. At the time of analysis, the majority of patients remained on LTHMV and were still of paediatrics age (n = 31; 47.7%), while others had transitioned to adult care (n = 15; 23.1%), had died (n = 11; 16.9%), or had discontinued ventilation due to clinical improvement (n = 8; 12.3%). The median age of children currently on LTHMV was 8.4 years (range: 2.58–17.58; 95% CI: 8.04–11.70).

Conclusions
Over the past two decades, the use of LTHMV in children in Lithuania has significantly increased. It has been applied to a clinically diverse paediatric population, most frequently in those with neuromuscular disorders. In recent years, NIV has emerged as the preferred treatment modality. More than half of the patients remained on LTHMV at the time of analysis, underscoring the chronic nature of their conditions. These findings emphasize the importance of long-term multidisciplinary care and coordinated transition planning to adult services.

PRESCHOOL WHEEZING: CURRENT UNDERSTANDING AND MANAGEMENT

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Objectives
Preschool wheeze is very common, and a source of considerable morbidity and even mortality. Management is challenging, and has recently been summarised in European Society Guidelines [Eur Respir J. 2024; 64: 2400624]. The first step is to understand what the family mean by “wheeze“, it is a term frequently used to describe noises other than true, expiratory, polyphonic whistling. Next is a diagnostic process; causes include (a) normal child; (b) a serious underlying condition (rare); (c) an asthma syndrome; (d) conditions which mimic or exacerbate wheeze (rhinitis, reflus); and (e) overanxious parents. Clinical skills are used to categorise the child, and determine which individuals need further investigation. Asthma is a

description not a diagnosis (wheeze, chest tightness, dyspnoea, and sometimes excessive cough) [Lancet 2018; 391: 350-400] and the next step is to determine which treatable trait the individual child has, specifically is there evidence of airway eosinophilia. Although there are no randomised trials of treatment, there is increasing evidence that the combination of a blood eosinophil count (BEC) >300 cells/μλ and sensitivity to food and aeroallergens predicts risk of acute attack of wheeze [J Allergy Clin Immunol. 2016; 138: 1608-1618; J Allergy Clin Immunol Pract. 2023; 11: 1485-1493] and response to inhaled corticosteroids (ICS) [Am J Respir Crit Care Med. 2025; 211: 263-265]. If BEC is used to guide treatment, and this is now practical with a point of care device [https://hemocue.com/en/contact/], it should be noted that it is variable over time and repeated measurements are likely to be necessary. Many preschool wheezing children are do not have airway eosinophilia and are not ICS-responsive. Cluster analysis has shown that at least some of these children have associated airway infection [Am J Respir Crit Care Med. 2021; 204: 523-53]. In addition to prescribing medications, attention must be paid to ensuring that an appropriate spacer is given, and also to the presence of allergens and irritants in the environment. There is compelling evidence that the combination of allergen sensitization, allergen exposure and respiratory viral infection are strongly associated with attacks of wheeze [Thorax. 2006; 61: 376-82] and reduction of the allergen burden improves outcomes [Am J Respir Crit Care Med. 2017; 196: 150-158.]. The adverse effects of exposure to irritants, especially tobacco and vaping, but also in some cultures, incense, should not be forgotten. The role of prednisolone in the treatment of acute attacks of wheeze is controversial; a recent meta-analysis showed reduction in wheeze at 4 but not 12 hours, and improvement in secondary endpoints, which were not dramatic [Lancet Respir Med. 2024; 12: 444-456]. Perhaps as well as phenotyping chronic symptoms, we should be phenotyping acute attacks as well – in adults at least some are eosinophilic and some are infective, non-eosinophilic [Allergy 2021; 76: 375-379; Lancet Respir Med. 2025; 13: 59-68], and if the same is true for preschool children, the eosinophilc acure preschool wheezers may be the group that respond to prednisolone, whereas this treatment is unnecessary for the non-eosinophilic wheezers. However this needs testing prospectively in future work.

Materials and Methods

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Results

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Conclusions

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THE CONCENTRATION OF TUNGSTEN IN DUST SAMPLES COLLECTED FROM KINDERGARTENS IS ASSOCIATED WITH THE INCIDENCE OF INFLUENZA IN PRESCHOOL CHILDREN

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Objectives

To evaluate the microelement composition of natural dust aggregates collected in Vilnius kindergartens and assess its relationship with the incidence of influenza in preschool-aged children.

Materials and Methods

Dust samples from Vilnius kindergartens were collected between November 2023 and January 2024. Dust as natural aggregation and sedimentation of aerosol pollutants was collected from areas typically missed during routine cleaning, such as high cabinet surfaces and central heating radiators. Pb, W, Sb, Sn, Zr, Rb, Cu, Ni, Mn, Cr, V, As, Ba, Br, and Zn concentrations were analyzed in dust samples using a SPECTRO XEPOS X-ray fluorescence (ED-XRF) spectrometer. The annual incidence of influenza for each kindergarten received from medical records collected by the State Institute of Hygiene.

Results

In 2023, influenza (ICD-10-AM J10–J11) incidence in preschool children across study kindergartens ranged from 0.7% to 6.3%. Using tungsten in cutting and drilling tools, welding electrodes, and vehicle parts like winter tire studs could have impacted the tungsten concentrations in kindergartens, ranging from 3.14 ppm to 29.37 ppm (median – 12.46 ppm). A statistically significant correlation emerged between tungsten concentration in natural indoor dust aggregates and influenza incidence in preschool-aged children (ρ=0,47, p=0,009). The linear regression model resulted in the equation: $y = 1.65 + 0.12 * x$ (F p=0,0074, R^2=0,23), where y is influenza incidence and x – tungsten concentration in ppm. Considering the highest tungsten concentration in dust samples from kindergartens and the maximum indoor mass concentration (193 $\mu g/m^3$) of particulate matter earlier found in kindergartens, we can calculate the maximal tungsten indoor air concentration of 0.0056 $\mu g/m^3$. Further research is needed to evaluate the cumulative effects of relatively low indoor concentrations of heavy metals in kindergarten and the child's home environment.

Conclusions
Increased tungsten concentration in the samples of natural dust aggregates taken from kindergartens is associated with a higher incidence of influenza among preschool-aged children. Measuring heavy metals in natural dust accumulations allows for assessing long-term aerosol pollution in preschool facilities and can support better planning of prevention strategies.
The study was funded by project S-MIP-23-128 of the Lithuanian State Research Council.

VANADIUM IN THE DUST OF KINDERGARTENS AND ITS RELATIONSHIP TO UPPER RESPIRATORY MORBIDITY IN CHILDREN

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Objectives
A significant and replicable correlation was found between the concentration of vanadium in samples of natural dust aggregates collected from primary schools and the incidence of acute upper respiratory infections in children (Prokopciuk N, 2024). Therefore, the aim of this work was to determine whether we would observe similar dependencies in kindergarten children.

Materials and Methods
In 22 kindergartens in Vilnius, a microelemental analysis of aerosol pollution was conducted using dust samples collected from areas of accumulation behind central heating radiators. The samples were analysed using a Spectro Xepos (XEPOS HE) and an ED-XRF spectrometer. The annual incidence of doctor-diagnosed acute infections of the upper respiratory tract (J00-J06) among children aged 3 to 6 years in each studied kindergarten was calculated for 2024

Results
The concentrations of heavy metals Pb, W, Sb, Sn, Zr, Zn, Cu, Ni, Mn, Cr, V, and As were determined in the dust samples to assess their potential impact on the occurrence of respiratory infections in children. A significant correlation was identified between vanadium concentrations and the incidence of acute infections of the upper respiratory tract in preschool children attending kindergartens. The F-statistic was 5.6, the p-value was 0.029, and $R^2 = 0.23$. The concentration of vanadium in the dust samples ranged from 8.86 to 27.58 ppm.

Conclusions
The concentration of vanadium in natural aggregates of aerosol pollutants taken from kindergartens is related to the incidence of acute infections of the upper respiratory tract among 3- to 6-year-old children, confirming the same findings from our previous studies at schools.
The study was funded by project S-MIP-23-128 of the Lithuanian State Research Council.

Critical Care and Emergency Medicine

TAKO-TSUBO CARDIOMYOPATHY IN A 17-YEAR-OLD AFTER CANNABIS INTAKE

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Objectives
Tako-Tsubo cardiomyopathy (TTCMP) is a well-known syndrome in adults, but a mystery in pediatrics, which is characterized by chest pain, left ventricular systolic dysfunction due to its apical ballooning, specific ECG changes, elevation of cardiac
enzymes and no visible changes in coronary angiography.
We report a case of a 17-year old patient with suspected stress cardiomyopathy after intake of cannabinoids.

Materials and Methods
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Results
A 17 year-old male patient was hospitalized due to chest pain, shortness of breath lasting for two days. He admitted smoking marijuana and experiencing a stressful break-up with his girlfriend several days before the onset of complaints. He was also known to be a professional athlete having no adjacent conditions.

Physical examination showed no pathological findings.
Blood tests showed highly elevated cardiac enzymes (CK-MB - 71,59mcg/l,Troponin T 2027ng/l), ECG showed elevated ST segment in V1-V4 derivations, multiple echocardiography evaluations showed transient hypokinesis of apex and lower part of left ventricle, low ejection fraction. No atherosclerotic plaques or stenosis were visualized on coronary angiography. All test results match Mayo Clinics' diagnostic criteria for TTCMP.
The treatment consisted of oxygen support, morphine infusion for pain management, NSAIDs, beta-blockers & ACEi.
The patient was discharged from the hospital with no cardiovascular complications.

Conclusions
It is important to consider TTCMP as a possible cause of acute chest pain in children, especially after suspected/proven use of narcotic substances.
Tako-tsubo CMP is a syndrome with a good prognosis, taking approximately 4 weeks for a recovery. Children should be followed up by pediatric cardiologists with a suspect of 10% recurrence and complications.

Endocrinology

"MYSTERY OF RAPID GROWTH AND HORMONAL WHISPERS": A CASE REPORT.

Olha Shevchenko [1]

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Objectives
To present a rare case of peripheral precocious puberty caused by a juvenile granulosa cell tumor of the ovary in a young girl, and to highlight the importance of early recognition, differential diagnosis, and the role of imaging and hormonal evaluation in distinguishing PPP from central causes.

Materials and Methods
Physical examination:
•4.8-y. o. girl, presented due to the thelarche (Tanner stage B2), obesity and rapid linear growth with no vaginal discharge, but estrogenized genitalia. No virilization.
No perinatal or morbid records of relevance, no use of medicine or products with phytoestrogens.
•The physical exam showed: height 125 cm (+5.7SD) and weight 47 kg (BMI 30.1).
•Pale skin,no light brown stains.
The following examinations have been done:
Bone age was advanced to 8 years(chronological age of 4.8 y.o)
Laboratory results: glucose – 4.6 mmol/L,HbA1c – 4.54%,insulin – 19.7 μIU/mL,HOMA index – 4.03,TSH – 2.58 mIU/L,T4f – 14 pmol/L,LH – <0.3 mIU/mL,FSH – 0.02 mIU/mL,Estradiol – 92 pmol/L,Prolactin – 18 ng/mL,17-OH-progesterone – 0.54 ng/mL.
After conducting a GnRH stimulation test with 0.1 triptorelin (Diphereline):
Stimulated LH – 1.02 mIU/mL, FSH – 3 mIU/mL.
Test was negative. No CPP.
A pelvic ultrasound was performed and revealed that the uterus and ovaries were bigger due to chronological age consistent with an 8-year-old, with visible follicles, but no tumor or cyst was found.
Some more laboratory tests for:
Alpha-fetoprotein (AFP)-28, and inhibin B-562 pg/ml (<80), AMH- 21 ng/mL.
Positive tumor markers
Granular cells secrete a large number of estrogens due to the overexpression of the aromatase enzyme, which stimulates the transformation of androgens into estrogens. The estradiol is responsible for symptoms of this disease; however, it is not useful as a marker in the follow-up due to the late increase .
The Inhibin B is synthesized by granular cells and it expresses itself in the follicles and can be used as a marker.

Results
Imaging:
The CT scan confirmed the abdominal expansive process (9x7x6cm) on the left, with a great effect of mass and a complex structure, probably from an ovarian origin. The uterus had an increased size (7cm long),a significant thickening in the endometrial cavity.
Treatment:
A left supine-oophorectomy was performed.
The histological study showed a tumor formed of big and medium cells, of moderate to abundant granular cytoplasm and nucleus slightly oval/pleomorphic, some showed the presence of follicles of different sizes.
The findings of the histology were compatible with juvenile granulosa cell tumor.
Prognosis:

2 months after the surgery, a control with CT showed no pathological findings.
Laboratory testing:estradiol-31pmol/L,inhibinB-55 pg/mlAMH-0.2ng/ml
A follow-up with Inhibin B or AHM every 6 months per 2 years, then annualy, without relapses in almost 4 years after the surgery.

Conclusions
A JGCT should be suspected in young girls with isosexual peripheral precocious puberty, especially when LH/FSH are suppressed and estradiol is elevated. Imaging and tumor markers(inhibin B) support the diagnosis. Surgery is the primary treatment and prognosis is excellent in localized disease.

4Q DELETION SYNDROME IN A TWO-YEAR-OLD BOY: A CASE REPORT

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Objectives
4q Deletion Syndrome in a Two-Year-Old Boy: A Case Report
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INTRODUCTION
Chromosome 4q deletion syndrome is a rare genetic disorder with highly variable clinical features, and it is estimated to occur in 1 in 100,000 people. Symptoms may vary significantly depending on the size and location of the missing chromosomal fragment, and whether it is terminal deletion or interstitial deletion. Among children with interstitial deletions, the most common features included abnormal skull, low-set ears, moderate to severe developmental delay, and postnatal growth failure.
To date, only a single case of a patient with 4q deletion syndrome treated with growth hormone has been reported in the literature. This index case described a 7-year-old boy with an interstitial 4q deletion, who presented with significant growth failure in early infancy. Growth hormone therapy was initiated at 13 months of age for small for gestational age indication. Following treatment, his height and weight improved significantly, rising from below the 1st percentile at 13 months to the 5th percentile by age 2. Since the age of 4 years, he has continued to grow along 10-12%.
CASE PRESENTATION
We present a case report of a 2-year-old boy with 4q deletion syndrome who was admitted to a paediatric endocrinologist for evaluation of short stature. He was born at 37 weeks of gestation after induction of delivery due to intrauterine growth retardation, which was detected on ultrasound examination at 34 weeks. The mother had no illnesses during pregnancy, no harmful habits, and no chronic disease. The child's birth weight was 1770 g (< 3rd percentile), length 43 cm, head circumference 30 cm, and chest circumference 28 cm. Nine days after birth, neurosonography findings were consistent with the child's age, and abdominal ultrasound showed no pathology. Due to mixed developmental delays and unspecified myopathy, brain MRI with spectroscopy was performed at the age of 1 year and 3 months. It demonstrated a small haemosiderin deposition in the left occipital horn following an intraventricular haemorrhage. Transthoracic echocardiography was also performed and had no pathological findings. Given motor delays, speech and language delays, and short stature, a 2-year-old boy underwent genetic analysis, which revealed an interstitial deletion of the long arm of chromosome 4. Chromosomal testing of the parents confirmed a normal karyotype in both.
Objectively, the boy presented with very dry skin, muscular hypotonia, and joint hypermobility. On examination, his weight was 7.7 kg (-4SD), and his height was 79.5 cm (-4SD), indicating significant growth retardation. He was at Tanner stage I, consistent with prepubertal status, with a testicular volume of 1 ml bilaterally.
DISCUSSION
Given the patient's current short stature and the limited evidence on the necessity and efficacy of growth hormone therapy in rare disorders such as 4q deletion syndrome, personalized care and a multidisciplinary approach, including thorough investigations prior to initiating therapy, are essential. It is particularly important to follow current growth hormone treatment guidelines as well as accepted national recommendations for growth failure. The patient meets the definition of SGA, so the current strategy would be expectant monitoring of growth parameters alone. The need for initiating growth therapy will be decided at 4 years of age according to currently accepted standards.

Materials and Methods
x

Results
x

Conclusions
x

DON' T FORGET CALCIUM

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Objectives

Disorders of calcium metabolism are relatively rare in children, and more often manifest with hypocalcaemia. Extremely low calcium levels may cause muscle aches, spasms, seizures, tingling and abnormal heart rhythm. These cases represent 2 different causes of hypocalcaemia in children.

Materials and Methods

x

Results

Case 1. A 9 years 4 months old girl with spastic hemiplegic cerebral palsy and epilepsy was first consulted by endocrinologist because of complains about dark spotting and initial hair growth in pubic region and on the face. She had history of 2 bone fractures at the ages 1 and 6 years. Bone age exceeded actual age by 2.6 years. Sex hormone tests did not show any pathology, but decreased calcium (1.77 mmol/l), increased phosphorus (2.69 mmol/l) with insufficient vitamin D (25-OH) level (25.4 ng/ml) were detected. Patient was recommended to start calcium and vitamin D substitution, and was assigned for follow-up with control laboratory tests (including PTH), but did not arrive. At the age of 10 years the patient was admitted to emergency room with seizure episode. Screening tests were performed, but secondary cause was not found. However, calcium levels were not checked. Later that year patient was consulted by orthopaedist because of left hand contracture in 4th and 5th fingers. Neurography was performed, but no signs of ulnar nerve damage were found. Patient was further sent to cranial, cervical and left brachial plexus MRI that revealed multiple, diffuse calcifications in basal ganglia, thalamus, cerebellum, nucleus dentatus, periventricular region, subcortical parts of cerebral hemispheres that were not present in previous examination in 2018. She was hospitalized for further investigations in department of neurology, and blood tests repeatedly showed hypocalcaemia (1.31 mmol/l), hyperphosphatemia (3.37 mmol/l), with vitamin D (25-OH) level of 29 ng/ml, as well as increased PTH level (219.3 pg/ml) that proves pseudohypoparathyroidism or PTH resistance. In addition, high-normal TSH and decreased thyroxine levels were detected that may indicate TSH resistance. Treatment with calcitriol, calcium and levothyroxine was initiated.

Case 2. A 9 years 7 months old girl was admitted to department of neurology because of repeated unprovoked fainting episodes, periodic muscle cramps in legs and arms (obstetrician’s hand), hoarseness of voice. Cranial and spinal MRI and echocardiography did not show significant pathology. EEG reflected multifocal epileptiform activity, but in ECG prolongation of QTc interval (485 ms) was observed. Blood tests demonstrated low calcium (1.32 mmol/l), high phosphorus (3.88 mmol/l) levels, low magnesium 0.64 mmol/l, insufficient vitamin D (25-OH) (28.75 ng/ml), high CPK (1191 U/l) and decreased PTH (0.4 pg/ml) level, confirming hypoparathyroidism. Autoimmune polyglandular syndrome and genetic causes were excluded. Electrolyte correction was first started intravenously with calcium gluconate and magnesium and further continued orally concomitantly with alfacalcidol and cholecalciferol.

Conclusions

These cases represent necessity of calcium and other electrolyte control in all cases of acute neurological symptoms, especially seizures and muscle spasms.

PHENOTYPING AND DEFINING DIFFERENCES BETWEEN METABOLICALLY HEALTHY AND METABOLICALLY UNHEALTHY OBESITY IN CHILDREN AND ADOLESCENTS - A PRELIMINARY DATA ANALYSIS

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Objectives

1.?To compare initial sociodemographic, anthropometric parameters and other clinical data, baseline laboratory parameters of cardiovascular health and glucose metabolism in children among different obesity phenotypes.

2.?To analyze the differences between metabolically healthy and metabolically unhealthy obesity in children in association with lifestyle and other risk factors.

Materials and Methods

This study included 50 children aged 10 to 18 years with BMI ≥95th percentile for age and sex. Included participants will be examined and sociodemographic, anthropometric parameters and other clinical data of weight, height, waist and hip circumference, blood pressure will be performed. The stage of sexual maturation will be assessed according to the Tanner scale, the signs of acanthosis nigricans will be determined. We will also assess body composition using bioimpedance analysis. MUO and MHO phenotypes will be determined according to the MHO 2018 Damanhoury et al consensus-based criteria: high density lipoprotein-cholesterol > 40 mg/dl (or >1.03 mmol/L), triglycerides ≤ 150 mg/dl (or ≤1.7 mmol/L),

systolic blood pressure ≤ 90th percentile, diastolic blood pressure ≤ 90th percentile, and fasting glucose ≤ 100 mg/dl (or ≤5.6 mmol/L).

Results
Results forthcoming

Conclusions
Conclusions forthcoming
1. Conflict of Interest:
All authors declare that they have no conflicts of interest to disclose.
2. Funding
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THE HYGIENE HYPOTHESIS IN THE DEVELOPMENT OF TYPE 1 DIABETES

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Objectives
There is a strong association between the incidence of immune-mediated diseases and improving standard of living and hygiene.
The aim of the DIABIMMUNE study, funded by EC 7FR Programme, was to assess the role of the hygiene hypothesis in the development of type 1 diabetes (T1D) using study populations from 3 neighbouring countries: Estonia, Finland and Russian Karelia.

Materials and Methods
The study comprised a birth cohort of ca 300 newborn infants from each country (Estonia, Finland and Russian Karelia) with increased genetic risk for T1D followed from birth up to the age of 3 years

Results
The dynamics of the human infant gut microbiome in 33 infants showed a marked drop in alpha-diversity in those who developed T1D in the time window between seroconversion and T1D diagnosis, accompanied by spikes in inflammation-favoring organisms.
Following the gut microbiome development from birth until age three in 222 infants we found that Bacteroides species are lowly abundant in Russians but dominate in Finnish and Estonian infants. Therefore, their lipopolysaccharide (LPS) exposures arose primarily from Bacteroides rather than from E. coli, which is a potent innate immune activator. We also showed that Bacteroides LPS is structurally distinct from E. coli LPS and inhibits innate immune signaling and endotoxin tolerance; furthermore, unlike LPS from E. coli, B. dorei LPS does not decrease incidence of autoimmune diabetes in non-obese diabetic mice.

Conclusions
The effects of the hygiene hypothesis in the development of Type 1 Diabetes the environmental effect is most likely mediated via the effect of environmental microbiota on the commensal microbiota influencing immunotolerance.

Gastroenterology and Nutrition

A CLINICAL CASE OF A PATIENT WITH INFLAMMATORY BOWEL DISEASE AND MULTIPLE COMPLICATIONS

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Objectives
Background
There is a growing global incidence of inflammatory bowel diseases (IBD), particularly among children, a pattern that is also observed in Lithuania. This case presents a patient with multiple complications and extraintestinal manifestations of IBD, that occur in about 30% of paediatric cases.
Case presentation
A 15-year-old male patient presented with complaints of bloody diarrhea occurring twice within a year. Colonoscopy revealed a few chronic ulcers in the ascending colon, histology demonstrated a chronic segmental granulomatous colitis,

and magnetic resonance enterography - a narrowed segment in the terminal ileum without stricture. Based on these findings, a Crohn's disease was diagnosed. The patient developed extraintestinal manifestations of IBD, including nasal septum defect, conjunctivitis, aphthous stomatitis, arthritis, and vasculitis during relapse periodes.
Management of this complex and severe condition was difficult to achieve. After induction with steroids, the first-line maintenance therapy with azathioprine had to be discontinued due to intolerance, which manifested as fever, vomiting, diarrhea, and neutropenia. Initiation of biological therapy with infliximab led to an anaphylactic reaction after the third induction dose. Switching to adalimumab resulted in only a partial response with short periods of remission that required weekly injections. Eventually, after another unsuccessful attempt with vedolisumab, treatment with ustekinumab was initiated and proved to be fully effective.
Surgical interventions were also required during the course of treatment. While on biological therapy, the patient experienced a relapse, presenting with symptoms of intestinal obstruction. An ileohemicolectomy was performed (40 cm. of ileum and 50 cm. of colon). Histological examination of the resected bowel revealed ulceration of colon with fissures surrounded by abscess-forming granulomas. Following the surgery, the condition worsened due to the formation of an abscess, which required external drainage.
Due to malabsorbtion and complicated condition parenteral and enteral feeding was introduced. As the patient experienced nausea both with oral intake and nasogastric tube feeding — a gastrostomy was performed and enteral nutrition was provided through it. At the age of seventeen, the patient achieved full persistent remission on ustekinumab treatment, surgical complications were resolved, the gastrostomy was closed, and the patient successfully reached adulthood.
At the age of 20, the patient was diagnosed with multiple sclerosis. Ustekinumab was replaced with a more suitable biological agent for multiple sclerosis — natalizumab — which has been successfully managing both multiple sclerosis and Crohn's disease for the past two years.

Materials and Methods

x

Results

x

Conclusions

Conclusion
The etiology of neurological extraintestinal manifestations in IBD is often immune-mediated and could be associated with brain–gut axis dysregulation.

ASSOCIATION BETWEEN SYMPTOM DURATION AND DISEASE ACTIVITY IN NEWLY DIAGNOSED PEDIATRIC INFLAMMATORY BOWEL DISEASE

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Objectives

Pediatric inflammatory bowel disease(IBD) is a chronic condition where early diagnosis is crucial to prevent complications and ensure timely treatment.We aimed to:
1.Describe the demographic characteristics of newly diagnosed pediatric IBD patients at our center.
2.Assess symptom duration prior to IBD diagnosis and mostly utilized diagnostic tests.
3.Evaluate disease activity using the Pediatric Ulcerative Colitis Activity Index(PUCAI) and Pediatric Crohn's Disease Activity Index(PCDAI)and its association with symptom duration and inflammatory markers,(CRP,fecal calprotectin).

Materials and Methods

A retrospective analysis of electronic health records of pediatric patients diagnosed with IBD between 2019 and 2024.Exclusion:incomplete endoscopic data or undocumented symptom duration.

Results

A total of 72 pediatric IBD cases were identified between 2019 and 2024,65 were included in the analysis after excluding 7 cases with incomplete endoscopic data.The median age was 14 years (IQR 12–16),with 53.8%(n=30) male and 66.2%(n=43) residing in urban areas.
Ulcerative colitis(UC) was the predominant diagnosis,accounting for 61.5% of cases(n=40),while Crohn's disease(CD) was diagnosed in 38.5% of patients(n=25),with no statistically significant differences in sex,age,or place of residence between the groups.
CRP and fecal calprotectin levels were available for the majority of patients (CRP:n=58,89.2%;calprotectin:n=60,92.3%). Median CRP levels were significantly higher in patients with CD (16.0 mg/L,IQR 5.0–28.0) compared to those with UC(5.0 mg/L, IQR 5.0–8.0), as determined by the Mann–Whitney U test(U=214.00,p=0.002).
Median fecal calprotectin levels were similar between diseases:930.0 µg/g(IQR 431.3–2100.0) in CD and 1000.0 µg/g(IQR 167.0–2100.0)in UC.No statistically significant difference was observed between the groups (U=379.00, p=0.477).
Symptom duration prior diagnosis was significantly longer in patients with CD compared to UC,at 14.24 months(standard deviation[SD] 16.68;range 1–72 months)versus 5.68 months(SD 9.72;range 1–60 months),respectively(p = 0.01).No

statistically significant correlations were found between symptom duration and CRP or fecal calprotectin levels.
Mean PUCAI and PCDAI scores indicated mild disease activity in UC(35.9±16.4) and CD(20.4±13.6),respectively.A Spearman's rank correlation analysis revealed a moderate,statistically significant negative correlation between symptom duration and PUCAI scores(ρ=−0.471,p=0.002,n=34),indicating that a longer duration of symptoms was associated with lower disease activity at presentation.Additionally,a moderate positive correlation was observed between CRP levels and PUCAI scores(ρ=0.361,p=0.036,n=34),suggesting that elevated CRP levels were associated with increased disease activity in patients with UC.No significant correlation was found between PUCAI scores and fecal calprotectin levels.PCDAI scores showed no significant correlations with symptom duration,CRP,calprotectin in CD group.

Conclusions
UC was more prevalent than CD with no demographic differences between groups.Crohn's disease was associated with longer diagnostic delays and higher CRP levels,though CRP did not correlate with disease activity.In ulcerative colitis,longer symptom duration was linked to milder disease,while higher CRP correlated with greater severity.These findings suggest that diagnostic delay may influence disease presentation in pediatric ulcerative colitis and highlight the need for timely recognition of IBD symptoms in children.

HELICOBACTER PYLORI ANTIMICROBIAL RESISTANCE IN LITHUANIAN CHILDREN: A 12-YEAR REVIEW

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Objectives
Background: Increasing rates of antimicrobial resistance in Helicobacter pylori (H.pylori) are a recognized global challenge, particularly in pediatric populations where treatment options are more limited. Eradication failure is closely linked to resistance, especially to clarithromycin and metronidazole, the main components of first-line therapy. Therefore, local surveillance studies are essential to support rational antibiotic use and formulating updated clinical guidelines.
Aim: To assess trends in antibiotic resistance of H.pylori strains isolated from children in Lithuania between 2013 and 2025 and to evaluate age-related susceptibility patterns.

Materials and Methods
This retrospective study analyzed microbiological data from pediatric patients aged 1 to 17 years who underwent upper endoscopy at a tertiary care children's hospital between 2013 and 2025. Gastric mucosal biopsies were collected and cultured for H.pylori. Bacterial identification was carried out via MALDI-TOF mass spectrometry (Bruker). Antimicrobial susceptibility testing was performed using minimum inhibitory concentration (MIC) gradient test strips (Lyophilchem), and results were interpreted according to EUCAST guidelines. The antibiotics tested included amoxicillin (AMO), clarithromycin (CLR), metronidazole (MTZ), levofloxacin (LEV). Statistical analysis was used to detect trends over time and assess potential age-related differences in resistance patterns.

Results
Out of 744 pediatric patients (1-17 years), H.pylori was successfully cultured in 458 cases (5-17 years). No statistically significant differences in resistance rates were observed between children aged <10 and those ≥10 years. The highest overall resistance was observed to clarithromycin (31.2%), followed by metronidazole (21.6%). Resistance to levofloxacin was low (5.0%), while amoxicillin resistance was negligible (<1%).
Longitudinal analysis demonstrated dynamic trends in antimicrobial resistance. Clarithromycin resistance was highest between 2013 and 2015 (46–48%), followed by a steady decline, reaching 31% by 2025. Metronidazole resistance increased markedly from 11% in 2013 to 31% in 2017 and remained at approximately that level through 2025, showing no further significant changes. In contrast, resistance to levofloxacin and amoxicillin remained consistently low throughout the study period, with minimal variation between years. Notably, levofloxacin resistance never exceeded 5% annually.

Conclusions
Over a 12-year observation period, resistance to clarithromycin and metronidazole remained alarmingly high among Lithuanian pediatric patients, underscoring the need for antibiotic stewardship and alternative treatment strategies. The consistently low resistance rates to amoxicillin and levofloxacin support their continued inclusion in therapeutic regimens. These findings highlight regular surveillance's importance in guiding effective, evidence-based treatment protocols for pediatric H.pylori infections.

IMPACT OF WAR-RELATED STRESS ON FUNCTIONAL GASTROINTESTINAL DISORDERS IN UKRAINIAN CHILDREN: A SURVEY-BASED STUDY

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Objectives

This study aimed to assess the prevalence of functional gastrointestinal disorders (FGIDs) in Ukrainian children aged 6 to 18 who experienced war-related stress due to displacement or continuous exposure to military actions. The primary objective was to evaluate correlations between stress factors and symptoms related to disorders of gut-brain interaction (DGBI).

Materials and Methods

An anonymous online survey was conducted among parents/guardians of Ukrainian children affected by war (n=205). Participants were categorized into four groups: 1 gr. refugees abroad (n=75), 2 gr. internally displaced (n=28), 3gr. temporarily displaced but returned (n=41), and 4gr. those who remained in their hometowns (n=61). Based on Rome IV criteria, the questionnaire assessed gastrointestinal symptoms (e.g., abdominal pain, nausea, constipation), psycho-emotional status, and stress-related environmental factors. Data were analyzed using descriptive statistics and correlation analysis. The two-sample Kolmogorov-Smirnov test was applied to compare distributions between groups, and Fisher's (φ-test) exact test was used to determine statistical significance.

Results

Refugee children (group 1) reported the highest number and intensity of symptoms associated with functional gastrointestinal disorders, including upper and lower abdominal pain, early satiety, nausea, bloating, and constipation. These symptoms were significantly more frequent than in other groups ($p \leq 0.01$-0.05). Notably, the highest proportion of parents reporting a general deterioration in their child's health was observed in the internally displaced group (group 2), where 71.42% noted worsening health after the onset of war. Children in groups 3 and 4 reported fewer symptoms overall, which may suggest a potentially protective effect of familiar environment.

Conclusions

The findings highlight a strong association between war-induced stress and DGBI symptoms in children, particularly among refugees and internally displaced populations. These results underscore the need for targeted psycho-emotional and medical support for children exposed to prolonged stress and displacement. Early identification of DGBI and implementation of trauma-informed care strategies are essential to mitigate long-term health consequences in conflict-affected pediatric populations.

SUCCESSFUL MANAGEMENT OF SHORT BOWEL SYNDROME – RELATED INTESTINAL FAILURE IN A 7-YEAR-OLD BOY: A CASE REPORT

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Objectives

Background

The incidence of intestinal failure due to short bowel syndrome (IF-SBS) is 24 cases per 100,000 live births. Although the survival rate currently reaches 95%, the management of this condition remains challenging [1]. This case report describes a successful treatment of a child with IF-SBS, resulting in the achievement of enteral autonomy and full recovery.

Case presentation

A 7-year-old male underwent small bowel resection due to intestinal thrombosis secondary to a coexistence of portal vein malformation and familial thrombocythemia. The residual bowel length was limited to 20 cm of jejunum from the ligament of Treitz, 20 cm of ileum to the ileocecal angle, and the entire large intestine. Jejunostomy and ileostomy were performed. The patient experienced a challenging recovery due to early infectious complications. Antibiotics, antifungals, and parenteral nutrition (PN) were initiated in the subsequent days.

Despite the efforts, the patient's condition deteriorated. Abdominal CT showed peritonitis and hemoperitoneum, prompting a relaparotomy. The colon appeared markedly inflamed, yet the surgical team opted against a high-risk resection. Fortunately, this episode resolved without the need for radical treatment.

PN with gentle introduction of enteral feeding was continued for 367 days post-resection. Despite having short bowel syndrome (SBS), the boy resumed a normal lifestyle and now sustains his needs by an oral diet supplemented with nutritional drinks.

Discussion

The suggested definition of SBS is 180-200 cm of residual small bowel in adults or less than 25% of small bowel that is expected for gestational age in neonates, along with prolonged need for PN [2]. However, the criteria remain unclear in

pediatric age groups. Intestinal failure is a reduction in functional intestinal mass insufficient to sustain life, requiring PN for at least 60 days within a 74-day consecutive interval [3]. The presented patient matches this definition. It is known that intestinal adaptation begins after a significant reduction of bowel mass and might be stimulated by a gradual introduction of oral and enteral nutrition [4]. Our patient successfully transitioned from PN after 367 days since resection. This duration is similar to or shorter than the ones documented in the literature [5–8]. Animal studies confirm that the adaptive response starts as early as 24 - 48 hours post-resection and is more effective in children compared to adults [9]. Adaptation involves villi elongation, deepening of crypts, increased enterocyte proliferation, and apoptosis. Multiple mediators contribute to this process: glucagon-like peptide-2, insulin-like growth factor-1, and growth hormone mostly exhibit proliferative activity, while epidermal growth factor stimulates cell differentiation [10–14]. Furthermore, small bowel length over 30 cm, preservation of the ileocecal valve, the ileum, and the entire colon are associated with faster enteral autonomy [15]. The ileum has more pronounced adaptive processes than the jejunum [16]. In this case, the episode of an inflamed colon resolved without radical treatment, hence the boy maintained all of these positive predictors, explaining his successful recovery.

Conclusion

This case report highlights the potential of positive outcomes in pediatric patients with IF-SBS by emphasizing the underlying pathogenetic mechanisms. More precise definitions and future research are needed for optimizing nutritional strategies.

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Materials and Methods
x

Results
x

Conclusions
x

Infectious Diseases, Vaccination and Immunology

CASE STUDY OF BCG VACCINE INOCULATION SITE HAEMORRHAGE AND ULCERATION

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Objectives
Bacillius Calmette–Guerin (BCG) is a live attenuated vaccine derived from a strain of Mycobacterium bovis, has been used to prevent tuberculosis since 1921. All children advised to receive BCG vaccination prior to discharge from maternity wards in Latvia, included in national immunization program. The vaccine is effective against the disseminated tuberculosis and severe forms such as meningitis in childhood. BCG Danish strain 1331 in dose 0,05 ml is used in recent years in Latvia.
Although the BCG vaccine is usually a safe vaccine, a number of complications can occur, more often local reactions, regional lymphadenitis or inoculation site abscess, with lymphadenitis being the most common complication. Technical factors are still considered the most significant in the development of vaccine-related complications.
We report a clinical case of BCG vaccine inoculation site haemorrhage and ulceration in an infant. The baby presented on 29.06.2025. with complaints of bleeding out of the inoculation site in the previous 2 days.

Materials and Methods
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Results
He is a second-born baby from twin pregnancy, dichorionic diamniotic, at 37th gestational week. The baby received BCG vaccine on a discharge day, 31.03.2025., as well as his twin-sister.
The baby was examined on following days after discharge, at 1 and 2 months of age. At the last appointment, 30.05. twins received their vaccines (Infanrix, Rotateq and Vaxneuvance) according to Latvian immunization program. No complaints of complications were received after that.
At the last appointment at 2 months BCG inoculation site looked like induration: 10 mm on brother's arm and 7 mm on sister's arm.
After that mother noticed some changes in the inoculation site in the baby-brother, about 2.5 months of age, induration began to grow to papula filled with blood. On 27.06.2025. the papula ulcerated and bled. The baby was then seen by a pediatrician on 29.06.2025. In the inoculation site there was a 12 mm ulcer with caseous content. No systemic symptoms were reported and noticed on examination, no lymphadenitis was present. No treatment was required.

Conclusions
Correct vaccination techniques remain the most important factor in the prevention of vaccine-related complications. Immunocompetent infant usually do not require any treatment for reactions like lymphadenitis, abscess of the inoculation site or haemorrhage and ulceration after BCG vaccination. Careful observation, parent information and wait-and-see tactics all is needed in similar cases.

COMPARING THE MICROBIOME OF THE ADENOIDS IN CHILDREN WITH SECRETORY OTITIS MEDIA AND CHILDREN WITH HEALTHY EARS

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Objectives
Our study aims to characterize and compare the surface microbiome of adenoid tissues in children with and without otitis media with effusion. By identifying specific microbial signatures, we seek to clarify the role of the adenoid microbiome in OME pathogenesis. We conducted a targeted, 16S rRNA-based sequencing analysis of adenoid surface samples collected

during routine adenoidectomies from children aged 3 to 7. We compared samples from children with OME to those from children without middle ear pathology.
By characterizing these microbial communities, we aim to lay the groundwork for microbiome-informed strategies in the prevention, diagnosis, and treatment of URT infections, particularly otitis media with effusion.

Materials and Methods
Adenoid tissue samples were collected from 40 children aged 3 to 7 years undergoing adenoidectomy between October 2022 and October 2023. Participants were divided into two groups: one with otitis media with effusion, and one without middle ear effusion
Microbial DNA was extracted using the FastDNA and the V3-V4 region of the 16S rRNA gene was amplified. Sequencing on Illumina MiSeq platform. Quality control, denoising, and taxonomic classification were conducted using QIIME2, with reference to the SILVA database. Statistical analyses included alpha diversity metrics, PCA, PERMANOVA, and differential abundance testing using Maaslin2.

Results
We analyzed 40 adenoid tissue samples from children, split evenly
Alpha diversity was measured using the Inverse Simpson, Shannon, and Piellou's indexes. Regarding The Inverse Simpson index and Shannon's index - neither showed statistically significant differences between groups ($p > 0.05$). However, Piellou's evenness was significantly lower in the OME group (0.45 vs. 0.57; $p = 0.036$), indicating a less even distribution of bacterial taxa.
Beta diversity analysis revealed broader microbial variation in the non-OME group. Despite this trend, PERMANOVA analysis showed no statistically significant group-level differences ($p = 0.46$, $R^2 = 0.025$).
Proteobacteria was the dominant phylum in both groups, more abundant in non-OME children. Firmicutes, Fusobacteriota, and Bacteroidota followed, with the OME group showing relatively higher levels of the latter three.
At the genus level, Haemophilus, Streptococcus, and Moraxella were most common in the non-OME group. In contrast, Fusobacterium was dominant in the OME group, followed by Haemophilus and Streptococcus. Genera such as Peptostreptococcus, Alloprevotella, and Burkholderia were more prevalent in OME cases, suggesting community shifts associated with middle ear effusion.
Maaslin2 analysis revealed six genera with significant differences between groups. Alloprevotella showed the strongest statistical association (coefficient = 2.71, p-adjusted = 0.054), while Bordetella showed the highest effect size (coefficient = 4.75, p-adjusted = 0.249). These genera may play a role in microbiome shifts linked to OME.

Conclusions
Our study revealed significant differences in bacterial diversity on children's adenoids linked to middle ear health. Patients with healthy ears showed greater evenness with dominance of Haemophilus and Streptococcus, while patients with otitis media with effusion had more variable microbiomes, abundant in Fusobacterium, Peptostreptococcus, and others. These microbial shifts may contribute to infection. Future studies should increase sample size and include healthy controls despite collection challenges.

MYCOPLASMA PNEUMONIAE INFECTION ASSOCIATED EXTRAPULMONARY MANIFESTATIONS IN CHILDREN

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Objectives
Background. Mycoplasma pneumoniae is a frequent bacterial pathogen causing community-acquired pneumonia in children 5 years and older. This pathogen is known to cause various extrapulmonary manifestations, including gastrointestinal, neurological, hematological, cardiovascular, and dermatological.
Case presentation.
Case 1: In October 2024, a 9-year-old girl started to experience cough and febrile temperature elevations. Two days later an X-ray of the chest was taken, which showed infiltrative opacity in the lower lobe of the left lung suggestive of pneumonia. She was prescribed 400 mg of Amoxiclav twice daily. The next day she started to experience cramping abdominal pain, vomiting 1 to 3 times a day, and loose stools. Her symptoms persisted, and three days later she was admitted to the Children's Clinical University Hospital. Crepitations were audible over the left lung during auscultation. Laboratory tests showed elevated titers of anti-Mycoplasma pneumoniae IgM. Biochemical tests revealed elevated lipase levels at 154.68 U/L. A day later, the lipase level reached 215.78 U/L, and the amylase level was elevated to 105.50 U/L. An abdominal ultrasound showed an enlarged pancreatic head measuring 2.4 cm in thickness. Viral antigen detection and stool culture were negative. The diagnosis of acute pancreatitis was made. She received therapy with Azithromycin and was discharged with follow-up recommendations.
Case 2: In March 2025, a two-and-a-half-year-old girl was admitted to a regional hospital with complaints of headache, nausea, and vomiting. During her hospitalization in the regional hospital, she experienced two seizures with generalization without regaining consciousness between episodes. The patient was intubated, received Ceftriaxone, Acyclovir, Fentanyl and Midazolam infusions and was transferred to the Children's Clinical University Hospital. Initially, the patient was admitted

to the Intensive Care Unit, but later that day she was transferred to the Neurology Department. In the Neurology Department the neurological examination was done and revealed no abnormalities. During hospitalization in the Children's Clinical University Hospital, seizures did not reoccur. Cerebrospinal fluid analysis showed no significant changes, the meningitis/encephalitis panel was negative. Diagnostic tests, including head CT, MRI, and EEG, revealed no pathological changes. Laboratory tests revealed a high titer of anti-Mycoplasma pneumoniae IgM, later a nasopharyngeal swab was also taken and tested positive for Mycoplasma pneumoniae. On the fifth day of hospitalization, on lung auscultation, diminished breath sounds were heard at the lung bases, with greater reduction on the left side. A chest X-ray showed initial left basal pneumonia. The patient was treated with Azithromycin and discharged with recommendations. It is important to note that other potential causes of seizures, including infections by other pathogens, febrile seizures, and epilepsy, were excluded.
Conclusion. Mycoplasma pneumoniae is primarily recognized as a respiratory pathogen; however, it can also be associated with various extrapulmonary manifestations, including gastrointestinal and neurological, as demonstrated in these clinical cases. Neurological presentations are considered one of the most common extrapulmonary manifestations, while Mycoplasma pneumoniae infection-associated acute pancreatitis is a rare manifestation with only five published cases in the past 25 years.

Materials and Methods

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Results

x

Conclusions

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TRENDS IN RESPIRATORY SYNCYTIAL VIRUS (RSV) INFECTIONS IN HOSPITALIZED CHILDREN OVER THE PAST DECADE

Piia Jõgi **[1], Berit Lomp [1], Eda Tamm [1], Krista Lõivukene [2], Hiie Soeorg [3]**

1. Children's Clinic of Tartu University Hospital, **2.** United Laboratories of Tartu University Hospital, **3.** Medical Microbiology, University of Tartu

Objectives

RSV is a leading cause of lower respiratory tract infections in infants and young children. COVID-19 measures affected the epidemiology of many infectious diseases. Recently introduced RSV prevention includes long-acting monoclonal antibodies (nirsevimab/Beyfortus) and maternal vaccination (Abrysvo).
This study aimed to describe demographic and clinical characteristics of children hospitalized with RSV at the Department of Acute Infections, Children's Clinic, Tartu University Hospital, focusing on the impact of the COVID-19 pandemic.

Materials and Methods

A retrospective analysis of patients aged 0–<19 years hospitalized with lab-confirmed RSV infection from 01.01.2016 to 01.06.2024 was conducted. Diagnoses were ICD-10-based, confirmed by antigen or PCR from nasopharyngeal/tracheal aspirates. Periods: pre-COVID (2015/16–2019/20), COVID (2020/21), post-COVID (2021/22–2023/24). Demographic and clinical data were extracted from electronic medical records.

Results

Of 17,425 patients, 598 (3.4%) had RSV infection. The mean age was 1.1 (±1.6 SD) years; 67% were <1 year. Of all study subjects, 12.2% had chronic conditions and 11.0% were born prematurely; however, none received prophylaxis with palivizumab during the same RSV season as their hospitalization. Bronchiolitis, bronchitis, pneumonia and RSV as the cause of diseases classified elsewhere was diagnosed in 64.0%, 27.3%, 4.2% and 4.5%, respectively. The mean time from symptom onset to hospitalization was 3.8 days (±0.9 SD), and the mean duration of hospital stay was 4.1 days (±2.54 SD). Respiratory support was required in 66.0% of patients, of whom 90.9% received supplemental oxygen, 8.1% required non-invasive respiratory support, and 0.8% were intubated. A total of, 4.3% of hospitalized patients required treatment in the intensive care unit (ICU), with a mean ICU stay of 4.0 days (±2.6 SD). No deaths occurred during the study period.
On average, 66 patients (±41 SD) were hospitalized per season, and the mean duration of each RSV season was 156 days (±83 SD).
The number of hospitalized patients was 285 in the pre-COVID period, 0 during the COVID year, and 313 in the post-COVID period. The average number of RSV hospitalizations per season increased from 57.0 (±11.3 SD) in pre-COVID to 104.3 (±49.3 SD) in post-COVID period, $p<0.05$. The RSV season lengthened from 145 days (±34 SD) in the pre-COVID period to 227 days (±74 SD) in the post-COVID period, $p<0.05$.
In the pre-COVID period, the time from symptom onset to hospitalization was shorter compared to the post-COVID period — 3.4 days (±0.7 SD) vs. 4.6 days (±0.4 SD), $p<0.05$. However, the duration of hospitalization was longer in the pre-COVID period — 4.2 days (±0.5 SD) vs. 3.9 days (±0.3 SD), $p<0.05$.
Post-COVID patients were older (1.3±1.9 SD vs. 0.9±1.2 SD years, $p<0.05$) and more often required respiratory support (71.3% vs. 60.0%, $p<0.05$) then pre-COVID patients. Although the number of patients requiring intensive care and the duration of intensive care were similar in both period.

Conclusions
The COVID-19 pandemic affected RSV-related hospitalizations. In the post-COVID period, children hospitalized due to RSV infection were older, and the course of illness was more severe compared to the pre-COVID period. As the majority of hospitalized patients were previously healthy infants, hospitalization rates due to RSV infection could potentially be reduced by using the new monoclonal antibody nirsevimab for RSV prophylaxis or by maternal immunization with RSV vaccine.o

Neonatology

ESTABLISHING FEEDING PATTERNS OF LATE PRETERM NEONATES IN RIGA MATERNITY HOSPITAL

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1. Riga Maternity Hospital, Daugavpils Regional Hospital, **2.** Riga Maternity Hospital, Children's Clinical University Hospital

Objectives
To define patterns of any possible feeding in late preterm neonate after delivery and beyond.

Materials and Methods
Statistical analysis of data from Riga Maternity Hospital from 2022 till 2024. We included late preterm babies that did not need any intensive care. We look for a group of randomly chosen 30 babies closer to define any particular patterns of assistance for feedings.

Results
In 2022, 2023, 2024 there were 247, 188 and 199 babies born late preterm that did not need any neonatal intensive care. It was 4.8%, 4.2% and 5.0% of all born babies in Riga Maternity Hospital accordingly. Nearly half of them started breastfeeding in the delivery room. Babies that discharged with exclusive breastfeeding were even less: 38% in 2022, 22% in 2023, and 19% in 2024. Most of the babies received formula as an additional supplement. In the group of the 30 selected babies only 2 babies discharged with exclusive breastfeeding (born at 36+6 gestational age (GA)) and 2 with breastfeeding and expressed mothers milk (36+2 and 36+6). Only 18 babies out of 30 were breastfed in the delivery room. Three babies received supplementation due to hypoglycemia (all 36 GA). Two babies achieved critical weight loss (-10.6 and -10.3%). There were 19 babies out of 30 that got expressed mother’s milk. Two mothers mentioned that they would like to feed a baby only formula. Median weight loss was -6.1% in 34GA, -6.15% IN 35GA, and -6.5% in 36GA.

Conclusions
Late preterm neonate and infant breastfeeding is a challenge in many hospitals and for many families as well as family physicians. Late preterm dyad (a mother and a baby) is prone to breastfeeding difficulties from the start due to diminished activity of a baby to wake up for feedings, stay active at the breast, prematurity health issues and necessity of additional breast milk expression by a mother. Most of these challenges could be addressed in the maternity wards.
For stable otherwise healthy babies, that were born late preterm, initial breastfeeding should be started in the first 2 (preferably 1 hour) of birth. Late preterm babies tended to receive too early and too often supplementation. Guidelines should be accepted to manage feedings of late preterm babies and additional support system for mothers of those babies. That would minimize sequential hospitalization for these babies due to poor feedings and weight loss.

Oncology/Hematology

WHEN PLATELETS TAKE A DIVE: UNDERSTANDING IMMUNE THROMBOCYTOPENIA IN CHILDREN

Elizabete Cebura [1]

1. Children's Clinical University Hospital

Objectives
To analyze the latest guidelines and compare them with existing practices in paediatric ITP management in order to develop an appropriate approach

Materials and Methods
A literature review of the latest sources on pediatric hematology, combined with a retrospective analysis of available data on pediatric ITP cases treated at the Children's Clinical University Hospital.

Results
Over the 5-year study period, 242 hospital episodes of ITP were recorded in 85 individual patients. Twelve patients experienced ≥3 hospital episodes, and 13 patients received at least a two-drug combination regimen during a single hospital stay. Seven patients developed chronic ITP; five of them required second-line therapy with thrombopoietin receptor agonists (TPO-RA). One patient with persistent ITP also received TPO-RA and achieved full recovery.Severe bleeding in ITP develops in very rare case and has not been observed at our centre.
According to guidelines in most cases, treatment is not necessary so the indications for treatment must be strictly evaluated. Only one of the criteria for treatment is platelet count, but the most important criterion is the presence and severity of bleeding signs. The proximity of a treatment centre, social conditions, and lifestyle must also be considered.

Conclusions
Most patients with ITP recover spontaneously and do not require treatment. According to current guidelines, for patients who do require treatment, the first-line treatment choice remains unchanged: intravenous immunoglobulins or corticosteroids.
Second-line treatment option TPO-RA that is a new opportunity to avoid therapeutic methods such as use of immunosuppressive drugs and splenectomy thereby lessen the treatment side effects and significantly improving their quality of life, especially in patients with chronic ITP.

Other

PRENATAL CONGENITAL HEART DISEASE DETECTION RESULTS IN LATVIA 2019-2024.

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1. RSU, Department of Pediatrics, Children university hospital, **2.** RSU Department of Pediatrics, Children university hospital

Objectives
Congenital heart disease (CHD) remains the most common pathology of birth defects, with significant implications for neonatal morbidity and mortality. Advances in prenatal imaging—second-trimester anomaly scans and targeted fetal echocardiography—have improved diagnostic accuracy, yet certain lesions remain challenging to identify before birth. High prenatal detection rates are critical, as early identification enables optimized perinatal management, delivery planning in specialized centers, timely initiation of medical or surgical interventions, and informed parental counseling. Improved detection directly correlates with better survival outcomes, reduced perioperative complications, and enhanced long-term quality of life.

Materials and Methods
Analysis of fetal echocardiography examinations in Latvia ovwer a six -year period (2019-2024)evaluation of 627 diagnosed CHD and comparison with postnatal diagnosis and autopsy findings.

Results
Evaluating the percentage of CHD detection over the past six years reveals a positive trend, increasing from 66.8% in 2019 to 70.5% in 2024. While this may not represent a substantial improvement, analysis of the gestational age at diagnosis shows that 92.7% of cases were identified before 24 weeks of gestation, and 25% before 16 weeks in 2024 . In 2019 only 45 % before 24 weeks and 14 % before 16 weeks of pregnancy.

Conclusions
Continued educational efforts and promotion of early diagnostic screening remain essential.

CLONIDINE AS AN EFFECTIVE TREATMENT FOR SLEEP DISTURBANCES IN A CHILD WITH NEUROFIBROMATOSIS TYPE 1: A CASE STUDY

Klinta Mozule [1], **Una Purviņa** [2], **Marta Celmiņa** [2]

1. Riga Stradiņš University, **2.** Children's Clinical University Hospital

Objectives
Background:
Sleep disorders affect one-quarter of the paediatric population and are even more common among children with

neurodevelopmental disorders (NDDs). In this group disturbed sleep is of particular significance, exacerbating functional impairments associated with the underlying disease and affecting the well-being of the entire family. Furthermore, these children often have multiple comorbidities, making standard treatment methods difficult to implement.
Aim:
To demonstrate the difficulties encountered in the treatment of severe sleep disturbances in a child with NDD and the importance of personalised and appropriate treatment.

Materials and Methods
A case study based on retrospective analysis of the patient's clinical history and medical documents, including sleep diary evaluation. Informed consent has been obtained.

Results
A four-year-old girl diagnosed with neurofibromatosis type 1 in infancy and associated comorbidities had difficulties in initiating and maintaining sleep, experienced restlessness during nocturnal awakenings and episodes of self-harm. The sleep disturbances negatively affected both the girl's and her mother's sleep and well-being during the day. Previous trials with an antihistamine, an anxiolytic, and immediate-release melatonin offered no relief. Prolonged-release melatonin was recommended, however, it is available only in tablets, which are not small enough to be administered through the percutaneous endoscopic gastrostomy feeding tube, which the girl has had since the age of one due to gastroesophageal reflux disease-related feeding difficulties. A low dose of clonidine was initiated instead, which proved effective, allowing the girl to sleep through the night, diminishing daytime manifestations of sleep disturbances, and providing relief for her mother.

Conclusions
This case illustrates that, especially for children with NDDs, the process of finding an effective treatment is often lengthy and may require multiple adjustments to therapy. However, the proper treatment promotes the quality of sleep and well-being of the child and their family.

HELPING ABUSED CHILDREN: CHANGES AND CHALLENGES

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Objectives
Child abuse severely disrupts a child’s development, leading to social instability, heightened aggression, and the perpetuation of violence across generations. Abused children face a higher risk of psychological and behavioural disorders, and those who are socially neglected may be at an increased risk of suicide. Infants who suffer physical abuse often develop serious neurological complications.
Children rarely disclose their abuse. They may not recognise that an adult’s behaviour is inappropriate and often feel guilty, intimidated, or ashamed. Many are unsure how to express their experiences and fear that they will not be believed.
The United Nations General Assembly adopted the Convention on the Rights of the Child in 1989. Lithuania signed the Convention in 1991 and ratified it on 30 January 1995. Article 19 of the Convention requires state parties to protect children from all forms of violence—physical, psychological, and sexual—regardless of whether the abuse is inflicted by parents, guardians, or other adults.
Despite ratification, implementing effective protective measures in Lithuania has been a long and complex process. Historically, only the most severely injured children—such as infants with shaken baby syndrome—were admitted to hospitals. Paediatricians often worked in isolation, without the support of social workers or psychologists. Reporting injuries was not mandatory and largely depended on the individual doctor’s initiative, while police responses were frequently slow and inadequate. Diagnosing shaken baby syndrome posed additional challenges because its signs were often subtle, leading to misdiagnosis or underreporting.
To address these issues, our Paediatric Intensive Care Unit established a shaken baby registry in 1994, which revealed alarming local data. In response, we began employing social workers and psychologists, improved our communications with police investigators, and expanded opportunities for forensic examinations.
Since 2001, changes in Lithuania’s legislative framework have enabled healthcare institutions to report suspected abuse without parental consent when necessary to protect a child’s rights. The situation further improved with the child protection reform initiated in 2017, which enhanced collaboration among hospital wards, child rights specialists, and law enforcement. Moreover, a university course on “Child Abuse Diagnostics” was developed in 2018 and will become mandatory for all healthcare providers serving children in Lithuania starting in 2025. Today, dedicated social workers in the emergency paediatric department and inpatient wards, along with psychologists focusing exclusively on children, provide crucial support. Extensive collaboration is underway with the non-governmental organisation „Volunteers for Children“, which assists in supervising children from socially at-risk families.
Despite these improvements, significant challenges remain. Access to qualified support is still limited, there is a shortage of psychologists, and societal attitudes toward abuse change slowly. A survey conducted in December 2024 revealed that 45% of Lithuanian parents continue to use physical punishment. Continued efforts are essential to further protect children and support those who have experienced abuse.

Materials and Methods
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Results
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Conclusions
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PARENTAL PERSPECTIVES IN PEDIATRIC PALLIATIVE CARE: EVALUATION OF PROVIDED SERVICES, FAMILY NEEDS, AND SELF-ASSESSED QUALITY OF LIFE

Anda Jansone [1], **Sindija Bergmane** [1]

1. Children's Palliative Care Society

Objectives
Introduction. In Latvia, pediatric palliative care (PPC) is provided by interdisciplinary teams based at the Children's Clinical University Hospital and Liepāja Regional Hospital, in collaboration with the Children's Palliative Care Society. These teams—comprising doctors, nurses, a social worker, chaplain, and psychologist—offer a holistic approach to caring for children with life-limiting or life-threatening illnesses and their families. The aim is to alleviate suffering, improve quality of life (QOL), and support all family members. Since 2021, a regular family satisfaction survey has been conducted to evaluate service quality and identify areas for improvement.
Objectives. To evaluate service quality from parents' perspectives, to identify key family needs, and to assess QOL in order to inform service improvement.

Materials and Methods
Materials and Methods. In 2024, an anonymous survey was conducted among parents and guardians of children receiving PPC. A total of 111 responses were received, 90.1% of which were from mothers. The questionnaire included both closed- and open-ended questions addressing care experience, satisfaction, support needs, and QOL.

Results
Results. The majority (78.4%) rated the service as "very good," and 17.1% as "good," with no negative ratings. Most respondents felt safe (98.2%), heard (91%), and involved in care decisions (89.2%). Trust in the team was high (98.2%), and 93.7% stated that their own needs were considered alongside their child's. Demand for social work and chaplaincy support increased compared to 2022. Although 72.9% rated their QOL as good or very good, 54.9% reported fatigue, 43% anxiety, and 36% headaches; 31.5% had little or no time for self-care. Only 12.6% felt supported by society, while 35.1% experienced stigma or isolation.

Conclusions
Conclusion. Latvian PPC teams are highly valued by families and provide accessible, interdisciplinary care. However, ongoing psycho-emotional strain and limited societal support highlight the need to strengthen respite services, expand psychosocial resources, and enhance public education initiatives.

SIMULATION-BASED ROTATIONS FOR PAEDIATRIC RESIDENTS IN LATVIA: PROGRAMME DESIGN, IMPLEMENTATION AND EVALUATION

Ieva Šlēziņa [1], **Jānis Kolbergs** [1], **Mareks Marčuks** [2], **Andreta Slavinska** [3], **Dace Gardovska** [3], **Ilze Grope** [3]

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Objectives
Junior paediatric residents in Latvia begin their duties in paediatric emergency departments early in training, where they are expected to manage acute, complex, and often high-pressure situations. Simulation-based training allows them to practice procedural skills, apply clinical algorithms, and address communication challenges in a safe, controlled environment. Previously, paediatric residency programs included simulation only intermittently, focusing mainly on CPR algorithms. The aim was to develop and implement mandatory simulation rotation weeks for 1st- and 2nd-year residents, covering clinical, communication, and legal aspects of real-life emergency cases.

Materials and Methods
The program was developed by a multidisciplinary team of paediatric, emergency, simulation, and legal experts. Its curriculum was based on the Latvian paediatrician professional standard, European needs assessments for paediatric simulation training, and structured feedback from current residents. Since its introduction in 2023, 26 first-year and 22 second-year residents have completed these rotations.

Each 40-hour, five days rotation took place at a simulation centre and combined interactive lectures, skill stations, and high-fidelity simulation scenarios with SimBaby, SimJunior manikins, as well as trained simulated patients. Sessions followed a prebrief–scenario–debrief framework.
First-year rotations concentrated on foundational paediatric emergencies: recognition and management of critically ill children, basic and advanced life support, core airway and circulatory procedures, and interdisciplinary scenarios involving patient rights, abuse recognition, treatment refusal, and parental conflict.
Second-year rotations progressed to advanced airway management, invasive procedural skills, targeted diagnostic techniques, and complex case management, including polytrauma, severe medical conditions, and ethically and legally challenging situations.

Results
Evaluation included structured feedback on topics, skills, and scenarios, along with self-assessment of knowledge, skills, and confidence. Survey results from 2023–2025 showed high perceived impact:
•Gained new knowledge: 100%
•Acquired new skills: 97%
•Gained confidence in my abilities: 87%
Confidence increased by 19% from 1st- to 2nd-year residents, indicating effective progression and skill consolidation.
Qualitative feedback emphasized the realism of scenarios, the value of structured debriefing, and the integration of legal and ethical dimensions into clinical cases. Many participants recommended extending simulation weeks to all residency years, while identifying the greatest benefit in the initial two.

Conclusions
Tailored simulation rotation weeks for junior paediatric residents in Latvia are both feasible and well-received. Early outcomes indicate significant perceived gains in knowledge, skills, and confidence. The curriculum's progressive design, combining technical, non-technical, and interdisciplinary elements, aligns with real-world demands in paediatric emergency care. These findings support the program's continuation and potential expansion, with further longitudinal evaluation planned.

SLEEP DIAGNOSTICS AT THE CHILDREN'S CLINICAL UNIVERSITY HOSPITAL: A RETROSPECTIVE REVIEW OF 2024 DATA

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Objectives
Sleep disorders represent a substantial healthcare challenge in both pediatric and adult populations, significantly impacting physical and cognitive development, as well as overall quality of life. Diagnostic modalities such as polygraphy (PG), capnography, polysomnography (PSG) and the multiple sleep latency test (MSLT) are essential tools for identifying a range of sleep-related conditions.
The aim of the study is to summarize the sleep examinations performed at the Children's Clinical University Hospital over a one-year period.

Materials and Methods
A retrospective data analysis was conducted on sleep examinations performed at the Epilepsy and Sleep Medicine Centre in 2024. Data were analyzed using Microsoft Excel, applying means, medians, and percentages to summarize waiting times and the prevalence of sleep disorders.

Results
A total of 644 examinations were performed (581 - PG, 37 - PSG, 26 – MSLT; capnography was used in 433 respiratory examinations). The youngest patient was 23 days old, while the oldest - 75 years old. The median waiting time for an examination was 95 days (0 – 469), and 66 days (1 – 141) to receive the report.
In total, 103 examinations were unevaluable due to lack of patient compliance (58 cases) or technical issues (45 cases). When evaluating sleep-disordered breathing, only 15.29% of the tests showed normal results. Capnography revealed normal results in 69.30% of cases, hypercapnia in 17.16%, and hypoventilation in 7.00%. Among the 255 examinations in which obstructive sleep apnea was diagnosed, moderate to severe forms were identified in 37.64% of cases.
PSG tests revealed periodic leg movements in 51.43% of cases and altered sleep phase distribution in 68.57%. Increased sleepiness was detected in 66.67% of MSLT, while narcolepsy was not identified in any of the cases.

Conclusions
These findings reveal that sleep studies are also essential for identifying serious illnesses and suggest insufficient testing frequency.

SLEEP MEDICINE: ANALYSIS OF KNOWLEDGE AMONG MEDICAL STUDENTS IN LATVIAN UNIVERSITIES.

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Objectives

Adequate amount and quality of sleep is a fundamental necessity for sustaining both physical and mental health, as well as its critical in maintaining high quality of life. Despite its recognized importance, sleep medicine as a medical field is insufficiently represented in many medical education programs.

It is well-known that awareness is half the solution, emphasising the importance for future medical professionals to understand sleep medicine as a whole, improving diagnosis, management, and prevention of sleep-related disorders.

This study aimed to evaluate the knowledge regarding sleep medicine among medical students in Latvia.

Materials and Methods

A cross-sectional study was carried out between October 2024 and May 2025. The data was collected through a self-administered online questionnaire disseminated to Latvian medical students in Rīga Stradiņš University and University of Latvia. The questionnaire included demographic information, details of the current curriculum, factual knowledge on sleep medicine (physiology, sleep disorders, diagnostic principles, and sleep hygiene), as well as opinions about the necessity of integrating sleep medicine into the curriculum. Responses were compiled using Microsoft Excel and statistically analysed using IBM SPSS 29.0.

Results

The final sample consisted of 78 respondents (87.2% female, 12.8% male), with a mean age of 22.7±3.2 years. Only 51.3% (n=40) of respondents reported receiving some education on sleep medicine, however 32.5% (n=13) of those received less than 1 hour of study, while 50% (n=20) received 1-5 hours. 80.0% (n=32) of these respondents indicated that the education they received about sleep medicine was at an insufficient amount.

Knowledge-based questions revealed significant gaps. Only 17.9% (n=14) correctly identified the duration of one sleep cycle of a 1-year-old, while 33.3% (n=26) accurately reported the number of sleep stages per cycle. A question assessing detailed knowledge of sleep stages was correctly answered by just 12.8% (n=10). Additionally, only 14.1% (n=11) correctly identified the difference between nightmares and night terrors.

In two short clinical example questions about basic sleep physiology, only 20.5% (n=16) answered correctly in a case about a 17-year-old, while just 12.8% (n=10) in a case about a 7-year-old.

Based on self-assessment, 92.3% (n=72) respondents agreed that sleep medicine should be included in the medical curriculum.

Conclusions

This research reveals substantial deficiencies in medical student knowledge about sleep medicine in Latvia. Given the important role of sleep in maintaining health and overall well-being, targeted integration of sleep medicine into undergraduate medical curricula is necessary. Strengthening sleep medicine education could increase future medical professionals' capacity to address sleep-related health issues, improving patient outcomes and public health.

THE ROLE OF THE OTOLATYNGOLOGIST IN RECOGNIZING SIGNS OF CHILD ABUSE AND NEGLECT. NEW CHILD PROTECTION INITIATIVE – REACT!

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Objectives

Context

According to UNICEF, child abuse and neglect remain a critical problem worldwide. The war conditions only worsen this situation. Our colleagues at the 17th Congress of the European Society of Pediatric Otorhinolaryngology (Stuttgart 2025) raised this topic by presenting the new “REACT!” algorithm. This child protection initiative is currently under development, but its basic principles have already been incorporated into pediatric otolaryngology practices in Germany, Sweden, the Netherlands, and the Czech Republic. Other countries are invited to collaborate.

Case presentation

Recognize Early - Act Fast - Protect Together! (REACT!) is a practical guide for clinicians, especially paediatric ENT specialists, audiologists, paediatricians, and frontline healthcare providers. The guidelines include warning signs to recognize in practice and action steps to follow in case of suspicion.

We would like to remind you that some of the body parts most affected by physical abuse in children are the head, neck and oral cavity. These are also the areas examined by an ENT specialist, so we take this topic very "personally". In some cases, an otolaryngologist may be the first person to detect signs of abuse.

Children with speech, hearing, and communication disabilities are the most vulnerable. These children often do not express clear verbal signals. It is the responsibility of healthcare professionals to recognize nonverbal cues, visible injuries, and signs of psychosocial distress.

Materials and Methods
We have reviewed and compared the recommendations of the Centre for Disease Prevention and Control of Latvia with those of the REACT Initiative and would like to present them to our Baltic colleagues.

Results
x

Conclusions
Preventing child abuse and neglect requires a multidisciplinary approach.
The earlier it is detected and prevented in a child's life, the more favorable the outcomes for their cognitive development, behavior, socialization, and education. As well as the impact on the whole society. Let’s work together and “react” to protect those who cannot speak for themselves. We can be a part of the solution.

WHAT CHALLENGES AND SUPPORTS INFLUENCE THE DELIVERY OF PAIN MANAGEMENT IN CLINICAL SETTINGS?

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Objectives
Pain management is often guided by various assessment scales, which are widely used to evaluate acute postoperative pain but may be interpreted differently by medical personnel. Additionally, individual variations in pain thresholds can influence both the perception and assessment of pain, ultimately affecting treatment decisions. As a result, inadequately treated pain after paediatric surgery remains a common issue.
The objective of this study was to investigate the impact of patient awareness, a critical non-pharmacological intervention, on reported pain levels and the frequency of requests for additional pain medication.

Materials and Methods
4-18 years old pediatric patients were enrolled in this study. Data were collected during first year of international pain registry PainOut in Childrens Clinical University Hospital in Riga, Latvia. This analysis derives from the European PainOut infant registry providing information on perioperative pharmacological data and patient-reported outcomes on the first day after surgery. Statistical analyses were performed by using IBM SPSS 29.

Results
Out of 2896 selected patients, 1318 were evaluated. Among them: 43% reported that their worst pain score was above 6, 7% reported pain above 6 even without movement, and 19% reported pain above 6 during movement. Despite patients self-reporting their pain scores, only 56% had their pain documented in the medical records. Regarding pain management: 82% of patients stated they received enough information about pain management. However, only 14% received individualized pain treatment. 15% reported a desire to receive more pain treatment. Many patients did not receive their full prescribed pain therapy. Additionally, patients who experienced fatigue, pain when coughing or taking a deep breath, and pain during the night had a significantly higher risk of requesting more pain treatment ($p < 0.005$), indicating a need for more accurate pain assessment and better-tailored management strategies.

Conclusions
Effective paediatric pain management necessitates interdisciplinary collaboration among anaesthesiologists, paediatricians, surgeons and nursing staff. Moreover, fostering a communication between healthcare providers and family members is fundamental to delivering patient-centered care. Adequate pain control in paediatric patients, particularly during the postoperative period, is critical and has significant implications for shaping children’s long-term attitudes and responses to pain.

Rare Diseases

4Q21 MICRODELETION SYNDROME – A CASE REPORT

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Objectives

4q21 microdeletion syndrome (OMIM: 613509) is a recently identified genomic disorder characterized by growth retardation, facial dysmorphism, intellectual disability, absent or severely delayed speech, hypotonia, and variable brain malformation. It is caused by the deletion of the chromosome 4q21 region ranging from 3.2 to 15.1 Mb. The minimal critical region for this disorder is 1.37 Mb and includes five known genes: PRKG2, RASGEF1B, HNRNPD, HNRPDL, and ENOPH1. To date, approximately 50 cases with defined genomic coordinates have been reported in the literature, with a prevalence of less than 1 / 1 000 000.

In this case report we describe one patient who was born at 38+1 GW, small for gestational age with birth weight of 2430g. This was the first pregnancy from a healthy couple without family history of birth defects or developmental delay. No significant changes were observed during pregnancy, except for poorer intrauterine growth and two vessels in umbilical cord. After birth the child's phenotype suggested osteochondrodysplasia due to large had and relatively short limbs, therefore an NSG panel for short stature and a skeletal dysplasia was performed.

The investigation revealed a pathogenic, nonrecurrent heterozygous deletion seq[GRCh3] 4q21.1q22.1(77605851_92590301)x1, which is approximately 14,98 Mb in size, which confirmed 4q21 microdeletion syndrome diagnosis. Additionally, a heterozygous deletion seq[GRCh38] 14q32.2(99271146_99271233)x1 of uncertain significance was revealed involving the 1st exon of gene BCL11B, this deletion is considered a variant of unknown significance.

Considering that many organ systems can be affected by this syndrome, a multidisciplinary team was involved in the patient's care, including a geneticist, neurologist, pediatrician, ophthalmologist, cardiologist, audiologist, nutritionist, rehabilitation doctor and physiotherapist. Additional examination such as CUS and brain MRI, skeletal X-ray, echocardiography, abdominal and renal US, and hearing assessment revealed no pathological findings.

At the age of nine month, the girl was observed to have delayed physical and psychomotor development. Her weight and height were at -3.0 SD, while her head circumference was at +3.0 SD. She also exhibited diffuse muscle hypotonia. She demonstrates hand-to-hand and hand-to-mouth contact, when an object is placed in her palm, she grasps it, thought she rarely reaches for objects independently. On the prone position she has an unstable forearm support, and is able to turn head from side to side. During traction by the hands, her head is strongly retracted, and she lacks head control in the sitting position. Upon verticalization, she shows no support on her legs. The girl also experiences difficulty with enteral feeding, especially with supplemental nutrition, but she is currently able to feed from a bottle well.

In summary, early diagnosis and comprehensive rehabilitation are crucial for patients with 4q21 microdeletion syndrome to support optimal growth and development. Collaboration among specialists, a multidisciplinary approach, and psychological support for the family, are also essential.

Materials and Methods

X

Results

X

Conclusions

X

A CASE OF SCIMITAR SYNDROME REVEALED THROUGH ADOLESCENT ARRHYTHMIA

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Objectives

Background

Scimitar syndrome – also referred to as congenital venolobar syndrome or hypogenetic lung syndrome – is a rare congenital condition involving partial anomalous pulmonary venous return. In this anomaly, pulmonary veins from the right lung drain into the inferior vena cava (IVC) rather than the left atrium, producing a left-to-right shunt. Common associated findings include right lung hypoplasia, cardiac dextroposition, and anomalous systemic arterial supply to the right lower lobe. Clinical presentation ranges from asymptomatic cases to severe forms in infancy with respiratory distress or heart failure. Diagnosis is typically established via echocardiography, CT angiography, or cardiac MRI. Treatment is individualized, with conservative management for asymptomatic patients and surgery for those with significant symptoms. Prognosis is favorable in most older children and adults, though it may be poor in infants with severe cardiopulmonary involvement.

Case presentation

A 15-year-old female with prior history of right renal agenesis, progressive scoliosis, and asthma was admitted for a

scheduled intravenous urography. During anesthesia, she developed marked bradycardia with rate of 27 bpm accompanied by polymorphic supraventricular and ventricular extrasystoles. Several doses of atropine increased the heart rate to 110 bpm. Following referral to a pediatric cardiologist, the patient denied palpitations but noted episodes of fatigue and brief episodes of rapid heart rate during the last few months. ECG showed sinus bradycardia and Holter monitoring demonstrated frequent atrial and ventricular extrasystoles. Echocardiography (Echo) revealed signs of moderate myopericarditis and dilated pulmonary artery (32mm). After one month a follow-up Holter study showed a polymorphic ventricular and supraventricular extrasystoles up to 30% of total heart beats. Lab tests, including Troponin-I, were within normal range. Follow up Echo revealed residual mild pericardial effusion, with a preserved left ventricular ejection fraction of 64%. Metoprolol therapy was initiated but later discontinued due to symptomatic hypotension and fatigue. Chest x-ray showed no abnormalities aside from progression of
scoliosis. Cardiac MRI demonstrated right heart dilation and significant pulmonary hypertension, raising suspicion for aortic isthmus narrowing and anomalous pulmonary venous return. Angiographic MRI confirmed partial anomalous venous drainage of the right upper and middle pulmonary veins into the IVC, consistent with a diagnosis of Scimitar syndrome. A hemodynamically significant left-to-right shunt was present, and the patient was referred to a pediatric cardiac surgery center, where the decision to abstain from surgical correction was made. Instead, pacemaker was implanted, and metoprolol therapy was reintroduced for ongoing management.
Conclusions
This case highlights the complexity and diagnostic challenges of Scimitar syndrome, particularly when the clinical presentation is atypical. In this 15-year-old patient, initial symptoms including bradycardia, frequent extrasystoles, and fatigue led to an extensive cardiac workup and pacemaker implantation.

Materials and Methods
X

Results
X

Conclusions
X

PARENTAL ANXIETY AND EMOTIONAL RESPONSE ASSOCIATED WITH NATIONWIDE NEWBORN SCREENING FOR CYSTIC FIBROSIS IN LITHUANIA: A SINGLE-CENTER REPORT

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Objectives
Nationwide newborn screening for cystic fibrosis (CF) was introduced in Lithuania in January 2023. Understanding parental awareness, information sources, and emotional responses to positive primary screening results is essential for improving communication and reducing anxiety.
Objectives:
To assess parental awareness of CF, the sources and timing of information about newborn CF screening, and emotional responses following a positive primary screening result.

Materials and Methods
A cross-sectional study was conducted using an anonymous 27-item web-based questionnaire completed by parents (n = 42) whose newborns had a positive primary screening result for CF, defined as elevated immunoreactive trypsinogen (IRT), between 2023 and 2024 and who provided informed consent to participate. The survey included questions on prior knowledge of CF, the provision and adequacy of information before and after childbirth, and emotional responses to screening outcomes. Only parents of hospital-born infants were included. Results are presented as percentages based on the total sample size (n = 42), and descriptive statistics were used for data analysis.

Results
Of the respondents, 95.2% were mothers and 59.5% were first-time parents. Prior to screening, 26.2% were aware of CF, and 57.1% reported receiving pre-birth information—most often from gynecologists (36.0%), obstetricians (32.0%), and nursing staff (28.0%). Post-birth, 85.4% received additional information, primarily from nursing staff (47.2%) and obstetricians (27.8%). Information was considered sufficient by 80.5% of parents.
The interval between notification of a positive primary screening result and the first pediatric pulmonology consultation was ≥7 days in 53.7% of cases, 4–6 days in 19.5%, and 1–3 days in 26.8%. At consultation, 82.9% reported receiving sufficient information about CF. Following notification, 78.6% reported high anxiety, 16.7% mild anxiety, 2.4% calmness, and 2.3% hope.
Final diagnoses showed that CF was not confirmed in 39 infants; 10 of these carried a single CFTR mutation, and 2 were diagnosed with CF. Overall, 83.3% of parents expressed satisfaction with newborn CF screening.

Conclusions
The implementation of nationwide newborn CF screening in Lithuania was positively received and facilitated early disease detection. However, significant gaps remain in prenatal and postnatal communication, as many parents reported insufficient or absent information. Strengthening public awareness initiatives and enhancing communication between healthcare providers and parents may improve understanding and reduce screening-related anxiety.

PHENYLKETONURIA AND THYROTOXICOSIS, CASE REPORT

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Objectives
Phenylketonuria (PKU) is an autosomal recessive inborn error of metabolism resulting from a deficiency of phenylalanine hydroxylase, an enzyme that catalyzes the hydroxylation of phenylalanine (Phe) to tyrosine (Tyr). Tyrosine, is essential for protein and dopamine synthesis in the brain and in thyroid hormne synthesis. Thyrotoxicosis is a clinical condition characterized by excessive thyroid hormone activity, primarily triiodothyronine (T3) and thyroxine (T4), typically resulting from abnormally high levels of circulating thyroid hormones.

Materials and Methods
Single case report.

Results
A 17-year-old girl was diagnosed with elevated Phe levels through newborn screening shortly after birth, and molecular testing subsequently confirmed the diagnosis of classic PKU. She was promptly started on a strict low-protein diet along with amino acid supplementation.
At the age of six, the patient was further evaluated due to complaints of hyperactivity and behavioral disturbances. Neurological consultation was obtained, and magnetic resonance imaging (MRI) of the brain showed no abnormalities. However, electroencephalography (EEG) revealed epileptiform activity, and treatment with levetiracetam was initiated, which she continues to receive.
The patient remains under the care of a specialized center for rare diseases. Despite ongoing management, long-term control of PKU has been suboptimal, as evidenced by persistently elevated serum phenylalanine levels and low serum tyrosine concentrations.
In 2021, at the age of 15, the patient experienced menarche and began to exhibit profuse sweating. She subsequently consulted a gynecologist, who ordered additional investigations due to clinical concerns. Laboratory tests revealed suppressed thyroid-stimulating hormone (TSH) levels and elevated thyroid peroxidase (TPO) antibodies. Thyroid ultrasonography showed features consistent with thyroiditis. Clinically, the patient appeared tearful, with observed exophthalmos. Her blood pressure was 123/69 mmHg, and her heart rate was 94 beats per minute. The girl had complaints of decreased appetite, fatigue, with a 2 kg weight loss over 4 months.
She was referred to a pediatric endocrinologist for further evaluation. Laboratory findings included:
- TSH: <0.014 mU/L (↓)
- Free T4: 48.79 pmol/L (↑)
- Free T3: >8.4 pmol/L (↑)
- TSH receptor antibodies: 9.14 U/L (reference <1.75)
- Thyroglobulin antibodies: 15.4 U/L (reference <4.5)
- TPO antibodies: 140 U/L (reference <60)

A diagnosis of chronic autoimmune thyroiditis and thyrotoxicosis was established, and treatment with Thyrozol 2.5 mg twice daily was initiated. The dose was changed depending on the results of the tests. The patient has been regularly monitored, and her general condition, as well as thyroid function parameters, have shown improvement with ongoing therapy.

Conclusions
This clinical case highlights a rare and unusual presentation: a patient with poorly controlled PKU, characterized by persistently elevated Phe levels and reduced tyrosine Tyr concentrations in plasma, who subsequently developed thyrotoxicosis. The occurrence of thyrotoxicosis in individuals with PKU is uncommon, as low Tyr levels—due to dietary restrictions and metabolic imbalance—typically limit thyroid hormone synthesis. This case underscores the need for careful endocrine evaluation in PKU patients presenting with new clinical symptoms, even when such manifestations are atypical for the underlying metabolic disorder.

SIFRIM–HITZ–WEISS SYNDROME WITH FAMILIAL PRESENTATION: A RARE CASE OF CHD4 VARIANT TRANSMISSION

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Objectives

Sifrim–Hitz–Weiss syndrome, also known as CHD4-related neurodevelopmental disorders, is a rare autosomal dominant genetic condition. A mutation in the CHD4 gene can be inherited from one of the parents or can arise de novo (new mutation). A person affected by this disorder has a 50% chance of passing it on to their children, while in 50% of cases, the mutation does not get inherited and the child may be healthy. Until now, scientific literature has not described any cases where CHD4-related neurodevelopmental disorders were inherited from a parent. All reported patients so far have had pathogenic variants that arose de novo.

Materials and Methods

Clinical Case:

In 2023, a boy was born at 38 weeks of gestation (6th pregnancy, 5th delivery) by planned cesarean section. Birth weight was 3760 g, length 55 cm, Apgar scores 6/7/8. Antenatally, there was suspicion of a rare congenital pathology—Vein of Galen malformation, confirmed postnatally by MRI and ultrasound. The malformation caused severe brain damage including stroke, epilepsy, visual impairment, and obstructive hydrocephalus. The boy was also diagnosed with psychomotor developmental delay and a large atrial septal defect.

To determine a possible genetic cause, next-generation sequencing (NGS) panel analysis was performed targeting genes associated with brain and vascular malformations. This revealed a pathogenic variant in the CHD4 gene (NM_001273.3: c.3227T>C (Thr1076Ile)), which strongly supports the diagnosis of Sifrim–Hitz–Weiss syndrome. Genetic testing of family members showed the identical CHD4 variant (c.3227T>C, p.Thr1076Ile) in heterozygous state in the mother, sister, and brother, confirming that they also have Sifrim–Hitz–Weiss syndrome. Family history revealed developmental disorders in two children (the older sister and brother), and social adaptation difficulties in the mother. Prior to the birth of the youngest child, there was no suspicion of a genetic disorder in the family.

Results

This clinical case is unique because Sifrim–Hitz–Weiss syndrome is present in multiple first-degree relatives—mother and three children. Until now, no reports in the literature have described inherited CHD4-related neurodevelopmental disorders from a parent; all reported pathogenic variants have been de novo. Clinical manifestations of Sifrim–Hitz–Weiss syndrome are diverse, which can significantly complicate early and accurate diagnosis. Therefore, thorough examination of all family members is crucial to timely identify potential genetic predisposition, clarify the diagnosis, and tailor an individualized multidisciplinary care and treatment strategy.

Conclusions

Conclusions:

Although Sifrim–Hitz–Weiss syndrome is described in the literature as sporadic, it can also present in familial form. If genetic abnormalities are detected in a child with congenital anomalies, siblings should also be tested. In families with several children exhibiting minor shared congenital anomalies, genetic testing should be performed to enable informed reproductive planning for adult family members. Detailed family history and symptom screening, even in mild clinical cases, can be decisive for accurate diagnosis. A carefully developed multidisciplinary model can help ensure early diagnosis and improve treatment efficacy, thereby significantly impacting patients' quality of life and offering new opportunities for medical advancement in this field.

SYSTEMATIC REVIEW: THE USE OF WHOLE-BODY VIBRATION THERAPIES IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY

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1. NRC "Vaivari"

Objectives

Duchenne muscular dystrophy (DMD) is a progressive X-linked neuromuscular disorder characterized by proximal muscle weakness and motor deterioration. With disease progression, the preservation of motor function and the prevention of secondary complications, such as bone fractures, are essential therapeutic goals. Whole-body vibration (WBV) therapy is a non-invasive intervention that has shown potential benefits in other populations for improving muscle function and bone health. This systematic review synthesizes current research on the safety and effectiveness of WBV in children with DMD.

Materials and Methods

A systematic review was conducted in June 2025 following PRISMA guidelines (studies published from 2000 to 2025 in PubMed, PEDro and Google Scholar) using keywords "Duchenne muscular dystrophy" or "DMD" and "vibration" or "WBV".

Results
Out of 73 search results 5 studies were included in the review and encompassed 2 randomized controlled trials and 3 prospective intervention studies.
WBV showed high tolerability across multiple studies. In patients with DMD, interventions lasting 4 to 14 weeks and conducted 2 to 5 times per week revealed no major adverse effects. While initial creatine kinase elevations were noted in some DMD participants, they were transient and not linked to functional deterioration.
Two small intervention studies using WBV, (8-week vibration training, 5 days a week and 3-month vibration training 2-3 times a week), found no significant improvements in muscle strength, time function tests, angular degree of dorsiflexion of the ankles or bone mass. Soderpalm et al, 2013, study noted a temporary, non-significant increase in a bone formation marker, which returned to baseline 3 months after stopping the intervention.
In a randomised control trial (Ali et al, 2024), two intervention groups were compared: one received trunk-oriented exercises with physical therapy, and the other received the same plus whole-body vibration three times per week for three consecutive months. Although WBV led to significant improvements in balance and abdominal muscle thickness, it was less effective than trunk-oriented exercises combined with physical therapy.
A randomized controlled trial (Bianchi et ap, 2022) assessed the effects of low-intensity vibration applied daily for 14 months. The study reported improvements in tibial bone density, maintenance of hip bone mineral density and content, better control of fat mass gain and fewer fractures in the low-intensity vibration group compared to placebo. A limitation of the study was that the low-intensity vibration group was on average 2.8 years older than the placebo group, potentially influencing the results.

Conclusions
While trunk-focused exercises may be more effective in improving balance and abdominal muscle thickness, vibration therapies, especially low-intensity, long-duration interventions, have promise for bone health preservation in DMD. Vibration therapy appears safe for children with DMD. Short-term WBV showed limited benefit, while long-term low-intensity vibration demonstrated potential for preserving bone health and reducing fracture risk in this population. Further long-term randomized studies of low-intensity vibration treatment with larger, age-matched cohorts are needed to confirm these findings.

UNMASKING SLEEP HYPOVENTILATION IN PEDIATRIC GUILLAIN–BARRÉ SYNDROME: THE CASE FOR EARLY SCREENING

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1. Riga Stradins University

Objectives
To underscore the clinical relevance of early screening for sleep-disordered breathing (SDB), particularly nocturnal hypoventilation, in pediatric patients with neuromuscular disorders. This case illustrates how delayed recognition of SDB in Guillain–Barré syndrome (GBS) can hinder recovery and mask ongoing respiratory insufficiency.

Materials and Methods
We present the case of a previously healthy male patient who developed rapid-onset flaccid tetraparesis and respiratory compromise, consistent with GBS. Cerebrospinal fluid analysis revealed albuminocytologic dissociation, and electromyography confirmed demyelinating polyneuropathy. The patient required mechanical ventilation, tracheostomy, and was later transferred to a neurology ward. During recovery, caregivers noted increased daytime sleepiness, interpreted as clinical improvement. Due to persistent neuromuscular weakness and suspicion of underlying SDB, overnight cardiorespiratory polygraphy combined with transcutaneous capnography was performed prior to tracheostomy decannulation.

Results
Despite appearing clinically stable, polygraphy revealed 62 obstructive apneas, 2 central apneas, and 4 hypopneas (AHI 7.8), with the longest apnea lasting 183.8 seconds. Transcutaneous capnography showed pCO_2 >50 mmHg for 12.6% of the night, indicating significant nocturnal hypoventilation. After initiating nocturnal BiPAP support, the patient became more alert, active in rehabilitation, and showed improved daytime function. The previously perceived “recovery” was reinterpreted as a manifestation of chronic hypercapnia and untreated SDB.

Conclusions
This case highlights the critical need for early and systematic SDB screening in all neuromuscular patients, including children with GBS. Nocturnal hypoventilation can remain unnoticed and mimic recovery, leading to delays in appropriate respiratory support. Early identification and timely initiation of non-invasive ventilation may prevent respiratory failure, improve outcomes, and facilitate successful weaning from tracheostomy. Capnography-based sleep studies are essential in guiding safe management and rehabilitation planning.

Rheumatology

TNFRSF13B GENE VARIANT DETECTED FOR A PATIENT WITH STILL'S DISEASE: A CASE REPORT

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Objectives

The tumor necrosis factor 13B gene TNFRSF13B encodes the protein TACI, a receptor involved in T cell-dependent antibody responses and the regulation of autoimmunity and lymphoproliferation. In this abstract, we present a case of pediatric Still's disease and TNFRSF13B gene variant, highlighting potential genetic contribution and diagnostic challenges, although the exact genes involved in Still's disease pathogenesis are still being investigated.

Materials and Methods

This abstract presents a case of pediatric Still's disease. Clinical data were collected retrospectively from medical records.

Results

We report the case of a 9-year-old girl with the gradual onset of disease, starting with a skin rash, positive Mycoplasma pneumoniae test, followed by fever and joint pain five days later, along with elevated CRP (75 mg/L) and ESR (45 mm/hour) levels. Treatment with clarithromycin and prednisone led to resolution of fever and joint pain. After seven days, the patient presented with recurrent high-grade fever (up to 40°C), salmon-pink skin rash, joint pain and stiffness, CRP and ESR were elevated to 135 mg/L and 83 mm/hour, respectively. Still's disease was diagnosed. Treatment included a 3-day course of intravenous methylprednisolone pulse therapy, followed by oral prednisone with tapering and the addition of tocilizumab due to remaining symptoms. Given the severity of systemic inflammation, additional inflammatory markers were assessed: ferritin and IL-6 were elevated (1077 mcg/L and 656.82 ng/L, respectively), soluble IL-2 receptor level was slightly increased (109 kU/L, normal range: 19.1-68.5 kU/L), but not diagnostically significant. Hemophagocytic lymphohistiocytosis (HLH) was considered. Bone marrow examination showed no hemophagocytosis, immunoassays (IgA, IgM, IgG) and lymphocyte phenotyping were within normal limits. Primary HLH was excluded. Trio whole exome sequencing with autoimmune disorders gene panel analyses was performed and a heterozygous, maternally inherited NM_012452.3(TNFRSF13B):c.[310T>C];[=] and NP_036584.1:p.[(Cys104Arg)];[(Cys104=)] variant in gene TNFRSF13B was detected and classified as pathogenic. This gene is associated with biallelic autosomal recessive common variable immunodeficiency type 2 (CVID 2) and immunoglobulin A deficiency. The contribution of heterozygous variants to impaired immune response was also suggested but was not finally approved. However heterozygous variants, likely due to a dominant negative effect, were reported to affect B cell activation and peripheral tolerance resulting in autoimmunity. Due to that we hypothesise that this TNFRF13B variant contributed to the symptoms of our patient throught the increased risk of autoimmunity.

Conclusions

This case report emphasizes that genetic testing in patients diagnosed with Still's disease is crucial, as it may help identify underlying gene variants contributing to disease mechanisms.

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