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ORAL PRESENTATIONS
OP01  Effects of Poor Eating Habits on Children’s Body Weight

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Aim: This study aims to investigate the association between poor eating habits and children’s body weight in Mostar city, Bosnia and Herzegovina.

Material and Method: A descriptive cross-sectional study was conducted between May and October 2022. Data on eating habits and anthropometric measurements were collected from the medical records of participants visiting five dental practices affiliated with the School of Medicine, University of Mostar, Bosnia and Herzegovina. The data for this study are part of a larger project investigating the correlation of oral and general health. Body mass index (BMI) was calculated and compared to age and gender-specific growth charts to categorize participants as underweight, healthy weight, overweight, or obese.

Results: The study included 267 children aged 2-18 years, with a mean age of 9.75±4.34 years. Of the participants, 136 (51%) were boys and 131 (49%) were girls. The first group consisted of 195 (73%) children who consumed sweet and salty snacks one or more times daily, while the second group comprised 72 (27%) children who consumed such snacks occasionally (1-3 times a week). There was a significantly higher prevalence of overweight and obesity in the first group (p<0.001). Additionally, boys in both groups exhibited a higher prevalence of overweight and obesity compared to girls (p=0.03). Among the first group, school-age children were more likely to be overweight and obese compared to preschoolers (p=0.02). Conclusions: Daily consumption of sweet and salty snacks contributes to the development of overweight and obesity in children, particularly among school-age boys.

Keywords: overweight, pediatric obesity, dietary habits.
OP02  
ROHHAD Syndrome - Focus on Endocrine Abnormalities and Precocious Puberty

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Aim: To present a case of ROHHAD (Rapid-onset obesity with hypoventilation, hypothalamic dysfunction, autonomic dysregulation) syndrome, a very rare disorder with multisystemic involvement and an unknown etiology.

Material and Method: We present the case of an 11-year-old girl with ROHHAD syndrome.

Results: The disease was diagnosed at the age of 3.5 years when she was hospitalized due to obesity, adynamia, hyponatremia and a convulsive seizure. An electroencephalogram (EEG) recording showed epileptic brain activity, and antiepileptic therapy was initiated. Diagnostic tests revealed the presence of a tumor mass in the abdomen, which was surgically removed. Histopathology confirmed malignant ganglioneuroblastoma. The girl received chemotherapy according to the protocol. Genetic analysis of the patient showed no genetic predisposition to the onset of the disease. At the age of 8.5, she was hospitalized again due to vaginal bleeding. Endocrinological evaluation revealed precocious puberty, absolute growth hormone deficiency (although growth was at the 25th percentile), and hypothyroidism. Growth hormone, luteinizing hormone-releasing hormone (LH-RH) analogue, and Levothyroxine were included in therapy. The precocious puberty episodes have been stopped, and she no longer receives LH-RH analog. Levothyroxine, growth hormone, and antiepileptic drugs are continued in the therapy. The girl has no cognitive disorders, achieves good results in school, and her current growth chart values are within the reference values for her age.

Preoperative (private archive)
Conclusions: ROHHAD syndrome is a rare disease, with around 100 cases described worldwide. The disease can lead to severe complications, including intellectual impairment and death. The diagnosis is made on the basis of clinical manifestations, and the therapy requires a multidisciplinary approach.

Keywords: hypothalamic dysfunction, precocious puberty, rare disease, obesity
OP03 Oxidative Damage of DNA and Medium-Molecular Weight Peptides in Girls and Boys with Constitutional Obesity

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Aim: was the analysis of the components of oxidative damage to DNA, medium-molecular weight peptides (MMWP) and thiol-disulfide system and in adolescent girls and boys with constitutional obesity (CO).

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Material and Method: The study included 24 girls (mean age-14.00±1.97 years) and 23 adolescent boys (mean age-13.86±2.21 years) with an established diagnosis of grade 1 CO. The control groups consisted of 23 girls (mean age-14.00±1.26 years) and 20 boys (mean age-13.89±1.41 years), respectively. Height, body weight, waist circumference were measured, body mass index was calculated, and the stage of puberty according to Tanner was determined. Spectrophotometric, fluorometric and enzyme immunoassay methods of analysis were used.

Results: In the group of obese adolescent girls showed elevated levels of DNA destruction index - 8-hydroxy-2’-deoxyguanosine (8-OHdG) (p=0.019), MMWP 238 (p<0.0001) and MMWP 280 (p=0.03) relative to the control groups. Level of 8-OHdG (p=0.007) and MMWP 238 (p=0.03) showed increased values in the group of boys with obesity relative to the control group. The activity of the thiol-disulfide system in adolescent patients with CO changed statistically significantly relative to the control parameters. Thus, there were reduced levels of GSH (boys (p=0.015), increased levels of GSSG (girls (p=0.0003), boys (p=0.012)) and GSH/GSSG ratio (girls (p=0.0002), boys (p=0.004)) in patients with CO compared with controls.
**Conclusions:** The study showed a significant increase in the parameters of oxidative damage to DNA, medium-molecular weight peptides and an imbalance in the thiol-disulfide system in adolescents with CO, regardless of gender. In connection with these changes, in adolescents with CO, it is recommended to carry out corrective measures to stabilize the indicators, with the appointment of drugs with antioxidant properties. This study was carried out with the financial support of the CPG Grants of the Russian Federation (NSh-3382.2022.1.4).

**Keywords:** constitutional obesity, adolescents, 8-hydroxy-2’-deoxyguanosine, medium-molecular weight peptides
Aim: Resistant epilepsies (RE) to regular antiseizure medications (ASM) in paediatric population have shown good results with add-on treatment of isolated cannabidiol (CBD), one of many constituents of the cannabis plant. Full spectrum cannabis extracts (FSCE), have been rarely used but in some studies have shown excellent results in treatment of severe spasticity in children with cerebral palsy (CP) and also for severe behavioral outbursts in children with autism. We will present our own results of treatment with different cannabis products for these severe neurological conditions.

Material and Method: Resistant epilepsies (RE) to regular antiseizure medications (ASM) in children have shown good results with add-on treatment of CBD while full spectrum cannabis extracts (FSCE), have been rarely used but in some studies have shown excellent results in treatment of severe spasticity in children with cerebral palsy (CP) and also for severe behavioral outbursts in children with autism. We will present our own results of treatment with different cannabis products for these severe neurological conditions.

Results: Of 66 children with RE, 20% were seizure-free and nearly 50% have had 50% reduction of seizures after CBD add-on treatment, no serious adverse effects were documented; the cohort of 42 children with severe forms of CP treated with FSCE revealed decreased spasticity, better sleep and appetite and being more calm in half of the patients, while in 3 adverse effects needed treatment and 5 stopped the treatments because no effects were seen. In the autism group 15 children were included and parents reported much improved or very much improved behavior in 10 children. There were no serious side effects reported while 3 stopped the treatment with no effect.

Conclusions: While CBD add-on treatment effect is well documented in children with RE, the effects of FSCE in CP and autistic children have still to be proven in other studies.

Keywords: children, cannabis extracts, resistant epilepsies, cerebral palsy, behavioral outbursts in autism
The Role of Lung Ultrasound in the Evaluation of Newborn with Respiratory Problems

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Aim: Recently, lung ultrasound (LUS) has been introduced more in neonatology because of being rapid, simple and radiation-free. Ultrasound examination provides a dynamic and accurate evaluation in newborns with respiratory distress with the unique findings of respiratory distress syndrome (RDS), transient neonatal tachypnea and pneumonia. Our aim in this study is to examine the lung ultrasound evaluations of patients followed in the Neonatal Intensive Care Unit (NICU) due to respiratory distress in one year.

Material and Method: Patients admitted to NICU between 2022 and 2023 due to respiratory distress were included in the study. The data of the patients were reviewed retrospectively, demographic data, lung ultrasound findings, diagnosis and treatments were noted.

Results: A total of 228 patients were included in the study. Pulmonary ultrasound was the first choice for lung imaging of patients admitted to NICU due to prematurity, respiratory distress or the need for respiratory support at birth. Among the participants, 131 (57.6%) were male and 97 (42.4%) were female. Mean gestational age was 34.3 weeks (±3.7 weeks), mean birth weight was 2262 g (±855gr). Ventilation support is needed for 51.3% (n=117) of the patients. There were 36 patients (15.7%) diagnosed with RDS and treated with surfactant, and 18 patients (7.9%) diagnosed with pneumonia. Fifty-four (23.6%) of the patients were diagnosed with transient tachypnea.

Conclusions: In our study, lung ultrasound was used as the first evaluation method in patients with respiratory distress in NICU. The diagnoses of the patients were decided upon the ultrasonographic evaluation and clinical findings. Using ultrasound can provide early diagnosis of RDS and surfactant decision on time and also prevent unnecessary antibiotic treatment and prolongation of hospitalization period in patients with transient tachypnea.

Keywords: newborn, respiratory distress syndrome, lung ultrasound
**Aim:** In our study, we aimed to evaluate the relationship between serum galectin-3 levels with disease activity and potential. Additionally, we compared the serum galectin-3 levels between patients diagnosed with Familial Mediterranean Fever (FMF) and those of healthy children who presented to the pediatric emergency service.

**Material and Method:** Between September 1, 2022, and February 1, 2023, we included 32 patients with FMF who presented to our pediatric emergency department in our study. We included 30 patients who matched the control group in terms of age and gender in the study. Simultaneous hemogram and CRP tests were performed.

**Results:** The control group and FMF patients were compared based on the Galectin-3 and Platelet variables, and no statistically significant difference was found (p>0.05). It has been determined that there was no statistically significant relationship among CRP and Galectin-3 values in the patient groups.

Comparison between Groups in terms of Galectin-3, CRP, Platelet and Hemoglobin Values of FMF patients and healthy group

<table>
<thead>
<tr>
<th>Group</th>
<th>n</th>
<th>Mean ± Std</th>
<th>Median (IQR)</th>
<th>Test Value</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Galectin-3 (ng/mL)</td>
<td>Patient</td>
<td>32</td>
<td>0,31 ± 0,24</td>
<td>0,27 (0,31)</td>
<td>-1,036</td>
</tr>
<tr>
<td></td>
<td>Control</td>
<td>30</td>
<td>0,38 ± 0,26</td>
<td>0,32 (0,31)</td>
<td></td>
</tr>
<tr>
<td>CRP</td>
<td>Patient</td>
<td>32</td>
<td>49,19 ± 66,93</td>
<td>10,75 (78,78)</td>
<td>-4,546</td>
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<tr>
<td></td>
<td>Control</td>
<td>30</td>
<td>2,7 ± 6,03</td>
<td>0,35 (1,88)</td>
<td></td>
</tr>
<tr>
<td>Platelets</td>
<td>Patient</td>
<td>32</td>
<td>291558,4 ± 78234</td>
<td>284500 (104000)</td>
<td>-.984</td>
</tr>
<tr>
<td></td>
<td>Control</td>
<td>30</td>
<td>350300 ± 114667,5</td>
<td>342000 (125000)</td>
<td></td>
</tr>
<tr>
<td>Hemoglobin</td>
<td>Patient</td>
<td>32</td>
<td>11,8 ± 1,5</td>
<td>11,7 (1,65)</td>
<td>3,912</td>
</tr>
<tr>
<td></td>
<td>Control</td>
<td>30</td>
<td>12,02 ± 1,4</td>
<td>12,05 (1,6)</td>
<td></td>
</tr>
</tbody>
</table>

**Conclusions:** There was no statistically significant difference in serum galectin-3 concentration between FMF patients and the healthy control group.

**Keywords:** Patients, Familial Mediterranean Fever, Serum, Control, Galectin-3
Aim: The aim of this study was to analyze pediatric oncology patients admitted to the pediatric intensive care unit (PICU) in terms of risk factors, admission, organ failures, and to determine risk factors for mortality.

Material and Method: Retrospectively analyzed patients between January 2019-December 2022. First admissions of the patients were included and recurrent admissions were not. Elective hospitalizations lasting less than 24 hours (biopsy, catheter, and bone marrow biopsy) were not included.

Results: During the study period, 1102 children were admitted to PICU, 170 of whom were oncology patients, 138 of them (81 girls, 57 boys) were included in the study. Twenty-eight % of them had central nervous system tumors, 13% leukemia and 10.9% had lymphoma. Highest PICU admission were post-operative follow-up (44.2%) and sepsis (25.4%) followed by neurological (9.4%) and respiratory problems (8.7%). High flow nasal cannula oxygen was applied to 41.3% of the patients having respiratory problem, non-invasive to 30.4%, mechanical ventilation(MV) to 33.3% and mean MV follow-up time was 3.76±11.50. Thirty-one % of the patients needed inotropes and the mean vasoactive inotropic score (VISmax) was 0.75±1.07. Nineteen (13.7%) of 138 patients died. When survived and non-survived were compared, no difference was observed in terms of age, gender, and oncological diagnosis. Cox proportional hazard regression models were used to quantify association between mortality and risk factors; both univariate and multivariate analysis showed sepsis, PELOD and MV statistical significant predictor factor for mortality (p < 0.05). Survival analysis was performed based on 6th-month mortality according to the admission categories. The “post-operative follow-up” had the longest survival, followed by admission due to “respiratory problem” and “sepsis”.

Conclusions: This study highlights the importance of early recognition and management of sepsis, as well as the severity of organ dysfunctions in predicting mortality. The findings contribute to the understanding of factors influencing outcomes in this specific patient population.

Keywords: pediatric intensive care unit, oncology patients, mortality
OP08 Investigation of the Effect of Using a Sleep Band and White Noise on the Time to Sleep in Pediatric Intensive Care Patients aged 3-12 months

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Aim: This study was carried out as a randomized controlled experimental study to examine the effect of sleep band and white noise use on the transition to sleep in 3-12 month old pediatric intensive care patients.

Material and Method: The study consisted of a total of 78 patients, between 01 October 2021-01 April 2022 in Adana Seyhan State Hospital Pediatric Intensive Care Unit. Data were collected with Baby Information Form, Infant Sleep Problems Diagnosis Form, and Vital Signs Follow-up Chart. Sleep band, white noise listening device (iPad), heart rate and oxygen measurement device, pediatric Glasgow score and Wong-Baker pain scale were used in the study. The groups were independent and homogeneous in terms of planned pregnancy, child’s gender, birth order, diet, sleep, sleeping position, pre-sleep activity, and falling asleep status of the child.

Results: The difference between the groups was significant in terms of falling asleep time, waking frequency and total sleep time (p<0.05). It was determined that 23 children (57.5%) in the intervention group did not wake up at all, and 20 children (50%) in the control group woke up three times or more. It was determined that those in the intervention group slept mainly 12-15 hours, while those in the control group slept less than 12-15 hours. The difference between the comparison of vital signs in terms of hours according to the groups was found to be significant in favor of the intervention group (p<0.05).

Conclusions: As a result, it is recommended that nurses listen to white noise, which is a non-pharmacological method, for 3-12 month-old children to sleep, because it prolongs sleep time, reduces the number of awakenings, and has positive effects on children’s vital signs.

Keywords: White Noise, Sleep Band, Sleep, Child, Pediatric Intensive Care Unit
**OP09**  
**Toxic Myocarditis in Children: Etiology, Treatment and Follow-Up Results**  
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**Aim:**  
This study aims to evaluate all toxic myocarditis cases followed up in a single center with cardiac biomarkers, electrocardiogram (ECG), and echocardiographic findings.

**Material and Method:** cases who applied due to intoxication to a tertiary hospital in eleven years were examined. Those with a troponin value above 0.06 ng/ml (in 124 of 2994 cases) were diagnosed with toxic myocarditis. The causative agent of intoxication, epidemiological features, clinical findings, ECG and echocardiographic findings, and prognosis were evaluated.

**Results:** The most common cause of intoxication was drugs (64.1%). 71.8% of toxic myocarditis cases were evaluated as possible subclinical acute myocarditis and 28.2% as probable acute myocarditis. 57.3% of the cases were female and 42.7% were male. 45% of the cases were adolescents. ECG findings were normal in 83.9% of the cases, ST segment-T wave changes (40%) were the most common pathological changes. The troponin value was significantly lower in asymptomatic cases compared to symptomatic cases (p=0.00). EF at admission (p=0.03). The median EF at admission was significantly lower in patients who died (p=0.00). 4% of the cases died.

**Conclusions:** Clinical or subclinical myocarditis may develop in pediatric patients followed up for intoxication. It is important to evaluate these cases with troponin value, ECG, and echocardiography.  
**Keywords:** Toxic myocarditis, pediatric, intoxication, troponin

**Keywords:** Toxic myocarditis, pediatric, intoxication, troponin
OP10  MIS-C Treatment: Is There Excessive Use of IVIG?

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Aim: Multisystem Inflammatory Syndrome in Children (MIS-C) is characterized by hyper inflammation and multisystem involvement following COVID-19 infection. This study aims to evaluate clinical characteristics, cardiac findings, long-term follow-up data, and treatments of MIS-C.

Material and Method: Pediatric patients (0-18 years) diagnosed with MIS-C between May 2020 and December 2021 in our clinic were included. Initial complaints, physical examinations, laboratory results, chest X-rays, echocardiography findings at diagnosis and one-year follow-up, and treatments were evaluated.

Results: Ninety-two patients were included, of them 41 (44.6%) were females. Median age was 42 months (3-214 months). Median time from COVID-19 infection to MIS-C diagnosis was 6 weeks (4-8 weeks). Nine patients (9.8%) had underlying chronic diseases. On chest X-rays, 8 patients (8.7%) had bilateral infiltrations, others were normal. All patients presented with fever. Median temperature was 39°C (37.5-40.1°C). Median duration of fever was 5 days (2-17 days). Echo cardiography was performed for all patients. Abnormal cardiac findings were observed in 31 patients (33%) [decreased ejection fraction/left ventricular dysfunction (n=12, 13%), myocarditis (n=8, 8.7%), coronary artery involvement (n=11, 12)]. Shock was present in 13% of patients at presentation (19.6% MAS, 8.7% renal failure, 20.7% liver failure, and 8.7% heart failure). Mechanical ventilation was required in 4.3% of patients. No ARDS cases observed. Intensive care unit monitoring was necessary for 13% of patients. All patients received 2 mg/kg steroids. Total duration of steroid treatment was 7.3±3.9 days. IVIG was administered only to patients with cardiac involvement (33.7%). There was no mortality observed, and at one-year follow-up, none of the patients had any complaints, and all echocardiography findings were normal.

Conclusions: Considering the side effects, long-term outcomes, and treatment success, steroid use is sufficient for cases without cardiac involvement, while IVIG administration is reliable and cost-effective only for cases with cardiac involvement. Further studies are warranted for conclusive evidence.

Keywords: Covid-19, MIS-C, Steroid, IVIG
Demystifying the Defibrillator

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Aim: Cardiac arrests are rare in paediatrics. This causes staff to have high levels of anxiety surrounding them. Skillet et al. analysed all cardiac arrests documented by the national cardiac arrest audit (NCAA) over seven years and noted 110705 events, of which only 1580 (1.4%) were paediatric events.¹ This quality improvement project aimed to analyse how confident staff felt using a defibrillator within the paediatric department in Morriston Hospital, Wales and then develop a resource that could be given before starting work. A similar study was performed in 2018, providing a training course to medical students with an improvement in confidence, highlighting the importance of this project.²

Material and Method: In this quality improvement, members of the paediatric team (nurses and doctors of all levels) at Morriston Hospital, Swansea, were asked how confident they felt about being part of a paediatric arrest and using the defibrillator. Participants were shown a PowerPoint and a video going through the defibrillator and the differences in its use within the paediatric population to see if this would increase confidence levels. They were given questionnaires pre and post-presentation to establish whether this would help before starting a job or rotation within the department.

Results: Overall, twenty-five team members, from nurses to junior doctors to consultants, completed both powerpoints. The average confidence of the staff before the PowerPoint was 2.76/5, which increased to 4.24/5 after working through the presentation and watching the video, a 1.5x increase in overall confidence. Table
Conclusions: Outcomes of paediatric cardiac arrest remain poor for many reasons, but by making staff more familiar and more confident with the defibrillators, we hope this can provide paediatric patients with the best chance of positive outcomes. One of the limiting factors of this project was the small sample size. However, the preliminary results show promise.

Keywords: Cardiac arrests, Defib, Quality Improvement
Investigation of the Effect of Listening to White Noise During the First Invasive Interventions of Newborns on Pain, Comfort, Crying Time and Physiological Parameters

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Aim: The study was conducted in a randomized controlled and experimental design to investigate the effects of listening to white noise during the first painful procedure of vitamin K injection, which is the first painful procedure, on pain, comfort, crying time, and physiological parameters.

Material and Method: The newborns in the white noise group were started to listen to white noise 5 minutes before vitamin K IM administration, IM was applied by the same midwife at the 5th minute, and 10-minute camera recording was taken until 5 minutes later. In the study; the action camera was placed on a tripod and the shots were taken. Data were collected using the Baby Information Form, the Family Information Form, the Newborn Pain Scale (NIPS) and the COMFORTneo Behavior Scale (COMFORTneo).

Results: It was determined that the mean crying times (24.14±15.43) of the babies who were listened to white noise (experimental group) were considerably lower than the control group (40.03±13.58) who were not listened to white noise (p=0.000). The crying time (sec.) in the experimental group was lower than those in the control group (Z=-4.428; p=0.000). While heart rates during and after the procedure in the experimental group were lower than those in the control group (p<0.05), SpO₂ values were higher than those in the control group (p<0.05). The Cronbach’s alpha coefficient values of the COMFORTneo and NIPS scales were found to be highly reliable for observers, respectively. As a result of the intra-class correlation analysis, it was determined that the agreement between the observers was significant and the degree of agreement was excellent (p<0.05).

Conclusions: Consequently; it is recommended that nurses listen to white noise, for newborns during invasive intervention, because of its positive effects on the physiological parameters, shortening the crying time, reducing the pain, increasing the comfort.

Keywords: Comfort, Invasive Processes, Newborn, Pain, White noise
Clinical Manifestations and Outcome of Anomalous Left Coronary Artery from the Pulmonary Artery (ALCAPA)

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Aim: Anomalous left coronary artery from the pulmonary artery (ALCAPA) is typically manifested in early infancy and causes easily misdiagnosed myocardial ischemia.

Material and Method: Retrospective analysis included twelve patients (5 males) diagnosed with ALCAPA in period from November 2008 to June 2023.

Results: Median age of presentation was 3.5 months (IQR 2.3-4.2), with bronchiolitis-like (83.3%), feeding difficulties (66.7%) and sepsis-like (16.7%) manifestations. All patients had cardiomegaly on X-ray, ECG abnormalities suggesting ischemia and elevated NT-proBNP levels (9545 ± 11116 pg/mL), while 45% had elevated troponin I level (4.11 ± 2.61 ng/mL). Initially, left ventricle end-diastolic diameter (LV-EDD) and left atrium diameter (LA) were significantly enlarged, 41.3 ± 6.0 mm (z-score 6.1 ± 1.1) and 21.6 ± 3.7 mm (z-score 3.1 ± 1.1), respectively. Ejection fraction (EF) (32.5 ± 9.3%) and fractional shortening (FS) (15.3 ± 4.9%) were diminished, with moderate mitral (MR) (2+, IQR 1.0-2.5) and aortic valve (AR) (2+, IQR 2.0-2.0) regurgitation. The diagnosis was established with echocardiography and cardiac catheterisation when needed. Surgical coronary transfer was performed at the age of 3.9 months (IQR 2.8-4.3) in 10/12 patients, while two patients had delayed diagnosis and surgery. Follow up period was 5.0 ± 4.8 years. Six months after surgery, normalization of systolic function was revealed, with EF of 62.1 ± 9.6% (p=0.001) and FS of 30.2 ± 4.7% (p=0.001). Final outcome proved normalization of all echocardiographic parameters (LV-EDD z-score 1.1 ± 1.6 (p=0.001)(Figure 1a), LA z-score 0.9 ± 1.8 (p=0.014), EF 66.0 ± 10.7% (p=0.001)(Figure 1b), FS 34.3 ± 7.4 (p=0.001), MR 1+, IQR 1.0-1.9 (p=0.098), AR 0+, IQR 0-0 (p=0.001)).

Figure 1. Left ventricle end-diastolic diameter z-score (a) and ejection fraction (b) in patients with ALCAPA before and after surgery.
EF: ejection fraction; FU: follow-up; LV-EDD: left ventricle end-diastolic diameter;
SD: standard deviation.

**Conclusions:** Timely suspicion of ALCAPA in infants with breathing and feeding difficulties, as well as cardiomegaly on X-ray, pathological ECG and elevated NT-proBNP, could lead to prompt echocardiographic diagnosis, surgical correction and favorable outcome.

**Keywords:** cardiac ischemia, infancy, anomalous left coronary artery from the pulmonary artery (ALCAPA)
Streptococcus Pyogenes Gr. A and Appearance of Scarlet Fever in Children from 2017-2023

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Aim: Streptococcus pyogenes gr.A is one of the most significant cause of upper respiratory infections in children up to 10 years of age. The bacterium, in addition to having an M protein that is responsible for its pathogenicity, also produces toxins and enzymes that causes significant activation of the immune system. The most significant are streptolysin-O according to which organism creates antibodies that can be proven in the body with an ASO test and pyrogenic toxins A, B and C that cause exanthema, enanthema and the appearance of scarlet fever. As a result of the creation of antibodies in the body, during the fight against the bacteria, autoimmune processes can occur in the body that affect the joints, heart and kidneys (migratory polyarthritis, rheumatic fever with endocarditis and post streptococcal glomerulonephritis).

Material and Method: A total of 1897 microbiological findings on nose and throat swabs in children aged up to 10 years were processed in the period from March 2017 to May 2023. Cases of scarlet fever are confirmed by a positive finding on a throat swab and/or an elevated ASO test value.

Results: Between March 2017 and March 2022, Streptococcus pyogenes gr.A was isolated in 23.2% of positive nose and throat swabs. Two cases of scarlet fever were recorded in January 2028. Between April 2022 to May 2023, Streptococcus pyogenes gr.A, was isolated in 53.5% of positive swabs. Scarlet fever was confirmed in 23 cases as well as one case of post-streptococcal glomerulonephritis.

Conclusions: the incidence of Streptococcus pyogenes gr.A and the complications it causes show a significant increase in children up to 10 years of age in the period after the COVID-19 pandemic. Taking strict hygiene measures in kindergartens and schools in order to reduce the spread of infection plays a significant role in preventing the occurrence of infections and complications caused by bacteria.

Keywords: Streptococcus pyogenes gr.A, scarlet fever, children, complication
Clinical Case of PFAPA Syndrome with 7p22 Microdeletion

Natalia Antonova
1Tallinn Children’s Hospital

Aim: The pathogenesis of the periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis (PFAPA) syndrome is unknown. It is regarded as an autoinflammatory process. Disease onset is usually before the age of five and generally resolves before puberty with no consequences for the patient. Children are asymptomatic between episodes and show normal growth. No specific diagnostic test for PFAPA is currently available. Syndrome has overlapping symptoms with other periodic fever syndromes with a known genetic cause. In addition, genetic variants that are known to cause other autoinflammatory syndromes have been found in PFAPA patients, but the impact of these genetic variants in PFAPA syndrome is still unknown. A 2-year Caucasian/Azerbaijan girl demonstrated repeated fever episodes with the high levels of C-reactive protein since 6 m.o. She was observed because of microcephalus, slight developmental delay, growth retardation and dysmorphic phenotype and diagnosed with 7p22 microdeletions. During a period of 10 months she was hospitalized 6 times with the high fever, cervical lymphadenitis and pharyngitis.

Material and Method: Laboratory tests and instrumental investigations were performed: abdomen ultrasound, chest X-ray, EKG and EHHOKG, ANA, HIV, Borreliosis serology and Quantiferron test, urine test, urine culture. Cervical ultrasound revealed increased lymphoid nodules with normal structure. Blood test showed no neutropenia, procalcitonin level and blood culture were repeatedly negatives. Brain MRI with spectroscopy was performed to exclude intracranial pathology. PFAPA was suspected because of typical clinical symptoms (repeated episodes of fever with aphthous pharyngitis, cervical lymphadenitis and high CRP, absence of neutropenia).

Results: Prednisolone treatment 1 mg/kg orally was used twice with excellent effect. Adenotomy with tonsillectomy was performed. After this treatment high fever episodes resolved. Sequencing of genes was performed to exclude MEFV, MVK, TNFRSF1A, IL1RN, and other gene abnormalities.

Conclusions: No monogenic fever syndrome was revealed.

Keywords: Periodic fever syndrome, PFAPA, 7p22 microdeletion
**Aim:** During the COVID-19 pandemic, we aimed to evaluate SARS-CoV-2 seropositivity in pediatric patients with chronic kidney disease before and after the adult vaccination program, and to measure the effect of parental vaccination in the home environment on the child.

**Material and Method:** Pediatric patients with CKD (kidney transplant recipients, patients receiving HD/PD, and stage 2-5 CKD) followed up regularly were included in the study. Patients were divided into two groups: the pre-vaccination period from January 2021-2022 and the adult vaccination period after January 2022. Anti-SARS-CoV-2 antibodies against the S1 domain of the SARS-CoV-2 spike protein were investigated with a micro ELISA kit.

**Results:** A total of 182 patients with a mean age of 11 years and 55% male were included in the study. 54.4% of patients were kidney transplant recipients, 9.9% were HD/PD patients, and 35.7% were stage 2-5 CKD patients. Of all the patients, 57 (31.3%) were seropositive and 125 (68.7%) were seronegative. Seropositivity rate is 30.3% in kidney transplant recipients, 38.8% in HD/PD patients and 30.7% in stage 2-5 CKD patients, respectively. The rates of primary kidney disease and the presence of immunosuppressive therapy were similar between seronegative and seropositive patients (p=0.865). Anti-SARS CoV-2 antibody positivity was 24.1% before community immunization period and 43.9% after immunization period (p<0.05). The proportion of patients with symptoms were higher after community immunization period compared to before immunization (95% vs. 54%; p<0.05). Symptoms that may be associated with COVID-19 were fever (40%), cough (33%) and GI symptoms (25%) in order of frequency. The rates of compliance with the isolation rules (mask, social distance, hand washing) and stay-at-home consciousness were significantly higher in the pre-immunization period(p< 0.05).

The serostatus, symptom and SARS-CoV-2 seroprevalence rate are shown in the period before and after community COVID-19 immunization.
Figure-1: Schematic representation of the distribution of patients included in the study according to serology, symptoms status. Figure-2: Anti-SARS CoV-2 antibody positivity was 24.1% (28/116) before community immunization period and 43.9% (29/66) after the community immunization period (p<0.05).

### Symptoms and risk factors for SARS-CoV-2 transmission

<table>
<thead>
<tr>
<th>Symptom that may be associated with COVID-19</th>
<th>Before community immunization (n:116) (%)</th>
<th>After community immunization (n:66) (%)</th>
<th>p value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fever</td>
<td>28 (24,1)</td>
<td>45 (68,2)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Cough</td>
<td>19 (16,4)</td>
<td>42 (63,6)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Myalgia</td>
<td>14 (12,1)</td>
<td>17 (25,8)</td>
<td>0.031</td>
</tr>
<tr>
<td>Headache</td>
<td>13 (11,2)</td>
<td>15 (22,7)</td>
<td>0.063</td>
</tr>
<tr>
<td>Throat ache</td>
<td>11 (89,5)</td>
<td>20 (30,3)</td>
<td>0.001</td>
</tr>
<tr>
<td>Anosmia</td>
<td>1 (0,9)</td>
<td>0.999</td>
<td></td>
</tr>
<tr>
<td>Ageuzia</td>
<td>1 (0,9)</td>
<td>0.999</td>
<td></td>
</tr>
<tr>
<td>GIS symptoms</td>
<td>22 (19)</td>
<td>23 (34,8)</td>
<td>0.017</td>
</tr>
<tr>
<td>Other symptoms</td>
<td>12 (10,3)</td>
<td>3 (4,5)</td>
<td>0.277</td>
</tr>
<tr>
<td>Risk factors for SARS-CoV-2 transmission</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Living in a crowded center</td>
<td>72 (62,1)</td>
<td>41 (62,1)</td>
<td>0.999</td>
</tr>
<tr>
<td>Travel story</td>
<td>82 (70,7)</td>
<td>48 (72,7)</td>
<td>0.903</td>
</tr>
<tr>
<td>Using public transportation</td>
<td>40 (34,5)</td>
<td>36 (54,5)</td>
<td>0.008</td>
</tr>
<tr>
<td>Being in crowded environment</td>
<td>16 (13,8)</td>
<td>40 (60,6)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Stay-at-home consciousness</td>
<td>86 (74,1)</td>
<td>38 (57,6)</td>
<td>0.021</td>
</tr>
</tbody>
</table>
Adherence to isolation rules (mask, social distancing, hand washing) | 105 (90,5) | 39 859,1 | <0,001
Hospital visits | 10 (8,6) | 44 (66,7) | <0,001
Patients with at least one family member with COVID-19 | 14 (12,1) | 12 (18,2) | 0,361
Close contact with COVID-19 patient | 19 (16,4) | 30 (45,5) | <0,001

**COVID-19 symptoms and Risk factors for SARS-CoV-2 transmission in the period before and after community immunization**

**Conclusions:** Seropositivity rate and symptom frequency in patients with CKD increased after community immunization. This can be attributed to the decreased sensitivity to the preventive measures introduced by the vaccination program.

**Keywords:** COVID-19, SARS-CoV-2, Pediatric chronic kidney diseases, Seroprevalence, community immunization
Retrospective Analysis of Children Diagnosed with Inflammatory Bowel Disease in Samsun

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¹University of Health Sciences Trabzon Kanuni Training and Research Hospital
²Ondokuz Mayıs University, Health Practice and Research Hospital

Aim: Inflammatory bowel disease (IBD) is a relapsing and remitting disorder, characterized by chronic inflammation of the intestine. Our study was planned to demonstrate the features of the patients with inflammatory bowel disease in our region.

Material and Method: Data of the patients in the Pediatric Gastroenterology Clinic between 2005-2015 were analysed retrospectively. 58 patients between 1 month-18 years of age diagnosed via colonoscopy, were included. Patients who were diagnosed in other facilities or ruled out with colonoscopy were excluded. Data gathering was through the archive and the automation system of the hospital. SPSS 23.0 Program was utilized and p values below 0.05 were significant.

Results: 58 patients, 46 of them were ulcerative colitis (UC) and 12 Crohn’s disease (CD) were included. 56.9% of the patients were female. Mean age was 15±4 years (UC) and 16±5 years (CD), respectively. Bloody diarrhea was the most common complaint among UC, weight loss and weakness were prominent among CD (p<0,05). Thrombocytosis, elevated erythrocyte sedimentation rate and C-reactive protein levels were significant in CD (p<0,05). The most common pathological finding on sonography was bowel wall thickness (in 18% of UC and 54% of CD). MRI results assisted the sonography. Colonoscopy mostly revealed edematous bowel loops (39.1% in UC and 41.6% in CD), crypt abscess in 46.6% of UC and 33.3% of CD. Meselazine was the drug of choice (in 97.8% of UC and 100% of CD). As PUCAI numbers increased, steroid resistance or dependence increased in UC (p=0.013).

The Presenting Symptoms of the Patients

<table>
<thead>
<tr>
<th>Symptom</th>
<th>UC n(%)</th>
<th>CD n(%)</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stomachache</td>
<td>39(84,8)</td>
<td>12(100)</td>
<td>0,150</td>
</tr>
<tr>
<td>Diarrhea</td>
<td>35(76,1)</td>
<td>10(83,3)</td>
<td>0,592</td>
</tr>
<tr>
<td>Bloody Diarrhea</td>
<td>25(54,3)</td>
<td>2(16,7)</td>
<td>0,020</td>
</tr>
<tr>
<td>Tenesmus</td>
<td>24(52,2)</td>
<td>8(66,7)</td>
<td>0,369</td>
</tr>
<tr>
<td>Nocturnal diarrhea</td>
<td>10(21,7)</td>
<td>5(41,7)</td>
<td>0,160</td>
</tr>
<tr>
<td>Weight loss</td>
<td>15(32,6)</td>
<td>9(75)</td>
<td>0,008</td>
</tr>
<tr>
<td>Weakness</td>
<td>20(44,4)</td>
<td>10(83,3)</td>
<td>0,017</td>
</tr>
<tr>
<td>Perianal Disease</td>
<td>5(11,1)</td>
<td>1(8,3)</td>
<td>0,781</td>
</tr>
<tr>
<td>Rectal Bleeding</td>
<td>34(73,9)</td>
<td>3(25)</td>
<td>0,002</td>
</tr>
</tbody>
</table>
Joint Pain 4(8,9) 2(16,7) 0,435
Fever 11(24,4) 4 (33,3) 0,534

**Conclusions:** The patients were mostly diagnosed in adolescence. Although studies report that CD incidence is increasing, studies from our country conclude that UC is more common. Therefore, prospective studies are required in order to determine the incidence of inflammatory bowel disease. Moreover, IBD may be accompanied by Familial Mediterranean Fever, as revealed by genetic analysis in half of our CD patients.

**Keywords:** colitis, ulcerative, inflammatory bowel disease, adolescence
Acute Appendicitis in Children Under Three Years: A Diagnostic Challenge

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1Children’s Hospital of Mexico, Federico Gomez

Aim: Acute appendicitis is the most common pediatric surgical emergency in the world, however, there are limited case series and with a small number of patients, which makes it difficult to standardize clinical criteria to suspect acute appendicitis in this age group. Our challenge today is to know the most frequent clinical characteristics of acute appendicitis in this age group and thereby reducing the complications associated with a late and/or erroneous diagnosis.

Material and Method: A retrospective, observational and descriptive study was carried out, from 2011 to 2021, with patients under three years of age diagnosed with acute appendicitis in a Tertiary Care Center in Mexico City.

Results: Of 421 reported cases of appendicitis, 10% were patients under 3 years old, with a male: female ratio of 1.5:1. The most frequent symptoms were: abdominal pain, vomiting, fever, hyporexia and absence of bowel movements. The evolution time was 4.4 days ± 3.5 days. Eighty-one percent of the patients received medication prior to their admission: 79% analgesics, and 50% antibiotics. Among the admission diagnoses were found: abdominal pain syndrome, intestinal occlusion, acute gas troenteritis and intestinal intussusception. The leukocyte count was normal in 48% of the cases. Fifty-two percent of the appendages were perforated. Thirty-six per cent of the patients presented complications, among which the following stand out: intestinal occlusion, surgical wound site infection, septic shock and wound dehiscence. The average number of days of hospital stay was 12 days ± 10 days. Mortality was 2.4%.

Most frequent symptoms of acute appendicitis in children under three years of age.

<table>
<thead>
<tr>
<th>Symptom</th>
<th>n=42</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Abdominal pain</td>
<td>37</td>
<td>88%</td>
</tr>
<tr>
<td>Vomiting</td>
<td>35</td>
<td>83%</td>
</tr>
<tr>
<td>Fever</td>
<td>33</td>
<td>79%</td>
</tr>
<tr>
<td>Hyporexia</td>
<td>21</td>
<td>50%</td>
</tr>
<tr>
<td>Absence of bowel movements</td>
<td>17</td>
<td>45%</td>
</tr>
<tr>
<td>Abdominal distension</td>
<td>16</td>
<td>38%</td>
</tr>
<tr>
<td>Diarrhea</td>
<td>15</td>
<td>26%</td>
</tr>
</tbody>
</table>

n=number of patients
Conclusions: Appendicitis in patients under three years old is a diagnostic challenge and a cause of high morbidity and mortality; since it is little suspected and it is similar to other infectious diseases, the excessive use of medications to alleviate symptoms delays timely diagnosis and increases the risk of complications and days of hospital stay.

Keywords: appendicitis, children, intestinal occlusion, perforated, abdominal pain
Aim: Low adherence is one of the reasons for the decrease in the effectiveness of treatment. Today, a number of approaches and techniques have been developed to assess adherence. But so far there is no "gold standard" for determining patient adherence to therapy. Aim: Comparative assessment of adherence to antiretroviral therapy in children with perinatal HIV infection according to the Morisky-Green questionnaire (MMAS-8).

Material and Method: A retrospective cohort study was conducted among HIV infected patients (age >18 years). Children receiving ART from January 2019 to March 2022 and for at least 6 months. The children and their parents or guardians were surveyed using the Morisky-Green questionnaire (MMAS-8). Multivariable logistic regression was employed to identify predictors of virologic failure.

Results: A total of 33 HIV-infected children for aged 7-17 years, were involved in the study. The mean age of the children was 12.2±2.44 years and 54% were girls. The prevalence of viral suppression (viral load < 1000 copies/mL) was 78.8%. Adherence of parents [adjusted odds ratio (AOR) = 11.1, 95% CI 0.165-3.879]) and children [(AOR) - 1.45, 95% CI (0.591-14.459)] to the Moriska Green questionnaire was not a significant predictor of virologic failure.

Conclusions: We have not found significant differences in the level of viral load in children with different degrees of adherence, both according to the results of a survey of children and parents or guardians. Further studies of the level of adherence and its determinants with multiple measures of adherence using prospective and multicenter studies were recommended. Low adherence is one of the reasons for the decrease in the effectiveness of treatment. Today, a number of approaches and techniques have been developed to assess adherence. But so far there is no "gold standard" for determining patient adherence to therapy.

Keywords: adolescents, children, adherence, antiretroviral therapy
OP19 Ophthalmological Examination of Children with ASD as a Part of Multidisciplinary Approach

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Aim: The solving the problem of comprehensive support for persons with disabilities is due to the provisions of the Universal Declaration of Human Rights, the UN Convention on the Rights of the Child, the UN Convention on the Rights of Persons with Disabilities. There are difficulties in conducting examinations of children with behavioral disorders, impaired verbal communication skills especially by doctors for ophthalmological examination. A multidisciplinary program is being implemented using the principles of a participatory approach. All physicians were trained in methods of approach to the examination of children with ASD. An ophthalmological examination was carried out.

Material and Method: 39 children from 1 to 17 y.o. diagnosed with ASD were included. An ophthalmologist examination: biomicroscopy of the eyeball, refraction by autorefractometry under natural conditions, visual acuity. All studies were conducted with the informed consent of parents and in their presence

Results: Ophthalmologist were trained on working with children with ASD. Middle age - 12,65+2,99 years. In 28%, heredity was revealed in violation of accommodation, myopia, phoria was diagnosed, in 23% - astigmatism. PINA as a precursor of accommodation spasm was diagnosed in 3% of patients. For the first time these 28% of children were fitted with spectacle correction, 10 at the age of 10 years and older.

Conclusions: To detect visual pathology in children with neurodevelopmental disorders, including ASD, additional training of specialists in the field of their examination is necessary. The absence of competencies, unidentified vision problems can affect behavioral disorders and even more maladjusted.

Keywords: ASD, Ophthalmological examination, Multidisciplinary approach
Hyperleukocytosis: A Rare Manifestation of Autoimmune Hemolytic Anemia

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2Department of Pediatrics Hematology and Oncology, Kartal Dr. Lütfi Kirdar Şehir Hastanesi, Istanbul, Turkey

Aim: CASE REPORT: Autoimmune hemolytic anemia (AIHA) is described by destruction of Red Blood Cells (RBCs) as a result of binding of antibodies to red blood cell surface antigens. White blood cells are usually normal and hyperleukocytosis is rare. The purpose of this study was to highlight hyperleukocytosis in AIHA caused by Mycoplasma pneumoniae.

Material and Method: The parents of a 4-year-old twin-boy rushed him to the emergency department in the middle of the night due to recent generalized fatigue, shortness of breath and occasional vomiting. His mother noticed sudden with yellow discoloration of his eyes two days ago. At the time of his admission, laboratory tests were performed that revealed a markedly elevated WBC of 37.6×103 cells/μL. In addition, with 75% neutrophils; significantly low hemoglobin level of 5.2 g/dL. The complement fixation test for M. pneumoniae was positive for IgM at 3.15 Index. After eliminating all other causes of autoimmunity and hemolytic anemia, leukemoid reaction due to warm type autoimmune hemolytic anemia caused by M. pneumoniae infection was diagnosed.

Results: Methylprednisolone (3 mg/kg/d) was given also intravenous immunoglobulin and antibiotics (clarithromycin 15 mg/kg/d) were empirically initiated for suspicion of M. pneumoniae. During treatment, the leukocyte count declined to normal values after day 5. He responded to red blood cell transfusion, steroid and intravenous immunoglobulin and remained well.

Graph

The hemoglobin levels and leukocyte count during hospital days.

Conclusions: Warm typed AIHA is mostly diagnosed after viral infection in children. Even though the significant relationship between infection and AIHA is rarely reported, M. Pneumoniae is the most important pathogen caused to AIHA. Even though very uncommon, M. pneumoniae may induce the occurrence of warm agglutinins and cause immune hemolytic
anemia. It is worth that multiple and atypical clinical manifestations of M. pneumoniae infection should be suspected, mainly in severe hemolytic anemia cases in order to start early specific therapy.

**Keywords:** Autoimmune Hemolytic Anemia, Leukocyte, Mycoplasma
Comparing Antipseudomonal Effects of Honey Samples with Different Geographical and Botanical Origin

Nemanja Petrović

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Aim: This research evaluated and compared different types of honey from various geographical origins for their antibacterial activity against Pseudomonas aeruginosa, using an agar-well diffusion assay.

Material and Method: The analyzed honey samples were raw, unheated, authenticated by pollen analysis and Codex Alimentarius methods, and obtained from Serbia (sunflower, acacia, organic buckwheat, honeydew, and two multifloral honeys), Russia (two buckwheat), Poland (buckwheat), New Zealand (manuka), and Australia (manuka). Two strains of bacteria Pseudomonas aeruginosa isolated from clinical specimen and a reference strain ATCC 10145, were used. Streptomycin at 20 µg/ml was used as positive control. Artificial honey and purified water were used as negative controls. The agar-well diffusion method was used according to the recommendation of CLSI and modified. Zone of inhibition surrounding the well was measured to evaluate the antibacterial activity, which was expressed as a minimum inhibitory concentration (MIC) of MIC80 and Break point.

Results: Sunflower, acacia, two multifloral honeys from Serbia - mountain Ravna gora, and buckwheat honey from Poland did not show antibacterial activity against tested Pseudomonas aeruginosa strains. Honeydew honey from mountain Zlatar showed the highest activity against the clinical specimen with MIC80 of 208.106 mg/ml and a Break point 260.133 mg/ml. Serbian buckwheat honey from mountain Zlatar showed similar results with MIC80 of 216.708248 mg/ml and Break point 270.88531 mg/ml, and a high potency against ATCC 10145 strain: MIC80 0.0909 mg/ml and Break point 1.5453 mg/ml.

Zone of inhibition surrounding the well was measured to evaluate the antibacterial activity, which was expressed as a minimum inhibitory concentration (MIC) of MIC80 and Break point.

<table>
<thead>
<tr>
<th>med</th>
<th>mic80</th>
<th>MIC90</th>
<th>MIC99</th>
<th>breakpoint</th>
</tr>
</thead>
<tbody>
<tr>
<td>MNZ</td>
<td>344.7396</td>
<td>387.83205</td>
<td>430.9245</td>
<td>430.9245</td>
</tr>
<tr>
<td>MA</td>
<td>258.019336</td>
<td>290.271753</td>
<td>322.52417</td>
<td>322.52417</td>
</tr>
<tr>
<td>R2</td>
<td>696.033448</td>
<td>783.037629</td>
<td>870.0418099</td>
<td>870.04181</td>
</tr>
<tr>
<td>Z1</td>
<td>216.708248</td>
<td>243.796779</td>
<td>270.88531</td>
<td>270.88531</td>
</tr>
<tr>
<td>Z2</td>
<td>208.106792</td>
<td>234.120141</td>
<td>260.13349</td>
<td>260.13349</td>
</tr>
</tbody>
</table>
Honeydew honey from mountain Zlatar showed the highest activity against the clinical specimen with MIC80 of 208.106 mg/ml and a Break point 260.133 mg/ml. Serbian buckwheat honey from mountain Zlatar showed similar results with MIC80 of 216.708248 mg/ml and Break point 270.88531 mg/ml.

Conclusions: The agar applied method demonstrated stronger antipseudomonal efficiency of honey samples from Serbia - region Zlatar compared with honey types having consistently higher bio-active properties presented in literature - manuka and buckwheat honey. Further research should focus on the antibacterial potential of honey from mountain region Zlatar.

Keywords: Honey; Buckwheat; Manuka; Antibacterial activity, Pseudomonas aeruginosa
OP22  Achieving Caloric Goal in Postoperative Management of Congenital Heart Disease

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²Koc University Hospital, Department of Dietetics and Nutrition, Istanbul, Türkiye
³Koc University School of Medicine, Department of Cardiovascular Surgery, Istanbul, Türkiye

Aim: Meeting caloric needs in congenital heart disease (CHD) surgery is an important factor in improving outcomes. This study will provide insight into the prevalence of malnutrition, time to achieve caloric goal, and risk factors affecting nutrition in patients after CHD surgery in cardiac intensive care unit (CICU).

Material and Method: Patients aged 1 month to 18 years old between 2021 and 2022 were included. History of major gastrointestinal surgery or severe postoperative feeding intolerance were excluded. Body mass index (BMI), height-for-age, weight-for-age, state of malnutrition, malnutrition z-score, Risk Adjustment for Congenital Heart Surgery-1 (RACHS-1), cardiopulmonary bypass (CPB) time, aortic cross-clamp time, Pediatric Risk of Mortality-3 (PRISM-3), Pediatric Logistic Organ Dysfunction-2 (PELOD-2), vasoactive inotropic score (VIS), total fluid balance, total duration of mechanical ventilation (MV), length of ICU stay (LOS), mortality, length of hospital stay, and time to achieve caloric goal were recorded.

Results: Of 75 included patients (boy=41, girl=34), malnutrition was detected on 22% (n=17) based on BMI z-score and 28% (n=21) based on weight-for-height z-score (WH). Sex, RACHS-1, mortality, CPB and aortic cross-clamp time, PRISM-3, PELOD-2, VIS, total fluid balance, MV duration, LOS and length of hospital stay were statistically similar in patients with and without malnutrition. Patients are compared according to achieve their caloric goal on fourth day versus extending fourth day. They are statistically different in mortality, VIS, MV duration CPB and aortic cross-clamp time, PRISM-3, PELOD-2, LOS and hospital length of stay (p< 0.05) however they were similar in terms of age, sex, RACHS-1, and total fluid balance (Table 1). Logit regression analyzes showed that MV duration, PELOD-2 and PRISM-3 seen as statistically significant risk factors affecting achieving caloric goal (Table 2).

Table 1 and Table 2
Conclusions: Malnutrition is prevalent in CHD patients, and concomitant organ failure along with MV duration play an important role in achieving caloric goal during postoperative period.

Keywords: Nutrition, Cardiac Intensive Care Unit, Congenital Heart Disease
OP23 Avoidant-Restrictive Food Intake Disorder in Children with Autism Spectrum Disorder

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Aim: Proper nutrition is one of the most important factors for the optimal development of the child. For children with autism, nutritional selectivity is typical. Leo Kanner, in his first publications about children with autism, mentioned that they are characterized by a limited diet (Kanner, 1943). It is important to develop the guideline for identification and management of ARFID (avoidant-restrictive food intake disorder) in children with ASD.

Material and Method: Single-stage non-comparative study. The research included 80 children with ASD (diagnosis verified with ADI-R), 59 males/21 females. The age ranged from 1,4 year to 17 years, (M=10+4,53). All patients were examined by multidisciplinary medical team (psychiatrist, pediatrician, allergist-immunologist, gastroenterologist, endocrinologist).

Results: The diagnostic clusters of ARFID were analyzed with the allocation of: behavioral, medical conditions, social dysfunction. Approaches to the assessment of somatic disorders due to malnutrition in children were presented. The guideline for diagnosing ARFID has been developed and piloted in children with ASD. A multidisciplinary team is required for the diagnosis and management of ARFID, also for diagnosing medical conditions that can mimic ARFID (allergies, GERD and others). In 71 out of 80 children with autism, signs of selective behavior were revealed, of which 68 children (85%) had additional signs - diagnostic criteria (negatively affected the physical health or social dysfunction), allowing to diagnose ARFID as co-occurring disorder.

Conclusions: ARFID is highly prevalent among children with autism. It is necessary to follow the guideline which include multidisciplinary examinations for the timely identification and management of ARFID in children with autism, to prevent negative consequences for their development and health.

Keywords: autism, ARFID, multidisciplinary team
OP24  Development of External Genitalia During Mini-Puberty: Is Related to Somatic Growth or Pubertal Hormones?

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²Department of Pediatric Endocrinology and Diabetes, Marmara University, School of Medicine, 34841, Istanbul, Turkey

Aim: Although hypothalamo-pituitary-gonadal axis activation and hormone levels in mini-puberty are similar to the pubertal period, its role in human development and reproduction has not been fully elucidated. We aimed to evaluate the effects of somatic growth and reproductive hormones on the development of external genitalia during mini-puberty.

Material and Method: The maternal, natal and postnatal history, anthropometric data, pubertal examination and hormone characteristics of healthy infants aged 1-4 months were evaluated. Serum FSH, LH, DHEAS, A4, SHBG, E2 and inhibin-B, testosterone (T) and AMH were studied. Free sex hormone index was calculated as T/SHBG for boys and E2/SHBG for girls.

Results: The mean age of 148 (74 female) infants included in the study was 2.31±0.76 months. Tanner stage 3 sex steroid and gonadotropin levels were reached in mini-puberty. A statistically significant difference was found between the weight, height, BMI, weight gain and serum FSH, LH, A4 measurements of girls and boys (p<0.05). A positive correlation was found between the weight and height of the boys and their penile length. Negative correlation was found between boys’ weights and serum LH and T/SHBG levels. Negative correlation between girls’ weight SDS, height SDS, weight gain and serum SHBG levels; positive correlation between weight gain and E2/SHBG levels, and between weight/height percentile z-scores and FSH levels was found. Mean FSH, LH and SHBG levels in girls and boys, mean estradiol levels in girls and mean testosterone levels in boys in relation to age.

Changes in serum hormone concentrations by age are shown.

Conclusions: External genitalia and breast development are related to somatic growth during mini-puberty. Peripheral effects of reproductive hormones in mini-puberty may be related to binding proteins and free hormone levels and/or receptor sensitivity rather than measured total hormone levels.

Keywords: Mini-puberty, hypothalamo-pituitary-gonadal axis, somatic growth, reproductive hormone, external genitalia
**OP25**

**Taxonomic Composition and Predicted Metabolic Functions in Gut Microbiome of Obese Teenagers**

Elizaveta Klimenko¹, Natalia Belkova¹, Anna Pogodina¹, Lubov Rychkova¹

¹Scientific Centre for Family Health and Human Reproduction Problems

**Aim:** The study aim was to describe the taxonomic composition of the intestinal microbiota and its potential metabolic profile in adolescents with obesity and normal body weight.

**Material and Method:** The study sample consisted of adolescents aged 11–17 years with obesity (n=18) and normal body weight (n=22), who were sent for examination to the Clinic of the SC FHHRP (Irkutsk). Fecal samples were processed according to the standard operating procedures of the International Human Microbiome Standards project. Amplicons of the 16S rRNA gene were sequenced. Raw data are deposited in the NCBI SRA as BioProject PRJNA604466.

**Results:** According to the alpha diversity indices, the intestinal microbiota is represented by a large variety of phylotypes and had a similar evenness of communities. Analysis of beta diversity using a weighted UniFrac did not reveal a significant difference for representatives of the main microbiome. According to the results of the unweighted UniFrac, all adolescents formed four metabolic groups, regardless of body weight. The first group consisted of 26 people who differed minimally in the composition of minor components. The other two groups included gut microbiomes that were slightly different from the first group. The fourth group was formed by adolescents whose microbiomes showed differences both from the main sample and among themselves. Representatives of the phyla Bacteroidetes (46.35±1.88%), Firmicutes (43.17±2.14%), Proteobacteria (7.45±0.83%) and Actinobacteria (1.9±0.0%) dominated in the intestinal microbiota. 57%). In intestinal microbiomes, biosynthetic pathways predominated over degradation, assimilation, and utilization pathways. The largest number of genes has been predicted for the pentose phosphate pathway, gondoic acid biosynthesis, starch degradation, and pyruvate fermentation to isobutanol.

**Conclusions:** The presence of adolescents with varying degrees of metabolic activity in the main group showed that even with differences in taxonomic composition, the intestinal microbiota compensates for the absence of certain microbes, realizing the necessary metabolic functions at the expense of other species.

**Keywords:** gut, microbiome, obesity, 16s rRNA
Genetic and Exogenous Factors of Mineral Metabolism Disorders in Inflammatory Bowel Diseases in Children

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²Research Clinical Institute of Childhood of the Ministry of Health of the Moscow Region

Aim: Genetic and exogenous factors of mineral metabolism disorders in inflammatory bowel diseases in children.

Material and Method: 42 children with IBD from 6 to 17 years were examined: 28 children with ulcerative colitis (UC, 11.71±4.92 years) and 14 children with Crohn’s disease (CD, 13.75 ± 3.66 years). Vitamin D availability was tested at level 25(OH)D in blood serum (ELISA). Molecular genetic analysis was carried out on total DNA isolated from whole blood leukocytes. The polymorphism of vitamin D biotransformation genes – CALCR, COL1A1; VDR gene - was studied by PCR. Assessment of bone mineral density (BMD) was carried out using dual-energy X-ray absorptiometry (DEXA) in the lumbar spine (L1 - L4) using Z-score, BMD, BMC, AREA indicators.

Results: Average security level 25(OH)D did not differ in the groups of UC (25.15±11.81) and BC (27.43±0.87) ng/ml (p>0.05), however, vitamin D deficiency (20<25(OH)D<30 ng/ml) was more often observed in BC (43% in BC, 21% in UC (p 0.03)), and deficiency (10<25(OH)D<20 ng/ml) is more common in UC (49% in UC, 28% in BC, p 0.04). When assessing MPC, the average level of Z-score did not differ in the UC groups (-0.87±1.16) and BC (-0.75±0.87) p 0.68. The frequency of decrease in MPC (WHO criteria, Z-score less than -1SD) was more often observed in the UC group (57%) than in BC (39.3%). When evaluating bone metabolism genes, polymorphisms of the genes were found: CALCR: TT genotype was found in 15/28 (BC) and 9/14 (UC) - 68.4% of IPC disorders. VDR vitamin D receptor gene mutations with replacement of thymine (T) with cytosine (C) (CC, TC, AA, GT): the most common GA genotype (20/42 (47.6%), 12/19, GG (15/42 (35.7%)), rare AA (4/42 (9.5%)) – predictor of IPC disorders; COL1A1 – collagen gene of the first type (genotypes GG, GT): the most frequent GG genotype (30/42 (71%)-100% of IPC disorders).

Conclusions: The frequency of vitamin D deficiency did not differ in UC and CD. Polymorphism of predictor genes of osteopenia in children with IBD: TT-genotype of CALCR gene, GA-genotype of VDR gene of vitamin D receptor (Taq), GG-genotype of COL1A1. The frequency of decrease in BMD did not differ significantly in UC and CD, reaching 57% (WHO criteria, UC), which is higher than in the population.

Keywords: mineral metabolism, DEXA, bowel diseases, vitamin D, Molecular genetic
Diabetes Management in Children with Type 1 Diabetes Mellitus

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Aim: In this study, diabetes management of children with Type 1 Diabetes Mellitus; It is aimed to evaluate diabetes behaviors by questioning the processes such as nutrition, insulin administration, etc. at school.

Material and Method: The cross-sectional study was conducted with 60 children who were diagnosed with Type 1 Diabetes and who wanted to voluntarily participate in the study, followed in the Balcalı Hospital Pediatrics Clinic/Policlinics in Adana, Turkey. The data were collected by face-to-face interview, a questionnaire form (Chicago Diabetes Research and Education Center) and the Diabetes Behavior Rating Scale (DDRS) between January 2018 and January 2019.

Results: The mean age of the children was 13.90±2.31 (years). 37 children (61.7%) diagnosed with type 1 diabetes stated that they used 4-6 drugs daily, 58 (96.7%) used drugs in the form of injections, and 50 (86.2%) administered the injection drug themselves. 46 children (76.7%) stated that they were successful in school, 35 (58.3%) stated that they were absent from school. 43 (71.7%) of them stated that they did not measure their blood sugar at school, and 52 children (86.7%) stated that they had insulin injections at their school, in the classroom or anywhere. The sub-dimension score of the use of medical equipment in diabetes care (frequency) of those whose mother's education level was primary/secondary school and high school or higher was statistically significantly higher than those who were literate/illiterate (p<0.05). Insulin administration (frequency) sub-dimension score, using medical equipment (frequency) sub-dimension score and record keeping (frequency) sub-dimension score of those who measure glucose at school were significantly higher than those who did not measure glucose at school (p<0.05).

Conclusions: Diabetes management is extremely important in the prevention of acute and chronic complications of Type 1 Diabetes Mellitus, which is a chronic disease. As the education level of the family increases, the diabetes management of children will be better.

Keywords: Child, Diabetes Education, Nursing, Type 1 Diabetes Mellitus
OP28  Infant Outcome After a Pregnancy Complicated with Sigmoid Colon Adenocarcinoma

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Aim: Background: To report a case of an infant born from an in vitro fertilization-conceived pregnancy complicated with sigmoid colon adenocarcinoma.

Material and Method: /

Results: Case presentation: A female infant was born at 35+6 gestational weeks after Cesarean Section with a birth weight of 2060 grams and Apgar Score 7 and 8 in the 1st and 5th minute. Mother was a carrier of MTHFR mutation and had a mitral valve prolapse. At the 11th gestational week mother was diagnosed with a sigmoid colon adenocarcinoma grade 2, with liver metastases. She was operated on two times, with a preformed colostomy. Additionally, the mother went through 5 cycles of chemotherapy with FOLFOX (folinic acid, fluorouracil and oxaliplatin). Near before delivery, the mother was diagnosed with anemia. Fetal development was normal, except for asymmetric intrauterine growth restriction. At birth, late preterm infant manifested signs and symptoms of respiratory distress syndrome, with hypotonia and incomplete reflex response. Venous blood gases analysis along with clinical appearance indicated the presence of perinatal asphyxia. Oxygen therapy in the incubator (oxygen fraction up to 50%) and prophylactic antibiotic therapy (Ampicilin, Amikacin) were started. After five days due to clinical deterioration, leucopenia and thrombocytopenia, a third antibiotic was introduced – Ertapenem. Indirect hyperbilirubinemia was treated with phototherapy. The newborn was discharged from the hospital on the 11th day after birth in good condition and afterward followed up for 21 months, revealing normal psychomotor development.

Conclusions: Growing incidence of pregnant women older than 40 years, present a risk factor for more frequent malignancy occurrence during pregnancy. Colorectal carcinoma in pregnancy is extremely rare with very inconsistent prognosis for both mother and the infant. A multidisciplinary approach is needed to provide the mother with the best therapy and care and to postpone the delivery thus contributing to better neonatal and long-term outcome for a child.

Keywords: sigmoid colon adenocarcinoma, pregnancy, infant outcome
Perinatal and Neonatal Outcomes of Refugee Infants: An Observational Study in a Tertiary Hospital in Turkey

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Aim: The immigration problem has been increasing worldwide due to conflicts. In recent years, Turkey has accepted more than 3.8 million refugees from many countries, principally Syria. In this study, we aimed to evaluate the prenatal features and neonatal outcomes of refugees and Turkish controls hospitalized in a tertiary neonatal intensive care unit in Turkey.

Material and Method: This retrospective case-control study included comparing data related to the populations based on whether they are refugees or not, and their perinatal and neonatal “outcomes” were compared.

### NICU admission diagnoses of Refugees and Turkish infants

<table>
<thead>
<tr>
<th>Diagnoses ,n(%)</th>
<th>Study group (n=127)</th>
<th>Control group (n=127)</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Respiratory distress syndrome</td>
<td>18 (14.1)</td>
<td>18 (14.1)</td>
<td>1.00</td>
</tr>
<tr>
<td>Transient tachypnea of newborn</td>
<td>36 (28.3)</td>
<td>47 (37)</td>
<td>0.141</td>
</tr>
<tr>
<td>Early-onset sepsis</td>
<td>8 (6.3)</td>
<td>10 (7.8)</td>
<td>0.625</td>
</tr>
<tr>
<td>Late-onset sepsis</td>
<td>9 (7)</td>
<td>7 (5.5)</td>
<td>0.605</td>
</tr>
<tr>
<td>Prematurity</td>
<td>25 (19.6)</td>
<td>12 (9.4)</td>
<td>0.021</td>
</tr>
<tr>
<td>Small for gestational age</td>
<td>1 (0.7)</td>
<td>0 (0)</td>
<td>0.316</td>
</tr>
<tr>
<td>Meconium aspiration syndrome</td>
<td>8 (6.3)</td>
<td>2 (1.5)</td>
<td>0.053</td>
</tr>
<tr>
<td>Bronchopneumonia</td>
<td>5 (3.9)</td>
<td>2 (1.5)</td>
<td>0.250</td>
</tr>
<tr>
<td>SARS-CoV-2</td>
<td>2 (1.5)</td>
<td>2 (1.5)</td>
<td>1.00</td>
</tr>
<tr>
<td>ABO incompatibility with jaundice 6</td>
<td>(4.7)</td>
<td>0 (0)</td>
<td>0.013</td>
</tr>
<tr>
<td>Rh incompatibility with jaundice</td>
<td>1(0.79)</td>
<td>0 (0)</td>
<td>0.316</td>
</tr>
<tr>
<td>Neonatal jaundice</td>
<td>16 (12.6)</td>
<td>15 (11.8)</td>
<td>0.848</td>
</tr>
<tr>
<td>Congenital heart disease</td>
<td>4 (3.1)</td>
<td>4 (3.1)</td>
<td>1.00</td>
</tr>
<tr>
<td>Polycythemia</td>
<td>1 (0.79)</td>
<td>0 (0)</td>
<td>0.316</td>
</tr>
<tr>
<td>Hypoglycemia</td>
<td>2 (1.5)</td>
<td>1 (0.79)</td>
<td>0.561</td>
</tr>
<tr>
<td>Syndromic disorders</td>
<td>5 (3.9)</td>
<td>10 (7.8)</td>
<td>0.183</td>
</tr>
<tr>
<td>Metabolic disease</td>
<td>2 (1.5)</td>
<td>1 (0.79)</td>
<td>0.561</td>
</tr>
<tr>
<td>Perinatal asphyxia</td>
<td>5 (3.9)</td>
<td>3 (2.3)</td>
<td>0.472</td>
</tr>
</tbody>
</table>
Results: Among 254 analyzed neonates, 127 were born to refugee mothers, and 127 controls were born to non-refugee Turkish mothers. The refugee rate in our hospitalized neonates was nine, a young mother’s age (p=0.010) with a higher rate of adolescent pregnancies (p=0.032) and consanguineous marriage (p=0.031) in comparison to non-refugees. The incidence of ABO incompatibility-related hemolytic jaundice (p=0.013) was higher in the refugees. The rate of formula feeding in the first month of life was significantly higher (p=0.027) in neonates born to refugee mothers. Despite lower perinatal care rates in refugees (<0.001), preterm morbidities did not differ between refugees and non-refugee preterm infants ≤32 gestational age (p>0.05).

Conclusions: The importance of breast milk must be supported strongly to initiate and promote exclusive breastfeeding in infants of refugees. Race is still an important risk factor for ABO incompatibility-related hemolytic jaundice. Providing high-quality health care is sufficient to prevent worse outcomes in refugee neonates.

Keywords: Neonate, outcome, refugee, Turkey
A rare cause of West Syndrome: SLC35A2 Mutations

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²Ankara University Faculty of Medicine, Department of Pediatric Neurology

Aim: SLC35A2 is a gene encoding the UDP-galactose transporter involved in glycosylation at Xp11.23. The role of SLC35A2 gene mutations in the etiology of epilepsy has been discovered relatively recently. Although glycosylation is a process that takes place in all tissues and organs, mutations in SLC35A2 gene predominantly affect the central nervous system and cause clinical findings such as early-onset epilepsy (often epileptic spasms), hypotonia, and developmental delay. We aimed to present a case of West syndrome with SLC35A2 gene mutation in its etiology.

Material and Method: Our presentation was designed as a case report.

Results: A four-year-old female patient with unrelated parents was admitted to our pediatric neurology outpatient clinic for the first time at the age of five months, with spasms of her bilateral upper extremities. Physical examination at admission was normal except for mild hypotonia and global developmental delay. The patient whose EEG was consistent with infantile spasm, was started on ACTH therapy. In the laboratory tests for the etiology, no specific finding was found for any neurometabolic disease. Cranial MRI showed thin corpus callosum. The whole exome sequencing analysis revealed a pathogenic heterozygous mutation of SLC35A2 gene. The patient, whose seizure-free state was achieved with ACTH and levetiracetam treatments, is still followed up under levetiracetam 40mg/kg/d antiepileptic treatment.

Conclusions: West syndrome is an epileptic encephalopathy with a wide heterogeneous etiologic spectrum including numerous genetic mutations. Therefore, genetic studies are very useful in the diagnostic processes of West syndrome cases, especially in the detection of rare etiologies.

Keywords: West syndrome, epilepsy, etiology, genetic, SLC35A2
Effectiveness of CFTR Modulator Elexacaftor/Tezacaftor/Ivacaftor in the Russian Population of Patients with cCystic Fibrosis (12-month)

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²Research Centre for Medical Genetics

Aim: The aim was to analyze the effectiveness of the CFTR modulator elexacaftor/tezacaftor/ivacaftor (ETI) in children with cystic fibrosis (CF) in real clinical practice during 12 months of observation.

Material and Method: The study was conducted based on the analysis of the database “Register of patients with CF of the Russian Federation” for 2021–2023 using a special application for the online platform. Research included 48 patients 6–18 years who received ETI. The indicators were analyzed: FEV1, FVC (% pred.), electrolyte conductivity of the sweat fluid, anthropometric data. The results were compared at the start of targeted therapy and after 12 months of treatment.

Results: In the group of children treated with ETI, an increase BMI (kg/m2) was registered from 17.1 (15.7–19.3) at the start of therapy to 19.0 (17.5–20.4) after 12 months of therapy, p < 0.001. FEV1 improved from 66.5% (53.3–78.3) before the start of therapy to 89.0% (64.1–104.3) after 12 months, p < 0.001, FVC 74.5% (58.4–86.3) before therapy and 87.0 (72.0–102.3) % after 12 months, p < 0.001. Sweat conductivity decreased from 116 (102.0–125.5) mmol/l at the start of therapy to 77.5 (55.8–86.8) mmol/ l after 12 months, p < 0.001.

Conclusions: After 12 months of therapy with ETI, there was a significant positive dynamic in BMI, spirometry and sweat test compared with the indicators before the start of targeted therapy.

Keywords: cystic fibrosis, CFTR modulator, observation, 12 months, sweat test
High Frequency Oscillations in Autism Spectrum Disorder: Is it Related with Clinical Severity?

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2Akdeniz University, Faculty of Medicine, Department of Pediatric Neurology, Antalya, Turkey
3Artificial Intelligence Engineering, Near East University, Nicosia, TRNC, Mersin 10, Cyprus
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5Ministry of Health, Ankara Research and Training Hospital, Department of Pediatric Neurology, Ankara, Turkey

Aim: Autism Spectrum Disorders (ASD) are characterised by impaired social communication and interaction, restrictive and repetitive patterns of interest, behaviour and activities causing developmental delays in children. High frequency oscillations (HFO) are oscillatory high frequency (>80Hz) signals that can be recorded by scalp EEG. They can be physiological or pathological. The aim of the study was to explore the presence of HFOs in children with ADS and its relation to clinical symptomatology.

Material and Method: Fifty two children with ASD and 49 healthy children enrolled to the study. N2 sleep records of at least 10 minutes were evaluated. EEG records evaluated by open-source Python library MNE-HFO for the detection of scalp HFOs. The sleep spindles were detected using the open-source YASA. Sleep spindles and HFOs evaluated by artificial intelligence.

Results: The mean ages were 63.8 ± 23.9 and 64.6 ± 25.3 respectively for ASD and healthy subjects. The duration of SS was shorter and the frequency was higher significantly in children with ASD. HFOs unrelated to SS were documented to be higher in children with ASD (p<0.01). Younger children with ASD had particularly higher HFOs unrelated to SS (p<0.01). Within ASD group of patients HFOs unrelated to SS were significantly higher in subgroups who had moderate-severe social interaction problems (<0.01), moderate-severe restricted interest (0.018) and severe linguistic delays (0.006). However no relation with stereotypies had been found.

<table>
<thead>
<tr>
<th></th>
<th>ASD (n:52)</th>
<th>ASD (n:52)</th>
<th>Controls (n:49)</th>
<th>Controls (n:49)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Duration of SS</td>
<td>778.9 ± 1315.3</td>
<td>295.3 (546.4)</td>
<td>1667.8 ± 6285.2</td>
<td>534.8 (620.7)</td>
</tr>
<tr>
<td>Frequency of SS</td>
<td>15.4±0.66</td>
<td>15.5 (0.32)</td>
<td>11.5±0.64</td>
<td>11.6 (0.22)</td>
</tr>
<tr>
<td>Amplitude of SS</td>
<td>1322.7 ±2095</td>
<td>389.3 (1145.1)</td>
<td>1125.3 ±2136.6</td>
<td>182.1 (115.4)</td>
</tr>
</tbody>
</table>

EEG parameters evaluated in the patient and control groups
<table>
<thead>
<tr>
<th></th>
<th>Density of SS</th>
<th>HFO percentage</th>
<th>HFO unrelated to SS</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0.81±1.22</td>
<td>46.3±31.6</td>
<td>14.7±3.1</td>
</tr>
<tr>
<td></td>
<td>0.29 (0.86)</td>
<td>41.4 (51.6)</td>
<td>10.2 (11.3)</td>
</tr>
<tr>
<td></td>
<td>0.67±0.55</td>
<td>34.7±28.9</td>
<td>4.5±1.8</td>
</tr>
<tr>
<td></td>
<td>0.63 (0.78)</td>
<td>33.3 (41.2)</td>
<td>4.3 (5.1)</td>
</tr>
<tr>
<td></td>
<td>0.204</td>
<td>0.071</td>
<td>&lt;0.001</td>
</tr>
</tbody>
</table>

**Conclusions:** The HFOs unrelated to SS were found to be significantly higher in children with ASD, particularly in subgroups with moderate-severe communication problems, restricted interest and severe linguistic delays. HFOs seems to be a reliable marker for ASD that can be used in the clinical settings. The HFOs may be the underlying pathology affecting symptomatology or ethiological factors might cause emerging of HFOs in children with ASD that needs to be clarified with the future studies.

**Keywords:** Autism spectrum disorders, HFO, sleep spindles, EEG, pediatric autism
Chronic Multifocal Nonbacterial Osteomyelitis: Is it Syndrome or Disease?

Irina Nikishina¹, Alia Arefeva¹, Svetlana Arsenyeva¹, Valeria Matkava¹
¹V.A. Nasonova Scientific Research Institute of Rheumatology

Aim: Chronic multifocal nonbacterial osteomyelitis (CMNO) is a condition that has been managed by surgeons/orthopedics for a long time. Adoption of an autoinflammatory conception of CMNO became crucial in the redirection of most patients (pts) to pediatric rheumatologist that proposed new therapy approach. We analyzed the experience of pediatric department in the Russian Federal Institute of Rheumatology regarding clinical/laboratory features and treatment in patients with CMNO.

Material and Method: We analyzed a single center retrospective six-year study, which includes 70 pts with CMNO. Infections, oncology and other reasons of bone damage were excluded. In addition to standard assessments, whole-body MRI and/or scintigraphy were done.

Results: Among our pts girls were prevalent (40:30). The median age at the disease onset was 9.6 years (Me 9.9 range 1.3;16.5). All pts had diagnosis according to rheumatologically nosology as juvenile idiopathic arthritis (JIA), poly-33, oligo-22, psoriatic including, juvenile ankylosing spondylitis (JAS) in 15. According to our data, bone lesions (osteitis, destruction) localized most common in pelvis (28%), femur (41%), tibia (56%), fibula (24%), lumbar spine (20%), clavicle (12%). Extra-skeletal manifestations were observed in 13 pts (acne conglobate-3, uveitis-1, inflammatory bowel disease-2, psoriasis-7). HLA-B27 antigen was found in 13 pts. Therapy included NSAIDs (all pts), methotrexate -32(46%), sulfasalazine – 15(21%), bisphosphonates 11(16%), glucocorticoids 10(14%). 49 pts were treated by Biologics, TNF-inhibitors as first-line. Persistent arthritis, severe course of multifocal CMNO, appearance of new bone lesions, were determinant factors for Biologics indication. Totally, there were 59 prescriptions (48 - 1-st line, 9 - 2nd, 2 - 3rd). Since 2022 we have started to prescribe JAK-inhibitor Tofacitinib as a new options of target therapy for the 1-st line. Decreasing of disease activity was reached in most of the patients.

Conclusions: We believe that bone lesion in CMNO could be a manifestation of the rheumatic disease, spondyloarthropathies mostly and need to use anti-rheumatic therapy including Biologics and JAK-inhibitors.

Keywords: Chronic nonbacterial osteomyelitis, Rheumatic disease, Biologics, Tofacitinib
Heterotopic Ossification - New Challenges for Pediatric Rheumatologists

Irina Nikishina¹, Svetlana Arsenyeva¹, Valeria Matkava¹, Alia Arefeva¹, Artyom Borovikov²
¹V.A. Nasonova Research Institute of Rheumatology
²Research Centre for Medical Genetics

Aim:
Heterotopic ossification (HO) is the process of pathological bone formation in soft tissues and divided into two categories: non-genetic and inherited genetic forms (fibrodysplasia ossificans progressive (FOP) and Progressive osseous heteroplasia (POH)). Aim of the study to describe variety of patients(pts) with HO and analyzed the main clinical features of FOP and POH.

Material and Method:
From 1998 to 2023, we identified 49 pts with FOP, 6-POH and 18-acquired forms of HO. All pts met typical clinical features of FOP/POH, mutations in ACVR1/GNAS genes respectively.

Results:
In all 49 FOP pts (27 male/22 female) the diagnosis was verified by “classic” phenotype: malformed great toes-47/96%pts; malformed thumbs-16/33%; peripheral osteochondromas-33/67%; abnormalities of the cervical spine–49/100%, HO–48/98%. Genetic test (ACVR1) was done in 40/82%pts and confirmed typical or ultra-rare mutation. We found a lot of similarities with rheumatic diseases such as synovitis, sacroiliitis, patterns of bone ankylosis. 6 pts with POH (4 male/2 female) characterized by superficial and deep HO that begins in dermis and extended into skeletal muscles. The average manifestation age was 22.1 months (from 2 weeks to 4 years). All pts had diagnostic delay from 2.5 to 20 years. Most pts were misdiagnosed with calcinosis and even dermatomyositis instead of HO. Skin biopsy/surgical manipulations led to progression of POH in 4 pts. Unilateral disorder of extremities with loss of mobility detected in 3/50%pts, 2 of them had severe skin damage and limb shortening because of HO. Bilateral asymmetrical involvement with superficial HO was detected in 3/50% pts with no functional impairment. All pts had different GNAS mutations, 4 pts–de novo. Non-hereditary HO included 18 pts with various causes of ossification: posttraumatic or inflammatory mostly.

Patient with POH
“Cocon-like” type of HO in patient with POH

**Conclusions:** It is important to establish the type of HO in order to choose the correct management strategy and avoid invasive diagnostic methods which can lead to progression of HO and severe disability. Early diagnosis and reposition of certain antirheumatic drugs may improve the prognosis for HO.

**Keywords:** Progressive osseous heteroplasia, Fibrodysplasia ossificans progressive, Heterotopic ossification, ACVR1 gene, GNAS gene
The Relationship of Sleep and Quality of Life in Children with Attention Deficit and Hyperactivity Disorder: A Meta-Analysis Study

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Aim: This meta-analysis study was conducted to determine the relationship between sleep and quality of life in children aged 6-12 years with ADHD.

Material and Method: Various databases, including “EBSCO Host”, “Web of Science”, “Pubmed”, “Ovid” and “Science Direct” were used to identify observational studies that included the relationship between sleep and quality of life in children with ADHD and published between 2011 and 2020. Comprehensive literature review and selection of appropriate articles were carried out by two independent researchers. Methodological quality assessment of studies was evaluated by three independent researchers using the JBI Critical Evaluation Checklist for Cohort Studies, JBI Critical Evaluation Checklist for Case-Study Studies, JBI Critical Evaluation Checklist for Cross-Sectional Studies. Eight studies meeting the inclusion criteria were included in this meta-analysis. Comprehensive Meta Analysis (CMA) 3.0 program was used to analyze the data. Hedge’s g was calculated to determine the effect size and Random Effects Model was used according to heterogeneity analysis. Funnel Plot, Classic Fail Safe N method with Orwin’s number, Begg and Mazumdar Corelation with Kendall’s tau and Egger Regression analyzes were used to examine publication bias. Cohen Kappa test was used by 3 independent researchers to evaluate the reliability of the study (k= 0.87).

Results: In the current study, the studies included in the meta-analysis had a positive “very large effect size” (g= 1.313, p= 0.006). In the subgroup analyzes made to explain the high heterogeneity in the study, it was aimed to explain the factors affecting the meta-analysis result. The quality of life and sleep scales used in the studies included in this meta-analysis were determined to be important moderators (p<0.05).

Conclusions: The meta-analysis, observational studies with a larger sample size, high methodological quality in different age groups is recommended to examine the relationship between sleep and quality of life in children ADHD in the future.

Keywords: Children with ADHD, meta-analysis, quality of life, sleep
The Effect of Nursing Empowerment Program Applied to Families of Children with Phenylketonuria on Coping Attitudes of Families and Phenylalanine Levels of Children

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Aim: The study was carried out experimentally to examine the effects of the strengthening program applied to the families of 0-3 year old children diagnosed with phenylketonuria (PKU), on the families’ coping attitudes, their knowledge about the disease, and the phenylalanine levels of the children.

Material and Method: The data of the study were collected in Family Health Centers affiliated to Van Provincial Health Directorate between 24.11.2021-28.02.2023. The study was completed with the families of 36 children with PKU. Nursing empowerment program (HGP) was applied to the families in the experimental group for 10 weeks. The scope of this program; It consists of face-to-face training on disease management, the delivery of the training guide (booklet) prepared for the families of children diagnosed with PKU, phone calls and counseling services during the education process. The families in the control group were given a routine practice (regular blood monitoring, taking into account the recommendations of the relevant physician/dietitian, etc.) during the application of the study, and the same training and booklet were given after the application.

Results: The mean level of knowledge about PKU of the families in the experimental group (93.15±40.15) was found to be statistically significantly higher than that of the control group (53.24±18.20) (p<0.05). The mean blood FA levels of the children in the experimental group (325.17±161.10 µmol/L) were found to be statistically significantly lower than the control group (525.59±330.76 µmol/L) (p<0.05). It was determined that the total mean score of the families in the experimental group on the post-test coping attitudes after HGP (96.95±8.08) was higher than the mean score of the families in the control group (83.29±9.03) (p<0.05).

Conclusions: It was determined that HGP was effective in keeping the blood FA value of children in the reference range, and it increased the level of knowledge and coping attitudes of families about the disease.

Keywords: Coping Attitudes, Education, Empowerment, Family, Phenylketonuria
OP37 Three-Component Participatory Multidisciplinary Health Care Model for Children with Autism Spectrum Disorder (Asd) in Pediatric Setting

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Aim: Adults and children with autism are often subject to stigma and discrimination, including deprivation of health care. The aim of our study is to develop a participatory model of health care to overcome barriers in the provision of medical care to children with autism.

Material and Method: Conducting a focus-group: 1) determination of the focus-group (parents-activists of children with autism were invited, known among parents and among the professional communities; 2) questions (about barriers to medical examination of a child with ASD and opportunities to overcome); 3) conducting a focus-group; 4) data analysis and strategies to overcome barriers. Approval of a participatory model aimed at overcoming barriers in medical care (40 children).

Results: There were indicated numerous problems associated with ASD health care. Health care barriers: 1) there are difficulties in attraction the attention of health care specialists to the health care problems of children with autistic behavior; 2) medical staff is not trained enough to conduct diagnostic and therapeutic interventions for children with ASD – lack of competencies of medical professionals; 3) children with ASD require special conditions during health care visits (silence, no waiting etc); 4) parents are not aware of the importance of a timely medical examination of a child with autism; 2) no training of skills for passing medical procedures by a child.

Conclusions: We have developed the three-component participatory multidisciplinary health care model for children with ASD using following strategies to overcome barriers: 1) increasing competences of health care specialists (professional and communicative skills); 2) autism friendly clinic (resource zone with silence and anti-stress strategies); 3) information for caregivers (importance of health care visit for child, peer-to-peer training for other parents to prepare child to pass medical examination, on the basis of applied behavioral analysis). As a result, 78 children with ASD passed medical examination, received diagnoses and appropriate treatment of co-occurring diseases.

Keywords: Adults, children, autism, participatory model, multidisciplinary
OP38  Assessment Caregivers of Children with Duchenne Muscular Dystrophy

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Aim: Duchenne Muscular Dystrophy (DMD) begins at a young age, progresses with degeneration of the muscles. In its advanced stages, the patient is totally dependent on the caregiver. The aim of this study is to determine the caregiver burden, sleep quality and anxiety levels of the caregivers.

Material and Method: This is a cross-sectional study that involved caregivers. Caregivers were invited to complete the Sociodemographic Information Form, Zarit Caregiver Burden Scale (ZBI), Pittsburgh Sleep Quality Scale (PSQI), State and Trait Anxiety Inventory (STAI-S and STAI-T). The ZBI used to assess the level of caregiver burden. The PSQI used to evaluate sleep quality. The STAI-S and STAI-T used to measure state and trait anxiety.

Results: The caregivers of 27 patients with DMD were included. 26 (96.3%) were male and the mean age 130 (±34.4) months. 15 (%55.6) of the caregivers were mother and 12 (%44.4) were father. The ZBI mean score was 42 (±9.9) (moderate-severe caregiver burden), PSQI median score was 3 (1-6) (severely affected sleep quality), STAI-S mean score was 42.5 (±5.2) (moderate momentary anxiety level), STAI-T mean score 43.4 (±7.5) (moderate continuous anxiety level). There was a statistically high positive correlation between caregiver burden (ZBI) and PSQI scores (p<0.001, r=0.64). There was statistically moderate positive correlation between PSQI scores and STAI-T scores (p=0.03, r=0.41).

Conclusions: Caregiver burden was found moderate to severe. While the sleep quality was severely affected, the state and trait anxiety levels were moderately impacted. The caregivers require physical, social, and psychological support for the caregiving process.

Keywords: Caregiver Burden, Duchenne Muscular Dystrophy, Sleep Quality
Aim: Duchenne muscular dystrophy (DMD) is a progressive neuromuscular disorder characterized by delayed walking and progressive muscle weakness. Despite the existence of guidelines for recognizing typical symptoms and signs and the increased acceptability of genetic tests, there is still a significant delay in diagnosis. Aim of the study is to determine the time from the onset of symptoms to the diagnosis of DMD along with evaluating factors that may contribute to the diagnostic delay.

Material and Method: This retrospective study involved a review of the medical records of children with genetically confirmed diagnosis of DMD at a tertiary pediatric center between January 2005 and June 2023.

Results: We included 29 patients with DMD. In 19/29 patients, motor signs and symptoms of disease were first observed at the mean age of 3.5 years, while mean age of diagnosis was 5.6 years. The duration between the symptoms onset and the diagnosis ranged from 3 months to 6.9 years (mean 2 years). In a subgroup of two patients exhibiting predominant cognitive delay and behavior changes, the delay in diagnosis was 3.7 years. Eight patients were hospitalized due to incidental findings of elevated creatine kinase levels. On examination, two infants were asymptomatic, one patient had calf pseudohypertrophy, while 5 out of 8 patients had delayed motor milestones or waddling gate and difficulty rising from the floor, neither observed by the family nor by physicians. The reasons for delay in diagnosis were consistent with those found in previous studies, including lack of awareness of DMD symptoms and lengthy diagnostic journey from initial parental concerns to examination by a pediatric neurologist.

Conclusions: Raising awareness among parents and physicians regarding the symptoms of DMD and reducing the time of referral to a neurologist are crucial for early diagnosis and access to the treatment that can influence the progression of the disease.

Keywords: Duchenne muscular dystrophy, diagnostic delay, muscle weakness, calf pseudohypertrophy, creatine kinase
Results of a Prospective Open Observational Study of the use of Dornase Alfa (Tigerase, Generium, Russia) as Part of Complex Therapy in Patients with Cystic Fibrosis

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Aim: Dornase alfa (DA) is the most commonly used mucolytic drug for cystic fibrosis (CF). A biosimilar of DA Tigerase (Generium) has been developed in the Russian Federation. Aim: to analyze the data of long-term use of Tigerase.

Material and Method: A post-registration prospective multicenter non-comparative open observational (non-interventional) study was conducted to evaluate the results of using DA. The study included 165 (88 female) CF patients from 11 Russian CF centers. There were 120 patients aged 5 to 18 years and 45 patients older than 18 years. The treatment with DA was 52 weeks.

Results: The number of exacerbations before the start of the study was 1.42 ± 1.38 times (Me 1 (IQR 0; 2)), during treatment with DA 1.28 ± 0.53 times (Me 1 (IQR 1; 1)). FEV1 data in the general group of patients were 85.4±22.62% (Me(IQR) 89 (71;103))%, after 52 weeks 84.62 ± 24.13 (Me(IQR) 86.5 (71;102)) %, p=0.35. In adult patients, FEV1 at the start of therapy was 67.96 ± 21.82% (Me(IQR) 65.4(46.3;82)) %, after 52 weeks of therapy - 65.8±23.84% (Me(IQR) 69.7(48.7;80.95)) %, p=0.07. In children, FEV1 at the start of therapy was 91.83±19.32%(Me(IQR) 93(77;106)) %, after 52 weeks of therapy - 92.05±19.91 %, p=0.7. Among the adverse events (AE) in adult patients were: cough (7%), bronchitis (4%), hemoptysis (4%), shortness of breath (4%), nausea (2%), dry mouth (2%), sore throat (2%), headache (2%), taste disturbance (2%). One child had dysphonia (0.8%). In 164 of 165 patients (99.9%), drug discontinuation was not required. Antidrug antibodies were detected in two patient

Conclusions: Patients in childhood tolerated Tigerase well. In adult patients, AE were noted, which in most cases did not require discontinuation of the drug. There no statistically significant decrease in lung function and no increase in the number of exacerbations of chronic pulmonary disease during the study period/

Keywords: dornase alfa, cystic fibrosis
Features of the Formation of Verticalization in Children with Albinism

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Aim: To study the effect of connective tissue dysplasia on the timing of the formation of verticalization skills in children with oculocutaneous albinism.

Material and Method: The study involved children with a genetically verified diagnosis of oculocutaneous albinism aged 2 to 17 years. A study of the perinatal anamnesis and data on the timing of the formation of motor skills, as well as a physical examination, were carried out.

Results: According to the results of the examination, it was found that in the neurological status, in addition to low vision and nystagmus, almost all children had a symptom complex in the form of a combination of muscular hypotension-hyporeflexia and increased elasticity of the connective tissue, which varied from mild to moderate severity. In the anamnesis, attention was drawn to the peculiarity of the pace of motor development, which consisted in a slowdown in the acquisition of motor skills starting from the second half of the first year of life - that is, skills mainly associated with verticalization. Of all albino children with a normal perinatal history and no additional concomitant neurological diseases, only about a quarter acquired the skill of independent walking before 11 months of age, and only about half of the total number of these children began to walk independently by 1 year of age. In the remaining half of the children, most of the children formed independent walking skills between the 12th and 14th months of life, and some children only by the 15th and 16th months of life.

Conclusions: The data obtained indicate the effect of muscle hypotension-hyporeflexia in combination with increased extensibility of ligaments and joint capsules in children with oculocutaneous albinism on postural stability, which, along with their reduced visual control due to low vision, is of key importance in slowing down the process of mastering verticalization skills.

Keywords: Albinism, hypotension-hyporeflexia, motor development
POSTER PRESENTATIONS
PP01 Allergy in Children with Autism Spectrum Disorder (ASD)

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Aim: Background: Co-occurring medical conditions, including allergy, could influence on behavior and emotions of children with ASD. The purpose of the study: To identify allergy among children with ASD.

Material and Method: Single-stage non-comparative study. The research included 64 children with ASD (diagnosis verified with ADI-R), 45 males/19 females. The age ranged from 1 year to 17 years, (M=10+4.679). All patients were examined by a psychiatrist, pediatrician, allergist-immunologist. All children underwent a bottom-up approach using ISAC allergy chips, containing 112 components from 51 allergen sources, and ALEX, which allows determining 282 analytes, 156 extracts and 125 components. Statistical methods were used to process the information (EAACI Molecular Allergology User’s Guide).

Results: Allergy was detected in 34 (53%) patients of the examined group. Allergic rhinitis was diagnosed in 16 patients (25%), atopic dermatitis - in 9 (14%), pollinosis - in 3 (5%), bronchial asthma - in 3 (5%), food allergy - in 3 (5%). Multimorbidity, which included 3 or more allergic diseases, was observed in 4 patients who had a more severe course of the underlying health condition. Sensitization was present in 28 (43.8%) cases. 28 patients (43.8%) had no symptoms of allergy at the time of examination, however, 13 (46%) of them were found to be sensitized to various (inhalation, food) allergens and wasp venom. Polyvalent sensitization was observed in 8 (12.5%). Moderate/high levels of IgE to inhaled allergens and cross-reactive components prevail among 18 patients. Sensitization to the allergen Bet v1 of birch is important, which is observed in 6 patients (33.3%). Sensitization to the allergen Phl p1 of timothy grass was detected in 5 cases (27.7%). Sensitization to inhalational allergen (cat) was detected among 7 patients (38.9%).

Conclusions: High prevalence of allergy among children with ASD was shown. It is necessary to optimize health care for children with ASD with awareness in allergy.

Keywords: Autism spectrum disorder (ASD), allergy, children, multimorbidity, sensitization.

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Aim: The hematological results data in children with 2019-nCoV infection is limited. The aim of our study is to investigate hematological parameters included WBC, neutrophil, lymphocyte, monocyte, eosinophil, basophil and platelet count. Moreover, we also wanted to study hematological indices such as NLR, MLR, ELR, BLR, PLR and MPV/PC levels whether they have conventionally significant value in children with 2019-nCoV infection.

Material and Method: The study was retrospectively conducted with hospital electronic medical information system between March and May 2020 in a university hospital. Two groups were classified as positive real time reverse transcriptase-polymerase chain reaction (RT-PCR) with bilateral patchy shadows or ground-glass opacity in their lungs and negative RT-PCR for 2019-nCoV infection. Children with 2019-nCoV positive and negative, aged ≤18 years old were included.

Results: A total of 204 children including 103 boys (50.50%) and 101 girls (49.50%) mean aged 107.7±69.6 months were recorded in the study. Of these 98 and 106 patients were enrolled 2019-nCoV positive and negative, respectively. Mean CRP and LDH levels were mainly increased. White blood cell, lymphocyte, neutrophil, monocyte, eosinophil, basophil and platelet count were lower in 2019-nCoV positive patients than that of negative patients. WBC, neutrophil, monocyte, eosinophil, basophil, and platelet count were strong significant in 2019-nCoV positive cases compared with negative ones.

Heatmap
Correlation coefficient and significance levels based on laboratory results were shown with heatmap. WBC and BLR results showed a good-high correlation.

**Conclusions:** The hematological parameters and ratios have not been analyzed before in children with 2019 novel coronavirus infection. Our study highlighted that WBC, ELR and BLR might be convenient parameters to express inflammatory situation of coronavirus infection. Presented parameters can be very readily estimated with total leucocyte count and other ratios for every patient. In addition, there is no necessity of another expense and technique. Hematological parameters and ratios could be necessarily taken into consideration in terms of diagnosis and predictable prognosis for children with 2019 novel coronavirus infection in clinical practice.

**Keywords:** Children, hematology, infection, lymphocyte, coronavirus
The Clinical Particularities of Long-Covid in Children

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Aim: During the pandemic period with the SARS-CoV-2 virus as well as after it ended, we observed a considerable number of children and adolescents, who still presented the symptoms of the COVID-19 disease even at 12 weeks after the diagnosis. The purpose of our research was to monitor pediatric children infected with the SARS-CoV-2 virus, who were at risk of developing long-covid, as well as to identify the clinical manifestations of the post-covid syndrome and its evolution.

Material and Method: We conducted an observational prospective study between August 2021 and June 2022, in the Children’s Clinical Hospital “Dr. Victor Gomoiu” from Bucharest, in which we included 25 patients between 10-18 years, who presented suggestive symptoms for long-covid. The data was analyzed using the SPSS statistical program, version 28.0.1.

Results: In a proportion of 64%, these patients did not have comorbidities, with the exception of 16% of them who had respiratory comorbidities, respectively 4% gastroenterological ones. It is observed that the post-COVID manifestations increase with age, thus those between the ages of 10 and 18 presented: cardiovascular manifestations (6%), digestive manifestations (12%), respiratory manifestations (48%) and neurological manifestations/neuromuscular (36%). The most frequent respiratory symptoms observed were: cough, dyspnea and nasal obstruction, while from the gastroenterological field the abdominal pain and nausea were encountered. The most persistent post-covid cardiovascular manifestations were tachycardia and chest pains, while from the neurological manifestations, headache was the most frequent symptom, followed by arthralgias and myalgias.

Conclusions: The particularities of Long-Covid, as similar as those of Covid-19, still requires a careful analyze of their evolution. We discovered that the post-COVID manifestations increases with age, especially at this category of patients and that we need to conduct more studies to see the evolution of this disease.

Keywords: Long-Covid, children, symptomatology, particularities
Ambiguity in Assessing Epileptiform Discharges in Children with Cerebral Palsy

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Aim: Although the electroencephalogram (EEG) of children with cerebral palsy (CP) has been studied for a long time, there remain questions about the diagnostics of epileptiform discharges (ED). Therefore an agreement between specialists in assessing the changes is important. The goal is to assess the degree of discrepancy in the results of EEG analysis of children with CP by different neurophysiologists, and to determine the reasons for disagreement.

Material and Method: Two neurophysiologists analyzed separately the EEG of 60 children aged 2 to 17 years with. Seven patients had a history of clinical epileptic seizures. EEG-videomonitoring was recorded on a Neurosoft device (Russia) in the awake and daytime sleep states. One of the specialists did not know the clinical data of the patients.

Results: The coincidence of these specialists’ opinions on the presence of ED occurred in 90% of cases. Discrepancies concerned different assessment of atypical phenomena. Generalized paroxysmal fast activity was ambiguous, due to frequent amplification of high-frequency activity in the EEG. In some children the type of ED corresponded to benign epileptiform discharges of childhood. The same assessment of these discharges occurred in 92% of cases. There was a discrepancy in assessment when ED of different types were recorded in the same EEG. There was no discrepancy in determining the brain areas involved in epileptogenesis, but the description of the dominant focuses and the prevalence of changes differed in 10% of cases, which can be explained by the multifocal ED.

Conclusions: Visual analysis of ED in children with CP when evaluated by independent specialists can be ambiguous and have an error in the frequency of changes detected (5-10% in our study), which is partly due to the difficulty of interpreting EEG data in children with CP. This finding indicates the importance of a collegial discussion of the contentious issues of clinical and neurophysiological diagnostics.

Keywords: electroencephalogram, cerebral palsy, epileptiform discharges, children
Echocardiographic Assessment of Intracardiac Hemodynamics in Children with Apical Hypertrophic Cardiomyopathy

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Aim: To demonstrate methods for assessing intracardiac hemodynamics by the example of one of the rarest variant for remodeling the left ventricular cavity in apical hypertrophy – Yamaguchi’s disease.

Material and Method: Echocardiography is the main diagnostic method that allows you to visualize the anatomical features and severity of remodeling of the heart chambers in various pathological conditions, to assess intracardiac hemodynamics. For the study, we used ultrasonic scanners Acuson SC 2000, Vivid E90. The studies included 2D, 4D echocardiography and Doppler echocardiography. Patient 13 years old, BSA 1,63m², complaints of increased fatigue, weakness, dizziness during exercises. Auscultation: systolic murmur at the apex of the heart.

Results: Local concentric hypertrophy of the apical segment of the left ventricle up to 18 mm was visualized during echocardiography. The left atrium was not changed, diastolic function was not impaired (fig.1). The left ventricle volume studies and intracardiac hemodynamics was carried out according to the Simpson biplane method: EDV 138 ml, SV 78,66 ml, EF 57% at a heart rate of 72 beats/min, cardiac output 5,66 l/min, cardiac index 3,47 l/min/m². Simpson 4D (semi-automatic mode): EDV 144,31 ml, SV 75,37 ml, EF 52,23% at heart rate 80 beats/min, cardiac output 6,03 l/min, cardiac index 3,7 l/min/m². Both variants of the study allow us to assess the pumping function of the heart, however, 4D echocardiography allowed us to assess the segmental contribution to the global systolic function of the left ventricle, which is important for estimation the mechanics of the apical segment, which is not available for standard techniques.

4D echocardiography

Conclusions: 4D echocardiography allows us to obtain more objective data on intracardiac hemodynamics and the severity of remodeling of the heart cavities without the need for invasive
and ionizing diagnostic methods, correctly interpret the patient’s condition and choose adequate treatment at the beginning of the disease.

**Keywords:** Echocardiography, Yamaguchi’s disease, apical hypertrophic cardiomyopathy, 4D echocardiography, ultrasound
Acute Transverse Myelitis Occurred After Upper Respiratory Infection: Case Report

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Aim: Background/Objective: Childhood acute transverse myelitis is a rare clinical syndrome of the central nervous system with an inflammatory involvement of the spinal cord characterized by acute onset of motor, sensory and autonomic dysfunction. Acute transverse myelitis cases visible in about 20% child by literature.

Material and Method: Case Report: The girl child was 13 years of age came no walking, hypokinesia and weakness in leg and complaint of urinary incontinence symptoms after 10 days from upper respiratory tract infection at pediatric emergency department. She could take 2 step without support. She had 4/5 muscle strength. Acute phase reactants was negative in blood. Spinal cord MRI revealed consistent with syringohydromyelia from under the C3 vertebra to the level of the C7 upper endplate. From T1 vertebra to mid-thoracic level; Lesions involving long segments extending from the lower thoracic level to the conus were observed. We thought might be an acquired demyelinating disease in the examinations performed. We were started on pulse steroid therapy on the patient. We referred to more comprehensive children’s hospital for investigation. Mycoplasma pneumonia IgM (1.522 +) and Chlamydia pneumonia IgM (1.549 +) were intermediate values, the patient was started on clarithromycin. Anti-MOG IgG negative, Neuromyelitis optica IgG negative. The patient was given pulse steroid therapy for the first 7 days of her hospitalization with the diagnosis of seronegative acute transverse myelitis. Then the patient was started with 35 mg prednisolone orally. The patient could walk without support 15 days after hospitalization. The patient taken IVIG treatment twice, recovered completely after 4 months

Long segment hyperintense signal seen on the T2WI
Spinal cord MRI revealed consistent with syringohydromyelia from under the C3 vertebra to the level of the C7 upper endplato. From T1 vertebra to mid-thoracic level; Lesions involving long segments extending from the lower thoracic level to the conus were observed.

**Results:** Discussion: If there isn’t infectious cause in acute transverse myelitis, high-dose intravenous corticosteroids or plasma exchange using immunomodulatory therapy recommended.

**Conclusions:** Acute transverse myelitis may develop after upper respiratory tract infection in child. The diagnosis of ATM and early induction of steroid therapy are important for prognosis in child.

**Keywords:** Acute transverse myelitis, pediatric, Spinal cord inflammation, Upper respiratory infection
PP07  Body Weight and Health-Related Quality of Life in Adolescents

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Aim: to Assess The Interconnection Between the Anthropometric Variables and Health-Related Quality of Life (Hrqol) in a Community-Based Sample of Schoolchildren and to Establish the role of Body Dissatisfaction (BD) in this Association

Material And Method: A Number of Tenth-Year Schoolchildren (N=413) Completed A Questionnaire Making it Possible to Describe Their Sociodemographic and Behavioral Characteristics And Assess Body Satisfaction And Health-Related Quality of Life (Hrqol) (Pedsql 4.0, France). The Anthropometric Measurements were Performed by Medical Professionals and, in Addition to BMI, the Waist Circumference (WC) and WC/ Height Indices were Assessed. The Hypothesis Of BD as Reflecting the Relationship Between Anthropometric Variables and Hrqol was Tested Using the Logistic Regression Method.

Results: No Significant Correlation was Found Between BMI and Integral Hrqol Scores. Waist Circumference Showed a Significant Negative Correlation with the Hrqol Scores in the Girls, but not in the Boys. The Frequency of BD was 60.3% (N=249). The Relationship Between BD and Anthropometric Variables Proved to be Differently Directed in the Boys and the Girls. BD had a Significant Negative Correlation with Psychosocial and Total Hrqol Scores in Adolescents of Both Sexes and with the Girls’ Physical Functioning Scores. The Relationship Between Hrqol and Anthropometric Indices in the Girls, After Making Adjustment For Sociodemographic, Behavioural Variables and BD in Multivariate Models, was No Longer Significant (Table).

Factors Associated with Low Total Hrqol Scores in Schoolgirls

<table>
<thead>
<tr>
<th></th>
<th>OR</th>
<th>95% CI</th>
<th>P-Level</th>
<th>OR</th>
<th>95% CI</th>
<th>P-Level</th>
</tr>
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<tbody>
<tr>
<td></td>
<td></td>
<td>Multivariate* (R2 = 0.34)</td>
<td></td>
<td>Multivariate # (R2=0.37)</td>
<td></td>
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<tr>
<td>Housing Conditions</td>
<td>9.8</td>
<td>3.5 - 27.6</td>
<td>0.001</td>
<td>10.1</td>
<td>3.5 - 28.7</td>
<td>0.001</td>
</tr>
<tr>
<td>Staying Outdoors Less</td>
<td>4.4</td>
<td>2 - 10</td>
<td>0.001</td>
<td>4.6</td>
<td>2 -10.6</td>
<td>0.001</td>
</tr>
<tr>
<td>Than 30 Minutes A Day</td>
<td></td>
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<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>PC Games Time On</td>
<td>1.3</td>
<td>1.1 - 1.7</td>
<td>0.007</td>
<td>1.3</td>
<td>1.05 - 1.6</td>
<td>0.017</td>
</tr>
<tr>
<td>Weekdays</td>
<td></td>
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</tr>
</tbody>
</table>
Another Screen Time On Weekdays & WC / Height & BD

<p>| | | | | | | |</p>
<table>
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<th></th>
<th></th>
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</tr>
</thead>
<tbody>
<tr>
<td>Another Screen Time</td>
<td>1.6</td>
<td>1.2 - 2.2</td>
<td>0.001</td>
<td>1.6</td>
<td>1.2 - 2.1</td>
<td>0.003</td>
</tr>
<tr>
<td>WC / Height</td>
<td>1.4</td>
<td>1.05 - 2</td>
<td>0.022</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>BD</td>
<td></td>
<td></td>
<td>2.2</td>
<td>1.3 - 3.6</td>
<td>0.002</td>
<td></td>
</tr>
</tbody>
</table>

* Adjusted To WC/ Height; # Adjusted To Body Dissatisfaction

**Conclusions:** This Study Has Shown that BD is Widespread Among Adolescents and has a Significant Impact on Hrqol. The Impact of Actual Weight Status On Girls’ Quality of Life is Entirely Mediated by their Dissatisfaction with their own Bodies, Thus Justifying the Approach to BD as a Stand-Alone Problem Having Important Psychosocial Implications and Requiring Active Identification and Targeted Intervention.

**Keywords:** Health-Related Quality of Life, Body Dissatisfaction, Body Weight, Adolescents
PP08 Risk Factors for Obesity in Rural Adolescents: A Case-Control Study

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Aim: The Aim of This Study was to Evaluate the Association Between Early Life Factors, Sociodemographic Influences, Behavioral Characteristics and Obesity in Adolescents Living in Rural Areas of Buryatia (Russia).

Material And Method: A Case-Control Study was Performed. A Total of 158 Adolescents Aged Between 11 And 17 Years Of Age (79 Adolescents with Normal Weight and 79 Adolescents with Obesity) were included in the Study. All the Adolescents had their Weight and Linear Growth Measured, and BMI was Calculated. Height and Weight Parameters were Based on the Reference Values Accepted by the WHO. Obesity was Diagnosed at BMI ≥95th Percentile the Data on the Variables of Interest were Obtained from Questionnaires Filled in by Parents and Adolescents and Their Medical Records from Local outpatient Clinics.

Results: The Results Showed that the Adjusted Ors and 95% Cis For Obesity were 1.15 (1.05–1.26), 4.87 (1.82–13.02), 3.08 (1.19–7.99) for Maternal BMI, Growth in Infancy and Unhealthy Eating Patterns, Respectively. Breastfeeding Longer than 12 Months Showed to Play a Protective Role for the Development of Obesity in Adolescence as Compared to a Shorter Period of Breastfeeding (OR=0.05, 95% CI: 0.006–0.40).

Conclusions: Maternal BMI, Rapid Growth in Infancy and Unhealthy Eating Patterns Are Associated with Increased Risk of Obesity in Adolescents in Rural Areas of Buryatia, While Long-Term Breastfeeding is a Protective Factor Against Obesity.

Keywords: Obesity, Breastfeeding, Growth In Infancy, Adolescents
PP10  Neuroendocrine Tumors, A Rare Entity in Children Age Case Report

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1University Hospital Center “Mother Teresa”, Tirana, Albania

Aim: The Aim of this Clinical Case Presentation is to Highlight the Symptoms of a Patient with a Neuroendocrine Tumor.

Material And Method: The 12-Year-Old R.B. has been Diagnosed with Acute Idiopathic Thrombocytopenic Purpura and has had Ecchymosis, Petechiae in the Limbs for 4 Days, Abdominal Pain Time After Time. An Abdominal Echo Revealed a Heterogeneous Mass with Distinct Outlines and Vascularization in the Inferior Pole of the Kidney and Spleen that was Considered to be a Neuroendocrine Tumor. To Confirm the Diagnosis, A CT-Scan With Ki/V and A Surgeon’s Consultation are Needed.

Results: The Patient Has Gone Through A Procedure of Removing the Tumor. The Entire Tumor Mass, The Tail of the Pancreas and a Splenectomy Were Completely Removed. A Well-Differentiated Pancreatic Neuroendocrine Tumor G1 with Capsular Invasion and Peritumoral Adipose Tissue were the Findings of the Histological Analysis. The Patient Had A PET-Scan, which Presented No Local Lesions That Hyper-Attract the Tracer.

CT

The CT with Contrast of the Patient

Conclusions: Since Neuroendocrine Tumors are a Rare Pathology, their Early Detection and Treatment are Crucial to the Excellent Prognosis of the Pathology. For the Early Phases of the Pathology, Surgical Excision Continues to be the Best Option. PET and CT-Scans Provide Great Specificity and Sensitivity For Identifying and Tracking Pathology.

Keywords: Surgical Excision, Thrombocytopenia, Biopsy, Neuroendocrine Tumor.
Computer Games: Importance in the Formation of Verbal-Logical Thinking of Adolescents

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²Pirogov Russian National Research Medical University, Moscow, Russia

Aim: to Identify the Features of the Level of Development of Verbal-Logical Thinking in Adolescents with Different Levels of Gaming Computer Activity

Material And Method: Participants: 83 People (44 Girls), Students of Russian Secondary Schools Age: 14 to 16 Years Old Methods: 1. Questionnaire Containing Questions About The Intensity of the Experience of Computer Games in Different Time Periods and Preferred Games 2. Methods for Studying the Features of Verbal-Logical Thinking:
   1) Subtest of Wechsler’s Verbal Scale
   2) Test of Elementary Logical Operations

Results: An Increase in the Intensity of Computer Gaming Activity on Weekends is Associated with Better Performance in Elementary Formal-Logical Operations, while Those Adolescents who Actively Play During The Holidays (> 3 Hours) Have Worse Results, and with Low Intensity (>1 Hour ) - The Most (P=0.019) The Significance of the Intensity of the Experience of Computer Games on Weekends and Holidays (P=0.008) And On Weekdays And Holidays (P=0.038) for the Level of Conceptual Synthesis is Noted. Moderate Intensity of Computer Game Experience (1-3 Hour) is Associated with the Best Performance in Conceptual Synthesis, High Activity With The Worst Results. Computer Gaming Activity Did Not Show Any Significance For The Ability To Categorial Generalization (Livin’s Criterion - 0.019). Adolescents Who Prefer Action Games Perform Elementary Formal-Logical Operations Worse (P=0.003) with a High Level Of Computer Gaming Activity, there is a Lower Academic Performance in All Subjects.

Conclusions: Intensive Experience Of Computer Games is Negatively Associated with the Ability to Conceptual Synthesis, as well as the Deterioration of Academic Performance in Basic School Subjects. For the Formation of Formal-Logical Operations, Computer Games can Become an Auxiliary Tool For Development. What Matters is the Genre of Preferred Games - Action Games Are Associated with Negative Results.

Keywords: Adolescents, Verbal-Logical Thinking, Computer Game, Cognitive Functions, Digitalization
The Possibility of Diagnosing Anticoagulation Disorders in Children

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¹Pediatrics and Child Health Research Institute of Petrovsky National Research Center Of Surgery, Moscow, Russia
²Research Centre For Child Psychoneurology

Aim: To Evaluate Changes in The Physiological Anticoagulant System in Children With Various Pathologies.

Material And Method: Deficiency Of Anticoagulant Factors in Children With Chronic Diseases is An Urgent Problem in Paediatrics And Neonatology, Especially When Using Venous Catheters, As The Risk Of Thrombotic Events, Disability And Death Increases. The Investigation Was Carried Out At The Consultative Diagnostic Center For Children Of The Research Institute Of Pediatrics And Children Health Protection Federal State Budgetary Scientific Institution “Academician B.V. Petrovsky” (Moscow) From 2021 To 2023. We included 228 Children With Various Chronic Diseases (Cardiovascular, Nervous, Musculoskeletal System, And Patients With Orphan Diseases). Blood Plasma Levels Of Protein C, Protein S And Antithrombin III Were Determined On An STA Compact Max Automatic Coagulograph (Stago, France).

Results: Protein C Deficiency Was Found In 28 Patients (12%) (Me 88, From 25% To 70%). Protein S Deficiency Was Detected In 41 Patients (18%) (Me 84, 42% To 70%). Decreased AT III Activity Was Detected In 7 (3%) (Me 116, 70% To 89%) Patients. Mild Protein C ...
Deficiency (50-60%) was found in 4 patients in combination with Protein S deficiency. These patients had a history of thrombotic episodes of lower limb veins, acute cerebral haemorrhage, transient cerebral impairment. At the same time, half of them had Leiden mutation (Coagulation Factor (FV) Leiden gene R534Q G>A, heterozygote), combined with polymorphisms of fibrinogen (FGB455 G>A) and plasminogen activator (SERPINE1/PAI-1, 675, 5G>4G) genes in homozygous state. Clinical manifestations occurred against a background of severe bacterial infection.

**Conclusions:** The results confirmed the significance of the study to assess the hemostasis system dysfunction in children with chronic diseases and the relevance of continuing the study. The results of the ongoing study will subsequently make it possible to identify at-risk groups of children in the population in order to prevent the development of complications of vascular and platelet haemostasis.

**Keywords:** Haemostasis, Protein C, Protein S, Anti-Clotting System
Venous and Capillary Point-of-Care Compared of Laboratory Bilirubin Measurement in Neonates

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¹University Of Ioannina, School Of Medicine, Ioannina, Greece

Aim: To Evaluate The Agreement Between Point-Or-Care Bilirubin Measurement in Venous And Capillary Samples And Laboratory Measurement.

Material And Method: We Conducted A Prospective Comparative Study For Diagnostic Test Validation, including Neonates ≥34 Weeks Of Gestational Age, And ≥72 H Of Age. The Agreement Between Point-Of-Care (Calmark Neo-Bilirubin) Venous And Capillary, And Laboratory Venous Bilirubin Was Examined With The Bland-Altman Plot, And The Passing-Bablok Regression Analyses.

Results: The Mean Bilirubin Was 12.68 (2.33) Mg/Dl in Reference Venous, 13.45 (2.69) Mg/Dl in Tested Venous, And 13.54 (2.79) Mg/Dl in Tested Capillary Samples. Bland-Altman Plots Showed Good Agreement For Tested Methods With The Reference Venous Method, And Between The Tested Methods. The Bias Between The Reference Venous And The Tested Venous Methods Was 0.771 [Levels Of Agreement (-1.814)-3.357], Between The Reference Venous And The Tested Capillary Methods 0.865 [Levels Of Agreement (-2.283)-4.014], And Between Tested Venous And Capillary Methods 0.094 [Levels Of Agreement (-3.118)-3.306]. Passing-Bablok Analyses Suggested No Proportional Or Systematic Bias Between The Reference And The Tested Methods. Figure 1.

Conclusions: The Point-Of-Care Venous And Capillary Bilirubin Levels Were in Good Agreement With The Reference Venous Measurements, And With Each Other, Suggesting The Point-Of-Care Measurement As Alternative For A Less-Invasive, More Rapid Evaluation Of Bilirubin.

Keywords: Diagnostic Test, Hyperbilirubinemia, Jaundice, Phototherapy, Point-Of-Care
Atopic Dermatitis – Correlation with Allergic Rhinitis

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Aim: Atopic Dermatitis, Better Known As Eczema, is The Most Common Skin Disease in Children. It Mostly Appears in the First Year Of Life, Most Often Between The Second And Fifth Months, is Chronic in Nature, With Periods Of Worsening And Improvement. In The Majority Of Children, it Lasts Until The Age Of Five, Or Sometimes it Can Last Until Puberty. Children With More Severe Atopic Dermatitis Are More Likely To Have Other Atopic Disease, Such As Allergic Rhinitis Or Asthma. Very Rarely Does A Patient Have All Three.

Material And Method: The Children’s Cards Aged 0–18 Treated in Our Ambulance in Own Research.

Results: The Research Has Demonstrated That 103 Children Have Atopic Dermatitis, 58 Boys(56,3%) And 45 Girls (43,7%). Allergic Rhinitis Have 207: 108 Boys And 99 Girls. The Researched 108 Boys With Allergic Rhinitis 58(53,7%) Have Atopic Dermatitis That Is 28% Of The All Children With Allergic Rhinitis. The Researched 99 Girls With Allergic Rhinitis 43(43,4%) Have Atopic Dermatitis That Is 21,7% Of All Children With Allergic Rhinitis.

Conclusions: Total Number Of Children With Atopic Dermatitis is 103; 58 Boys And 45girls. A Degree Of Correlation Between Allergic Rhinitis And Atopic Dermatitis is Little Bit Higher Among Boys(28%) Then Among Girls (21,7%). In This Small Sample There Is A High Degree Of Correlation Between The Two Diseases.

Keywords: Atopijski Dermatitis, Rinitis, Korelacija, Deča
PP15 Retinoblastoma; Albanian Data

Donjeta Alia\Balj, Denisa Dyrmishi, Rajna Kanina, Mirzana Kapllanaj, Mirela Xhafa, Enkeleda Duka, Eneda Balliu, Alketa Tandili, Anila Godo, Iren Maloku, Fabian Cenko, Massimo Pettoello-Mantovani

University Hospital Center “Mother Teresa”-Tirana
Catholic University “Our Lady Of Good Counsel“, Tirana
University Of Foggia-Italy

Aim: To Present the Distribution of Retinoblastoma, The Characteristics of This Distribution, in Albania Pediatric Population, as well as the Different Clinical Forms of its Presentation.

Material And Method: The Study is Retrospective and Includes the Identification of Patients with Retinoblastoma, Born From 1997 to 2022, while the Cases Analyzed in Terms of Clinical Characteristics were Diagnosed from 1 January 1998 Until 28 February 2023, at the Pediatric Oncohematology Department of the UHC “Mother Teresa”-Tirana.

Results: From the Results Found, for This Period, there are 30 Cases in the Hospitalization Registers. At The Same Time, Work Was Also Done with the Clinical Records of The Day Hospital of our Service, on the Clinical Characteristics and Their Follow-Up. The Description of the Sample According to Age is Presented as Follows: Average Age is 1.7 Years, Standard Deviation - 1.5, and Lower and Higher CI = [1month; 6 Years]. The Largest Distribution Was Observed in Female 19/11. The Median Age is 1.2 Years, which Explains that the New Diagnosis is Mainly Made in Young Children. Regarding Localization, 44% were Bilateral, 39% Dexter and 17% Sinister. Out of a Total of 22 Cases, 19 Were Presented After Enucleation of One of The Eyes, of Which 3 Were Bilateral Forms. The Most Frequent Initial Complaints Result in Leukocoria and Strabismus. Regarding Geographical Distribution the Most Cases Belong to Central Albania. Retinoblastoma Distribution in Albania

Conclusions: Retinoblastoma, Although a Disease with a Low Incidence, Continues to be one of the Most Disabling (Mutilating) and Life-Threatening, in Countries with Unfavorable Social and Economic Conditions and with Poor Recognition of the Seriousness of this Disease. The History of our Cases Shows Late Presentations to the Doctor, or when they
are Presented Early, the work in the Multidisciplinary Group is Delayed. A Very Simple Routine Ophthalmological Examination, even by a Basic Pediatrician, Prevents Major Dramas.

**Keywords:** Prognosis, Ophthalmological Examination, Pediatrician, Distribution, Retinoblastoma
Prevalence of Cow’s Milk Sensitization in Children with Allergies

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¹Pediatrics And Child Health Research Institute Of Petrovsky National Research Center Of Surgery, Moscow, Russia
²Pirogov Russian National Research Medical University, Moscow, Russia

Aim:
Cow’s milk is one of the main allergens in childhood. It is important to consider regional characteristics when determining sensitization profiles, as this can help develop personalized management measures for children. The aim was to determine the prevalence of sensitization to cow’s milk allergens in children with allergies living in the Moscow metropolitan area.

Material and Method:
A cross-sectional study was conducted on 160 children (0 months - 17 years 11 months) with symptoms of allergic rhinitis/atopic eczema/food allergy/asthma, living in the Moscow metropolitan area. Sensitization to cow’s milk extract was detected using ImmunoCAP; component-resolved diagnostics was conducted using ImmunoCAP ISAC or ALEX assay.

Results:
Sensitization (0-6 class) to cow’s milk allergen extract was found in 77 (48%) patients. Among the components of cow’s milk sensitization to alpha-lactalbumin Bos d 4 was detected in 14 (8.7%), to casein Bos d 8 and beta-lactalbumin Bos d 5 in 6.8% patients each. Asymptomatic sensitization to cow’s milk allergen was observed in 50 (65%) children. 35 (22%) patients reported symptoms related to cow’s milk: with 22 experiencing rash, 8 - diarrhea, 7 - urticaria, 3 – vomiting, 4 – sneeze and itchy throat, 1 - anaphylaxis, out of them only 13 (3.7%) had sensitization to cow’s milk allergen extract, 22 (63%) patients reported symptoms (rash, diarrhea or sneezing) related to cow’s milk, but the sensitization to cow’s milk extract/components was not detected.

Conclusions:
The frequency of sensitization to cow’s milk extract was found to be 5 times higher than to components in children with allergies living in the Moscow metropolitan area. More than half of the children had asymptomatic sensitization to cow’s milk allergen. Only in 1/3 of patients whose parents reported complaints the sensitization was detected.

Keywords: cow’s milk sensitization, children, component-resolved diagnostics
PP18 Treatment of Speech Disorders in Children Using Transcranial Magnetic Stimulation.

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²Pirogov Russian National Research Medical University, Moscow, Russia

Aim: To evaluate the efficacy and safety of transcranial magnetic stimulation in the treatment of children with speech disorders.

Material and Method: 46 children were examined, matched by sex and age, the structure of speech diagnoses: 26 children received treatment with TMS, 20 children received treatment with a nootropic drug. All children underwent psychological and pedagogical assessment of speech and cognitive development, EEG before and after treatment. A comparative analysis of the effectiveness of TMS and the use of nootropic therapy in patients with speech development disorders was carried out.

Results: Treatment of speech developmental disorders in preschool children with transcranial magnetic stimulation showed statistically significant positive dynamics and safety, with an efficiency exceeding drug treatment in terms of the main parameters. In the main group, the positive dynamics in speech development was 15.4% of the initial level (in the comparison group - 10.3%), in the average number of words in a phrase - 74.1% (in the comparison group - 48.9%). No adverse reactions were recorded in the treatment of TMS in any case.

Dynamics of parameters of speech development in the studied groups

<table>
<thead>
<tr>
<th>Parameters of speech development</th>
<th>Main group</th>
<th>P value</th>
<th>Comparison group</th>
<th>P value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Initial level</td>
<td>30.2±4.5</td>
<td></td>
<td>32.1±2.9</td>
<td></td>
</tr>
<tr>
<td>Average number of words in phrase</td>
<td>7.3±1.3</td>
<td></td>
<td>6.7±1.2</td>
<td></td>
</tr>
<tr>
<td>Initial level</td>
<td>861.2±120.4</td>
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<td>952.4±121.3</td>
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</tbody>
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...
Conclusions: TMS in the treatment of children with speech development disorders is a non-invasive and safe technique. The study demonstrated the potential of using TMS in the treatment of children with speech development disorders.

Keywords: transcranial magnetic stimulation, speech disorders, impaired cognitive development, children, effectiveness
Clinical Case of Stickler Syndrome in a Patient with Juvenile Arthritis

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²Research Centre For Medical Genetics

Aim: Stickler syndrome (SS) is a genetic disorder of type II collagenopathies which connected with pathogenic variants in 6 different genes and may lead to severe disability due to multiple system involvement: skeletal/joints disorderes, ophtalmological complications, hearing loss. We describe a clinical case of newly diagnosed SS which was recognized because of unusual course of Juvenile Idiopathic Arthritis (JIA).

Material and Method: Case report: Boy, 9 y.o. was admitted to our clinic with polyarticular course of JIA. Since 6 y.o. he had suffered from progressive disability due to limitation of motion in hip joints, knees and pain in lumbar spine. Instrumental findings revealed kyphoscoliosis, hypoplasia of the iliac bones, signs of thoracic and lumbar vertebrae destruction (platyspondylia and wedge-shaped deformation). Infectious/oncologic diseases were excluded. His mother has diagnosis of ankylosis spondylitis with total hips replacements. HLA-B27 was negative in both.

Results: According to detailed examination we observed definite signs of multiple synovitis, active bilateral sacroiliitis and foci of bone destruction, regarded as Chronic non-bacterial osteomyelitis (CNO), osteonecrosis of acetabulum. But there were also many features unusual for JIA: valgus deformity of lower extremities, waddling type walking, short stature, strabismus, progressive myopia and conductive hearing loss, lytic lesions of thoracic/lumbar vertebrae. Genetic disorder suggested. Target sequencing confirmed heterozygous pathogenic variants in COL2A1 gene. Patient was treated by NSAIDs with mild positive effect. Given the ambiguous situation, we decided to prescribe Tofacitinib, avoiding the use of methotrexate and received very good response-relief of pain, increasing of motion in hips and knees.

Pelvic X-Ray Of Patient With Stickler Syndrome

Severe Destruction Of The Acetabulum As A Manifestation Of Spondyloepiphyseal Dysplasia
**Conclusions:** In this case we came across with definite diagnosis of SS(COL2A1+) on the one hand and all the grounds to make a diagnosis of JIA associated with CNO on the other hand. We couldn’t say yet whether this is a case of a special inflammatory phenotype of SS or its combination with JIA. The universal anti-inflammatory effect of JAK-inhibitors Tofacitinib seems to be the preferred alternative. It allows us to hope for the prevention of joint destruction.

**Keywords:** Stickler syndrome, COL2A1 gene, Spondyloepiphyseal dysplasia, Chronic non-bacterial osteomyelitis, Juvenile Idiopathic Arthritis
Allergic Pathology in Children with Recurrent Upper Respiratory Tract Diseases

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2Pirogov Russian National Research Medical University, Moscow, Russian Federation

Aim: Recurrent upper respiratory tract diseases in children are a major concern in pediatrics. There are various coexisting conditions that contribute to the recurrence of inflammatory processes in the upper respiratory tract in children, and one of these conditions is allergic pathology. The aim of the study was to determine the frequency of concomitant allergic pathology in children with recurrent upper respiratory tract diseases.

Material and Method: The study included 65 children aged 3 to 18 years: 58% boys (mean age of 11.13 ± 3.4 years) and 42% girls (mean age of 10.41 ± 3.2 years) diagnosed with recurrent tonsillitis, rhinosinusitis, adenoiditis, and otitis media. The examination involved instrumental methods such as endoscopy of the ENT organs and tympanometry, as well as laboratory methods to determine total and specific IgE levels. The patients were consulted by a pediatrician, ENT doctor and allergist.

Results: According to our study 57 (88%) out of the examined children had allergic diseases as a comorbid pathology: 35 (54%) children had allergic rhinitis, 11 (17%) - food allergy, 6 (9%) - atopic dermatitis, 5 (8%) – asthma. Allergic rhinitis was newly diagnosed in 18%, asthma - in 3% of the cases.

Conclusions: Children with recurrent upper respiratory tract diseases often experience a high frequency of allergic reactions. However, the clinical symptoms of these allergic diseases are often underestimated. Therefore, it is important to include allergen testing and consultation with an allergist in the examination process for these patients. This will help determine personalized management tactics, prevent recurrence of symptoms, and the development of chronic inflammation in the upper respiratory tract.

Keywords: Recurrent Upper Respiratory Tract Diseases, Children, Allergic Pathology
Assessing the Health-Related Quality of Life in Adolescents Using EQ-5D.

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²Pirogov Russian National Research Medical University, Moscow, Russia

Aim: One of the important indicators of the patient's health status and their interaction with the doctor is their quality of life. Assessing health-related quality of life (HRQOL) can help identify aspects of social functioning, psychological and physical states, and the degree of adaptation to illness. The aim of the study was to estimate HRQOL and the incidence of allergy manifestations in adolescents.

Material and Method: A total of 300 conditionally healthy adolescents living in Moscow with median age of 15.00 [15.00; 17.00] years, were interviewed using the standardized tool EQ-5D and GA2LEN questionnaire.

Results: An analysis of the health profiles of the respondents showed that the majority of adolescents indicated no problems (level 1) in the following components: mobility (96%), self-care (98%), usual activities (94 %), pain/discomfort (86%), anxiety/depression (68%). However, 30% of respondents indicated some problems (Level 2) and 2% indicated significant problems (Level 3) when assessing the anxiety/depression component. Additionally, girls reported having some/extreme problems associated with anxiety/depression significantly more often than boys (p=0.008). The odds of girls having problems associated with anxiety/depression were 2.3 times higher than boys (OR 2.43; 95% CI 1.35; 4.61, p=0.004). The average self-assessment score on the EQ visual analogue scale (EQ VAS) was 84.8 points (min 15, max 100). There was no significant difference in self-assessment of health on the EQ VAS scale between boys and girls (p=0.062). It was found that the majority of children (86%) who indicated significant problems when assessing the anxiety/depression component had any allergy symptoms: 71% - manifestations of allergic rhinitis, 43% - asthma-like symptoms and 14% - eczema symptoms.

Conclusions: Adolescents with asthma-like and allergic rhinitis symptoms were significantly more likely to have reduced HRQOL, including problems related to anxiety/depression, compared to children without these symptoms (p=0.032 and p=0.024, respectively).

Keywords: adolescents, allergy, HRQOL, EQ-5D
The Pediatrician’s Role in the Detection of Congenital Diseases

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Aim: Congenital diseases constitute a significant part of pathologies. The role of a pediatrician in the diagnostic process is especially crucial. This specialist should estimate the presence of dysembryogenesis stigmas, which may be clinical markers of hereditary and congenital diseases. If there are five or more stigmas, the presence of a genetic syndrome should be suspected.

Material and Method: The parents of a 4-month-old child have reported asymmetry in the child's body. They noticed this problem during the child’s early growth. An instrumental screening was conducted at two months of age, which revealed a 5mm atrial septal defect with a left-right shunt, signs of right atrium overload, and a level I aortic insufficiency on the echocardiogram. It was also discovered that the child's father has a flattened nape, and the paternal grandfather has asymmetrical auricles that differ in size. Through palpation and visual observation, it was found that the left leg and left hand are larger and longer than the right limbs, eyes OS>OD, pupils OS=OD, the left auricle is larger than the right, and there is an appendage at the level of the left tragus, while the left external acoustic meatus is narrower than the right. There is plagiocephaly combined with a flattened nape and a high-arched hard palate. No pathology was observed in other organs and systems.

Results: The child with five stigmas of dysembryogenesis, was referred for a genetic consultation. The geneticist suggested a diagnosis of Goldenhar syndrome due to the combination of facial dysmorphia and cardiovascular abnormalities. Consequently, the child was recommended for genomic sequencing.

Conclusions: A pediatric physical examination is the initial step in diagnosing all diseases, including congenital and genetic conditions. During the propaedeutic examination, it is important to identify stigmas of dysembryogenesis, as their number and combination can provide direction for further diagnostic investigations.

Keywords: congenital diseases, Goldenhar syndrome
Clinical Case of Complicated Varicella in a 5-Year-Old Child

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Aim: To present a clinical case of a complicated course of varicella in a 5-year-old child.

Material and Method: The child’s medical history was analyzed, and a clinical examination, ultrasound and MRI of the hip joint were conducted. The child fell ill acutely with a body temperature of 37.3°C, vesicles appeared on the scalp, face, and torso. On the third day of the disease, the child complained of severe pain in the gluteal region, followed by pain around the hip joint and restricted leg movement.

Results: Upon examination, the patient exhibited a forced posture, restricted leg movement, and painful internal and external rotation. Ultrasound revealed signs of effusion in the hip joint and infiltrative changes in the surrounding soft tissues. MRI showed signs of sciatic neuropathy, swelling of the gluteal muscles, psoas major muscles, and sacroiliac joint. The diagnosis was “Varicella, typical form, moderate course, complicated by sciatic neuropathy.”

Conclusions: Varicella is a disease that can lead to serious complications, including peripheral neuropathy. In 2022, more than 648 thousand cases of varicella were registered in Russia, with the majority of them in children (95,6%). Not all countries have included the varicella vaccine in their national immunization calendar. In Russia, varicella vaccination is carried out according to epidemic indications. Currently, the Union of Pediatricians of Russia has developed the “Ideal Vaccination Calendar” which recommends regular varicella vaccine prophylaxis. In this presented clinical case, the child was not vaccinated prophylactically, resulting in a complication of the current disease. This highlights the need for varicella immunization to reduce the risk of severe forms of the disease, complications, and death. It is necessary to promote adherence to varicella vaccine prophylaxis and work towards its introduction in the general population, starting from the first year of life.

Keywords: Varicella vaccine, Complicated varicella, Varicella prevention, Sciatic neuropathy
PP24  Quality of Life of Children with Inflammatory Bowel Disease

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Aim: Inflammatory bowel diseases (IBD) - chronic, recurrent inflammation of the gastrointestinal tract. The estimation of quality of life (QoL) in children with IBD is significantly important as the course of the disease affects many aspects of life (not only physical health). The objective is to study the QoL of children with IBD.

Material and Method: We examined 116 children with IBD. I group included 59 patients with Crohn’s disease (CD), II group – 57 children with ulcerative colitis (UC). Median age of patients was 15.3 years. Control group (CG) included 124 apparently healthy children. The estimation of QOL was performed using various age-related blocks of PedsQL questionnaire forms for children on-scale from 0 to 100 pointson dimension: “physical functioning” (PF), “emotional functioning” (EF), “social functioning” (SF), “school functioning” (SchF), “general score” (GS). We also have performed correspondence analysis of PedsQL questionnaire general score and PUCAI, PCDAI.

Results: Children from the I group have rated their QoL significantly higher than in II group on SF dimension, (p<0,05). Children with IBD have estimated their QoL lower than children from CG in almost all dimensions: PF – 75 [61; 88] against 88 [75; 95] points (p<0,05); EF – 64 [60; 88] against 75 [63; 81] points (p=0,05); SchF – 67 [47; 78] against 79 [62; 89] points (p<0,05), and GS–71 [59; 83] against 84 [68; 91] pointsrespectively (p=0,000). The higher is the CD activity according to PCDAI the lover is the PF in children, there were no significant correlations in patients with UC(p<0,05).

Conclusions: The significant differences for marked decline of QoL in children with IBD in relation to healthy children in almost all dimensions were revealed. Though the QoL in patients with UC and CD differs only on PF dimension. The CD activity correlates well with the activity according to PedsQL questionnaire. This demonstrates the more severe course of CD in comparison to UC in children.

Keywords: Children, quality of life, Inflammatory bowel disease, Crohn’s disease, ulcerative colitis
PP25 Ultrasound Diagnosis of the Intestinal Wall in Inflammatory Bowel Disease in Children
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Aim: Assess the possibilities of ultrasound examination in the diagnosis of ulcerative colitis, Crohn’s disease to assess the effectiveness of therapy.

Material and Method: Examined 59 children (aged 7 to 17 years) with inflammatory bowel disease with clinical manifestations, as well as in remission using an ultrasound scanner Acuson Sequoia (Siemens) multi-frequency convex, linear transducers with scanning frequency ranges of 1-10 MHz. The study was performed on an empty stomach without contrast. The presence or absence of free fluid was assessed, thickening of the walls of the large intestine throughout (img.1), in places accessible by the location; presence or decreased differentiation of intestinal layers wall, its vascularization in the color flow mode, the volume of the lesion, omentum reaction; lymphadenopathy of the mesenteric lymph nodes. Verification was carried out according to colonoscopy and clinically.

Results: In patients with clinical manifestations of varying severity sonographic changes correlate with the severity of clinical patterns and disease activity. The main echographic signs of IBD: intestinal wall thickness more than 2.5 mm, uneven thickening intestinal walls were determined in 93% of the examined, a decrease in the differentiation of the layers of the intestinal wall (up to loss) – in 22% of the examined, pathological vascularization of the intestinal walls - 19% of the examined, inflammatory infiltration of the mesentery in 8% of the examined, the presence of free fluid in the abdominal cavity in 7% of the examined, an increase in the size of the mesenteric lymph nodes in 83% of the examined. Localization and severity of intestinal lesions correlated with the results of colonoscopy and MRI.

Conclusions: Ultrasound is a highly informative, non-invasive, highly mobile method for visualizing inflammatory bowel diseases, including in pediatric practice, which makes it possible to widely use the method for diagnosing the disease, clarifying the localization of the lesion, and monitoring the effectiveness of therapy.

Keywords: Ultrasound, inflammatory bowel disease, diagnosis of ulcerative colitis, Colon imaging, Crohn’s disease
Characteristics of Newborns with Perinatal Infection Born From Covid 19 Positive and Non-Covid Positive Mothers: A Cohort Study

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Aim: To determine clinical usefulness, we assessed sensitivity, specificity, positive and negative predictive values for mean IgG to assess the clinical effect of the IgG in perinatal infected infants born from Covid-infected and noninfected mothers.

Material and Method: The Covid-19 pandemic created the biggest challenge till today. The human immune system varies as a consequence of both heritable and nonheritable influences that lead to an inter-individual variation that has consequences on immunological condition and diseases. In the clinical laboratory, measurement of IgG is common practice for the screening of immune system dysfunction. The measurement of exact level of IgG can be complicated by specific ability of human immune system by placental transferring of maternal IgG, meaning that IgG levels may appear relatively high at birth due to the influence of maternal IgG. The measurement of immunoglobulin concentrations is a helpful method in many clinical conditions, in both cases, when we have normal or elevated levels of IgG to confirm or exclude the abnormalities. Congenital defects in one or more components of the humoral immune system may effect on a dysfunction of immune system. The clinical data of infants were compared and analyzed in LTD „Imedi Clinic“, Neolab(Ig G-ELISA).

Results: 72 neonates were admitted to LTD „Imedi Clinic“ from January 13.2022 up to day. The neonates were divided into two groups according to mothers’ conditions. Neonates born from the mothers with confirmed Covid-19 (25%) and from no infected mothers (47%). From both groups 63,8% of patients had to take treatment in intensive care department. According to blood culture pathological pathogens were found 4,2% and sterile were in 95,8%. High level of Ig G were found in 36% newborn, Neonates born from the mothers with confirmed Covid-19 and 59,5% born from no infected mothers.

Conclusions: Most patients with abnormal levels of IgG are easy to predict the severity of infection.

Keywords: Newborn, IgG, Perinatal Infection, Covid-19, Outcome
PP27

Diagnostic Value of Biomarkers for Vincristine-Induced Neuropathy in Children

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Aim:
To determine the correlation between biomarkers and VIPN in children with ALL.

Material and Method:
A single-center prospective cohort study was conducted involving 199 children (3 – 17 years old) with ALL receiving chemotherapy according to protocol ALL-MB 2015. All patients were genotyped for a single-nucleotide polymorphism in the CEP72 gene (rs924607) by polymerase chain reaction, and the levels of cytokines in blood plasma and cerebrospinal fluid were evaluated by multiparametric immunofluorescence analysis method.

Results:
VIPN was registered in 80.9% (161/199) of patients. The frequency of the risk allele (T) in this group was 87.5% (147/168), and children without VIPN - 12.5% (21/168). Information on the allele frequency is summarized in Table 1. It was found that the presence of the allele (T) is associated with VIPN and increases the risk of its development by 2.2 times (95% CI 1.27–3.81, p=0.004). The comparative analysis of cytokines levels and VIPN development revealed that patients with VIPN had a 2.3-fold increase in baseline IL-13 levels (p=0.042). An increase in cerebrospinal fluid levels of CXCL10/IP-10 and CXCL12/SDF-1 was detected at the completion of remission induction (p=0.005 and p=0.054, respectively), while there was a 3-fold decrease of VEGF-A (p=0.011) in the blood plasma during the consolidation phase and a 2-fold increase in HGF levels (p=0.029) during the maintenance phase of chemotherapy (Figure 1).

Comparative analysis of cytokines in children with ALL depending on the development of VIPN during chemotherapy stages (pg/mL)
Table 1. Comparative Analysis Of Allele Frequency In Children Depending On The Development Of VIPN

<table>
<thead>
<tr>
<th>The Fact Of VIPN</th>
<th>Allele T N=168</th>
<th>Allele C N=230</th>
<th>OR (Odds Ratio)</th>
<th>95% CI (Confidence Interval Of OR)</th>
<th>P-Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>VIPN «+»</td>
<td>147</td>
<td>175</td>
<td>2.20</td>
<td>1.27-3.81</td>
<td>0.004</td>
</tr>
<tr>
<td>VIPN «-»</td>
<td>21</td>
<td>55</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Conclusions: It has been established that the presence of the CEP72 risk allele (the T allele of rs924607) should be considered as a promising biomarker for predicting VIPN in children with ALL. Cytokines IL-13, VEGF-A, HGF in blood plasma and CXCL10/IP-10, CXCL12/SDF-1 in cerebrospinal fluid may serve as laboratory diagnostic criteria.

Keywords: vincristine, acute leukemia, neuropathy, CEP72, cytokines
Correlation Between Lactoferrin Concentration in Biological Substrates and Intestinal Microbiota Characteristics

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Aim: Composition of infantile intestinal microbiota is one of the most important health formation factors so it is important to study nutrition components which influence its characteristics. The purpose of our study is to study correlation between lactoferrin sufficiency in full-term and preterm infants (during the first month of life) and indigenous bacteria concentration in their microbiota.

Material and Method: lactoferrin sufficiency in human milk and infantile fecal masses was measured using ELISA test and quantitative bacteriology analysis of intestinal microbiota. 42 full-term and 15 preterm infants were studied twice – at 3rd day of life and 30th day of life.

Results: women after premature delivery and birth in time had no significant differences in human milk lactoferrin sufficiency, and it was, respectively, 7,6 ±3,6 / 7,06 ± 5,2 mg/mL n 2,09 ± 1,01 / 2,17 ± 1,9 mg/mL. Lactoferrin concentration decrease during the first lactation month is statistically significant (p<0,05). Lactoferrin concentration in faeces increased from 977,7±260,3 to 3040,9±886,2 ug/mL (for full-term infants) and 1848,5±1406,5 to 5257,9 ±1798,6 ug/mL (for preterm infants). Linear positive correlation between lactoferrin concentration in human milk and infantile faeces after the first month of life was found out (R=0,44, p=0,030). Bifidobacteria concentration in full-term and preterm infants’ faeces almost doubled. Positive correlations between lactobacilli and bifidobacteria levels in intestinal microbiota and fecal lactoferrin concentration in preterm infants at the 3rd day of life (R=0,62; p=0,02 n R=0,68; p=0,015 respectively).

Conclusions: changes of lactoferrin concentration in infants’ faeces is contrary to lactoferrin concentration in human milk. Found out correlations between fecal lactoferrin concentration and indigenous bacteria levels show that lactoferrin plays a significant role in infantile microbiota formation.

Keywords: infant, lactoferrin, human milk, microbiota, gut
The Early Formation of Food Sensitization

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Aim: The rapid increase in the prevalence of allergic diseases indicates the necessity to detect modifiable risk factors in the early ontogenesis period. Such factors can be dietary preferences of mothers during pregnancy and lactation, features of baby’s feeding. The aim of our study was to analyze the nutritional characteristics in the mother-child dyad and nature of infants’ food sensitization, which in the future will allow us to create management algorithms of feeding for children at risk of atopy.

Material and Method: 50 mother-child dyads were observed. The mother-child diet and levels of slgE in infants (ISAC IMMUNOCAP) were studied. The data were processed in the Statistica 11 program.

Results: 72% of children had a family history of atopy. The average age of infants was 2.0 [1.0–3.0] months. Various skin allergy symptoms were identified in 100%. A significant number of pregnant women had different food preferences: 52% - cow’s milk; 40% - eggs; 34% - fish; 30% - nuts; 28% - seafood; 6% - soy; 4% - peanuts. During lactation: 40% - cow's milk; 24% - eggs; 10% - fish. By the 3rd month of life, 48% of infants were exclusively breastfed. The levels of slgE were increased to 8 antigens of food allergens in 10% of children: cow’s milk (Bos d 6, Bos d 8), egg white (Gal d 1, Gal d 2, Gal d 3), soy (Gly m6), shrimp (Pen m 1, Pen m 4). Moderate/high levels of slgE were also registered. The infants’ diet did not include egg white, shrimp, soy. Probably, these antigens penetrated into the body with breast milk or through skin.

Conclusions: The risk of food sensitization in infants at risk of atopy remains high and often requires to decrease levels of slgE. Science based recommendations for the mother-child dyad will help to reduce risks of clinically significant sensitization.

Keywords: infants, IgE, food allergy, nutrition
Gut Dysbiosis and Sensitization In Infants at Risk of Atopy

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Aim: Early dysontogenesis of the gut microbiota in children with atopic heredity has not been studied enough. The aim of our study was to research associations between the initial changes of gut microbiota and levels of slgE to food antigens in children with allergy manifestation.

Material and Method: We observed 50 full-term infants 6-12 months old with primary allergy symptoms. The composition of gut microbiota (culture method) and molecular sensitization profiles (ISAC IMMUNOCAP) of the researched group were studied simultaneously. The data were processed in the Statistica 11 program.

Results: 72% of children had a family history of atopy, often in combination with risk factors for gut microbiota dysontogenesis: cesarean section - 24%, use of antibiotics - 50%, formula fed based on cow’s milk - 52%. First complementary foods were introduced at the age of 4-6 months old to 54% of children. Various deviations in the composition of gut microbiota were found in 94%. Reduced amount of Bifidobacterium spp. was noted in 23%. Increased growth was registered: Clostridium spp. – in 77%, Proteus spp. – 49%, Staphylococcus aureus – 31%, Klebsiella spp. – 46%, Citrobacter spp. – 57%. Sensitization was detected in 36%, slgE were increased to 10 antigens of food allergens (cow’s milk/meat, egg, peanut, wheat, soy, kiwi). The analysis revealed positive correlations between: Clostridium spp. and total IgE, R = 0.31; Proteus spp., Citrobacter spp., Raoutella spp. with slgE to egg antigens (Gal d 1, Gal d 2, Gal d 2), R = 0.49; R = 0.34; R = 0.52, respectively.

Conclusions: The increased level of some representatives of Clostridiaceae, Enterobacteriaceae, Enterobacter in the microbiota of an infant can be considered as a prognostic criterion for risks of atopy formation. Individual differences in the microbiota of an infant with initial manifestations of allergy are the basis for the development of methods for correcting gut dysbiosis.

Keywords: infants, IgE, food allergy, microbiota
Effect of Probiotic Supplementation (Lactobacillus Reuteri) in Patients with Cystic Fibrosis

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Aim:
Probiotics have gained significant attention in recent years due to their potential therapeutic applications in various health conditions. They are known to modulate the immune system, improve intestinal barrier function, and exert anti-inflammatory effects. Probiotics are proven to reduce intestinal inflammation in children with cystic fibrosis (CF) and may reduce the duration of respiratory and gastrointestinal diseases. We want to determine the effects of the probiotic Lactobacillus reuteri on symptoms of respiratory and gastrointestinal diseases in CF.

Material and Method:
38 patients with CF (2-24y, mean age 9.6±4.2), received probiotic Lactobacillus reuteri for 6 months. Main outcome parameters were forced expiratory volume (FEV1), FVC (forced vital capacity) and change in anthropometric parameters.

Results:
In the study were measured FEV1 and FVC in CF patients before and after they received probiotic Lactobacillus reuteri. We found significant difference for FEV1 (p<0.05) and FVC (p<0.05), in both children and adolescents. We didn’t find significant difference for body weight, body height and for BMI, but the patients were heavier after 6 months.

Conclusions:
Probiotics may delay respiratory impairment and gastrointestinal inflammation. However, while the potential benefits of probiotics in CF are encouraging, further research is necessary to fully understand their mechanisms of action, optimal dosages, and long-term effects.

Keywords:
Cystic Fibrosis, Probiotics
Kabuki-Syndrome, Type 1: Renal and Urinary Tract Findings in 13 Russian Patients

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Aim: Kabuki syndrome, type 1 - is a rare inherited disease, characterized by short stature, microcephaly, distinctive facial features, intellectual disability, hearing loss, cardiovascular, gastrointestinal, endocrine, kidneys anomalies and caused by heterozygous mutations in the KMT2D gene.

Material and Method: We followed 13 patients with Kabuki syndrome, type1 aged 1 to 11 years. The median age at diagnosis was 6 years [1;11]. Pathogenic nucleotide variants in KMT2D was detected in heterozygous state in all cases. Growth retardation, facial features, look like kabuki-make up, skeletal anomaly, mental retardation and developmental delay, endocrine problem was in all cases, immunodeficiency - in 75 %.

Results: Renal and urinary tract findings was detected 10 cases (83%) and include horseshoe kidney (3), renal dystopia (3), renal hypoplasia (10), renal segment hypoplasia (1), renal duplication (1), L-shaped kidney and cyst dysplasia (1), renal cysts (1), additional renal vessels (3), hydronephrosis (1), ureteral duplication (2). Five cases (50%) with renal and urinary tract malformations was diagnosed hyperechogenic pyramid syndrome, in one case - medullar nephrocalcinosis, 4 patients of this group had chronic pyelonephritis.

Conclusions: Our results underscore the high prevalence renal and urinary tract findings in Kabuki syndrome, type1 patients. Notably, hyperechogenic pyramid syndrome and nephrocalcinosis was observed to be a common and prognostically unfavorable symptom in Kabuki syndrome, type1

Keywords: Kabuki syndrome, KMT2D, renal and urinary tract, nephrocalcinosis
Rett Syndrome: Clinical Characteristics of 45 Russian Patients

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Aim: Rett syndrome is one of the most common causes of intellectual disability in females, and it leads to severe cognitive and physical impairments. The majority of Rett syndrome cases are associated with genetic variants in the MECP2 gene. The aim of our study - to identify the most common symptoms of the disease and to investigate the association between disease onset and the genetic variant type in the MECP2 gene among patients with Rett syndrome in our study

Material and Method: The study included 45 female patients aged 1 to 13 years with molecularly confirmed Rett syndrome. The missense variants were present in 44% of the patient cohort (n=20), nonsense variants were detected in 29% of the cases (n=13). Additionally, frame-shift variants were identified in 27% of the cases (n=12).

Results: After normal development, first symptoms occurred at first year of life. Onset of the disease in patients with nonsense mutations and frame-shift mutations occurred on average in 10 months, IQR - 4;24. Patients with missense mutations had a median age of onset in 11 months, IQR - 6;24. All patients with Rett syndrome showed delayed psychomotor development, autistic behavior, loss of acquired skills, stereotypical hand movements, loss of purposeful use of the hands, truncal ataxia, microcephaly and seizures. Eye contact with parents preserved in 37% cases (n=17), 35% patients (n=16) had separate sounds and words. Scoliosis was identified in 49% of cases (n=22).

Conclusions: All patients included in this study showed delayed psychomotor development, stereotypies, loss of acquired skills, microcephaly and seizures. No correlation identified between the type of genetic variant in the MECP2 gene and the clinical presentation of the patient.

Keywords: Rett syndrome, MECP2 gene, delayed psychomotor development, stereotypies
A Case of Sertoli-Leydig Cell Tumor of the Ovary in an Adolescent

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Aim: A case of Sertoli-Leydig cell tumor of the ovary in an adolescent. Sertoli Leydig cell tumors are rare sex-cord stromal tumors of the ovary with a good prognosis and low recurrence rate.

Material and Method:

Results: A 12 years adolescent girl was admitted to the Pediatric Surgery Department in June 2021 with progressive abdominal distension during the last five months. She was in Tanner’s stage 3 at clinical examination, with a very distended abdomen and obesity (97th percentile). Abdominopelvic CT and MRI scans revealed a mixt tumor, with cystic and tissular components, arising from the right ovary, peritoneal carcinomatosis and massive ascites. Chest and brain CT showed no distant metastasis. Serum markers CA 125, beta-HCG and AFP were assessed preoperatively. CA 125 was high (683.3 U/ml; normal range 0-35U/ml) and AFP showed a moderate increase (68.63 U/ml; normal range 0.5-8 U/ml). Beta-HCG levels were normal. Endocrinologic studies were performed, with testosterone, FSH, LH, estradiol, thyroid function within normal range. The case was classified as a stage IIIC tumor, based on FIGO staging system for ovarian tumors. Surgery was performed and consisted of tumor resection with right salpingectomy, omentectomy and evacuation of about 10 liters of transudate ascites fluid. Histopathology examination completed with immunohistochemical analysis were compatible with the diagnosis of a Sertoli-Leydig cell tumor, moderately differentiated. Adjuvant chemotherapy was initiated in the Oncopediatrics Department, according to MAKEI 05 protocol and 6 cycles of PEI regimen were administered. Serum marker CA 125 was monitored before every cycle of chemotherapy and showed a constant decrease and normalization. Follow-up imaging studies and serum tumor markers were normal at periodical evaluations after the completion of chemotherapy.

Conclusions: We presented a rare case of ovarian Sertoli-Leydig cell tumor in an adolescent, diagnosed in an advanced stage, who is in complete remission after a 24-month follow-up period.

Keywords: Sertoli-Leydig, Tumor, Ovary, Adolescent
Gestational Trophoblastic Neoplasia in Adolescent – Case Report

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Aim: Gestational trophoblastic neoplasia in adolescent. Gestational trophoblastic disease results from the abnormal proliferation of trophoblastic tissue and encompasses a spectrum of benign and malignant gestational tumors.

Material and Method:

Results: We present the case of a 14 years old adolescent-girl, with a history of a complete hydatidiform mole evacuated in 2021. She was admitted to the Gynecology Department in October 2022 for metrorrhagia and pelvic pain. Beta-HCG value at admission was greater than 200000 U/ml. The abdominopelvic MRI revealed a uterine tumor mass invading the left adnexa; no distant metastases were found when completing imaging studies. Surgery was performed and consisted of tumor excision, left salpingectomy and partial omentectomy. Histopathological analysis was highly suggestive for the diagnosis of choriocarcinoma. The disease was classified as stage II, according to FIGO staging system for gestational trophoblastic neoplasia, with a prognostic score index totalizing 10 points (based on the high preoperative beta-HCG value, tumor dimensions and the interval from index pregnancy), which denoted a high-risk score. Multiagent chemotherapy was initiated, using the EMA-CO regimen. Biological response (normalization of beta-HCG) was noted after 3 cycles of chemotherapy, which was continued for another two cycles. Imaging studies performed after completing chemotherapy showed a complete response and Beta-HCG values remained within the normal range at periodical evaluations. The patient was initiated on combined oral contraceptives for one year after the end of chemotherapy to prevent a potential pregnancy.

Conclusions: Although choriocarcinoma is known to have the worst prognostic among gestational trophoblastic neoplasia entities, our case was diagnosed in a localized stage and showed a complete response to the specific chemotherapy regimen.

Keywords: Neoplasia, Choriocarcinoma, Adolescent, Uterine, Metrorrhagia
VDR Gene is the Formation of Clinical Manifestations and Course in Asthma

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Aim: The effect of low vitamin D levels on lung function, the frequency of exacerbations of asthma and the use of medications continues to be studied. The contribution of exogenous factors and genetic regulation, including those involving genetic variants (GV) of the VDR gene, are not clear. The aim of the study was to carry out an associative search between genetic variants (c.1206T>C, c.1175-9G>T, c.152T>C, c.1174+283G>A) of the VDR gene with clinically significant manifestations of the disease and complications.

Material and Method: Polymorphic variants of the VDR gene were identified in 160 patients (11.7 ±3.9 years) with asthma. The analysis of GV of the VDR gene was carried out by restriction analysis (RFLP).

Results: The course of asthma and rhinitis was more often observed in patients with AA and GA genotypes of BsmII polymorphism (c.1174+283G>A), TT and CT genotypes of the GV c.1206T>C(A>G) TaqI, T and TC genotypes of the c.152T>C FokI of the VDR gene. The carriers of the CC genotype of the c.1206T>C(A>G) TaqI were less likely to have a severe course of asthma and did not need biological therapy (p<0.05). The carriers of the TT genotype of the genetic variant c.1206T>C(A>G) TaqI, carriers of the genotype CC of the c.152T>C FokI and carriers of genotypes AA and GA of the BsmI polymorphism (c.1174+283G>A) more often implemented all three components of the “atopic march” (p<0.05).

Conclusions: The contribution of the studied genetic variants: c.1206T>C(A>G) TaqI, c.152T>C FokI, BsmI (c.1174+283G>A) of the VDR gene in the formation of course in asthma was confirmed.

Keywords: VDR gene, asthma, complications, manifestations, genetic variants
Aim: Targeted therapy is currently the most effective approach to the treatment of cystic fibrosis (CF). The CFTR modulator Lumacaftor/ivacaftor, which is a combination of two small molecule drugs aimed at restoring the function of the chloride (CFTR) channel in CF, is approved for the treatment of patients with the F508del/F508del genotype. The aim of the study was to analyze the effectiveness of therapy with the CFTR modulator lumacaftor/ivacaftor in children and adolescents with cystic fibrosis after 12 months of treatment.

Material and Method: The study included data from 135 children over 6 years of age who received the CFTR modulator lumacaftor/ivacaftor, presented in the Register of Patients with Cystic Fibrosis of the Russian Federation for 2021-2022. Anthropometric parameters, the results of a sweat test, and parameters of respiratory function (RF) were analyzed before the start of therapy and after 12 months of therapy.

Results: According to the registry data, after 12 months of therapy in patients treated with lumacaftor/ivacaftor, there was an increase in body weight from Me (Q1-Q3) 36.2 (27.5-46.1) to 40.5 (30.8-50.0) kg, p<0.001, height from 148.8 (134.8-159.3) to 155.0 (137.9-162.5) cm, p<0.001, body mass index (BMI) from 16, 4 (14.7-17.8) to 17.4 (15.9-19.1) kg/m², p<0.001. Sweat conductivity decreased from 113.0 (104.0-122.0) mmol/l NaCl at the start of therapy to 90.0 (75.5-103.0) mmol/l NaCl after 12 months, p<0.001. The respiratory function parameters did not undergo statistically significant changes during therapy.

Conclusions: In children with CF older than 6 years, during 12 months of lumacaftor/ivacaftor therapy, there were statistically significant positive changes in weight, height, BMI and sweat test.

Keywords: CFTR-modulator, cystic fibrosis, children, effectiveness, 12-month
PP39  Characteristics of Patients with Primary Ciliary Dyskinesia

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Aim: The purpose of this scientific study is to analyze the clinical, laboratory and instrumental characteristics of patients suffering from primary ciliary dyskinesia (PCD).

Material and Method: The study collected and analyzed data from a group of 90 patients, which included 22 adults and 68 children treated in various medical centers. The study used methods such as collection of anamnesis, spirometry, microbiological examination of respiratory tract sputum, video microscopic analysis of the functional activity of the ciliated epithelium of the nasal mucosa, transmission electron microscopy of the ciliated epithelium, DNA diagnostics.

Results: A total of 90 patients participated in the study, including 22 adults and 68 children. Kartagener’s syndrome was detected in 23 patients, including 6 adults. The anamnesis of patients by recurrent bronchitis (68 adults and 46 children), pneumonia (21 adults and 48 children), otitis media (44 adults and 33 children) were dominated. Hearing loss was noted in 5 adults and 15 children. Lung function was reduced as in adults, and in children, but a significant decrease was noted in adult patients. In microbiological crops, Pseudomonas aeruginosa prevails - 21.3% (intermittent inoculation - in 13.2%, chronic inoculation - in 9%), which has high resistance to antibacterial drugs (ABD). Light microscopy of the ciliated epithelium showed in 12 patients (3 adults and 9 children), motor activity of cilia was not registered. The complete absence of external and internal dynein handles (51%) were more common in transmission electron microscopy. Molecular genetic study in 55 patients (16 adults and 38 children) was performed.

Conclusions: it was found that the problems of timely diagnosis, effective management and adequate treatment remain relevant for Russian patients. The creation of a PCD patient registry will address these issues and contribute to the development of genetic diagnostic algorithms, the determination of optimal antibiotic therapy strategies, the improvement of overall patient care.

Keywords: ciliopathy, axonema, electron microscopy, DNA diagnostics, primary ciliary dyskinesia (PCD)
Reference Intervals of Acylcarnitines for Newborns in North Asian

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Aim: Establish reference intervals (RI) for 13 acylcarnitines (AC) in blood of healthy newborns from North Asian and assess their differences from corresponding reference values for neonates from other populations.

Material and Method: A cross-sectional study of 5321 healthy newborns was conducted. Dried blood spots (DBS) collected from filtered paper were used to analyze and measure derivatized LC-MS/MS.


Conclusions: Reference values of long-chain acylcarnitines (C10, C12, C14, C16, C18), medium-chain acylcarnitines (C6, C8), short-chain acylcarnitines (C2, C3, C4, C5) in DBS of newborns are multidirectional in different countries of the world.

Keywords: acylcarnitines, reference intervals, health newborns, North Asian
Kallmann Syndrome: An Unusual Presentation with Mirror Hands

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Aim: Kallmann syndrome is a rare genetic disorder characterized by hyposmia/anosmia and congenital hypogonadotropic hypogonadism. It is most commonly caused by mutations in the ANOS1 gene in the Xp22.3 region of the X chromosome. Undescended testis is usually the first finding in boys. They present with the short stature, absence of axillary/pubic hair, no increase in testicular volume and micropenis in pubertal period. Girls present with absence of breast development and axillary/pubic hair growth, primary amenorrhea. Moreover, renal agenesis and bimanual synkinesis can be seen rarely in some patients.

Material and Method: A 15-year-old male patient has visited to the General Pediatrics outpatient clinic of our hospital with the complaint of mirror movement in the hands (bimanual synkinesia). Mirror movement was noticed when he started writing at the age of 7. On physical examination, the patient's Height for Age Z score was under -3 standard deviation. The testicular volume was 2 milliliter and the testicles were bilaterally undescended. Penis size was small for his age. When hand movements were evaluated, it was observed that any movement made with one hand was made involuntarily simultaneously with the other hand. In addition, he has impaired sense of smell.

Results: He was diagnosed with Kallmann syndrome after further investigations.

Conclusions: The detailed anamnesis and physical examination of the patient enabled the diagnosis. It is necessary to evaluate the growth and puberty of all children who apply to the outpatient clinic. In children with delayed puberty findings, Kallmann syndrome should be considered, and the presence of hyposmia/anosmia should be questioned. With this case, we emphasize the importance of full physical examination together with the evaluation of the complaint in outpatient clinic applications.

Keywords: kallmann, hypogonadotropic hypogonadism, bimanual synkinesia
Chronic Kidney Disease in TARS2-Related Mitochondrial Disease – A Case Report

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**Aim:** Mitochondrial disease caused by mutation in TARS2 gene is rare. To date, only four cases were reported in the literature presenting primarily with mitochondrial encephalomyopathy. Renal involvement has not been described in this particular mitochondrial disorder so far.

**Material and Method:** We report four years old boy with developmental delay, seizures, hypotonia, and terminal chronic kidney disease (CKD).

**Results:** He is the first child of non-consanguineous parents, from uneventful pregnancy. Birth weight was 4.2 kg and perinatal period was normal. In early infancy poor weight gain was observed. Thorough diagnostic evaluation was initiated at six months of age due to failure to thrive, hypotonia, motor delay and recurrent bronchiolitis. Laboratory findings included hyperlactatemia and hypokalemia without other electrolyte abnormalities in serum and urine. Estimated glomerular filtration rate were normal at the time. Ultrasound showed hyperechoic kidneys. After discharge he was lost to follow-up until two years when he was readmitted with elevated creatinine level (111 umol/l), reduced glomerular filtration rate (30 ml/min), anemia, hyperkalemia, hypomagnesemia, hyponatremia, low level of parathyroid hormone and urine abnormalities consistent with Fanconi syndrome. Two heterozygous pathogenic variants in TARS2 gene were detected by WES (c.1298T>G (p.Phe438Cys) missense variant derived from the mother and the c.1931A>T (p.Asp644Val) missense variant derived from the father). Since 2.5 years of age he had occasional seizures while brain MRI examination detected pathologically increased signal of the brain parenchyma, primarily in the posterior aspects of the pons and white matter of the cerebellum. Treatment included dietary modification, erythropoietin, calcitriol, “mitochondrial cocktail” of supplements and vitamins, antiseizure medication. Currently, he has 4 years, body weight of only 11 kg, severe motor and verbal delay and fifth stage of CKD requiring preparation for renal replacement therapy.

**Conclusions:** This is the first reported case of a patient with TARS2 mutations and chronic kidney disease as the key clinical feature.

**Keywords:** mitochondrial disease, TARS2 mutation, chronic kidney disease
Management of Hypersensitivity Reaction in a Patient with Hunter Syndrome

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Aim: Patients with mucopolysaccharidosis type II (MPS II) require lifelong enzyme replacement therapy (ERT) using recombinant forms of iduronate-2-sulfatase (idursulfase or idursulfase beta) to compensate for the deficiency of the endogenous enzyme. There is a significant risk of drug hypersensitivity reactions (DHR) associated with ERT, necessitating the development of new therapeutic approaches that minimize the risk for life-threatening side effects.

Material and Method: This is a case report of a 9-year-old boy diagnosed with MPS II at the age of 2.

Results: From the age of 2 the boy was administrated with weekly intravenous idursulfase 0.5 mg/kg. At the age of 5, he experienced urticaria on his upper limbs for the first time during infusion. Subsequently, every infusion was accompanied by urticaria and sometimes dyspnea. He was treated with intravenous antihistamines and corticosteroids. At the age of 6 he was administered oral corticosteroid for one month, ERT was discontinued. Upon resuming the therapy, he experienced DHR again. His ERT was switched to idursulfase beta, but the side effects persisted despite premedication with corticosteroids and antihistamines, decreased infusion rate. Even between infusions, the urticaria persisted, ERT was discontinued. Due to the critical necessity of ERT he was administered with systemic therapy: daily cyclosporine - 2.5 mg/kg and omalizumab at a monthly dosage of 300 mg subcutaneous for a period of 6 months. At the age of 7 ERT was reintroduced, but DHR occurred once again during infusion. Subsequently, a personalized desensitization protocol for idursulfase beta was developed. Since that time the child has been receiving ERT in full dosage according to a personalized regimen weekly along with omalizumab monthly without experiencing any adverse effects.

Conclusions: The development of a personalized protocol has effectively prevented DHR during ERT in this patient with MPS II.

Keywords: mucopolysaccharidosis type II, hypersensitivity reaction, Hunter syndrome, iduronate-2-sulfatase, personalized desensitization
Symptoms of Anxiety and Stress in Psoriatic Children.

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Aim: Investigating the effect of Psoriasis on the occurrence of symptoms expressing stress and anxiety, compared to healthy children.

Material and Method: 218 children who were diagnosed with psoriasis vulgaris from January 2021 - March 2023 participated in this study. The control group was also included in this study, where 250 healthy children without psoriasis vulgaris were selected. The champion is chosen at random. In both groups, the control group as well as the psoriasis group, the survey was anonymous. The survey was administered for symptoms of stress, anxiety (DAAS survey). The tau-b Kendall correlation coefficient was used in the statistical analysis between two proportional variables; Chi-square test to analyze proportional data; binary and multiple logistic regression to see the relationship of a dependent variable to several independent ones.

Results: Symptoms of stress (≥15 points) and anxiety (≥8 points) are statistically more frequent in children with psoriasis compared to the control group (P < 0.001). Psoriatic children have a risk OR = 2:42 compared to children without psoriasis to have anxiety attacks. For stress phenomena in psoriatic children, there is an OR=3:53 risk of occurring compared to children without psoriasis. The severity of psoriasis has a significant relationship with the severity of stress symptoms (P < 0.001), as well as with that of anxiety. There was no significant difference between anxiety and stress symptoms in female and male children (p>0.05). There is no significant difference between the symptoms of anxiety and stress in children who lived in rural areas and those in urban areas (p>0.05). The children’s economic income did not affect the appearance of symptoms of anxiety and stress (p> 0.05).

Conclusions: Conclusions: Psoriasis in children gives psychological complications and can negatively affect the quality of life of children who suffer from it. Psychological care is needed for children suffering from psoriasis.

Keywords: Psychological care; psoriasis; children; stress; anxiety;
The Case of Lung Affection in 8-Year Old Girl with Diastematomyelia

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Aim: To demonstrate some difficulties in lung involvement diagnoses in patients with Diastematomyelia

Material and Method: case report

Results: Diastematomyelia (D.) (split cord malformation) is a type of spinal dysraphism (spina bifida occulta) - a congenital abnormality with longitudinal split in the spinal cord. D. features may include scoliosis with lung dysfunction and sometimes it may be difficult to differentiate if symptoms caused by lung disease or by chest deformation. The 8 year old girl with severe chest deformation (scoliosis, thoracic spine lordosis, lumbar spine kyphosis), occasional cough with breathlessness and wheezing caused by physical activity from 4 y.o. The girl’s father had asthma. The girl was diagnosed with acute bronchitis on repeated occasions. Treatment: nebulised ipratropium bromide + fenoterol and budesonide with partial effect. Spirometry: FVC and other parameters were significant decreased, salbutamol test – negative. Chest X-ray revealed vertebrae malformation, CT-scan of chest - multiple thoracic spine malformation, picture similar to tracheobronchomalacia plus significant left main bronchus narrowing causing by chest deformation, pulmonary Consolidation focus in inferior lobe of left lung. MRI of spine and spinal cord revealed multiple malformations of chest vertebrae and spinal cord (D. I and II type). Trachebroncoscopy - trachea and bronchi deformation and diffuse endobronchitis at the left side. The girl underwent surgery by neurosurgeon with correction of D. of Th2-Th5 and liberation of spinal cord.

Conclusions: This is a rare case of D. in child. It demonstrates some difficulties in diagnoses lung involvement in patients with chest deformations.

Keywords: children, Diastematomyelia, rare diseases, lung, chest deformation
Schinzel-Giedion Syndrome – A Rare Cause of Psychomotor Delay and Refractory Seizures

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Aim: Introduction: Schinzel-Giedion syndrome is a rare genetic syndrome characterized by severe developmental delay, facial dysmorphism, seizures, and multiple congenital malformations. Affected children have increased risk of developing neuroepithelial brain tumors.

Material and Method: Case report: We are reporting a case of a 15 months-old boy with profound psychomotor delay, distinctive facial features (prominent forehead, deep set eyes, low set ears, midface retraction) and drug-resistant seizures.

Results: He is the first child from a pregnancy complicated by gestational diabetes. The delivery was on term, the newborn experienced moderate perinatal asphyxia and soon after birth he was referred to our hospital due to cyanosis and respiratory distress. On admission, facial dysmorphism and moderate hypotonia were prominent. He experienced frequent seizures accompanied by apnea, which were treated with phenobarbital and levetiracetam. Extensive metabolic and genetic investigations, including karyotyping, microdeletion screening, and chromosomal microarray analysis, yielded normal results. Further examination revealed associated congenital heart defect (atrial septal defect) and hydronephrosis. Serial brain MRI scans showed progressive brain atrophy. During infancy he had multiple hospital admissions due to daily seizures (mostly focal, then from the seventh months of life, combined with infantile spasms). Despite attempts with various anti-seizure medications, corticosteroids, and ketogenic diet, the effects on seizure reduction were limited. The child exhibited severe developmental delay, with hearing and vision impairment. At the age of 12 months, whole exome sequencing (WES) was performed, identifying a pathogenic variant in SETBP1 gene, which further confirmed the diagnosis of Schinzel-Giedion syndrome.

Conclusions: Implementation of new genetic methods (WES) enabled determination of genetic basis for numerous rare diseases and syndromes, crucial for providing adequate information to parents regarding the nature and expected course of the disease. Given the association of this syndrome with an increased risk of tumors, accurate diagnosis enables regular monitoring, facilitating early detection and treatment of potential complications or associated disorders.

Keywords: Schinzel-Giedion syndrome, developmental delay, seizures, whole exome sequencing, facial dysmorphism
Clinical Case of Early Maple Syrup Urine Disease Detection in an Infant

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Aim: Since 1 January 2023 neonatal screening in Russia is extended from 5 to 36 disorders. The purpose of our work is to show a clinical case of early leucinosis detection in infants.

Material and Method: Neonatal screening for 36 disorders was conducted at the first day of life. Moreover, detection of ammonium concentration level in blood and DNA testing were conducted.

Results: A boy was born in a family burdened with genetic anamnesis: intermarriage, first two pregnancies with acrania. The relevant pregnancy was burdened with anemia, hypertonia, CS at the 41st w GA. BW 4130 g. Apgar score was 8/9. The infant received mixed feeding. In the first days of life there was bad sucking, jaundice. After discharge on the 6th day of life - apathy, salivation, convulsion, hyponatremia, hypoglycemia, lactic acidosis. At the hospitalization day following neonatal screening results were obtained: leucine - 455.0 µmol/l (N 55-266 µmol/l), valine - 324.0 µmol/l (N 44-247 µmol/l). Enteral nutrition was suspended, phenobarbital and infusion therapy were assigned. Repeated screening results at the 9th day were as follows: leucine - 3036.423 µmol/l, valine - 763.747 µmol/l. Increased levels of ammonium in the blood were noted (from 94 to 238 mmol/l), sodium benzoate was included in the therapy. From the 10th day of life, enteral nutrition was introduced with a formula without leucine, isoleucine, valine. Subsequently, to correct protein deficiency, milk formula 1 was introduced in a minimum volume until protein availability of 1.6 g / kg / day was achieved, partial parenteral nutrition was carried out, the child also received thiamine and levocarnitine. The result of DNA diagnostics is a homozygous mutation in the BCKDHA gene, nonsense variant chr19-41423071-C-T:NM_000709.4: c.1069>T p.(gln357Term), leucinosis type 1. Following treatment of the condition with positive dynamics.

Conclusions: The case demonstrates the importance of early detection of a severe hereditary disease.

Keywords: newborn, rare diseases, Maple syrup urine disease, neonatal screening
Co-Occurring Hearing Loss and Other ENT Problems in Children with Autism Spectrum Disorder

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Aim: identifying of ENT problems and hearing loss in patients with ASD for timely initiation of additional intervention.

Material and Method: 79 children aged 18 months to 17 years (M=10±4.679) with an established diagnosis of “autism spectrum disorder” underwent a complete audiological examination, examination by an otorhinolaryngologist as part of the research work. The diagnosis was made by a psychiatrist and verified using Autism Diagnostic Interview-Revised (ADI-R). Examination methods: otoscopy, pharyngoscopy, anterior rhinoscopy, nasopharyngeal endoscopy, registration of otoacoustic emission, tympanometry, registration of short-term auditory evoked potentials, auditory steady state response (ASSR-test).

Results: ENT disorders were revealed, such as adenoid hypertrophy was detected in 44 (55%), chronic otitis - in 8 (10%), acute adenoiditis - in 4 (5%), chronic tonsillitis - in 4 (5%), hearing loss was detected in 3 (4%) cases, 1 (1.3%) had a cleft of the soft palate. At the moment, the study is ongoing.

Conclusions: The high prevalence of ENT pathology and cases of hearing loss in children with ASD has been shown. It is important to provide special examination with objective methods to identify hearing loss and ENT disorders.

Keywords: ENT pathology, hearing loss, ASD, autism spectrum disorder
PP49  Some Features of Cognitive Function Formation in Children with WAGR Syndrome

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Aim: To study the features of cognitive function formation in children with WAGR syndrome. WAGR syndrome is a rare syndrome characterized by Wilms tumor (W), aniridia (A), genitourinary anomalies (G), and intellectual disability (R). This rare condition is caused by changes in contiguous genes, characterized by a de novo deletion of chromosome 11p13, including the WT1 gene (Wilms tumor and genitourinary anomalies) and the PAX6 gene (aniridia). The BDN gene is considered to be involved in the development of intellectual disability and obesity in WAGR syndrome, and the SLC1A2 and PRRG4 genes are also considered.

Material and Method: The study involved Russian children with a genetically verified diagnosis of WAGR syndrome. The WISC-IV (Wechsler Intelligence Scale for Children, 4th edition) was used for intelligence assessment, as well as neuropsychological testing using the A. Luria method.

Results: Given the extreme rarity of the pathology, 4 children with WAGR syndrome were examined, aged 6, 7, 14 and 16 years old, including 2 girls. Two of the examined children had intellectual disability, while the other two did not. In children with preserved intelligence, non-verbal intelligence scores averaged 106 (average range); verbal intelligence scores averaged 114 (average range); and overall intelligence scores averaged 109 (average range). In cases of intellectual disability, verbal and non-verbal intelligence scores were below 50 (severe degree of intellectual disability). According to additional neuropsychological examination data, children with normal intelligence showed a decrease in neurodynamic components of mental activity: slowed pace of work, fluctuations in attention, increased fatigue, inertia, and emotional instability.

Conclusions: In our study, it was found that children with WAGR syndrome can have normal variants of cognitive function development. In children with normal intelligence, there is no significant difference between verbal and non-verbal intelligence.

Keywords: Children, WAGR syndrome, WISC-IV, neuropsychological testing
PP50  Dysen’s Dystrophy and the Possibility of Early Detection of Heart Damage

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Aim: Dysen’s dystrophy (Duchenne) is a sex-linked, inherited muscle disease caused by a mutation in the dystrophin Xp21 gene. This is a progressive muscle disease, which is caused by a complete lack of the dystrophin protein. As a primarily muscular disease, it affects the striated muscles and the heart muscle. Almost all patients with Duchenne dystrophy develop cardiomyopathy in their third decade of life at the latest. Early detection of cardiomyopathy associated with muscular dystrophy is important, as cardioprotective therapy can slow negative cardiac remodeling and alleviate symptoms of heart failure in these patients.

Dysen’s dystrophy and the possibility of early detection of heart damage

Material And Method: /Case report

Results: A boy aged 10 years and 8 months with a molecularly-genetically confirmed diagnosis of Duchenne’s dystrophy-deletion of the muscle promoter Dp427m, was examined cardiologically. A clinical cardiology examination showed that the heart was normal. An electrocardiogram showed sinus rhythm, normal heart rate. The laboratory determined the value of NT pro BNP (190.6 pg/ml). Standard two-dimensional transthoracic echocardiography showed normal dimensions of the left ventricle, as well as its contractility (LVD 47, LVS 30, FS 0.36, EF 65%). Magnetic resonance of the heart (CMR) obtained normal dimensions and volumes of the left ventricle. EF 62.5%. The contrast method with gadolinium showed significant subepicardial fibrosis of the entire basal and part of the lateral wall of the left ventricle. After receiving the CMR findings, therapy with an ACE inhibitor (Enalapril) and a beta blocker (Presolol) was included, with the addition of Calcort. With further clinical follow-up, control echocardiographic findings show mild globulation of the left chambers, with both circumferential and longitudinal function preserved. Control examinations by magnetic resonance of the heart, the field of fibrosis increases in terms of involvement of deeper layers myocardium, and retains approximately the same distribution.

Conclusions: Conclusion: Cardiac magnetic resonance (CMR) is invaluable in the early detection of cardiac damage in Dysen’s dystrophy patients.

Keywords: Dysen’s dystrophy, heart damage, Magnetic resonance of the heart (CMR)
Russia had Implemented an Expanded Newborn Screening for 36 Diseases

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Aim: The expanded newborn screening is a basis for early diagnosis of hereditary diseases. Since 2006, the newborn screening had been conducted for 5 diseases in Russia, and since 2023 - for 36. Aim is to show the first experience of organizing and implementing the expanded newborn screening in Ural Federal District, covering 10.6% of Russia’s territory.

Material and Method: The blood samples were analyzed using tandem mass spectrometry and real-time polymerase chain reaction.

Results: From January 1st, 2023 the expanded newborn screening was implemented in Russian hinterlands. The Ural Federal District includes 6 regions with a population of over 12 million people, the largest of which is the Sverdlovsk region, where over 4 million people live. For the first half of the year 2023, 58 262 newborns were born in the district. The examination was carried out on 56 408 infants (96.8%). 56 497 samples were delivered to a laboratory in Ekaterinburg. The risk group consisted of 1655 children (2.9%). The diseases were confirmed in the federal genetic center in 22 infants (1.3%). There were 9 cases of inherited metabolic disorders, 7 of spinal muscular atrophy, 6 of primary immunodeficiency.

Conclusions: The expansion of newborn screening is a priority trend of public policy directed to early detection of hereditary disorders, timely treatment of children with severe life-threatening, including rare (orphan) diseases.

Implementation of a newborn screening

Keywords: screening, newborn, pathogenetic treatment, diagnostics, hereditary diseases
Differences in Neonatal Outcomes Between Normal and Preeclampsia-Complicated Pregnancies

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Aim: To investigate possible differences in neonatal outcomes between normal and preeclampsia-complicated pregnancies in one tertiary gynecology and obstetric center in Serbia.

Material and Method: This case-control study included 50 preeclampsia patients and 50 healthy normotensive pregnant women followed up in Clinic for Gynecology and Obstetrics, Clinical Center of Serbia. Neonatal outcomes in both groups were monitored until discharge from the hospital. Appropriate statistical tests were applied for experimental and clinical data analysis.

Results: Cesarean Section was significantly more frequent in preeclampsia than in the normal pregnancy group. The mean gestational age was approximately 4 weeks lower in newborns of preeclamptic mothers than in healthy control (p<0.001). Apgar scores were significantly lower in the experimental than in the control group. Hospitalization was longer after preeclampsia-complicated pregnancies than uncomplicated controls. Admission to the neonatal intensive care unit was observed in 56% of newborns in the cases group and 6% in the control (p<0.001). Perinatal asphyxia was reported in 40% of cases from the preeclampsia group and 4% in controls (p<0.001). Respiratory distress syndrome had one-third of newborns from the experimental group and none from normal pregnancies (p<0.001). Neonatal convulsions, intracranial hemorrhage, jaundice, anemia, and infection were significantly more common in the cases group than in the controls. In the whole study group, only one newborn from a pregnancy complicated with early and severe preeclampsia had a lethal outcome.

Conclusions: The frequency of Cesarean section was significantly higher, whereas gestational age and Apgar Scores were lower in preeclampsia than in the normal pregnancies group. This study demonstrated that overall morbidity was significantly higher among newborns from pregnancies complicated with preeclampsia compared to those from healthy pregnancies.

Keywords: preeclampsia, infant, newborn, morbidity
The skin’s primary function is to provide protective barrier that maintains internal hydration and electrolytes while preventing entry of exogenous substances such as bacteria, allergens, particulates, pollutions, etc. The outer – most layers of the skin, the stratum corneum (SC), are often described as bricks (cells) and mortar (lipid) structure. Baby skin care is arguably an area of maternity service provision considered to be of relatively lower priority compared to antenatal and intrapartum care. However, with the rising uncertainties amongst health care professionals and parents about effective and safe baby skin care practices, current baby skin care advice given to parents by health professionals may be a contributory factor.

There are structural differences between baby and adult skin. The epidermis in babies is 20% thinner and the stratum corneum is 30% thinner1, increasing susceptibility to permeability and dryness. The ratio of baby body surface to body weight is higher than that for adults2, which means that topical agents may have a more intense effect on baby skin. Baby skin also has a propensity to greater trans-epidermal water loss [TEWL] and reduced stratum corneum hydration, reflecting a less effective skin barrier function3,4. Babies have a higher skin surface pH (low acidity) which amplifies protease activity and the breakdown of corneodesmosomes, the supportive connective components of the stratum corneum5,6. At birth, baby skin barrier is adequately developed to tolerate extraterine environment; however, it continues to develop throughout the initial years of life7,8,9.

In most of the research when the topic is baby bath, qualitative research indicates that parents and health professionals believe that water alone is best10. There was some evidence to suggest that daily use of full-body emollient therapy may help to reduce the risk of atopic eczema in high-risk babies with a genetic predisposition to eczema; however, the use of olive oil or sunflower oil for baby dry skin may adversely affect skin barrier function11. Maintaining the health of a newborn’s skin is an essential part of infant care and the choice of nappies and hygienic nappy-care plays an important role in achieving this. The critical care points where compromise can occur are identified as the time the infant spends in a wet nappy, leading to mechanical, chemical and microbial trauma, the cleansing routine of the skin at nappy change time, the steps taken to preserve the skin’s own defence mechanisms, and control of infection through avoiding contamination of clothes, bedding and carers by soil and urine.
An important consideration in reducing skin irritation are innovative diaper care products which can mitigate key causes of DD development. Recommendations to prevent diaper dermatitis from paediatric and neonatal professionals include the use of superabsorbent diapers, frequent diaper changes, good skin hygiene using appropriately formulated wipes, and prophylactic use of emollient to protect the skin from external irritants like bacteria, viruses, fungi, and allergens. Superabsorbent diapers, like Pampers Active Baby are, trap urine and stool to prevent overhydration while sequestering stool away from skin. Emollients can provide a protective layer similar to that imparted by vernix to the fetus during late gestation. Pampers Active Baby is the diaper with a triple absorbent layer that protects the natural moisture of the baby’s skin, ensuring dryness and air flow for up to 12 hours. In addition to three absorbent layers, the diaper features a double leg cuff that provides improved protection around the legs, as well as flexible wings that expand as baby moves, providing a comfortable and secure fit around the waist, preventing chafing or irritation and improving leakage protection. Pampers disposable baby wipes effectively clean the skin by removing fecal material and may be engineered to deliver solutions which can positively impact skin physiology, including skin pH. Many fecal enzymes (eg proteases and lipases) are maximally active at neutral or slightly alkaline pH, but have reduced activity at acidic pH.

Children’s skin, especially babies, has special characteristics that make it more vulnerable and less resistant to bacteria, irritants and allergens that can easily penetrate the skin, so babies and children are more susceptible to skin irritations and infections. That is why good skin hygiene and right diaper selection is necessary.

References


