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ABSTRACTS

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ACUTE HEMIPARESIS AS PRESENTING SYMPTOM OF NEUROBORRELIOSIS IN PAEDIATRIC PATIENT

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Objectives

Lyme disease is an infectious, tick-borne illness primarily caused by Borrelia burgdorferi (B. burgdorferi). It remains common in endemic regions with an incidence of 15.0 per 100 000 Latvians in 2023. 10-15% infected patients develop Lyme neuroborreliosis (LNB). Most common symptoms of LNB in children are facial nerve palsy and subacute meningitis. Rarely, LNB can manifest as cerebral vasculitis. The aim is to report a case of acute hemiparesis being the first presenting symptom of LNB due to secondary vasculitis and arterial ischemic stroke (AIS) in a 12 year-old female and a positive outcome after treatment.

Materials and Methods

A retrospective study of the patient's data was done from time of initial hospitalisation in Children's Clinical University Hospital in Riga in July 2023 until time of follow-up in the outpatient centre in December 2023. Data was collected from medical history data system.

Results

A 12 year-old girl presented to the Emergency Department with complaints of right-sided weakness, which led to inability to stand up after awakening. Upon examination, she had mild hemiparesis and hypoesthesia on the right side. It was later revealed that the patient had been experiencing gait instability and unusual sensations in the right leg for 2 days. Vital signs were stable. The patient reported no chronic diseases and was not taking any medication. History was negative for tick bites. The patient was fully immunized. Acute magnetic resonance imaging (MRI) and angiography of the brain revealed an AIS in the left frontal lobe and gyrus cinguli, arteria cerebri anterior (ACA) sinistra (sin) A2 occlusion, stenosis of arteria carotis interna sin supraclinoid segment and blood vessel irregularities in arteria cerebri media sin M1 segment. The patient was started on aspirin. While in the Neurology department, comprehensive workup was initiated. Other causes of AIS were excluded. A lumbar puncture was done and cerebrospinal fluid (CSF) analysed: lymphocytic pleocytosis 183.0x1 u/L, elevated protein 1.67 g/L and decreased glucose 1.62 mmol/L. Empiric antibacterial and antiviral therapy was initiated. Two weeks after admission the patient started complaining of nausea and diplopia. MRI was repeated and showed new ischemic lesions in left basal ganglia and right frontal lobe. Intravenous methylprednisolone was initiated. Low molecular weight heparin was added to therapy. In CSF, immunoglobulin M (IgM) and immunoglobulin G (IgG) for B. burgdorferi and antibody index was positive confirming the diagnosis of LNB. In serum, IgG for B. burgdorferi was positive and IgM was negative. The patient received 2 weeks of intravenous ceftriaxone followed by a week of oral doxycycline. She was discharged with National Institute of Health Stroke Scale Score 0 and received rivaroxaban for 3 months, followed by aspirin. A follow-up MRI at 6 months showed improvement in ACA sin A1 and A2 blood flow.

Conclusions

We report a case of paediatric acute cerebral stroke with hemiparesis as a first manifestation of Lyme neuroborreliosis due to secondary vasculitis in an otherwise asymptomatic patient. Early recognition and treatment of Lyme disease can prevent complications of LNB. CSF testing is an integral part of the diagnostic workup for vasculitis. Etiologic treatment should be initiated for secondary vasculitis whenever possible. Antithrombotic therapy or anticoagulants may be effective for patients with vasculitis although research in the paediatric population is lacking.

CEREBRAL VENOUS THROMBOSIS IN CHILDREN: CLINICAL EXPERIENCE FROM LATVIA

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Objectives

Cerebral venous thrombosis (CVT), although rare in children, represents the second most common type of venous thromboembolism in this age group. It is characterized by total or partial occlusion of blood flow in the cerebral venous system and has an estimated annual incidence of 0.3 - 0.6 cases per 100,000 children. Our aim was to evaluate the prevalence, diagnostic methods, clinical manifestations, potential etiologies, and treatment modalities of CVT in Latvian children aged between 28 days and 18 years.

Materials and Methods
A retrospective study was conducted by analyzing medical records of individuals diagnosed with CVT and receiving treatment at Latvia's Children's Clinical University Hospital (CCUH) from January 2015 to December 2023. The collected data underwent analysis using IBM SPSS.

Results
The study population included 20 patients diagnosed with CVT. Between 2015 and 2016 only 1 case was documented in the database while 19 cases occurred between 2017 and 2023 with an average of 2.7 cases (1 - 8 cases per year) annually in this time period. The mean age of CVT occurrence was 8.4 years (11 months - 17 years), with equal distribution between male and female patients at 50.0% (N=10) each. Intra-hospital events accounted for 45.0% (N = 9), while 55.0% (N = 11) of the patients experienced first symptoms outside the hospital. The majority of patients, 80.0% (N = 16), were initially admitted to CCUH, while 20.0% (N = 4) were transferred from regional hospitals. The most common initial symptoms were a new onset headache (N=9, 45.0%), drowsiness (N=6, 30.0%), and focal neurological deficit (N=6, 30.0%), with the most common being abducens nerve (cranial nerve VI) palsy (N=4, 20.0%). Motor seizures were observed in 2 patients. Among all, brain magnetic resonance imaging with venography was the primary neuroimaging method in 90.0% (N = 18) of cases. Out of the 18 patients who had their D-dimer levels measured, 16 (89.0%) exhibited elevated levels, with a median of 1.21 mg/L (IQR = 0.99). In most cases (N=14, 70.0%), thrombosis in multiple venous sinuses was observed, with the sigmoid sinus (N=14, 35.9%) and transverse sinus (N=11, 28.2%) being most commonly affected. In 2 patients venous infarction occurred. Ophthalmologists consulted 12 patients, among whom 3 patients (25.0%) had optical disc oedema, but only 1 had complaints about blurry vision . In the acute phase, the majority of patients (N=19, 95.0%) received anticoagulant treatment with low molecular weight heparin. Admission to the intensive care unit was required for 35.0% (N=7) of patients. Otomastoiditis (40.0%, N = 8) and concurrent oncological diseases with patients receiving chemotherapy (25.0%, N = 5) were the most common presumed etiologies. In total, the etiology was specified for 90.0% (N=18) of all patients. The mean hospital stay was 27 days ± 16 SD, with 1 patient having a lethal outcome. Among survivors, after the acute phase, anticoagulation with rivaroxaban (N=8, 42.1%) or warfarin (N=4, 21.1%) was most commonly continued.

Conclusions
Cerebral venous thrombosis (CVT) is a rare yet severe condition observed in pediatric patients. Our study demonstrates that it may manifest with nonspecific clinical symptoms, posing challenges for early diagnosis. Utilization of laboratory tests and neuroimaging studies aids in achieving accurate diagnosis. Identifying the underlying etiology is crucial as it enables mitigation of the risk of recurrent events. Anticoagulation is the treatment of choice for CVT

PEDIATRIC ARTERIAL ISCHEMIC STROKE: INSIGHTS FROM LATVIAN CLINICAL PRACTICE FROM 2012 TO 2023

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Objectives
Arterial ischemic stroke (AIS) represents a significant contributor to childhood disability and mortality, with an estimated annual incidence of 1-2 cases per 100,000 children. Our aim was to evaluate the prevalence, diagnostic methods, clinical manifestations, potential causes, and treatment modalities of arterial ischemic stroke in Latvian children aged between 28 days and 18 years.

Materials and Methods
A retrospective study was conducted by reviewing medical records of individuals diagnosed with arterial ischemic stroke (AIS) and receiving treatment at Latvia's Children's Clinical University Hospital (CCUH) from January 2012 to December 2023. The collected data underwent analysis using IBM SPSS.

Results
In total, 43 cases were included, averaging 3.6 ± 1.6 SD cases per year, of which 74.4% (N = 32) occurred outside the hospital, but 25.6% (N = 11) were intra-hospital events. The majority of patients, 65.1% (N = 28), were initially admitted to CCUH, while 32.6% (N = 14) were transferred from regional hospitals, with one patient arriving from abroad. The mean age of stroke onset was 101 ± 59.6 SD months, with 55.8% (N = 24) being male and 44.2% (N = 19) female. The most common initial symptoms were hemiparesis (48.8%, N = 21) or seizure (18.6%, N = 8). Among cases occurring outside the hospital, the majority (75.0%, N = 24) of patients were admitted on the same day as symptom onset. Among all patients, brain magnetic resonance imaging was the primary imaging method in 55.8% (N = 24) of cases. Only 3 patients have received acute treatment - 2 patients (4.7%) received intravenous thrombolysis and 1 underwent thrombectomy. Identified risk factors were present in 79.1% (N = 34) of patients, with cerebral arteriopathies (27.9%, N = 12) and congenital heart disease (25.6%, N = 11) being the most common presumed etiologies. The median hospital stay was 17 days (IQR = 16).

Conclusions
Our results shows that Arterial ischemic stroke (AIS) regularly affects Latvian children, yet only a small number receive immediate treatment. Despite the age-specific causes and symptoms of childhood stroke, similar to adults, it necessitates prompt and accurate diagnosis and treatment to minimize long-term complications. Establishing a nationwide protocol for diagnosing and treating arterial ischemic stroke in childhood could potentially aid in achieving this objective.

PEDIATRIC STROKE RESEARCH IN ESTONIA

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Objectives
The aim of the presentation is to give an overview of pediatric stroke research in children in Estonia.

Materials and Methods
To summarize the most important findings published by Estonian Pediatric Stroke Workgroup since 2007.

Results
Incidence rate of perinatal stroke in Estonia is 63.4/100,000 or 1:1578 live births (1/3 neonatal and 2/3 presumed cases). 1/3 of the perinatal stroke cases have neonatal stroke and 2/3 have presumed perinatal stroke. Most children (95%) with presumed perinatal stroke are born at term and 2/3 of the cases have periventricular venous infarction (PVI). Incidence rate of childhood stroke in Estonia is 2.73 per 100,000 person-years. Sixty percent of childhood stroke cases are arterial ischemic stroke (AIS), 30 % of cases are hemorrhagic stroke (HS) and 10% of cases are cerebral sinovenous thrombosis (CSVT).
Children with presumed periventricular venous infarction had significantly more often maternal bacterial infections compared to the control group (47% vs 20%, respectively, $P < .001$). Mothers with bacterial infection in the presumed periventricular venous infarction group had significantly more often pyelonephritis compared to the control group (50% vs 3.4%, respectively, $P < .001$). Factor V Leiden and prothrombin 20210G>A mutations were risk factors for venous stroke, but not in case of arterial stroke. Genetic testing using exome or large gene panel (n = 6700 genes) sequencing found pathogenic variants associated with stroke in 11 of 85 (12.9%) children with periventricular hemorrhagic infarction/PVI. COL4A1/A2 and COL5A1 variants were found in 7 of 11 (63%) children.
The global neurodevelopmental outcome of children with perinatal stroke is poor: assessed with Pediatric Stroke Outcome Measure 95% of children with AIS and 94% with PVI have moderate to severe neurological impairment in. Combined deficits of motor, language and cognitive/behavioral functions are significantly more common among children with AIS (90%) compared to children with PVI (53%, $p=0.007$). A significant increase in default mode network connectivity (FDR 0.1) and lower cognitive functions ($p < 0.05$) were found in children with AIS compared to the controls and the PVI group in resting-state functional MRI study. Volumetric MRI study revealed that smaller volume of ipsilesional thalamus was associated with poor affected hand function regardless of the perinatal stroke subtype. Tasked-based functional MRI study of right-handed children with stroke demontsrated that most children with large perinatal stroke lesions had language activation reorganized to the right hemisphere, but language reorganization to the unlesioned right hemisphere did not ensured normal language outcome.
The 18-year cumulative poststroke epilepsy risk according to the Kaplan-Meier estimator was 40.8% (95% CI 20.7-55.9%). Epilepsy and interictal epileptiform discharges (IEDs) without clinical seizures were more often expressed in children with AIS (55%) than in children with PVI (21.2%), $p = 0.0057$. Both patients with AIS and PVI with severe damage to the basal ganglia and the thalamus have a higher risk of developing poststroke epilepsy.

Conclusions
Pediatric stroke includes several vascular syndromes, each with unique timing, risk factors, and outcomes. In order to consult parents and plan rehabilitation and follow-up care, child neurologists should be aware of these differences.

SUCCESSFUL MECHANICAL THROMBECTOMY FOR BASILAR ARTERY OCCLUSION IN A PEDIATRIC PATIENT

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Objectives
Studies have shown the benefits of endovascular treatment (EVT) in adult stroke cases, but its application in pediatric stroke remains controversial. Despite evidence of improved outcomes in adults, there are no established recommendations for EVT in children. Conducting individual case reports and case series is vital to understanding its potential advantages and

disadvantages in this context.

Materials and Methods
In this case report we describe a 9-year-old male who was initially admitted to a regional hospital due to a persistent complaintof diarrhea, exceeding ten episodes in two days, accompanied by a single episode of vomiting, tension headache, and mild vertigo. On the subsequent day after admission the patient suddenly experienced weakness localized to the left side of his body.
Urgent imaging, consultation of a neurologist and interventional radiologist, and transfer of the patient to a specialized center followed.

Results
Upon admission, the patient displayed a Glasgow Coma Scale (GCS) score of 15 and presented with left-sided hemiparesis, as assessed by the National Institutes of Health Stroke Scale (NIHSS), which recorded a score of 6.
Comprehensive imaging revealed acute ischemia in the cerebellum, indicating a basilar artery thrombus. Urgent endovascular treatment (EVT) was performed 8.5 h after the onset of neurological symptoms. Throughout the procedure, multiple left vertebral arteriograms were performed, consistently illustrating blockages in the middle segment of the basilar artery. After six meticulous attempts, a final angiogram conclusively confirmed the complete removal of the obstructions in both the basilar artery and the posterior cerebral arteries, achieving a TICI scale score of 3, indicating full and successful revascularization.
The patient underwent rehabilitation and was later discharged with improved neurological status.
During the six-month follow-up, the patient exhibited a complete neurological recovery, as evidenced by an NIHS score of 0 and an mRS score of 0.

Conclusions
Effective treatments for the pediatric ischemic stroke population lack robust evidence from randomized clinical trials, with insights primarily drawn from isolated cases or case.
This clinical case of a pediatric patient with basilar artery thrombosis who underwent successful mechanical thrombectomy shows a favorable clinical outcome, indicating that procedure is safe and effective event in patients with late time-window (8.5 h after the onset of neurological symptoms).

UNRAVELING COMPLEXITY: LYME DISEASE, ARTHRITIS, CRANIAL NEUROPATHY AND CEREBRAL VENOUS SINUS THROMBOSIS IN PEDIATRIC PATIENT - A CASE STUDY

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Objectives
Lyme disease presents as a multifaceted infectious condition, that have the potential to affect multiple organ systems, including the skin, nervous system and musculoskeletal system. Albeit uncommon, cerebral venous sinus thrombosis represents a serious complication of otomastoiditis. The objective of this study is to examine the clinical manifestations, diagnostic procedures, treatment strategy, and outcome in a pediatric patient diagnosed with concurrent Lyme disease, displaying symptoms of acute arthritis and cranial neuropathy, alongside with asymptomatic cerebral venous sinus thrombosis secondary to otomastoiditis.

Materials and Methods
Retrospective analysis of a pediatric patient case was conducted. Data was collected from medical history data system and included clinical observations, diagnostic imaging results, laboratory findings, treatment regimens, and follow-up assessments.

Results
1-year and 11-month-old male initially presented with symptoms of subfebrile body temperature, difficulty placing his left foot and unstable gait. Hip magnetic resonance imaging (MRI) imaging unveiled signs of septic arthritis in the left hip joint, leading to arthrotomy. Following the surgical procedure, on next day the patient developed clinical signs of left facial nerve neuropathy. Further investigation via head MRI unveiled neuritis affecting the 7th and 8th cranial nerves, labyrinthitis, left-sided mastoiditis, and partial thrombosis of the sigmoid sinus. Analysis of cerebrospinal fluid (CSF) revealed an increase in cell count with lymphocytes being predominant. Alongside IgM antibodies against B. burgdorferi in both, CSF and serum, was detected using ELISA and Immunoblot assays. With bacteriological cultures from the hip joint and middle ear being negative, a comprehensive assessment led to a diagnosis of Lyme disease, presenting with acute hip arthritis and cranial neuropathy as well as concurrent asymptomatic otomastoiditis, and cerebral venous sinus thrombosis. Therapy plan included a 21-day antibacterial therapy with intravenous ceftriaxone for B. burgdorferi infection and anticoagulation, initially with low molecular weight heparin, followed with a 3-month course with rivaroxaban for venous sinus thrombosis, as well as tympanotomy to address otomastoiditis. Follow-up brain MRI performed after 3 months revealed improvement with no observed thrombotic masses. The symptoms of facial nerve paralysis were completely alleviated. Child maintained stable health with age-appropriate psychomotor development throughout the duration of therapy and did not develop any side

effects from therapy.

Conclusions
This case illustrates a patient with two rare diseases simultaneously - acute Lyme disease with manifestations of acute arthritis, cranial neuropathy, and cerebral venous sinus thrombosis. Timely identification of both, Lyme disease and cerebral venous sinus thrombosis, is crucial for prompt and successful treatment, leading to potentially positive patient prognosis. Both conditions can exhibit a wide range of symptoms. In our case study, we detail a patient presenting with Lyme disease, displaying symptoms of acute monoarthritis and cranial neuropathy, alongside an asymptomatic occurrence of cerebral venous thrombosis.

Autoimmune Neurological Disorders

EPIDEMIOLOGY OF PEDIATRIC AUTOIMMUNE ENCEPHALITIS IN LATVIA

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Objectives
Autoimmune encephalitis (AE) is one of the most common causes of encephalitis, after infectious aetiologies and acute disseminated encephalomyelitis. Due to diverse, complex clinical presentations and various investigation results, diagnosis can be challenging. Clinicians should be aware that there is high rate of diagnostic mimics that are even more common than AE itself. The aim of this study was to determine the incidence of pediatric AE in Latvia.

Materials and Methods
This was a retrospective study and included Children's Clinical University Hospital (CCUH) patients with diagnosed definite, probable, and possible AE starting from 2014 till 2022. Sociodemographic data, investigation findings regarding autoantibody testing were retrospectively collected from medical history data system.

Results
Our study group consisted of 18 patients. There were 10 male and 8 female patients, the median age of the patient group was 8 years (interquartile range 3 – 13), the youngest patient was 9 months old and the oldest - 17 years. Majority of patients (66.6%, n = 12) had seronegative AE, the rest were classified as seropositive (33.3%, n = 6). In seropositive group most were (n = 4) anti - NMDA receptor AE and two patients had other antibodies (anti GAD-65 or anti ZIC-4). The mean incidence of AE was 0.56/100 000 children. Incidence range was from 0 to 1.68/100 000, number of patients diagnosed with AE varied from 0 to 6. Over the studied years incidence remained relatively stable (except for 2017, n = 6), however in the last studied year it was above the mean (0.84/100 000 children, n = 3).

Conclusions
Pediatric AE incidence in Latvia is similar as previously reported in other studies and remains stable in the studied years. Some patients might be misdiagnosed, since a high proportion of seronegative AE was present. Awareness of possible misdiagnosis should be raised.

FEBRILE INFECTION RELATED EPILEPSY SYNDROME TREATMENT OVER YEARS LATVIAN EXPERIENCE

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1. Dr.

Objectives
Febrile infection related epilepsy syndrome (FIRES) is a subcategory of NORSE (new-onset refractory status epilepticus) and is characterized by sudden refractory status epilepticus in previously healthy children without an identifiable cause. The annual incidence and prevalence of FIRES among children and adolescents are estimated to be, respectively 1:100000 and 1:10000. (Reppucci et al, 2022) The outcome of FIRES is poor with death rate up to 30% and those surving having neurological sequel and refractory epilepsy. (Serino et al, 2019)

Materials and Methods
This was a retrospective study and included Children's Clinical University Hospital patients diagnosed with FIRES tarting from 2011 to 2024. Clinical and demographic data were collected from medical history data system.

Results
In our study group 3 patients with FIRES were included with mean age 10 years, ranging from 4 to 17 years. FIRES ncidence in the studied years was 0 - 0.53 per 100 000 children. There were two males and one female. Mean time in intensive care unit 65 days. All the patients underwent extensive diagnostic workup. In the acute phase all patients received antiepileptic drugs (AEDs) immunotherapy with IVIG, steroids, plasmapheresis, Rituximab, two patients received Anakinra and one patient received Tocilizumab. All patients received ketogenic diet. No patients died in this period. All patients had refractory epilepsy, mental retardation or behavioral disturbances. For seizure control in the chronic phase patients received different AEDs. Two of the patients has several seizures per day. Only one patient was seizure free after discharge of ICU and continues rehabilitation.

Conclusions
Based on the current ILAE classification, the clinical presentaion, seizure type, EEG findings, CSF, and MRI findings collectively characterized FIRES as an epilepsy syndrome with progressive neurological deficits starting in childhood. A suspected diagnosis should be made as early as possible to modify the status epilepticus treatment and initiate immunotherapy as early as possible. As early treatment can improve the outcome of the patients.

MEASLES AND SUBACUTE SCLEROSING PANENCEPHALITIS: A CASE REPORT FROM LITHUANIA

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Objectives
Objectives. Subacute sclerosing panencephalitis (SSPE) is a rare, progressive, and fatal degenerative encephalitis caused by slow measles virus infection in the central nervous system. SSPE poses significant challenges in pediatric neurology due to its rarity, progression, and severe outcomes.

Materials and Methods
Aim. To report the only diagnosed case of SSPE in Lithuania so far and to discuss the diagnostic, and treatment challenges.

Results
Case Report. We present a case of a 5-year-old male. The SSPE started with progressive behavior changes and instability while walking, jerking movements of the hands accompanied by head nodding, followed by tonic positioning of his hands in flexion, hyperkinetic movements in his legs, intention tremor, and cognitive regression within a few weeks. The case history revealed that the patient had measles at the age of 16 months. Investigation showed periodic complexes of repeated high-amplitude delta waves and epileptiform activity in electroencephalogram (EEG), pathological high fluid-attenuated inversion recovery (FLAIR) signals on MRI of the patient, and increased serum measles antibody titer. Despite antiseizures medications and immunomodulatory therapy, the patient's condition continued to deteriorate, with an increase in the frequency of jerking movements and severe encephalopathy. When the diagnosis of SSPE was confirmed, he was in a persistent vegetative state already. He was managed symptomatically, treated with Isoprinosine, and a ketogenic diet with no significant effect. The patient remained in a vegetative state.
Discussion. An acute fulminant presentation of SSPE can confuse with acute encephalitis, and SSPE is often not considered because of its rarity. Whenever there are unusual clinical manifestations and neuroimaging pictures, the cerebrospinal fluid should be examined for anti-measles antibodies. Our case also showed the limitations of currently available treatment options that may delay but do not prevent the outcome of those affected by the disease.

Conclusions
Conclusions. This case report and discussion provide insights into SSPE diagnostics, and treatment, focusing on possible solutions to emerging difficulties.

PEDIATRIC ANTI-NMDA RECEPTOR ENCEPHALITIS IN LATVIA: CASE SERIES

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Objectives
Anti-N-methyl-D-aspartate receptor (anti-NMDAR) encephalitis is the most common autoimmune encephalitis in children after acute demyelinating encephalomyelitis. Children with anti–NMDAR encephalitis often present with movement abnormalities, agitation, insomnia, seizures, ataxia and/or hemiparesis. Diagnosing anti-NMDAR encephalitis can be

challenging due to complexity of evaluating behaviour in a developing child. Most patients with anti-NMDAR encephalitis respond to immunotherapy, however delayed treatment is associated with poor outcome. Aim of this study was to summarise the clinical characteristics, diagnostic findings, treatment and outcomes of anti-NMDAR patients in Latvian paediatric population.

Materials and Methods
A retrospective study was conducted and included Children's Clinical University Hospital patients diagnosed with anti-NMDAR encephalitis from 2014 till 2023. Information about symptoms, investigations, therapy and outcomes were collected from medical history data system.

Results
Five patients were enrolled in this study with a median age of 9.0 (6.0-16.0) years. Majority of patients were male (n = 4). Median time from symptom onset to hospitalisation was 6.0 (1.0-7.0) days. One patient had preceding herpes simplex virus encephalitis. In majority of cases presenting symptoms were behavioural changes (n = 4). All patients experienced behavioural changes during course of disease, furthermore common (n = 4) symptoms were movement disorders, autonomic dysfunction, disorders of consciousness, followed by seizures (n = 3). Majority of patients (n = 4) were admitted in intensive care unit (ICU), median time spent in ICU was 28.5 (12.5-45.3) days. Cerebrospinal fluid (CSF) analysis showed pleocytosis in all patients, median leukocyte count was 16.0 (14.0-36.0) /mm3, CSF protein was elevated in one case. Anti-NMDAR antibodies were positive in all cases; however, one patient was tested only in serum. Oligoclonal bands were negative in all patients in serum and CSF. Electroencephalography was abnormal in all cases: generalised slowing (n=5), frontotemporal slowing (n=2), epileptiform activity (n=2). In two patients Magnetic resonance imaging showed changes consistent with encephalitis. On average, treatment was initiated 6.0 (4.0-7.0) days after symptom onset. Most patients (n = 4) received treatment with intravenous methylprednisolone (IVMP) combined with intravenous immunoglobulin, one patient received IVMP with plasmapheresis. Median time from first to second-line therapy was 22.0 (18.8-28.5) days. Second-line therapy was indicated in majority of patients (n = 4): rituximab (n = 3), rituximab combined with cyclophosphamide (n=1). Median time till improvement after first-line treatment initiation was 32.0 (22.0-37.0) days. Most patients (n = 4) had long-term sequelae, such as learning and concentration difficulties (n = 3), motor deficit (n = 3), seizures (n = 1). Median anti-NMDAR encephalitis one-year function status (NEOS) score was 3.0 (3.0-4.0). Patient with lowest NEOS score 1.0 had no long-term sequelae.

Conclusions
In 10 – year period there were five pediatric patients diagnosed with anti-NMDAR encephalitis in Latvia. All patients had multiple clinical symptoms, the most common being behavioural changes. Most patients had severe clinical course with necessity of ICU admission. All patients received first-line treatment, in most second-line treatment was also necessary. Majority of patients had long-term neurological and psychiatric sequelae.

RELAPSE PROBABILITY AND CHARACTERISTICS AFTER ACUTE DISSEMINATED ENCEPHALOMYELITIS

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Objectives
Acute disseminated encephalomyelitis (ADEM) is an autoimmune inflammatory disease of the central nervous system (CNS), which may follow by a previous infection or vaccination. Usually, ADEM has monophasic disease course, however relapses following ADEM are reported in up to 25% of cases. In most cases patients after ADEM relapse are diagnosed with either multiple sclerosis (MS) or other demyelinating disease.

Materials and Methods
This was a retrospective study and included Children's Clinical University Hospital patients with first diagnosed ADEM starting from 2010 to 2022. Demographic, investigation, and clinical data were retrospectively collected from medical history data system.

Results
Five patients were enrolled in the study, however, overall 5 patients were diagnosed with 8 ADEM episodes. Mean age of study group was 8.2 ± 4.32 years, ranging from 3 to 14 years, there were 80% (n = 4) males and 20% (n = 1) female patient. The relapse rate of ADEM in our study group was 60% (n = 3), however it was 80% (n = 4), if relapse of other demyelinating event (optic neuritis) was included. Within relapsing disease group one patient was diagnosed with multiphasic ADEM, two patients were diagnosed with myelin oligodendrocyte glycoprotein antibody disease (MOGAD) and experienced relapse of ADEM or optic neuritis, one patient was diagnosed with pediatric onset multiple sclerosis (POMS). Patients with MOGAD were younger (4.5 ± 2.1 years; n = 2) compared to other patient group (10.7 ± 3.5 years; n = 3), p = 0.09, patient with monophasic ADEM was the oldest – 14 years old. The mean time till relapse was 107.3 ± 65.1 days, it was shorter (51.5 ± 12.0 days; n = 2) in MOGAD group compared to other patient group (163.0 ± 11.3 days; n = 2), p = 0.01.

Conclusions
Majority of ADEM patients were younger than 10 years and predominantly were males. Most ADEM patients had another demyelinating episode and were diagnosed with POMS or MOGAD. MOGAD patients were younger and had shorter time till relapsing event. Patients should have monitoring after initial ADEM event to prevent late diagnosis of relapsing demyelinating diseases.

Biomarkers in neurological disorders

EVALUATING AXONAL DAMAGE PLASMA BIOMARKERS NFL AND GFAP, AS INDICATORS OF DISEASE SEVERITY IN CHARCOT-MARIE-TOOTH PATIENTS

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Objectives
Charcot-Marie-Tooth disease (CMT) represents the most prevalent form of inherited neuropathy. Monitoring its progression and assessing the severity of the condition requires the identification of potential molecular markers. Potential biomarkers suggested to access nerve damage are neurofilament light chain (NfL) and glial fibrillary acid protein (GFAP). Our primary goal is to determine whether these biomarker levels are related to the severity of the disease.

Materials and Methods
Initially, 44 patients with CMT and 44 controls participated in this study. Disease severity was assessed through clinical evaluations using the CMT Neuropathy Score version 2 (CMTNSv2). NfL and GFAP were measured using a single-molecule array.
Funding
This research has been developed with funding from the Latvian Science Council, Project Discovering biomarkers of disease progression and variability in Charcot-Marie-Tooth neuropathy, No lzp-2021/1-0327.

Results
The group of CMT patients was stratified into subgroups, namely demyelinating CMT and axonal CMT, based on the findings of nerve conduction studies. There were no significant difference in sex ($\chi 2 = 0.0455$, $p = 0.831$) or age ($p = 0.95$) between the CMT and control groups. In the group of patients with CMT, the concentrations of NfL, and GFAP were significantly higher than in the control group ($p<0.05$). NfL and GFAP levels were correlated with the CMTNSv2 score ($rs=0.46$, $p=0.002$; $rs=0.31$, $p=0.04$).

Conclusions
Our study has provided confirmation that plasma concentrations of NfL and GFAP are significantly elevated in patients with CMT compared to controls. Furthermore, NfL and GFAP levels were correlated with the clinical severity of CMT. These findings suggest that NfL and GFAP could be used as reliable disease indicators in future studies.

SEARCHING FOR METABOLIC MARKERS OF CHARCOT-MARIE-TOOTH DISEASE

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Objectives
Charcot–Marie–Tooth disease (CMT) is the most common hereditary neuromuscular disorder. It is a clinically and genetically heterogeneous group of disorders with the phenotype of chronic, slowly progressive neuropathy affecting both the motor and the sensory nerves. Currently, there are no effective pharmacological treatments for CMT available. Biomarkers that could detect the effect of treatment on disease progression are crucial for successful clinical trials. Metabolome analysis is the characterization of small molecules (<1500 Daltons) in biological matrices using analytical chemistry techniques. It has been widely used for discovering diagnostic and prognostic markers. This study aimed to analyse selected plasma metabolite concentrations in CMT cohort and compare them to healthy controls.

Materials and Methods
A cohort of 84 patients with CMT and 34 healthy controls were recruited to this study. All CMT individuals underwent genetic testing. As a control group, our study included healthy individuals without known neurological diseases or symptoms. Targeted plasma metabolic analysis was performed by ultrahigh performance liquid chromatography-mass spectrometry

(UHPLC-MS) to determine plasma levels of 55 selected metabolites. Statistical analysis was performed with Prism 9 and MetaboAnalyst 6.0. The study was approved by the Central Medical Ethics Committee of Latvia (No. 3/18-03- 21). Written informed consent was obtained from all participants in the study.

Results
A total of 33 metabolites were analysed in plasma. We found that plasma ratio of acetylcarnitine was elevated and plasma ratio of glycine was decreased in the CMT group compared with controls. Next we subdivided CMT group according to the genetic findings: CMT1A (n=37), CMTX1 (n=17), CMT2A (n=4), HINT1 (n=5), other genetic subtypes (n=14), unknown genetic type (n=7). Consequently, we screened for differential plasma metabolites between separate genetic CMT groups. We discovered that acetylcarnitine in the CMT1A group and glycine and valine in the CMT1X group are different from the controls. However, prediction analysis showed poor accuracy for predicting the disease.

Conclusions
In this study we performed targeted plasma metabolic analysis in CMT patients and healthy controls. We have identified that CMT patients have significantly higher levels of acetylcarnitine and decreased glycine levels compared to controls. In addition, the CMTX1 subgroup has decreased valine levels compared to controls. However, our predictive models suggest no good predictive power of the detected plasma metabolites for any CMT group. In general, above mentioned metabolites have been reported before as contributors of pathogenesis in peripheral neuropathies. However, more data and longitudinal evaluation is needed to establish whether these metabolites could potentially become specific CMT biomarkers.

Demyelinating disorders

CHILDHOOD MULTIPLE SCLEROSIS: ARE INITIAL MANIFESTATIONS AND DISEASE COURSE SPECIFIC?

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Objectives
Multiple sclerosis (MS) starts quite rarely in childhood, comprising just 3–10% of all diagnosed cases of MS population. The age of onset of the disease may be related to the initial phenotype and the prognosis of MS. Our objective was to assess the specific characteristics of childhood onset of multiple sclerosis.

Materials and Methods
Two groups of patients were analyzed: those diagnosed with MS in childhood (0<18 years of age) in 2005–2021 (group A), and those diagnosed in adulthood (≥18 years old) (Group B). The data were collected from the database of the Lithuanian University of Health Sciences Kauno Klinikos.

Results
For the analysis, 105 patients were selected: 35 children (group A) and 70 adults (group B). The initial symptoms of MS in Group A patients appeared with a similar frequency in boys and girls at median (Q1-Q3) age of 16 (14–17) years, and at 36 (29–44) years in group B. At the onset of the disease, 28.6% of children and 55.7% of adults experienced sensory symptoms ($p<0.05$); accordingly, in children and adults, visual disturbaces were experienced by 62.9% and 70.0% ($p>0.05$), motor symptoms by 34.3% and 48.6% ($p>0.05$), dyscoordination by 25.7% and 40.0% ($p>0.05$). Cognitive disorder as well as fatigue or pelvic dysfunction were not documented in children as compared to adults (accordingly, 4.3%, 5.7%, 5.7%). Isolated symptoms were more common in children (65.7%) as compared to adults (28.6%), $p < 0.001$. Optic nerve and cerebral hemispheres were mostly affected in group A ($p < 0.05$). During the first year after the diagnosis, the median number of relapses in group A was higher (3, range 1–5) as compared to group B (1, range 1–2) ($p < 0.001$). Recovery time after a relapse was shorter in children as compared to adults ($p < 0.001$). Oligoclonal bands were found in 85.7% of children and in 98.6% of adults ($p = 0.007$).

Conclusions
In most of the childhood MS the initial symptoms were limited to the dysfunction of a single part of the nervous system. Children usually started with visual disorders, while sensory, coordination or motor disorders were less common. The course of the disease in juvenile patients with MS was more aggressive in the first year of the disease as there were more relapses, but the functional impairment recovered faster as compared to adults.

Epilepsy and sleep medicine

ASSESSING SLEEP DISORDERED BREATHING IN PEDIATRIC PATIENTS WITH SMA AND DMD: INSIGHTS FROM A RETROSPECTIVE COHORT STUDY

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Objectives
Sleep-disordered breathing (SDB) is a common concern in pediatric patients with neuromuscular disorders (NMD) due to respiratory muscle weakness and declining lung function. The current practice guideline recommends polysomnography as the preferred option for assessing the need for noninvasive ventilation (NIV) in symptomatic NMD patients when pulmonary function tests (PFT) and overnight oximetry (ONO) show normal results. However, there is low certainty of evidence supporting this recommendation, highlighting the need for careful evaluation of individual patient needs and circumstances. Our objective was to compare the efficacy of spirometry and polygraphy with transcutaneous capnography (PG+trCO2) in detecting SDB and assess PG+trCO2's role in diagnosing SDB in patients with DMD and SMA.

Materials and Methods
A retrospective study at Children's Clinical University Hospital, Riga, Latvia involved evaluating the PG+trCO2 and spirometry data of DMD and SMA diagnosed patients.

Results
70 patients were included; 37% had PG+trCO2, 64% spirometry, and 20% both. 77% of those with PG+trCO2 showed alterations such as sleep tachypnea and hypoventilation. Of the 45 who had spirometry, 47% demonstrated alterations in their FEV1 and FVC, ranging from mild to severe restrictive breathing. Of the 14 patients who had both evaluations (PG+trCO2 and spirometry), 21% had alterations only in PG+trCO2. This resulted in a change in their treatment regimen, with the addition of NiV.

Conclusions
Polygraphy and transcutaneous capnography, when combined with spirometry, offer enhanced detection of SDB compared to using spirometry alone. The study underscores the importance of incorporating PG+trCO2 into routine diagnostics for early SDB detection in pediatric NMD patients. A proposed "PG+trCO2 for All" policy suggests annual screenings for NMD patients, but further research is imperative to determine optimal monitoring strategies.

EXPANDING HORIZONS: THE KETOGENIC DIET BEYOND EPILEPSY - APPLICATIONS IN TREATING OTHER NEUROLOGICAL DISORDERS

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Objectives
The Ketogenic Diet Therapy (KDT) has been applied worldwide for over a 100 years. The efficacy and safety of this treatment method for refractory epilepsy and GLUT1 deficiency syndrome have been extensively described and are beyond doubt. Recently, there has been an increasing amount of research and accumulating data on the application of the KDT in the treatment of other neurological disorders.

Materials and Methods
Literature review.

Results
Although there is no treatment for autism spectrum disorders, studies indicate that the KDT can help normalize social behavior in patients through various mechanisms, such as reducing inflammatory and oxidative stress, modulating gut microbiota, and thereby remodeling the gut-brain axis.
The effect of the KDT on migraine treatment has been described since 1928, but in recent years, with increasing interest in non-pharmacological treatment methods, various research groups have been examining and formulating recommendations on how to apply this treatment method for migraine, cluster headaches, or other types of headaches to reduce the frequency of pain days, improve productivity, and quality of life.
More and more studies are being published discussing the potential benefits of the KDT in treating various psychiatric conditions, such as mood or anxiety disorders, and successful cases of treating schizophrenia have been described. Metabolic therapy not only directly affects psychiatric disorders but often has a positive impact on disease mechanisms and

comorbid conditions.
There is an increasing amount of preclinical and clinical research on the effect of the KDT on reducing brain tumor growth and increasing survival, reducing seizure frequency, and improving overall patient well-being. It is recommended to apply the KDT alongside chemotherapy and radiotherapy for tumor treatment.
The KDT can affect Parkinson's disease progression through several different mechanisms, such as modifying factors affecting nervous system inflammation, mitochondrial dysfunction, disrupted dopamine synthesis, accumulation of damaged proteins, formation of Lewy bodies, and others.
In Alzheimer's disease, brain metabolism is impaired, which can be corrected with the help of the KDT. Positive effects of the diet on the functioning and quality of life of patients with dementia have been observed in studies.

Conclusions
The increasing interest of patients and their relatives in non-drug treatment methods, along with the rapidly growing amount of scientific research, opens up much broader possibilities for applying the KDT not only for epilepsy treatment. However, more detailed and long-term studies are still needed to evaluate not only efficacy but also safety, economic costs, and changes in quality of life.

FAT AND CBD AGAINST FIRES

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Objectives
We present a case of a 6-year-old patient with febrile infection-related epilepsy syndrome (FIRES) treated with various antiseizure medications, including cannabidiol (CBD), as well as a ketogenic diet (KD) and immunotherapy.

Materials and Methods
A case study was conducted by summarizing the data of the patient based on medical records from the Hospital of Lithuanian University of Health Sciences Kauno klinikos database.

Results
A 6-year-old patient was admitted to the pediatric intensive care unit (PICU) due to intractable focal and generalized tonic-clonic seizures. Autoimmune, metabolic, genetic, infectious, structural, and other etiologies were unremarkable and remain to be determined. Immunotherapy, including intravenous steroids and immunoglobulin, tocilizumab, and anakinra, were ineffective. Various antiseizure medications were trialed; however, the introduction of CBD and reaching its dose of 15 mg/kg/day at day 72 of the disease reduced seizures significantly; therefore, no further treatment in the PICU was required. The ketogenic diet was introduced within two weeks; however, it had to be discontinued due to hypertriglyceridemia as a side effect of coadministration of propofol or tocilizumab. It was reintroduced after 4 months of treatment and is successfully continued in conjunction with antiseizure medication and remains effective. The patient currently has up to 6 tonic seizures per day, severe encephalopathy, and brain atrophy evident in MRI scans.

Conclusions
Current evidence does not support the early introduction of CBD in cases of FIRES, although it might be useful in conjunction with other antiseizure medications in the acute or chronic phases of FIRES. It is vital to anticipate the side effects of different therapies in order not to deviate from currently proposed treatment strategies.

GENETIC EPILEPSY LANDSCAPE IN PAEDIATRIC PATIENTS IN LATVIA

Ieva Mičule [1], **Madara Mašinska** [1], **Gita Tauriņa** [1], **Dana Apela** [1], **Jurģis Strautmanis** [1], **Mikus Dīriks** [1], **Agnese Zariņa** [1], **Olga Šterna** [2], **Aigars Dzalbs** [1], **Inna Iņaškina** [3], **Baiba Lāce** [4]

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Objectives
Epilepsy is the most frequent chronic neurologic condition in childhood, and genetic diseases are estimated to cause up to 30% of childhood epilepsy cases. Although several studies have reported genetic causes detected in epilepsy patients, analysis based on epilepsy as the indication for genetic testing misses many patients, who have reached their genetic diagnosis due to different presenting symptoms before the onset of seizures.
Our aim was to analyse the landscape of different genetic epilepsies encountered in Latvian paediatric epilepsy patients, including patients with different presenting symptoms.

Materials and Methods
The retrospective analysis was applied to data from paediatric patients consulted in Children's Clinical University Hospital (CCUH) during the years 2019-2022 who met the criteria of having both a diagnosis of epilepsy and a genetic diagnosis, regardless of the presenting symptoms or the time of reaching the diagnosis.

Results
1345 epilepsy patients were consulted during the selected period, and we identified 195 patients (188 probands and 7 siblings) who had a genetic diagnosis. In these we have identified 103 different genetic diagnoses. Most (79, 77%) are unique (occurred in one family), but 24 (23%) are recurrent and encountered on at least two occasions. Recurrent genetic causes account for majority of cases – 109 families (58%), still, non-recurrent diagnoses, 79 families (42%), comprise a considerable part of our patients. The 10 most frequently encountered genetic epilepsies are Tuberous sclerosis complex (25 cases, 13.3%), SCN1A related epilepsy (19,10.1%), Angelman syndrome (9, 4.8%), PRRT2 related BFIS (5, 2.7%), Proximal 16p11.2 deletion syndrome (5, 2.7%), KCNQ2 related epilepsy (4, 2.1%), Down syndrome (4, 2.1%), 15q11.2 microdeletion syndrome (4, 2.1%), Neurofibromatosis 1 (3, 1.6%), SYNGAP1 related DEE (3, 1.6%). Of all the detected causes 141 (75%) were monogenic diseases and 47 (25%) were chromosomal or imprinting disorders. Genetic diagnosis was established before the start of seizures in 47 patients (24%), 16 (34,8%) of them presented with developmental delay.

Conclusions
Our study is the first to aggregate data on causes of heritable epilepsies in Latvia. The results may be biased towards cases with more severe presentation due to patient selection in a tertiary care centre. Tuberous sclerosis is a recognized cause of genetic epilepsy but does not appear among the most prevalent in other studies. Frequency in our patient cohort could possibly be attributed to existence of clinical diagnostic criteria allowing the diagnosis to be made even in the absence of molecular confirmation.

KCNQ2 GENE RELATED SELF-LIMITED FAMILIAL NEONATAL EPILEPSY PRESENTING WITH MOTOR SEIZURES AND POSSIBLE OCULOGYRIC CRISES

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Objectives
KCNQ2 gene-related disorders includes a wide range of epileptic conditions, ranging from the milder self-limited familial neonatal epilepsy (SeLFNE) to the more severe neonatal-onset developmental and epileptic encephalopathy. The objective of this study was to describe an infant with early-onset motor seizures and presumably oculogyric crisis who was later diagnosed with KCNQ2-related SeFLNE. There are very limited information about oculogyric crisis being related to this gene mutation.

Materials and Methods
A retrospective patient case study was conducted. Data was collected and analysed from patient medical history.

Results
The patient, a term-born female infant delivered via planned cesarean section at 40 weeks gestation, experienced her first motor seizure on the fourth day of life, characterized by seizure onset with upward deviation of gaze, arm flexion, and leg extension, followed by clonic movements in both arms and legs lasting under one minute. On the fifth day of life similar event repeated. Electroencephalography (EEG) and head magnetic resonance imaging (MRI) showed normal results, and the patient was discharged from the hospital. At 3 months of age, the patient experienced 2 episodes with brief gaze upward deviation and arm flexion, lasting few seconds, leading to admission to a regional hospital. Three days post-admission, a motor seizure with gaze fixation followed by clonic arm and leg movements lasting 2 minutes occurred and ended after administration of Midazolam. Subsequently, the patient was transferred to the Children's Clinical University Hospital (CCUH) in Riga. On the same day at CCUH, another seizure featuring eye upward deviation, arm flexion, and subsequent asymmetric clonic movements in the limbs was observed, lasting 2 minutes. Treatment with Valproic acid (20 mg/kg/day) was initiated, and the patient was admitted to the intensive care unit (ICU) for further monitoring. Normal results were obtained from EEG performed during this period. Over the next 24 hours, no similar events occurred, and the patient was transferred to a regular ward. However, on the following day, the patient experienced 4 brief events with gaze upward deviation and arm flexion. Video-EEG captured one of these events, revealing no epileptiform brain activity during it. As there was no epileptiform acitivity, these episodes were interpreted as possible oculogyric crisis. Over the next 7 days, the frequency of these events gradually declined until they were no longer observed, also no motor seizures were seen, leading to the patient's discharge from the hospital. At 7 months of age, no similar events recurred, and genetic analysis revealed a mutation in the KCNQ2 gene, consistent with the diagnosis of SeFLNE. The patient continued therapy with valproic acid until 9 months of age, after which it was discontinued as no paroxysmal events repeated and first follow-up EEG was normal. Next EEG was repeated at age of 12 months and continued to show no epileptiform activity. Patient was screened for aromatic l-amino acid decarboxylase (AADC) deficiency and results came back negative. At follow-up at 1 year and 3 months of age, the patient had age appropriate psychomotor development, with no recurrence of motor seizures or presumed oculogyric crises.

Conclusions
This case illustrates a patient with genetically confirmed KCNQ2-related self-limited familial neonatal epilepsy presenting with early-onset motor seizures and presumably oculogyric crises, with a favorable outcome being observed.

KETOGENIC DIET FOR EPILEPSY TREATMENT. EXPERIENCE IN CHILDREN'S CLINICAL UNIVERSITY HOSPITAL, RIGA, LATVIA

Dana Apela [1], **Dace Pretkalniņa** [1], **Jurģis Strautmanis** [1]

1. Children's Clinical University Hospital

Objectives
The aim of this observational study was to assess the efficacy of ketogenic diet for epilepsy treatment in Children's Clinical University Hospital, Riga, Latvia.

Materials and Methods
The data about paediatric epilepsy patients, that had started ketogenic diet prior to 30th April, 2024, were collected retrospectively from medical records and patient diaries. There were 72 patients, including 37 boys. Median age at diet initiation was 3.25 (IQR 1.58; 6.38) years. The seizure outcome was assessed after 6 and 12 months. The patients with at least 50% seizure reduction were called responders. Global impression of improvement was also used as an outcome. Data were gathered in RedCap database. Statistical analysis was performed using R (version 4.1.2) and RStudio software.

Results
The diet is still continued by 21 patients, the median duration of the diet for them is 42.2 (23.2; 65.0) months. The median diet duration for the rest of patients has been 6.0 (3.0; 21.5) months. The diet was used for at least 6 months in 43/72 (60%) patients. At 6 months timepoint, there were 27/43 (63%) responders, i.e., 27/72 (38%) of all treated patients [missing data for 14 patients]. There was an overall impression of improvement in 38/43 (88%) cases at 6 months, i.e., 38/72 (53%) of all treated patients [missing data for 2 patients]. The diet was used for at least 12 months in 35/72 (49%) patients. At this timepoint, there were 17/35 (49%) responders, i.e., 17/72 (24%) of all treated patients [missing data for 16 patients]. There was an overall impression of improvement in 27/35 (77%) cases at 12 months, i.e., 27/72 (38%) of all treated patients [missing data for 2 patients].

Conclusions
There has been quite good proportion of patients with overall improvement. Nevertheless, the number of patients with at least 50% seizure reduction is not so satisfying. It must be noted that the results might be affected by missing data – for many patients only global assessment without precise seizure count was available.

PARENTAL NONADHERENCE TO THEIR CHILDREN'S EPILEPSY TREATMENT PLAN

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Objectives
Parental nonadherence to their child's epilepsy treatment plan may result in reoccurrence of seizures. It may be unintentional, such as healthcare inaccessibility, or intentional, parents not following the treatment plan in fear of side effects, poly-therapy and multiple dosing being too much of a burden.
Our aim was to examine adherence to antiepileptic drugs (AEDs) in a cohort of parents, whose children have epilepsy, and assess the barriers to treatment from their perspective.

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

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

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

PHENOTYPE DIVERSITY IN PATIENTS WITH PRRT2 GENE VARIANT: CASE SERIES REPORT

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Objectives
Pathogenic variants in the PRRT2 (proline-rich transmembrane protein 2) gene have been identified as the main cause of an expanding spectrum of disorders, including paroxysmal kinesigenic dyskinesia and benign familial infantile epilepsy, which places this gene at the border between epilepsy and movement disorders. The clinical spectrum has largely expanded to include episodic ataxia, hemiplegic migraine, and complex neurodevelopmental disorders in cases with biallelic mutations. In this case series report we would like to explore clinical symptoms for patients with PRRT2 gene variant.

Materials and Methods
We report 8 patients (1 male and 7 females) from 4 different families. Each family had one member diagnosed with early onset epilepsy undergo whole exome sequencing with epilepsy gene virtual panel. Other family members were tested by targeted Sanger sequencing. The most common PRRT2 gene pathogenic variant NM_145239.3:c.649del was confirmed for all patients.

Results
6/8 patients had early onset epileptic seizures (median age 5 months) which responded well to standard treatment. 4 patients are on medication at the analysis time and 3 patients were prescribed valproic acid and one patient uses phenytoin. 6/8 of these patients had appropriate development for age. 2/8 patients have autistic traits and intellectual disability with psychiatric disease possibly caused by other reasons. Seizures resolved without treatment in one case. 2 adult patients have symptoms of hemiplegic migraine that started in the teenage years. One patient had seizures as a baby and now involuntary movements were noticed in adulthood.

Conclusions
We confirm that PRRT2 gene variants cause wide range of symptoms: familial benign epilepsy, hemiplegic migraine and movement disorder. Treatment with valproic acid was effective for our patients. It is very important to properly recognize families with possible PRRT2 gene variants to hasten and optimize genetic testing.

SLEEP AND EPILEPSY: A KIND OF A TWO-WAY STREET RELATIONSHIP

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1. Lithuanian University of Health Sciences

Objectives
We aim to describe the reciprocal relationship and its various aspects between sleep and epilepsy.

Materials and Methods
A thorough overview of the available literature about sleep and epilepsy was conducted. The various aspects of the relationship between sleep and epilepsy are illustrated by clinical case examples.

Results
The recently published consensus review of standard procedures for the diagnostic pathway of sleep-related epilepsies and comorbid disorders by the European Academy of Neurology, the European Sleep Research Society, and the International League Against Epilepsy has highlighted sleep-related epilepsies based on how sleep affects epilepsy in regards to clinical symptoms and electroencephalographic findings. On the contrary, epilepsy, along with antiseizure medications, in turn, has various effects on sleep structure, cognitive functioning, and the sleep-wake cycle. It is also more prevalent in people with epilepsy to have comorbid sleep disorders such as parasomnias, sleep-disordered breathing, or insomnia. Poor control of seizures has a negative impact on sleep, while poor sleep, in turn, has negative effects on seizure control, albeit these effects differ in different types of epilepsies. However, current studies often involve mixed samples of patients and are

scarce. Based on current data, it is difficult to provide high-quality recommendations regarding the evaluation of patients with sleep-related epilepsy.

Conclusions
Epilepsy has varying and complex effects on sleep, and vice versa, although the true impact in multiple different aspects is yet to be determined.

THE BENEFICIAL OUTCOME OF SUBSEQUENT TREATMENT WITH ANAKINRA DURING THE CHRONIC PHASE OF FEBRILE INFECTION-RELATED EPILEPSY SYNDROME (FIRES): A CASE REPORT

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1. University of Latvia, **2.** Children's Clinical University Hospital

Objectives
Febrile Infection-Related Epilepsy Syndrome (FIRES) is a rare disorder characterized by refractory status epilepticus following febrile infections, affecting previously healthy children. This study aims to elucidate the limited therapeutic approaches for FIRES and presents a case study of an 8-year-old with intractable epilepsy, emphasizing the favorable response to anakinra during the chronic phase.

Materials and Methods
A comprehensive case report was conducted, detailing the patient's medical history, treatment regimen, and outcomes during anakinra therapy.

Results
Anakinra, initially administered during the acute phase in 2019, was temporarily discontinued after 29 days. In 2022, facing a chronic-phase exacerbation, the patient underwent a second course of anakinra treatment, demonstrating a notable positive impact on seizure activity. Over one year of anakinra therapy, a significant improvement in both seizure frequency and severity was observed.

Conclusions
This report contributes to existing evidence supporting the potential efficacy of anakinra in treating FIRES, particularly during the chronic phase. The presented case underscores the positive outcomes achieved with extended anakinra therapy, suggesting its continued benefits. The findings underscore the importance of further research to solidify anakinra's role in FIRES treatment and optimize its administration during different phases of the disorder.

Genetics

CHARCOT-MARIE-TOOTH DISEASE AND INTELLECTUAL DISABILITY

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Objectives
This study aims to investigate the association between Charcot-Marie-Tooth disease and intellectual disability, focusing on the role of recently identified genes.

Materials and Methods
Literature review was conducted using PubMed (MEDLINE) and Google Scholar databases. Six articles were selected for analysis. Key data points included the age at symptom onset, development impairment, findings in MRI scans, genetic data.

Results
Through literature review, we analyzed the relationship between Charcot-Marie-Tooth disease and intellectual disability. Six papers were included in our initial review. There are several case reports, presenting patients with mild to moderate intellectual impairment and Charcot-Marie-Tooth disease. After performing the whole exome sequencing a few novel genes were identified, for example, MCM3AP. Nerve conduction studies showed that mutations in MCM3AP gene cause sensorimotor axonal neuropathy characterized by early onset and severe symptoms: skeletal deformities, loss of ambulation before adulthood, second most common finding being intellectual disability. Most of the patients had normal MRI scans. We highlight the impact of intellectual disability on patients' quality of life, emphasizing the need for tailored interventions and comprehensive management strategies.

Conclusions
Recognizing the association between Charcot-Marie-Tooth disease and intellectual disability in addition to understanding the role of genetic etiology, is crucial for clinicians to provide holistic care and support for affected individuals. Further research should be carried out to improve outcomes and enhance the well-being of patients with Charcot-Marie Tooth disease and intellectual impairment.

CLINICAL PHENOTYPE OF SCNXA GENE MUTATION: CASE SERIES

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Objectives
Voltage-gated sodium channels (SCNs) gene group consist of 13 gene subgroups. Different genetic variants of four human brain-expressed SCN genes SCN1A/2A/3A/8A have been associated with heterogeneous epilepsy phenotypes and neurodevelopmental disorders.
Objective: In order to get better understanding of clinical phenotype of SCN-related epilepsies we determined and analyzed all pediatric patients with SCNxA mutations attending the Hospital of Lithuanian University of Health Sciences Kauno klinikos (KK).

Materials and Methods
From December 2019 to May 2023, 234 children with epilepsy were referred to genetic testing at KK. Some kind of chromosomal anomaly or gene mutation were found in 107 (45.7%) children. The most frequent monogenic cause of epilepsy was associated with SCNxA mutations. Overall, 11 patients with SCNxA gene mutation were determined: 7 - SCN1A, 2 - SCN2A, 1 - SCN3A, and 1 - SCN8A. Clinical phenotype of these patients was analyzed: onset and type of seizures, epileptic syndrome, neurodevelopment, EEG and MRI findings, response to the treatment.

Results
Six of 7 patients had likely pathogenic SCN1A gene variant, and 1 had variant of uncertain significance (VUS); 6 of 7 SCN1A patients had early onset epilepsy (average age at seizure onset was 5.3 months), in 1 case epilepsy was suspected but not confirmed, though the patient had positive family history of epilepsy. All SCN1A patients except one had febrile seizures (FS), prolonged in 4 cases. Initial EEG was normal in all cases, later with variable non-specific (generalized or focal) abnormalities. Four patients with SCN1A mutation later developed ataxia, 4 - speech delay, 1 - global developmental delay (GDD), 1 was suspected with autism spectrum disorder. Brain MRI of SCN1A cases were unremarkable.
One of 2 patients with SCN2A was born premature with hypoxic-ischemic brain injury and developed seizures on the 2nd day of life, diagnosed with Ohtahara syndrome, later evolving to Lennox-Gastaut syndrome; he also had GDD, spastic tetraplegia, gastrostomy, optic nerve atrophy. This patient had extremely drug-resistant epilepsy, refractory to multiple medications and ketogenic diet, with sodium channel medications being partially effective. In this case, it is difficult to determine the clear impact of genetic mutation and epigenetic factors though the genetic mutation seems to play a role in the course of the disease. Another patient identified with SCN2A VUS developed seizures at age of 33 months which manifested as prolonged FS, further turning to non-febrile focal seizures. Now seizures are infrequent with medication (1 time per year). The patient with SCN3A gene mutation had early onset epilepsy, which manifested with controllable infantile spasms, the patient is seizure-free and medication-free at age 3 years old, though with cognitive disorder. SCN8A patient presented with absence seizures with eyelid myoclonia from 8 years of age. Epilepsy is difficult-to-treat, the patient has memory and attention deficits.

Conclusions
Cases with SCN1A mutation seem to be related to a quite specific recognized phenotype though with variable course and prognosis, while other SCN-related mutations may have different epilepsy phenotypes and the course of the disease. Further experience and multicenter investigations may lead to search for better treatment and comprehensive care options.

DIAGNOSTIC ODYSSEY OF UNEXPLAINED PAROXYSMS: CASE REPORT OF TANGO2 DEFICIENCY

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Objectives

TANGO2 deficiency is a rare autosomal recessive condition, affecting 1:1 000 000 individuals. It is characterized by developmental delay, intellectual disability, gait incoordination, speech difficulties and TANGO2 spells (non-life-threatening paroxysmal neurological episodes, including sudden onset of hypotonia, ataxia with loss of balance, head and body tilt, increased dysarthria, drooling, lethargy and disorientation), as well as acute metabolic crises and risk of cardiac crisis. Additionally, patients may have hypothyroidism, seizures, exotropia and constipation.

Materials and Methods

We report a clinical case of a single patient with typical symptoms and genetic diagnosis of TANGO2 deficiency disorder.

Results

A patient is a 10-year-old female who was referred to geneticist due to unclear neurological disease and hypothyroidism. Her early motor development was normal – she started sitting at 7 months of age, standing at 8 months, walking at 1 years of age. The first paroxysmal neurologic episode appeared at 1 years and 9 months of age, during which loss of balance, incoordination, drooling and speech impairment occurred and lasted about 2 hours. After the first episode, progressing motor and speech difficulties with recurrent paroxysmal neurological episodes were observed. Several MRIs of brain and spine were performed, however no structural abnormalities were found. Several EEG did not show any signs of epileptiformic activity, and the diagnosis of epilepsy was not confirmed. The patient was diagnosed with an unspecified paroxysmal movement disorder. At the age of two, clinical diagnosis of hypothyroidism was confirmed and therapy with L-thyroxine was started. At the age of 5 years and 10 months, after getting sick with several infections, an episode of acute metabolic crisis occurred, with worsening of neurological symptoms and characteristic laboratory findings - elevated liver enzymes and creatine kinase. No cardiac complications were observed. Symptoms progressed over time with increasing frequency and duration of paroxysmal neurological episodes, also with progressing movement and gait problems, accompanied by speech and learning difficulties, short attention span and emotional control deficiency. Despite of the progressing symptoms, the diagnosis was not confirmed until the age of ten, when the whole exome sequencing (WES) performed and revealed a pathogenic homozygote variant NM_152906.7:c.4del, creating a frame shift and early termination codon p.Cys2AlafsTer35. The diagnosis of TANGO2 deficiency (#MIM 616878, ORPHA:480864) was confirmed.

Conclusions

The accurate diagnosis of TANGO2 deficiency allows health care specialists to stop irrelevant investigations and to prevent acute metabolic crises and cardiac complications by vitamin B-complex supplementation; this treatment was suggested to our patient as well. In addition, when the diagnosis is clear, other possible targeted organs may be evaluated before any symptoms occur. A definite genetic diagnosis also allows to evaluate risk of disease recurrence in future pregnancies.

GENOTYPIC AND PHENOTYPIC SPECTRUM OF 956 CASES OF PDHA1-RELATED PYRUVATE DEHYDROGENASE DEFICIENCY

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Objectives

Pathogenic variants in X-linked PDHA1 cause pyruvate dehydrogenase complex (PDHc) deficiency with lactic acidosis and various neurological findings amenable to ketogenic diet and/or thiamine. This study aimed to analyze genotype and phenotype of PDHc deficiency cases related to disease causing PDHA1 variants.

Materials and Methods
The genotypic and phenotypic details of individuals with PDHA1 deficiency were retrospectively collected via literature review or online survey.

Results
956 individuals (480 females) were included, of whom 430 were unpublished. 324 different PDHA1 variants were identified, of which 115 previously unpublished. Many variants were recurrent, with p.R263G (n=72) being the most frequent. Most individuals (612/956) had missense variants. Loss-of-function variants were more common and randomly distributed in females (158/480=33%, OR 4.11, p<0.001), while in males they were mostly restricted to the C-terminal domain. Phenotypically, the three most frequent characteristics were developmental delay (402/466, 86%), muscular hypotonia (352/452, 78%) and intellectual disability (255/324, 79%). Half of the individuals (126/253) achieved independent walking, 39% (86/223) communicated with whole sentences, and 26% (48/186) attended mainstream schools. Common neuroimaging findings included basal ganglia alterations (217/558, 39%), cerebral atrophy (204/558, 37%) and corpus callosum abnormalities (139/558, 25%). Treating physicians considered ketogenic diet successful in 69% (145/210) and thiamine supplementation in 48% (147/309).

Conclusions
Our study is consistent with findings of previous smaller studies, namely the equal male:female ratio of affected individuals, and the common clinical and neuroradiological findings. This study adds new insights on the mutational spectrum of this disorder (including 115 previously unreported variants) and survival rates, and has important counseling implications. Furthermore, the availability of treatment options for this condition underscores the importance of early genetic investigations in children presenting with developmental delay.

INFANTILE APNEA TREATED WITH PARENTERAL VITAMIN B12: A CASE REPORT FROM LITHUANIA

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Objectives
Apnea of prematurity (AOP) is a common presenting symptom of different diseases. Inborn errors of metabolism are a group of disorders that may cause AOP, therefore it is important to search for potentially treatable conditions in order to improve patient outcomes.

Materials and Methods
Here we report a rare case of intracellular cobalamin disorder in an infant treated with parenteral vitamin B12 injections.

Results
Our patient is a 10-month-old boy who presented with hypotonia, apnea and anemia from the neonatal period. He was born at 34 weeks of gestation and these symptoms were explained by prematurity, but they did not improve with time. Metabolic screening showed low methionine level in dried blood spot specimen, but plasma methionine level was within normal range. A very slight elevation of methylmalonic acid in urine was detected. Whole exome sequencing revealed hemizygous variant in HCFC1 c.2795C>T, p.(Ser932Leu), which was classified as a variant of unknown significance.
The patient is a third child to the family. His eldest brother has mild developmental delay and he also had apnea until the age of 5 years. A 35-year-old maternal uncle has mild intellectual disability as well, and he experienced apneic episodes in infancy and early childhood.
The segregation analysis showed that patient's mother is a carrier of the HCFC1 variant. Also, patient's brother and maternal uncle are hemizygous for this variant.
Considering clinical symptoms, metabolic profile and family history, the patient was suspected to have an intracellular cobalamin disorder. After multidisciplinary consultation the treatment with parenteral vitamin B12 (hydroxocobalamin) was initiated. The treatment was started with intramuscular injections of vitamin B12, and apnea disappeared after the first injections. Patient's development improved, his hemoglobin level increased, and biochemical profile became normal. As intramuscular route caused painful injections, we decided to switch to subcutaneous administration of vitamin B12 as a less invasive procedure for daily injections.

Conclusions
This case illustrates the importance of newborn screening for possibly treatable conditions associated with severe apneas. Our patient's case and his family history show that disorders of intracellular cobalamin metabolism have a variable phenotype and age of onset, and most of them are inherited in an autosomal recessive or X-linked manner. As our case demonstrates, accurate diagnosis and specific treatment can prevent life-threatening complications.

MULTIFACETED MITOCHONDRIOPATHIES: A REVIEW OF 3 CLINICAL CASES

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Objectives
Mitochondrial diseases are a diverse set of disorders that arise from abnormalities in the function of mitochondria. These conditions present a wide spectrum of clinical manifestations, which can make diagnosis challenging. Moreover, genetic diagnosis of mitochondrial disease can be complex because genetic variants can be found in mitochondrial DNA or nucleus DNA. In this review, we will explore three clinical cases to illustrate the complexities of mitochondrial diseases.

Materials and Methods
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Results
Case 1. A 15-year-old girl has been experiencing various impairments since childhood, including growth retardation, hypothyroidism, cataracts, optic nerve atrophy, neurosensory deafness, mitral valve insufficiency, cardiomyopathy, chronic kidney disease, delayed sexual maturation, and a lipoma in the brain. Despite treatment, her symptoms have been progressively worsening, with episodes of metabolic acidosis occurring more frequently. Additionally, she has experienced deteriorating gait, tremors, and fatigue. Her mother and grandmother died at a young age. Both of them had kidney failure and hearing impairments. The whole exome sequence (WES) was performed and after additional bioinformatics analysis, a likely pathogenic variant in the MT-ND1 gene m.3761C>A was found in our patient which causes mitochondrial complex I deficiency. This diagnosis explains her symptoms and possibly the premature deaths of her female family members.
Case 2. A 19-month-old boy was first admitted to the hospital due to status epilepticus. His condition worsened over time as he developed encephalopathy, tetraparesis, polyneuropathy, and epilepsia partialis continua. From the onset of the disease, he had mildly elevated transaminases, which later progressed to liver failure. WES confirmed compound heterozygous pathogenic in POLG gene: heterozygous pathogenic NM_002693.3(POLG):c.2243G>C (p.Trp748Ser) variant and heterozygous likely pathogenic variant NM_002693.3(POLG):c.2666C>T (p.Ala889Val). The Alpers-Huttenlocher syndrome diagnosis was confirmed.
Case 3. A 13-year-old boy presents with progressive tetraparesis, cognitive decline, and seizures every few years. Brain MRI findings at 9 years old indicate non-specific changes suggestive of mitochondrial leukoencephalopathies and bilateral optic nerve atrophy. Holter monitoring revealed supraventricular extrasystoles, and he developed hypothyrosis. Genetic testing revealed MT-ND5 gene heteroplasmic pathogenic variant NC_012920.1:m.8344A>G.

Conclusions
Mitochondrial diseases can manifest in a variety of ways, making early diagnosis challenging. Nevertheless, the common thread among these disorders is the impairment of organs sensitive to energy requirements. A holistic approach to patient care and interdisciplinary collaboration is essential to facilitate accurate diagnosis. With the advancement of genetic testing capabilities, it is now possible to diagnose mitochondrial diseases more accurately and provide timely and high-quality patient care.

THE FIRST REGISTERED TYPE 0 SPINAL MUSCULAR ATROPHY PATIENT IN LATVIA: CALL FOR CHANGE IN PRENATAL DIAGNOSTIC PROCEDURES

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Objectives
Spinal Muscular Atrophy type 0 (SMA 0) is a rare genetic disorder characterized by the degeneration of anterior horn cells and motor nuclei in the lower brainstem. This study aims to elucidate the clinical manifestations, genetic factors, and challenges associated with prenatal detection and management of SMA 0, presenting the first registered case in Latvia.

Materials and Methods
Case report.

Results
We present the first registered patient in Latvia with type 0 spinal muscular atrophy (SMA). During the first-trimester ultrasonography of the unborn patient, an increased thickness of the nuchal fold was detected. The mother reported decreased foetal movements during the pregnancy. Following birth, the infant's general condition was extremely severe, indicating a suspected neuromuscular disorder. A precise diagnosis of type 0 SMA was established 7 days after birth through newborn pilot-screening, conducted with parental consent. The infant's condition deteriorated, experiencing severe respiratory distress and multiple events leading to his unfortunate demise.

Conclusions
While existing literature is sparse on the correlation between increased nuchal translucency (NT) measurement and Spinal Muscular Atrophy type 0 (SMA 0) diagnosis in fetuses, this study highlights the clinical significance of elevated NT levels, which may indicate associations with genetic syndromes, fetal malformations, disruptions, and dysplasias. Given the current absence of a cure for infants with SMA 0, early prenatal detection becomes paramount for delivering optimal care, emphasizing the importance of incorporating palliative measures.
The imperative for early prenatal detection is further underscored by the limited treatment options available for SMA 0, emphasizing the necessity for appropriate counseling and palliative care planning. Although increased nuchal translucency (NT) demonstrates clinical relevance in various fetal anomalies, its association with SMA remains confined. The integration of extensive panel genetic testing, such as next-generation sequencing, and sequencing fetal exomes, including SMN1, has the potential to refine diagnostic precision, offering timely counseling to parents confronted with unexpected fetal anomalies. This early recognition provides parents with the essential time needed to make informed decisions, including the option of pregnancy termination.

THE MILDEST DESCRIBED 17P13.3 MICRODELETION SYNDROME: A CASE REPORT

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Objectives
Microdeletions in chromosomal region 17p13.3 are associated with neuronal migration disorders, with PAFAB1H1 being the major gene affected. The genomic imbalances, including the YWHAE and CRK genes, cause more severe structural brain malformations. The spectrum ranges from an isolated lissencephaly sequence to Miller-Dieker syndrome (MDS). Patients carrying only YWHAE and CRK deletions but sparing PAFAH1B1 may have severe growth restriction, neurodevelopmental delay, common craniofacial features, structural brain abnormalities.

Materials and Methods
Case report.

Results
We describe the case of a 2 years and 7-months old girl with 17p13.3 microdeletion syndrome. The patient is a carrier of YWHAE and CRK deletions, but she lacks PAFAH1B. Her current height is 78 cm (>-3SD), weight 8kg (>-3SD). She began walking at 14 months, her gait is consistent with her age. Her communication skills partially correspond to her age.
The first signs appeared right after birth – she had developed stigmas, including, low-set ears, a short neck, wide eye gap. She had an enlarged large fontanel (3 x 3 cm), umbilical hernia, diffuse hypotonia, and a prolonged bleeding episode after biopsy. Hirschsprung's disease was suspected. The first visit to the geneticist was at 2 months of age. Noonan, DiGeorge syndrome, inherited metabolic disorders, were excluded. Later on, whole exome sequencing confirmed 17p13.3 microdeletion syndrome. Although patients with 17p13.3 microdeletion syndrome have relevant structural brain abnormalities in most cases, this case was different. Her MRI findings showed wider liquor spaces in the basal ganglia and slightly wider lateral ventricles. However, no characteristic MRI changes for neuronal migration disorders were found.

Conclusions
We describe a rare case in which a patient with 17p13.3 microdeletion syndrome has severe growth restriction but no characteristic structural brain abnormalities. Thus, our experience shows that confirmed genetic analysis is not always consistent with all described malformations and helps to broaden the phenotype of 17p13.3 microdeletion syndrome.

Neuromuscular diseases

AN EXPANDED ACCESS PROGRAM OF RISDIPLAM FOR PATIENTS WITH TYPE 1 OR 2 SPINAL MUSCULAR ATROPHY IN LITHUANIA

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Objectives
Introduction. In Lithuania, treatment for spinal muscular atrophy (SMA) has been available since August, 2018. However, intrathecal administration of nusinersen is not feasible in all SMA patients. Risdiplam was the first oral medication approved for the treatment of SMA, and it became available in September, 2020, in the Expanded access program (EAP) for SMA

type 1 and 2 patients.
Objectives. To review the results of the EAP of risdiplam for patients with type 1 or 2 SMA in Lithuania.

Materials and Methods
A total of 20 patients (10 adult patients and 10 children) were enrolled and 18 patients completed the EAP at the Center for neuromuscular diseases at the Hospital of LUHS Kauno klinikos. Retrospective review of their medical records was performed.

Results
All ten adult patients were diagnosed with type 2 SMA. Two and eight pediatric patients had type 1 and type 2 SMA, respectively. Ten patients were previously treated with nusinersen injections. The reasons of switching from nusinersen to risdiplam were scoliosis surgery, difficult intrathecal injections requiring sedation, and desire to avoid hospital admission during COVID-19 pandemic.
Ten patients were treatment-naïve before the EAP: two children (one was diagnosed in September, 2020, when risdiplam became available, and another had severe scoliosis) and eight adults (due to scoliosis and severe general condition).
Two adult patients refused to take risdiplam: one prior to the start of the EAP, and one after one year of treatment. The reason for discontinuation of treatment was perceived lack of efficacy.
In general, the drug was well tolerated, and side effects were rare and mild: skin reaction after sun exposure in one pediatric patient and gastrointestinal issues in one adult patient.
After the closure of the EAP in December, 2023, all 18 patients continue to take risdiplam.

Conclusions
Risdiplam is safe and well tolerated, suitable for all SMA types despite the age and functional status of the patient.

BILATERAL IDIOPATHIC CARPAL TUNNEL SYNDROME IN A 5-YEAR-OLD BOY

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Objectives
The objective of this study was to to examine the clinical manifestations, diagnostic procedures, treatment strategies, and results in a case of 5 year old pediatric patient diagnosed with idiopathic bilateral carpal tunnel syndrome (CTS). CTS emerges as the most common entrapment neuropathy, predominantly affecting middle-aged patients, with bilateral symptoms prevalent in a notable proportion of cases. While CTS is a common condition in adulthood, its occurrence in childhood is rare. Notably, mucopolysaccharidoses and wrist trauma are cited as a frequent cause of CTS in children within existing literature.

Materials and Methods
A retrospective analysis of the pediatric case was conducted. Data was collected from medical history encompassing clinical observations, nerve conduction study results, diagnostic imaging results, laboratory findings, genetic testing, treatment regimens, and follow-up assessments.

Results
A 2 years and 10 months old boy presented to a pediatric surgeon with symptoms of pain in his right palm and restricted extension in his right fingers III-V. Ultrasound examination revealed tendinosynovitis of the right wrist joint. At the age of 2 years and 11 months, the patient underwent surgical treatment comprising tendosynovectomy, neurolysis, and median nerve decompression at the right wrist level, resulting in the resolution of pain and finger movement restriction. Histological examination of the surgical specimen revealed nonspecific chronic inflammation with lymphocytic infiltration, without evidence of specific granulomatous inflammation or neoplasia. At the age of 5, the patient returned for outpatient consultation to a Child neurologist, reporting tingling sensations in his left palm extending to fingers II-V after physical activities involving hand grip, particularly following bicycle riding and writing over the past 4 months. Also, pain in the palm after sleep was present. Pain during night was denied. Neurological examination revealed age-appropriate psychomotor development and no other neurological deficits. Nerve conduction studies demonstrated prolonged motor and sensory latencies with signs of axonal loss in the left median nerve at the carpal tunnel level and prolonged sensory latencies in the right hand. Nerve ultrasound revealed slight nerve volume enlargement at the carpal tunnel level bilaterally, with no evidence of synovial tissue changes. Metabolic causes, such as mucopolysaccharidoses, were excluded, and genetic testing for mutations in the PMP22 gene came back negative. Patient had no traumatic wrist injury in anamnesis. The patient commenced nonsurgical treatment with regime adjustment and nighttime wrist bracing, resulting in complete symptom resolution after 3 months.

Conclusions
Carpal tunnel syndrome, traditionally recognized as a condition of adulthood, can also occur among the pediatric demographic. Despite its rarity in this population, pediatric presentations of CTS manifest clinical signs and symptoms akin to those observed in adults. This case study presents an instance of idiopathic bilateral carpal tunnel syndrome, notable for its first onset before the age of three years. While idiopathic CTS is uncommon during childhood, it's important to consider it

in the list of possible causes when evaluating wrist pain in children.

NEWBORN SCREENING FOR SMA: FIRST YEAR IN LITHUANIA

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Objectives
2023 was the year when Spinal Muscular Atrophy (SMA) has been included in Lithuania's newborn screening practice, alongside other rare disorders. SMA is a neuromuscular disorder is characterised by progressive muscle weakness and atrophy. Timely treatment plays a crucial role in maintaining muscle strength and the improving the quality of life. Given the incidence of SMA, which varies from 0.005% to 0.013% of all newborns, and considering the latest birth rates in Lithuania, there should be 1 to 2.6 newborns diagnosed with SMA each year. This is precisely what the updated scope of newborn screening has led to in 2023: two infants with early diagnosed SMA continue their outcome-changing treatment plan and are able to achieve their developmental milestones.
The primary objective of this study is to evaluate the impact of integrating Spinal Muscular Atrophy (SMA) into Lithuania's newborn screening program.

Materials and Methods
We conducted a retrospective analysis of infants identified with SMA through Lithuania's newborn screening program in 2023. Genetic test results and clinical data were collected for each patient. Developmental milestones and outcomes were monitored during follow-up assessments.

Results
Two infants diagnosed with SMA through the newborn screening program were included in the analysis. Both patients exhibited homozygous pathogenic deletions of exon 7 in the SMN1 gene, confirming SMA diagnosis. In both cases, the diagnosis of SMA was established prior to the manifestation of any clinical symptoms indicative of the disease. Treatment with Nusinersen was initiated in both cases, resulting in positive outcomes, and later updated to Risdiplam. Motor and respiratory function remained stable, and developmental progress was within the normal range for both patients.

Conclusions
The findings of this study underscore the importance of early diagnosis and intervention in SMA. Integrating SMA into newborn screening programs enables specialists to diagnose the condition in its subclinical stage. With prompt treatment initiation, clinical symptoms of SMA can be prevented from developing, resulting in improved outcomes and quality of life for affected infants. These results advocate for the ongoing implementation and improvement of comprehensive newborn screening programs for rare disorders such as SMA. Further research is warranted to investigate the long-term outcomes of early-treated SMA patients and to address potential obstacles in making the diagnosis-to-treatment route as efficient as possible.

GENE THERAPY FOR SPINAL MUSCULAR ATROPHY: FIRST EXPERIENCE IN LITHUANIA

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Objectives
To present the first case of spinal muscular atrophy (SMA) treated with gene therapy in Lithuania.

Materials and Methods
Here we report a patient with SMA type 1 who received onasemnogene abeparvovec in Lithuania.

Results
Our patient is a 2,8 year old girl, who was diagnosed with SMA type 1 at the age of 15 weeks. She received the first injection of nusinersen at the age of 18 weeks. Her motor functions were assessed using CHOP INTEND scale and the scores improved from 14/64 before treatment up to 35/64 and 46/64 points after four and five doses of nusinersen, respectively. Nevertheless, her bulbar function was declining. At 7 months, due to respiratory insufficiency and frequent respiratory infections NIV was established. She also had feeding difficulties and failure to thrive, and a G-tube was inserted at the age of 13 months. At the same time the family initiated their own fundraising campaign, attracting a lot of attention from the media and politicians. Finally, with the partial reimbursement from the government of Lithuania, the girl was able to get dosed with onasemnogene abeparvovec at the age of 20 months (body weight was 9,2 kg). After the infusion, blood tests were performed regularly, and the only side effect we observed was elevation of transaminases. According to the protocol, the patient was put on steroids for three months. After one year after gene therapy, the patient can sit independently, she can stand with support. No major changes in bulbar function were noticed so far. Her CHOP INTEND score increased by two points (47/64 points).

Conclusions
To our knowledge, this is the first case of gene therapy for SMA in the Baltic countries. We consider it to be successful, as the patient was almost two years old, pretreated with nusinersen, and the risk of complications was higher than in young patients.

THE FIRST CHARCOT-MARIE-TOOTH DISEASE TYPE 1A PATIENT IN LATVIA WITH PXT3003 TREATMENT, CASE REPORT

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Objectives
Charcot-Marie-Tooth disease type 1 (CMT1) is a peripheral neuropathy, predominantly caused by a duplication of the PMP22 gene, resulting in abnormal levels of PMP22 protein and subsequent myelin production failure. This study examines the therapeutic potential of PXT3003, a novel fixed-dose combination of baclofen, naltrexone, and sorbitol, in improving neuromuscular function in CMT1A patients.

Materials and Methods
Case report

Results
A 12-year-old boy clinically and genetically diagnosed with Charcot-Marie-Tooth disease type 1A (CMT1A) presented with profound motor limitations, necessitating wheelchair use since early childhood. Despite normal cognitive development, delays in motor milestones were evident, and genetic testing confirmed the presence of the characteristic PMP22 gene duplication on chromosome 17. Initiation of PXT3003 therapy demonstrated notable improvements in physical activity without significant adverse effects, despite the patient's limited engagement in physical rehabilitation.

Conclusions
This case underscores the potential benefits of PXT3003 therapy for CMT1A patients. The promising results warrant further research to validate its efficacy and safety in a broader patient population. Developing effective therapies for CMT1A holds significant potential for improving the lives of affected individuals and their families.

Neurorehabilitation

CHILD- ROBOT INTERACTION PATTERNS: A COMPREHENSIVE OVERVIEW

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Objectives
The aim was to investigate characteristics of child-robot interaction (CRI), focusing on differences in robot perception between healthy children (control group CG) and children with neurological diagnoses (ND) and between boys and girls; in addition to describe the specific emotional and behavioral patterns used by children during interact with a social robot.

Materials and Methods
Study was conducted at Tartu University Children's Hospital, involving randomly selected 89 children aged 4-16 yrs. (median age 9 years): 39 CG and 50 ND, 31 girls and 44 boys (14 children had no sex reported). We selected humanoid Pepper as a more companion-like and motivating social robot (height 120 cm, weight 28 kg). Interaction (8-10 min) was examined in 3 ways: therapists' observations, child's own rating of his/her emotional state and by survey based on four socio-cultural concepts.

Results
Data from the observational part confirmed a promising potential of CRI. All children quickly engaged with child-like robot, with a median contact time of 2.0 seconds; 93.8% maintained eye contact during the whole interaction. Children found robot friendly, cheerful, smart and safe and rated their own and Pepper's emotional levels highly. Most of the children smiled (90.6%), nodded (82.8%) and listened with interest (98.4%).
We found that interaction process was influenced by neurological illness. Children with ND had difficulties maintaining long-lasting eye contact and they tended to use non-verbal communication more often ($p<0.05$). While they perceived Pepper more anthropomorphic, [OR 4.1 (0.87-25); $p=0.053$] two-times safer ($p=0.076$), and they assessed own mood

higher (p=0.08) compared to CG.
Boys and girls reported similarly high anthropomorphistic qualities in Pepper. Girls found Pepper more intelligent (p<0.05), made eye contact quicker (p<0.05), and reported Pepper's likability higher than boys (p=0.052). No differences were found in mood assessments between genders, 78.8% of boys and 85.2% of girls assessed their mood as happy or very happy. No difference in contact time between girls and boys (p=0.38) was found. To conclude: Robot Pepper was an attractive and motivating communication partner for both - boys and girls.

Conclusions
The study assessed the characteristic features of CRI via dialogue with humanoid. CRI showed perfect suitability of humanoids in future for pediatric neurorehabilitation: children of all ages, both boys and girls found Pepper safe (particularly children with ND), made contact quickly, used more social skills, and expressed positive mood. Pepper was perceived by the children as a peer which enhanced their emotional and social interaction.The presence of characteristics such as anthropomorphistic design, friendliness and truthfulness helps children to see the social robot as an equal companion, which in turn encourages pleasant conversation similar to human-to-human with active dialogue. Our findings support the idea and suitability of using Pepper in medical applications, for example in social deficit therapy for children with neurological disorders.
Study was funded by Estonian Research Counsil PRG789.
The study was approved by The Research Ethics Committee of the University of Tartu (approval number 302/M-11, 2021).
The authors have no conflicts of interest to declare

SELECTIVE DORSAL RHIZOTOMY IN SPASTIC CEREBRAL PALSY PATIENTS IN LATVIA: A CASE SERIES

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Objectives
Selective dorsal rhizotomy (SDR) is a surgical procedure for treating spasticity in primarily ambulant children with cerebral palsy (CP), which may also lead to improvements in motor function and gait. However, due to lack of studies on long-term effects of SDR in children, controversies remain regarding indications, techniques and outcomes. Extensive rehabilitation is mandatory following SDR with the goal of strengthening muscles and increasing endurance. Aim of this study was to summarise epidemiologic data of SDR patients in Latvia and analyse improvement of motor function 6, 12 and 18 months post-operatively.

Materials and Methods
A retrospective study was conducted and included 17 patients from Children's Clinical University Hospital (CCUH) diagnosed with spastic CP who had SDR surgery between 2019 and 2023. Information about motor function evaluation pre- and post-operatively (post-op) was collected from medical history data system.

Results
SDR was performed in 17 patients. 64.7% (n=11) patients were male. All patients were diagnosed with spastic CP: majority 76.5% (n=13) had lower extremity paraplegia, 17.7% (n=3) had tetraplegia, 5.9% (n=1) had hemiplegia. Mean age of patients at time of operation was 7.1 ± 2.3 years. Pre-operatively, all patients were evaluated using the GMFCS and most 52.9% (n=9) were level III, followed by level II in 35.3% (n=6) cases, and levels I and IV 5.9% (n=1) each. Modified Ashworth Scale (MAS) and Gross motor function 88-item measure (GMFM-88) were most commonly used to assess spasticity and motor function pre-operatively in 88.2% (n=15) and 82.3% (n=14) patients respectively. Nearly half patients 47% (n=8) were also assessed using Selective control assessment of the lower extremity score (SCALE) and Amsterdam gait classification system was utilized in a third of patients 35.3% (n=6).
Within 18 months post-op, majority of patients 52.9% (n=9) received rehabilitation exclusively in a non-affiliated outpatient rehabilitation center; less than half patients 41.2% (n=7) had rehabilitation at CCUH. No post-op rehabilitation data is available for one patient. After SDR, one patient was diagnosed with hereditary spastic paraplegia 3A with a mutation in the ATL1 gene.
At 6 months post-op, a third 35.3% (n=6) patients were evaluated at CCUH using GFMCS: no change in level (n=4), increase from level II to level III (n=1) and decrease from level II to level I (n=1). At 12 months post-op 17.7% (n=3) patients were evaluated using GFMCS: no change in level (n=2), increase from level II to level III (n=1). At 18 months post-op, 11.8% (n=2) patients underwent evaluation using GFMCS and both did not have change in level. MAS, GMFM-88, SCALE and Amsterdam gait classification system were used in evaluation irregularly, therefore data could not be analyzed.

Conclusions
We report a single center study including 17 patients with spastic CP receiving SDR surgery from 2019 till 2023. Although nearly all patients received post-op rehabilitation, less than half underwent rehabilitation at CCUH. In those patients who were evaluated post-op at CCUH, GFMCS did not improve in most patients 6, 12 and 18 months after surgery. At this time,

data is insufficient to make certain conclusions on the efficacy of SDR. Limitations of our study include lack of follow-up for patients after SDR, which needs to be further investigated. Future directions could include analysis of subjective improvements of parent and child quality of life before and after SDR using validated scales.

SOCIAL ROBOTS AS COMPANIONS IN REDUCING CHILDREN'S HOSPITAL FEAR AND PAIN

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Objectives

Young patients who undergo venipuncture often experience high levels of fear and pain. Hospital fear (HF) is a critical modifier of patient compliance and medical care quality. Distraction techniques are considered one of the most effective non-pharmacological methods in reducing HF and pain. The aim of the study was to investigate the intervention effect of robot Pepper on children's HF and pain during blood draw.

Materials and Methods

The study, conducted at Tartu University Hospital's Children's Clinic from February to April 2024, involved 27 randomly selected outpatients aged 2-13 yrs (median 8.0 yrs; SD 3.3), including 12 boys and 15 girls. Child-like robot Pepper was used to reduce children's HF through talking, showing videos, singing, playing music and dancing during the procedure. Nurses examined the child's (under the age of 7) pain level before and during the interaction using FLACC scale. Children over 7 yrs HF and pain level was assessed by the VAS scale. The scale consisted of 5 different emotions and a parallel numeric scale: very happy (0-1), happy (1-3), neutral (4-6), sad (7-8) and very sad (9-10). Parents completed questionnaires with Likert scales about their child's experience.

Results

According to the VAS analysis, 86.6% of the children over 7 yrs felt little or no fear (0-3 point out of 10) during the procedure, 27.3% under 7 yrs felt severe HF (7-10 points of 10), 36.4% little or no fear (0-3p/10). 66.7% of the older children rated Pepper's likability highly/very highly, 33.3% neutral (4-6p/10), younger children 100.0% rated Pepper highly/very highly. The mean reduction of pain by FLACC among 12 children under 7 yrs (mean age 4.67, SD 1.50) was 1.5 points (95% CI: 0.0-3.0) therefore, after using the robot, the children's pain scores decreased significantly ($p=0.049$). Parents evaluated their children's HF before the intervention with a mean score of 4.4 points out of 10, after the intervention 2.9 points; the mean reduction of reported fear level was 1.5 points (95% CI: 0.32-2.68, Paired Samples T-Test, $p<0.015$). Satisfaction with robot Pepper among parents was evaluated highly - 76.0% strongly agreed, 8.0% agreed and 16.0% partially agreed. In addition, Pepper was requested by the parents for the next procedure by 88.0% (strongly agreed/agreed).

Conclusions

We found that robot Pepper increased children's comfort and cooperation during blood sampling. Pepper was an effective distraction tool in pain and HF management in children of different ages, being most effective under the age of 7. Comparing these results to the study "Characteristics of Pediatric Hospital Fear and Efficiency of New Distraction Technique Holographic Display for Reducing Fear and Pain in Children" (2022), we found that both technical intervention tools lowered pain and fear scores. Feedback from children and parents supports the use of Pepper as a positive companion during hospital stays and procedures.

The authors have no conflicts of interest to declare

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Ethical approval number 302/M-11, 2021

Others

DOES TREATMENT WITH METHYLPHENIDATE CORRELATE WITH TREMOR SEVERITY FOR PATIENT WITH POSSIBLY GENETIC DYSTONIA: CASE REPORT

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Objectives
Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive movements and/or postures. Dystonic movements are typically patterned and twisting, and may be associated with tremor. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation. Most forms of dystonia tend to worsen initially. There are various types of dystonia and one of isoleted dystonia type caused by ANO3 gene variants.

Materials and Methods
We report a teenage patient case with progressive tremor and dysarthria treated with methylphenidate.

Results
Our patient is a 17-year-old female, the main complaints being progressive hand and head shaking. The tremor is more prominent after physical and emotional exertion. Progressive dysarthria was also observed during the recent years, starting from 10 years of age. The patient's mother had intellectual disability and died while giving birth to the patient's sister. The patient has 4 sisters with developmental problems and/or intellectual disability. No tremors or other movement disorders have been documented in the family. The patient lives in a social community home and is under the supervision of a child and adolescent psychiatrist due to her intellectual disability, schooling difficulties, attention-deficit/hyperactivity disorder (ADHD) and hair self-picking from 10 years of age. She has also been diagnosed with ulcerative colitis and is taking mesalasine for the treatment. Hand tremor has been progressing during follow-up time: the patient experiences difficulties in writing or handling a fork and a spoon, she also drops and spills out food while eating. The girl's intellectual abilities (IQ) were assessed with Wechsler Intelligence Scale for Children–III (WISC-III) twice in 5-year period: verbal IQ showed increasing scores (70 --> 82), and nonverbal IQ showed regression (80 --> 73), total IQ remaining quite stable (72 --> 76). Neurological examination showed unvoluntary tongue movements, hyperreflexia in lower limbs and mild tremor in hands. Muscle tone was found to be normal so far. No structural
Whole exome sequencing (WES) was performed and heterozygous variant of unknown significance NM_031418.4(ANO3):c.[1943A>G];[1943=], p.Asn648Ser was found. Unfortunately, it was not possible to do segregation analysis.
The patient was prescribed methylphenidate XL 18 mg/d for ADHD, and improvement of tremor was noticed. Unfortunately, due to elongated QTc interval on ECG, now methylphenidate treatment is contraindicated for this patient.

Conclusions
The detected variant of ANO3 gene may be the cause the patient's dystonia phenotype. Further follow-up of the patient is planned. We hypothesize that methylphenidate possibly improved dystonia symptoms, but more studies are needed. We are also seeking for other treating modalities to control the symptoms.

PEDIATRIC VISUAL SNOW SYNDROME: A CASE SERIES

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Objectives
Visual snow syndrome (VSS) is a neurological (and neuro-ophthalmologic) disorder characterized by persistent positive visual disturbances. The proposed criteria consist of visual snow defined as constant flickering dots in the entire visual field and the presence of some additional visual phenomena. The disease is most likely related to dysfunctional central visual processing and/or hyperexcitability of the visual cortex. It has high comorbidity and shares several common features with migraine. Evidence for efficient pharmacological treatment remains insufficient. Although VSS is increasingly diagnosed by neurologists and ophthalmologists, it remains underrecognized and rarely described in pediatric patients. The objective of this presentation is to review the characteristics of VSS and to emphasize the importance of its recognition in children.

Materials and Methods
Analysis of case series.

Results
We present four patients – three boys and a girl who started to have or realized having VSS symptoms at different ages. All four complained of monochromatic and constant visual snow as well as other persistent visual phenomena such as bright flashes, colorful shapes, strong blue field entopic phenomenon, palinopsia, nyctalopia, and photophobia. One patient had migraine with visual and sensory aura. Ophthalmological and radiological findings were normal in all. Centro-temporo-parietal spikes on EEG without clinical seizures were observed in the girl, whose mother has VSS as well. Treatment with sulthiame markedly reduced epileptiform activity but had only a minor effect on some visual phenomena. Lamotrigine and acetazolamide attempted in two patients was without obvious effect. One patient chose not to receive a pharmacological treatment. Impact on daily and academic activities ranged from merely perceptible to bothersome. Recognition of the diagnosis of VSS enabled two patients to reduce anxiety and to better cope with their symptoms.

Conclusions
A detailed medical history and appropriate additional exams have to be performed in patients with persistent visual phenomena. Subjective origin of the symptoms, difficulty in depicting them in younger patients, and lack of awareness about VSS can mislead diagnosis and lead to unnecessary investigations and treatments. Reassuring and sharing the knowledge about this benign condition may provide efficient relief to both – patients and family members.

SUBACUTE SCLEROSING PANENCEPHALITIS OUTCOMES BASED ON TREATMENT INITIATION STAGE: A SYSTEMATIC LITERATURE REVIEW

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Objectives
Analyze subacute sclerosing panencephalitis (SSPE) outcomes (improvement, stabilization, progression) based on treatment initiation stage (early (I-II), advanced (III-IV)) and different treatment types.

Materials and Methods
A systematic PubMed search was conducted using keywords "SSPE", "Subacute Sclerosing Panencephalitis", filters "English", "Full text", "Abstract", time frame: 1994-2024. Inclusion criteria: specified SSPE treatments, stages before treatment, treatment outcomes. Exclusion criteria: literature reviews, immunocompromised or pregnant patients, animal or in vitro studies, unrelated, published before 1994, unfinished studies. 28 articles, including 80 cases (52 in st. I-II, 28 in III-IV) were analysed. Cases were classified by stage before treatment (I-II, III-IV) and treatment outcomes (improvement, stabilization, progression) to assess treatment outcomes in different stages. Classifying cases by different treatment types reflected diverse medication combinations most commonly used for SSPE and their efficacy individually. We compared outcomes in stages I-II and III-IV to assess overall outcomes based on SSPE severity. Additionally, we compared progression rates across treatment groups in both early and advanced stages to assess effectiveness of each treatment.

Results
Treatment outcomes and progression rates in SSPE patients varied between early and advanced stages. Fisher's exact test, conducted on all patients regardless of treatment type, revealed a statistically significant association ($p = 0.02728$) between disease stages (early and advanced) and treatment outcomes (improvement, stabilisation, progression). The most effective early-stage treatments, such as ribavirin and lamivudine in combination with isoprinosine and interferon-α (IFN-α), as well as IFN-α/β alone, showed moderate effectiveness. Similarly, treatment outcomes varied among patients in advanced stages, with interferon-α and isoprinosine showing the most promising results. Isoprinosine monotherapy exhibited high progression rates across all stages.

Conclusions
There is significant association between SSPE stages before treatment and treatment outcomes. Although starting treatment in early stages does not guarantee improved outcomes, tailored treatment approaches based on disease stage are important. Treatment efficacy varied depending on the disease stage, with certain treatments showing promising effectiveness in early stages and others - in advanced stages. Since there is no definitive cure for SSPE, disease-modifying therapies play an important role in improving outcomes. Furthermore, measles vaccinations remain crucial in preventing SSPE development.

VISUAL IMPAIRMENT IN THE OUTCOME OF STROKES IN CHILDREN

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Objectives
to trace the types and dynamics of visual impairments in children in stroke outcomes

Materials and Methods
By visual impairment we meant visual field limitations acquired after a stroke, decreased vision, amaurosis and strabismus. We also determined the presence of visual impairments 1 year and 3 years after the stroke to identify the dynamics of these impairments. A year later, the children were examined, which amounted to a total of 346 patients, of which visual impairment was detected in a total of 178 (49.1%). Almost every second child had visual impairments of one kind or another. 178 children who had suffered ischemic (IS) and hemorrhagic (HS) strokes were examined. In the children we observed, visual acuity and visual fields were checked, and the presence of strabismus (secondary) in the dynamics after a stroke was taken into account. An analysis of the dynamics of visual impairments was also carried out after 3 years, which revealed a slight increase in visual impairments, which amounted to 195 (57.1%) people.

Results

more than half of children who have suffered an ischemic stroke have visual impairment as a result (51.5%); children with visual impairment as a result of hemorrhagic stroke make up a slightly smaller number, but quite high 46.3%; in ischemic stroke with hemorrhagic transformation, considered the most severe, children with visual impairment accounted for 48.2%. We also analyzed specific types of visual impairment depending on the type and duration of the stroke. Thus, out of 178 children with visual impairments (identified 1 year after the stroke), secondary strabismus occurred in 107 cases, which amounted to 60% of the total number of children with visual impairments. Next in frequency of occurrence, we noted myopia to varying degrees (33.7%), visual field defects deserved attention in 20.78% of cases. Myopia occurred in 65 children 3 years after the stroke. Observations have shown that the dynamics of visual impairments are most marked in children with perinatal stroke and early childhood stroke compared with strokes in older children.

Conclusions

Thus, visual impairment after stroke in children is an important but poorly studied area in pediatric neuro-ophthalmology. Based on the literature describing the adult population, we should recommend that children who have suffered a stroke undergo a functional assessment of the state of the visual analyzer at least once a year, followed by appropriate therapeutic correction of the identified disorders.

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