subjects could sit unassisted. At 24 months after gene therapy, 64% of subjects achieved head control, 50% could sit unassisted, and 18% could stand with support. At 60 months after gene therapy, 75% of subjects achieved head control, 67% of subjects could sit unassisted, 25% of subjects could stand with support, and 8% subjects could walk with support. In contrast, only 4% of the subjects in the NHDB control group achieved key motor milestones ($P<0.0001$ ). These results demonstrate that the attainment of milestones is sequential in nature and that the percentage of patients achieving more advanced milestones (eg, standing with support, walking with assistance) increases over time.

The number of patients achieving full head control and sitting unassisted was significantly higher in patients treated with eladocagene exuparvovec compared to the NHDB control group. The results indicate that patients with AADC deficiency treated with eladocagene exuparvovec show significant improvements in achieving motor milestones, impacting the natural history of disease.

## 391 FEBRILE CONVULSIONS AND INFLUENZA A OR B- ARE THERE DIFFERENCES?

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10.1136/archdischild-2021-europaediatrics.391

Influenza viruses may cause predominantly respiratory illness, but could also be responsible for numerous neurologic symptoms and complications. All children who are admitted in hospital with symptoms and changes in their neurological status, especially during the flu season, should be tested even if their respiratory problems are mild or non-existent. The most common neurological complications are febrile convulsions.

In this study, we retrospectively reviewed patients with febrile seizures over two periods from 2008-2011. and 2018.-2020. during a flu season (November-March) who had a proven influenza A or B virus and who were hospitalized at the Department of Neuropediatrics of the UHC Sestre milosrdnice. We aimed to find out if there were differences in the type of influenza depending on the period and what features of febrile convulsions were associated with influenza type A or type B and to emphasize the need for optimized prevention.

In the period 2008-2011. at our Department there were 480 patients hospitalized, 99 of them with febrile seizures. In the period 2017.-2020.

(end of February) there were 659 patients, 102 of whom with febrile seizures. The first period had 37 influenza positive patients (15 patients influenza type A, 5 influenza type B, 7 unknown) and the second period had 44 patients with proven influenza (36 patients had influenza type A, 8 patients had influenza type B).

In hospitalized children during both periods, influenza type A was associated with a higher incidence of febrile seizures than influenza type B. The reason could be that influenza type A is more neurotropic than influenza type B and more often causes febrile seizures. Our finding of similar incidence of febrile seizures during both periods can be explained by continued poor prevention and fear of vaccination. This study indicates that there is a need to raise awareness of better prevention of influenza virus transmission.

## 392 HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSY

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10.1136/archdischild-2021-europaediatrics.392

**A Case Report** I.Šeparović[1], M.Kukuruzović[1], M.Malenica[1] 1.-UHC Sestre milosrdnice, Zagreb, Croatia Hereditary neuropathy with liability to pressure palsies (HNPP) is a rare, autosomal dominant disease, with prevalence estimated between 0.84 and 16 per 100,000, affecting peripheral myelin, which manifests as recurrent, and usually transient, painless motor and/or sensory neuropathies. Neuropathies are triggered by minor trauma or repetitive movements with compression and traction of peripheral nerves. The most frequent forms of presentation affect the peroneal and the ulnar nerves. Brachial plexopathy occurs in 11%-20% and is an unusual clinical manifestation; bilateral presentation is even rarer. Genetic tests are available to aid in diagnosis as molecular analysis has identified a deletion in the chromosome 17p11.2 in the majority of these patients. The deletion of chromosome 17p11.2 in hereditary neuropathy with liability to pressure palsies appears to be the reciprocal meiosis product of the 17p11.2 duplication seen in Charcot-Marie-Tooth disease Type 1a. We present the clinical case of a 16-year-old boy, football player. It was his first episode, HNPP presenting with foot drop and palsy both nerves peroneus after distortion articulation talocruralis.

Electromyography studies showed subacute axonal lesions of both nerves peroneus. The diagnosis of hereditary neuropathy with liability to pressure palsy was confirmed by PMP22 deletion of chromosome 17p11.2. He started motor rehabilitation and avoidance of stressing factors with progressive recovery. These palsies generally resolve without surgical intervention but orthopedist decided to make decompression of nerve. The recovery followed quickly. After one-year follow up, he was completely asymptomatic. A case report is presented of a patient with this disorder to promote awareness and recognition that this entity should be considered in patients with multiple nerve palsies.

## 393 NEUROLOGICAL PRESENTATION OF WILSON'S DISEASE IN A PEDIATRIC PATIENT WITH SILENT CIRRHOSIS

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10.1136/archdischild-2021-europaediatrics.393

A previously healthy 13-year-old girl with a 5-month-history of hypersalivation, dysarthria, tremor, thrombocytopenia, and leukopenia was admitted to our hospital. On examination we noticed hypersalivation with an incomplete closing of the mouth, dysarthria, splenomegaly, resting and action tremor of the upper extremities, and slightly weakened hand grip. Jaundice, palmar erythema, or spider-like nevus were not present. Her body mass index was in the 1st percentile (Z-score -2.21). Magnetic resonance (MRI) of the brain showed abnormal T2 hyperintensity in the basal ganglia, mesencephalon, and pons. Abdominal ultrasound indicated diffuse changes in liver parenchyma with circular edges, regenerative nodes, splenomegaly, and suspected portal hypertension, without ascites. Fibrosis was confirmed by liver fibroscan and abdominal MRI, which corresponded to laboratory findings (lower prothrombin

time, levels of coagulation factors and albumin, bicytopenia, but with normal ALT and bilirubin; only AST and GGT were minimally above the upper normal limit). Esophagogastroduodenoscopy revealed esophageal varices grade I and portal gastropathy due to portal hypertension. Kayser Fleischer ring was present. Low ceruloplasmin levels and positive penicillamine test further confirmed the suspicion for Wilson's disease which was confirmed by genetic testing that showed homozygous H1069Q mutation. Once the diagnosis was established, we gathered a multidisciplinary team which included neurologist, gastroenterologist, hematologist, cardiologist, nephrologist, rheumatologist, endocrinologist, dietitian, and psychologist. There were no signs of renal tubular damage and the heart was structurally healthy.

Penicillamine was gradually introduced, but not to the maximum dose recommended by the European Society for Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN). One week later zinc acetate was added into the therapy. We have chosen this scheme because of the risk of hematological complications at the penicillamine full dose. Vitamin D and calcium supplementation was introduced due to reduced bone density. Other supportive therapy included a copper-free diet, high-energy oral nutritional supplement adjusted for patients with liver disease, MCT oil, and gastroprotection. Two months after initiation of therapy cupriuria is threefold increased as compared to the initial values suggesting efficacy of therapy. She has not had side effects with this combination therapy.

Although the most common presentation of Wilson's disease in childhood includes liver disease, we should be aware of its possibility to present with neurological symptoms without obvious clinical signs of liver disease, despite the existence of cirrhosis. A multidisciplinary team is required to monitor possible complications of the disease, side effects of the therapy and offer psychological support to the patient and their family.

## 394 AUDIT ON FIRST PAEDIATRIC ASSESSMENT OF CHILDREN REFERRED WITH SUSPECTED EPILEPSY BEFORE AND DURING PANDEMIC

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10.1136/archdischild-2021-europaediatrics.394

Diagnosing epilepsy can be complex, and prone to be misdiagnosed between 5–30% of the time. It is therefore important to have specialist review early in all cases of suspected epileptic seizures to facilitate subsequent care and management, as well as to reduce parental anxiety. However, Covid-19 pandemic has added extra challenge for healthcare providers to achieve and maintain this standard of practice.

The objectives are;

1) To audit health care for children with suspected epilepsy against NICE recommendation; NICE guideline recommends all children and young people presenting with a suspected epileptic seizure to be seen by a specialist in the diagnosis and management of the epilepsies within 2 weeks of presentation.

2)To determine the effect of the pandemic on the number of referrals. 3)To look at the outcomes following first assessment for suspected epileptic seizures.

1) Identification of 2 cohorts of children presenting to outpatient service for suspected seizure between March-May 2019 and March – May 2020.

2) Retrospective case notes analysis following first paediatric assessment.

1) Number of referrals declined by more than 20% during pandemic, especially from general practitioners.

2) In 2019 cohort (pre-pandemic), 55% of the cases were seen within 14 days of referral compared to 42% in 2020 cohort ( during pandemic).

3) More than half of the referrals were diagnosed as non epileptic events after specialist review. However, the outcome was better in 2020 cohort compared to the previous year.

1) The pandemic is likely to have contributed to the decline in number of referrals and resulted in more delays to clinic appointments due to limited clinic slots imposed by the pandemic restrictions.

2) Local measures to enhance referral pathway to ensure suspected epileptic seizure cases to be seen or assessed within 14 days as per guideline;

– Clear signpost to secretaries for clinic allocations.

– Creating a group email for epilepsy team as one of the pathways for referral. This will make correspondence easier for both ends and aides in filtering process as well as expediting clinic appointment.

– Encourage a phone triage in cases where the diagnosis of epileptic event isn't obvious.

3) Liaise with IT department to add a few prompts for filtering and checklists before providing the option of 'first seizure clinic' when electronic referral is made. This is meant to facilitate in obtaining relevant information, referral checklists prior to appointment and to ensure referrals are allocated to the right clinic.

3) This audit can be used as a feedback tool for the local healthcare providers both in term of referral outcomes and raising awareness on first seizure referral.

## 395 SEVERE NEUROLOGICAL SYMPTOMS IN A 7.5-MONTH-OLD GIRL WITH MEGALOBLASTIC ANAEMIA AND METHYLMALONIC ACIDURIA – CASE REPORT

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10.1136/archdischild-2021-europaediatrics.395

The case report focuses on a 7,5-month-old girl, who was admitted to our hospital becouse of vomiting, failure to thrive, pathological somnolence and developmental regression. The girl was exclusively breastfed and mother tried to introduce new foods many times with failure. Routine and commonly used laboratory tests showed megaloblastic anaemia and vitamin B12 deficiency. Further investigation revealed methylomalonic aciduria and elevated levels of homocysteine and lactic acid, which provides additional evidence of a functional measure of intracellular B12 levels. After starting vitamin B12 supplementation, a significant improvement in the clinical condition was observed and all symptoms gradually disappeared. Further treatment included supplementation of liposomal vitamin B12, folic acid and