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. 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.

In a pediatric patient with silent cirrhosis

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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 -3.0). 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 nodules, 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.

2) Retrospective case notes analysis following first paediatric assessment.
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.
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.
4) 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.

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

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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.

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

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The case report focuses on a 7.5-month-old girl, who was admitted to our hospital because 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 methylmalonic 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...