time, levels of coagulation factors and albumin, bictopenia, 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 gas-
troathy 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
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 expedit-
ing 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 fil-
tering 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 terms of referral outcomes and
raising awareness on first seizure referral.

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**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 misdiag-
nosed between 5–30% of the time. It is therefore important
to have specialist review early in all cases of suspected epilep-
tic seizures to facilitate subsequent care and management, as
well as to reduce parental anxiety. However, Covid-19 pan-
demic 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 epi-
leptic 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 assess-
ment for suspected epileptic seizures.

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**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 regres-
sion. The girl was exclusively breastfed and mother tried to
introduce new foods many times with failure. Routine and
commonly used laboratory tests showed megaloblastic anae-
mia and vitamin B12 deficiency. Further investigation
revealed methylmalonic aciduria and elevated levels of
homocysteine and lactic acid, which provides additional evi-
dence 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
vitamin B6. The control follow – up at 14 months of age showed normal psychomotor development.

Severe infantile vitamin B12 deficiency is rare and usually due to maternal nutritional deficiency (vegetarian and vegan diet) or malabsorption (gastric resection, pernicious anaemia, Crohn’s disease, celiac disease and other).

In this case the origin of mother’s normochromic and normocytic anaemia during pregnancy was not diagnosed and as usual, she only was supplemented with folic acid and iron. In conclusion possible mechanisms of the influence of vitamin B12 metabolism on the nervous system based on the literature data are presented. The aim of this case is to draw attention to the importance of adequate supplementation to prevent potentially irreversible neurologic damage.

## Frontal Lobe Epilepsy with Gelastic Seizures

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Gelastic epilepsy are an uncommon seizure type most often symptomatic associated with hypothalamic hamartomas, with a prevalence rate of about 0,5 per 100 000. However, idiopathic and cryptogenic cases with no evidence of cortical structural lesions have also been described. The term gelastic comes from Greek word gelos meaning laughter. Laughter is pathological in nature and can be spontaneous without obvious cause.

We present a 13-year-old girl without structural lesions, manifesting gelastic seizures. The seizures were not associated with feelings of mirth.

Repeated 1,5T MRI revealed no structural abnormality. Interictal EEG showed paroxysms in the right frontal region. Ictal EEG demonstrated diffuse attenuation, followed by fast activities and spike-wave complexes predominantly over the right hemisphere. Unlike the seizure from temporal lobe, semiological investigations revealed that the laughter in our case was not accompanied by a subjective feeling of mirth, and an interictal EEG showed frontal spikes. Results of neurological examination were normal. The Wechsler intelligence scale for children IV (WISC-IV) revealed a mild mental retardation with the intelligence quotient score <70. An interictal FDG-PET CT showed hypometabolism over the right superior frontal lobe and right medial temporal lobe. The seizures were resistant to oxcarbazepine, levetiracetam, valproate and lamotrigine and were suppressed by topiramate monotherapy. Further clinical examinations (high-resolution 3T MRI, ictal and interictal SPECT) will be done.

In conclusion, gelastic seizures are commonly associated with hypothalamic hamartomas but ictal laughing, has been reported with temporal lobe and frontal lobe seizures. Although gelastic seizures have been described as intractable, a few medications including valproic acid, lamotrigine, levetiracetam and steroids were reported to be effective in patients without hypothalamic hamartoma. In ours patient topiramate monotherapy proved to be effective in the treatment of gelastic seizures without hypothalamic hamartoma.