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). Esophagastroduodenoscopy revealed esophageal varices grade 1 and portal gas- tropathy 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, dietician, and psychologist. There were no signs of renal tubular damage and the heart was structurally healthy.

Penicillamine was gradually introduced, but not to the max- imum dose recommended by the European Society for Paedi- atric 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 dis- ease in childhood includes liver disease, we should be aware of its possibility to present with neurological symptoms with- out 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 ther- apy and offer psychological support to the patient and their family.

2) Retrospective case notes analysis following first paediatri- cian assessment.
3) 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 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 seiz- ure 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.

### Audits

#### 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.

#### Severe Neurological Symptoms in a 7.5-Month-Old Girl with Megaloblastic Anaemia and Methylymalonic 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 methylylmalonic 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
Frontal Lobe Epilepsy with Gelastic Seizures

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Gelastic epilepsy is 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. Intercital 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 our patient topiramate monotherapy proved to be effective in the treatment of gelastic seizures without hypothalamic hamartoma.

Long-Term Monitoring and Outcome of High Neuromisk Preterm Infants

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Perinatal brain damage is non-progressive and the processes of maturation and plasticity along with medical procedures can lead to functional recovery. About 3% of newborns are high neuromisk and especially susceptible to brain damage.

The aim of this study is to show the neurodevelopmental outcomes of 77 preterm infants born ≤32 nd week of gestation, at the age of nine.

We studied and habilitated 170 high neuromisk children born in Zagreb’s largest maternity hospital between 2007 and 2008. At the age of nine, 132 children have remained continuously studied, 77 of whom were preterm infants born ≤32 nd week of gestation. All the children were assessed using the Touwen examination. Children with CP were classified according to motor function classifications. Furthermore, all children have undergone EEG, early neonatal brain ultrasound, whereas children with CP had MRI as well.

Infants were classified as ‘high risk’ according to factors of risk.

At the age of nine years, out of 77, 17 children (22.1%) developed CP. As for associated disabilities, 6 children (35.3%) had an IQ below 70, severe speech impairment had 8/17 (47%), visual impairment 14/17 (82.4%) and 4/17 (23.5%) severe sight loss. 12/17 (70.6%) had refractive error and strabismus. Four children (23.5%) developed epilepsy. Brain ultrasound showed the most severe forms of periventricular leukomalacia for 8 children, another eight children had the most severe forms of peri – intraventricular haemorrhage, while only one child had no abnormality detected. MRI was performed with 13 children, 12 of which suffered from predominant white matter damage, and one of them had additional thalamic haemorrhage. A normal motor outcome was observed in 60/77 children (78%). Of the children with normal motor outcome, one third had refractive error or strabismus, 25/77 (32%) suffered from specific language development disorders, while a almost a quarter had pathological EEG.

High neuromisk children are particularly susceptible to brain damage in the pre- and perinatal period. Because of that, they need to be detected early, their development needs to be monitored even at school age and they must be included in habilitation programmes. The process of selecting, registering and habilitating children is simple and easily applicable. For that reason, it should be carried out with all preterm infants.

Long-Term Outcome of Epilepti Seizures and Adaptive Behavior in Patients with West Syndrome - Experiences from Our Center

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Abstracts