P.071

Case report: cheiro-oral syndrome secondary to thalamic ischemic stroke

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Background: Patient with small thalamic infarct may present with a variety of subtle and seemingly disconnected sensory deficits which can be easily missed. Methods: Here we report a case of an 80 years 'old Chinese female presenting sudden onset persistent sensory symptoms over perioral area and right hand. The patient underwent further investigation with Brain MRI. The patient underwent further investigation with Brain MRI. A literature review was conducted and the patient's clinical finding was compared with the literature. Results: The patient's presentation was consistent with clinical manifestation of type 1 Cheiro-Oral syndrome. Brain MRI performed the day after admission revealed small non-hemorrhagic infarction involving the left thalamus. The diagnosis was type 1 cheiro-oral syndrome secondary to left thalamic ischemic stroke. Conclusions: This report highlights both clinical presentations of cheiro-oral syndrome and correlating clinical symptomatology with anatomic localization. It is important for physicians to recognize this rare condition with subtle presentations for complete work-up and definitive treatment.

Child Neurology (CACN) Epilepsy and EEG

P.073

Diagnostic utility of specific abnormal EEG patterns in children

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Background: CTS and PPR are common EEG findings that are classically associated with CECTS and GGE respectively. PPD and sleep spindles are physiologic phenomenon that occur in respond to intermittent photic stimulation and sleep respectively. Methods: We reviewed EEG studies with CTS, PPR, asymmetric PPD, or asymmetric sleep spindles. For CTS, we determined sensitivity, specificity, PPV and NPV for a diagnosis of CECTS. For PPR, we determined the same diagnostic outcome measures for a diagnosis of GGE or JME. For each of asymmetric PPD and asymmetric sleep spindles, we determined the same diagnostic outcome measures for the presence of a structural abnormality on brain MRI. Results: CTS had 83% specificity and 75% PPV in children with normal neurological examination. PPR had high specificity of 92% and NPV 92% for GGE; for JME, PPR also had high sensitivity (92%). Asymmetric PPD had low sensitivity for structural brain abnormalities (17%), with specificity 80%. In contrast, asymmetric sleep spindles had higher sensitivity and specificity, 44% and 97%, respectively. Conclusions: CTS are seen with CECTS and other conditions. PPR is highly indicative of a GGE, though may be seen other conditions. Relative attenuation of sleep spindles is a more reliable indicator of structural brain malformation than asymmetric PPD.

P.074

An assessment of next-generation panel testing in epilepsy

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Background: With the now routine use of next-generation sequencing it is important to know the baseline outcomes as they relate to clinical care for pediatric epilepsy in Ontario. We sought to assess the diagnostic yield of genetic epilepsy panel testing and characterize the impact on patient care. Methods: We conducted a retrospective chart review of patients with epilepsy seen at CHEO between 2012-2020 with genetic testing. 227 patients met our inclusion criteria. Patient charts were reviewed for clinical details, co-morbidities, genetic testing results, and changes to management. Results: Diagnostic yield was 19% for multi-gene epilepsy panel testing. A further 10% received a diagnosis from additional genetic testing. The diagnostic yield was significantly higher in patients with a younger age of onset of seizures. A direct change in clinical management as a result of the molecular diagnosis was evident for 9% of patients; however, all diagnoses impacted prognosis and family counselling. Conclusions: The diagnostic yield of genetic epilepsy panel testing conducted at CHEO is comparable to other reported rates. Genetic testing resulted in clinical benefits of recurrence risk counselling, prognostic information and though a direct change in management was advised in a minority of individuals, targeted treatment recommendations will continue to increase with ongoing testing.

P.075

EEG features reflecting the neurodevelopmental assessment at term equivalent age in preterm born infants

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Background: In Canada, 7% of children are born preterm between 29 and 36 weeks gestational age (GA). Electroencephalography (EEG) provides a bedside evaluation of brain activity, yet the clinical significance of several EEG patterns requires further study. The goal of this study is to determine the EEG features at term equivalent age (TEA) that correlate with

neurodevelopmental evaluation at TEA in infants born between 29-36 weeks GA. Methods: Prospective cohort study of preterm infants born 29-36 weeks GA with 1 hour EEG recording at TEA. EEG discontinuity index (proportion <25mcV amplitude) and spectral power densities were calculated as well as the mean and maximum values of interburst intervals. At TEA, neurodevelopment was evaluated using the *General Movement Optimality Score (GMOS)*. Linear regression analyses were used to evaluate the association between EEG features and neurodevelopmental assessment. Results: Eighty-two children (median GA 33.6 weeks) were included (47 males). Median GMOS was 29.0 (IQR 24.3-35.0). A greater EEG discontinuity index was associated with reduced GMOS (B -6.85; 95% CI -12.13,-1.57; p=0.012). Conclusions: At TEA, a greater EEG discontinuity index was associated with a more abnormal neurodevelopmental assessment. Ongoing longitudinal neurodevelopmental assessments are needed to better evaluate the prognostic potential of TEA EEG.

P.077

Response to the ketogenic diet in refractory epileptic spasms at BC Children's Hospital

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Background: Epileptic spasms (ES) are a devastating seizure type with poor neurodevelopmental outcome; 1/3 are resistant to treatment with first line therapies. Recently attention has been drawn to the ketogenic diet (KD) as a potentially effective therapy, though data regarding optimal time of initiation, and its sustained effectiveness, are lacking. Methods: Retrospective chart review of all patients with ES treated with KD at BC Children's Hospital between 2002 and 2020 (n=28) with comparison of spasm response based on age of initiation of KD in two groups: < 12 months (n=11) and ≥ 12 months (n=17). Results: Comparing the <12 months and ≥ 12 months groups showed: unknown etiology in 9% vs 25%; spasm freedom for 3 months on KD in 18% vs 41%; median time to spasm freedom was 2 vs 6 weeks; relapse after a period of spasm freedom occurred in 66% vs 70%. Conclusions: Although more effective in children ≥ 12 months of age in the first 3 months, spasm freedom in either group was not sustained with KD. KD is recommended as early therapy for refractory ES, but this study suggests clinicians be aware the KD has limited efficacy in long-term control of ES and must be used with other therapies.

P.078

Children with Trisomy 21 and Lennox-Gastaut Syndrome with predominant myoclonic seizures

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Background: Background: Lennox-Gastaut syndrome (LGS) is a severe form of pediatric epilepsy that is classically defined by a triad of drug-resistant seizures, characteristic EEG patterns, and intellectual disability. Long-term prognosis is generally poor with progressive intellectual deterioration and persistent seizures. At present, there are few reported cases of LGS and Trisomy 21 (T21) in the literature. To further delineate the spectrum of epilepsy in T21, we reviewed children with T21 and LGS at one center over 28 years. Methods: Methods: This is a retrospective case series. At our institution, all EEG results are entered into a database, which was queried for patients with T21 from 1992-2019. Pertinent electro-clinical data was obtained from medical records. Results: Results: 63 patients with T21 and epilepsy, 6 (10%) had LGS and were included in the study. Four of the six patients were male and 5/6, had neuro-imaging, which was normal. Follow-up ranged from 3-20 years. Notably, 5/6 had predominant myoclonic seizures throughout the course of their epilepsy, associated with generalized spike-wave discharges. Conclusions: Conclusions: Myoclonic seizures appear to be a predominant seizure type in patients with T21, suggestive that T21 patients may have a unique pattern of LGS.

Metabolic Disease

P.079

MT-TA: A mitochondrial genome cause of developmental and epileptic encephalopathy

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Background: *MT-TA* (OMIM 590000), one of 22 mitochondrial transfer-RNA (mt-tRNA) genes, encodes the mt-tRNA for alanine. Pathogenic variants in mt-tRNA genes affect the translation of respiratory chain complexes I, III, and IV; which leads to mitochondrial dysfunction and a clinically variable phenotype. *MT-TA* pathogenic variants have been described in only seven patients, all of whom had isolated myopathy Methods: Case report. Results: Our patient initially presented with drug-resistant West syndrome, later evolving towards a Lennox Gastaut phenotype. Although she had hypotonia, serum creatine kinase and electromyography were normal. Brain-MRI showed bilateral symmetric hypointense T1, hyperintense T2-fluid-attenuated-inversion-recovery and restricted-diffusion signal changes in the dentate nuclei. Mitochondrial genome testing identified a previously published pathogenic variant in *MT-TA* (m.5591G>A) with 14% blood heteroplasmy and 16% urine heteroplasmy. The variant was absent in serum sampled from the patient's mother Conclusions: Our case extends the phenotypic spectrum of *MT-TA* variants to include developmental and epileptic encephalopathy, in the apparent absence of muscle disease. We hypothesize that our patient may have the greatest degree of heteroplasmy in brain tissue; however, animal models and induced pluripotent stem cell (iPSC) models are needed to identify the precise mechanism by which *MT-TA* dysfunction results in variable phenotypes with variable degrees of heteroplasmy.