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

Y Xie (Singapore)*

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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

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Diagnostic utility of specific abnormal EEG patterns in children

M Ashour (Montreal)* *E Minato* (Montreal) *A Alawadhi* (Montreal) *S Berrahmoune* (Montreal) *E Simard-Tremblay* (Montreal) *C Poulin* (Montreal), *K Myers* (Montreal)

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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.

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An assessment of next-generation panel testing in epilepsy

H Leduc-Pessah (Ottawa)* *T Hartley* (Ottawa) *D Pohl* (Ottawa), *D Dyment* (Ottawa)

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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.

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

A Dufour (Montréal)* *M Gagnon* (Montréal) *B Marandyuk* (Montréal) *Z Mahdi* (Montréal) *G Côté-Corriveau* (Montréal) *A Nuyt* (Montréal) *M Dehaes* (Montréal) *T Luu* (Montréal) *M Simard* (Montréal), *EF Pinchefsky* (Montréal)

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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