Child Neurology (CACN)

C.1

Utility of genetic testing in the pre-surgical evaluation of children with drug-resistant epilepsy

*S Alsubhi (Montreal) S Berrahmoune (Montreal) RW Dudley (Montreal) D Dufresne (Sherbrooke) E Simard Tremblay (Montreal) M Srour (Montreal) KA Myers (Montreal)**

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Background: All patients with drug-resistant focal epilepsy, should undergo an evaluation to determine if non-medical options, including surgical intervention, are appropriate. This evaluation involves a thorough work-up, typically including some or all of neuropsychological evaluation, prolonged video EEG monitoring, and advanced neuroimaging. The utility of genetic testing as part of this evaluation has not been thoroughly investigated. Methods: In this retrospective study, we reviewed the charts of pediatric patients referred for epilepsy surgery evaluation over a 5-year period. We extracted and analyzed results of genetic testing as well as clinical, EEG, and neuroimaging data. Results: 125 patients were referred for epilepsy surgical evaluation, 86 of whom had some form of genetic testing. Of these, 18 had a pathogenic or likely pathogenic variant identified. Genes affected included *NPRL3*, *TSC2*, *KCNH1*, *CHRNA4*, *SPTAN1*, *DEPDC5*, *SCN2A*, *ARX*, *SCN1A*, *DLG4*, and *ST5*. One patient had ring chromosome 20, one a 7.17p12 duplication, and one a 15q13 deletion. A specific medical therapy choice was allowed due to genetic diagnosis in three patients who did not undergo surgery. Conclusions: Obtaining a molecular diagnosis may dramatically alter management in children with drug-resistant focal epilepsy. Genetic testing should be incorporated as part of standard investigations in the pre-surgical work-up of such patients.

C.2

An in-depth analysis of pediatric inflammatory myopathies: findings from a comprehensive tertiary care hospital

E Mostofi (Edmonton) H McMillan (Ottawa) R Jurencak (Ottawa) L Hamilton (Ottawa) A Yaworski (Ottawa)**

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Background: Pediatric inflammatory myopathies (PIM) are a rare, heterogenous group of disorders requiring prompt diagnosis and treatment to reduce complications and improve long-term outcome. This study reviews the clinical characteristics, management, and outcomes in PIM. Methods: A retrospective analysis of pediatric patients diagnosed with PIM at CHEO from January 2009 to December 2023 was performed. Patient data, including age at symptom onset, diagnostic testing performed, treatment, and follow-up durations, were evaluated. Results: A total of 25 patients with juvenile dermatomyositis (JDM), overlap syndromes, and necrotizing myopathy (HMG-CoA reductase and anti-SRP myositis) were identified. Symptoms began at an average age of 8.37 years (1.10-14.11), with formal diagnosis occurring at 8.57 years (2.02-16.11). Initial symptoms included skin changes, muscle weakness, joint pain, and fatigue. Diagnosis involved laboratory testing (CK, myositis antibodies), muscle MRI, electromyography, and/or muscle biopsy. Treatments included corticosteroids, IVIG, and steroid-sparing agents (methotrexate, mycophenolate mofetil, rituximab, hydroxychloroquine). Follow-up averaged 4.23 years (range: 0.5 to 13). Most patients displayed only mild residual symptoms with the exception of an anti-SRP myositis patient who became wheelchair-dependent, requiring ventilatory support. Conclusions: Inflammatory myopathies require prompt treatment to prevent complications. Most patients require multiple treatment modalities, however with early diagnosis and treatment the majority of patients' symptoms resolve.

C.3

Retrospective study of sulthiame in treatment of pediatric epilepsy

A Laliberté (Montreal) KA Myers (Montreal)*

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Background: This retrospective study assessed the efficacy and tolerability of sulthiame as a treatment in children with epilepsy. In Canada, sulthiame is only available through Health Canada's Special Access Program. Methods: Patients who received sulthiame at the Montreal Children's Hospital from April 2012 to March 2023 were included. Patients' medical charts were reviewed, and clinical data was extracted from neurology clinic notes and electroencephalogram (EEG) reports. Efficacy was assessed by comparing seizure frequency and frequency of EEG epileptiform abnormalities before and after initiating sulthiame, while also noting any reported changes in cognition or behaviour. Results: Sixteen patients were included (10 males, 6 females), all of whom had drug-resistant epilepsy and continuous spike-wave in sleep (CSWS) on EEG. Sulthiame starting dose ranged from 0.74 to 6.75 mg/kg/day. Improvement, either in terms of seizure control, cognition, or reduction in EEG epileptiform abnormalities, was reported in 8/16 children (50%). Two patients (13%) became seizure free, while three more (19%) had reduced seizure frequency. Three other patients (19%) had reported improvements in concentration, learning abilities or behaviour. No serious adverse event was reported. Conclusions: These data indicate that sulthiame is effective and well-tolerated in children with CSWS, regardless of the etiology and type of epilepsy.

C.4

Neurologic injury in pediatric patients cannulated for rescue extracorporeal life support

SG Buttle (Calgary) K Woodward (Calgary) L Foster (Calgary) P Yee (Calgary) A Kirton (Calgary) S Jamal (Calgary) A Mineyko (Calgary) J Blackwood (Calgary) MJ Esser (Calgary)*

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Background: Historical literature suggests the risk of neurologic injury in children supported by extracorporeal life support (ECLS) is between 10-20%, however recent studies suggest the