The development of a rigorous protocol has reduced potential adverse effects, inadvertent complications from errors have improved and parent satisfaction enhanced. Conclusions: This poster will demonstrate how an interdisciplinary approach for a ketogenic diet protocol, involving an advanced Practice Nurse, nutritionist, neurologist and parent, resulted in improved care.

P.058

Ketogenic diet for medication refractory infantile spasms in patients with Down syndrome: experience at the Children's Hospital of Eastern Ontario

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doi: 10.1017/cjn.2025.10229

Background: Ketogenic diet can be an effective alternative therapy for medication refractory infantile spasms. Infantile spasms are more prevalent in children with Down syndrome compared with the general population and often medication refractory. Methods: Charts of infants who presented to the Children's Hospital of Eastern Ontario with Down syndrome and refractory infantile spams treated with ketogenic diet from 2012 to 2025 were reviewed. Clinical response defined by cessation of epileptic spasms and resolution of hypsarrhythmia. Diet ratio,tolerance, side effects, concomitant medications, and diagnostic tests were evaluated. Results: 5 infants were treated with ketogenic diet after failing first line anticonvulsant mendications: viagabatrin and corticosteriods. Ketogenic diet was viable only via G-tube in 1 patient and by NG tube in 3 due to risk of aspiration. Diet was compatible with second line anticonvulsants. Complete electroclinical response occurred in 2 infants after 4 weeks. Partial seizure reduction and electrographic improvement was observed in 1 infant. 1 patient died due to unrelated respiratory illness. Conclusions: Ketogenic diet is a viable potentially effective therapeutic option for infants with Down syndrome and medication refractory infantile spasms. These infants present challenges inherent of Down syndrome such as hypotonia, higher risk for aspiration which need to be considered before diet introduction.

P.059

Exome-based testing for seizure indications captures a broader range of diagnostic genes and more diagnostic variants than provincially-funded epilepsy panels

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Background: Ontario and other Canadian provinces fund multi-gene sequencing panels as the initial testing approach for patients with epilepsy. However, genetic testing guidelines issued by the US-based National Society for Genetic Counselors and endorsed by the American Epilepsy Society recommend exome as a first-line test. We explored the theoretical improvements in diagnostic yield when selecting exome over provincially-funded panels (PFPs). Methods: Our comparative analysis used a list of 768 diagnostic genes and 4474 diagnostic variants identified in diagnostic exome cases involving clinical indications of seizure. We compared these lists to the genes included in two PFPs (190 genes and 474 genes) to see which exome-identified genes and variants would have been captured by the PFPs. Results: Most exome-identified diagnostic genes may have been missed by the PFPs (82% and 65% for the 190 and 474-gene PFPs), and close to half of the exome-identified diagnostic variants (62% and 43% for the 190 and 474-gene PFPs) may have been missed. Conclusions: Exome-based testing captures a broader range of diagnostic genes and more diagnostic variants than PFPs. The adoption of exome over panels as a first-line test may lead to improved diagnostic rates and permit earlier treatment for individuals with seizures.

P.061

Exploring emergency department visits in adolescents with epilepsy and mild intellectual disabilities (MID)

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Background: In Canada, individuals with intellectual disabilities (ID) make up approximately 25% of the epilepsy population. Despite making up only a small portion, adult hospitalization data in Canada shows that individuals with ID are significantly more likely to be seen in the ED, be hospitalized, and to die as a result of epilepsy and epilepsy complications, than individuals with typical cognitive development. Data looking at ED visits in adolescents with epilepsy and varying cognitive abilities is extremely limited. Methods: To address this, a retrospective chart review of 122 adolescents (42 MID and 80 typical cognitive development) with epilepsy between the ages of 14 and 18 was done. Results: Results showed that adolescents with typical cognitive development had significantly more ED visits (p=.006), and seizure related ED visits (p= .008) than adolescents with MID. Despite the reasons for ED visits not significantly differing between the two groups, adolescents with MID had significantly longer ED visits (p=.023). Finally, when looking exclusively at the MID group, results showed that females were significantly more likely to be seen at the ED than males (p=.001). Conclusions: Results suggest that ED visit frequencies differ among adults and adolescents with ID, potentially suggesting the presence of unique protective factors for adolescents.

Movement Disorders

P.063

Pediatric status dystonicus: 10-year experience at a single tertiary children's hospital

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Background: Status dystonicus is characterized by frequent or prolonged severe episodes of generalized dystonia. The