stimulation (VNS). Methods: A retrospective study was completed examining the effectiveness of VNS and CC in children with SYNGAP1-DEE using the SynGAP Research Database and an additional child followed at our centre. Results: Fifteen patients from the SynGAP Database were included. Of those who had VNS (n=11), 7 children had an >50% reduction in seizure frequency (n=7/11, 64%), 2 had worsening (n=2/11, 18%), 1 had no change (n=1/11, 9%), and 1 had an unknown response (n=1/11, 9%). Two children had CC only, 1 had complete seizure freedom, and 1 had a >50% reduction. Two children underwent VNS and CC, 1 had a >50% reduction in seizure frequency and the other had no change. One child followed at our centre experienced a sustained >80% reduction in seizure frequency following CC (i.e., after 1.5 years). Conclusions: We provide the first in-depth description of the response to VNS and CC in children with *SYNGAP1*-DEE, and provide insight into the use of of palliative surgical procedures in this population.

P.053

Biallelic SCN1A variants with divergent epilepsy phenotypes

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Background: Dravet syndrome and genetic epilepsy with febrile seizures plus (GEFS+) are associated with pathogenic variants in *SCN1A*. While most such cases are heterozygous, there have been 16 reported homozygous cases. We report two new biallelic cases associated with divergent phenotypes. Methods: We performed a chart review for two patients with different homozygous SCN1A variants and reviewed all previously published biallelic *SCN1A* pathogenic variants. Results: Our first patient exhibited early afebrile seizures and severe developmental delay, without febrile seizures or status epilepticus. A homozygous c. 1676T>A, (p. Ile559Asn) variant of uncertain significance was identified, carried by asymptomatic parents. The second patient exhibited early, recurrent, and prolonged febrile seizures, moderate developmental delay, and motor dysfunction; a homozygous pathogenic c. 4970G>A, (p. Arg1657His) variant carried by asymptomatic parents was identified.

Of 18 known cases of biallelic SCN1A pathogenic variants, 15/18 (83%) have diagnoses of Dravet or GEFS+. The remaining 3/18 (17%) had pharmacoresponsive epilepsy with prominent GDD. Cognitive phenotypes ranged from intact neurodevelopment to profound developmental delay. Eleven out of 18 cases (61%) had motor concerns. Conclusions: These cases expand the phenotypic spectrum of biallelic *SCN1A* variants. While some patients present typically for Dravet/GEFS+, others present with developmental delay and controllable epilepsy.

P.055

Novel pipeline for triage of variant of uncertain significance reclassification in epilepsy genomics

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Background: Increased availability of genetic testing has led to increased burden of follow up of variants of uncertain significance (VUS). As of January 2025, 327 VUS were identified patients at BC Children's Hospital. We propose a pipeline to triage and follow up of patients with identified VUS to clarify diagnosis through paternal testing. Methods: Of the 327 patients with VUS, 13 patients with high clinical suspicion for a genetic disorder were identified by their neurologist. Initial chart review for each patient was performed. Clinical phenotype data and the patient's variant were inputted into the online tool Franklin. This program generates a variant interpretation based on 17/ 28 criteria in ACMG scoring. For each patient the variant would be assumed to be de novo in order to determine if parental testing could change variant classification. Results: 5/13 of the patients had suggested reclassification of variants. 6/13 of the patients would have reclassification of variant to likely pathogenic/pathogenic if the variant was found to be de novo, suggesting a need for paternal testing. Conclusions: This highlights a novel clinical pipeline to improve expediency and triaging of VUS reclassification for paternal testing in epilepsy genomics.

P.057

A review of a twenty-seven year experience with the Ketogenic diet: lessons learned and moving forward

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Background: Although the history of the ketogenic diet dates back centuries, with the advancement of anti-seizure medications, the use of the diet for epilepsy declined. It was not until the early 1990s that there was a resurgence of the diet as an adjunct therapy to anti-seizure medication. In 1998, the Montreal Children's Hospital introduced the ketogenic diet to a child with drug resistant epilepsy. Shortly after, a presentation of the ketogenic diet at hospital Grand Rounds met much skepticism. However, over time the diet has developed into a well-established treatment option for children with drug resistant epilepsy. Two hundred children have since utilized the diet at the Montreal Children's Hospital. Methods: A review of patient files since the initiation of the program was undertaken. Data was extracted regarding adverse effects, common errors in both hospital and home setting, risks for unfavorable outcomes and parental concerns Results:

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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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