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.

Metabolic Disease

P.109

Diagnostic Yield of Targeted Exome Sequencing in West Syndrome

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Background: West syndrome (WS) is characterized by the onset of epileptic spasms usually within the first year of life. Global developmental delay with/without regression is common. Advances in high-throughput sequencing have supported the genetic heterogeneity of this condition. To better understand the genetic causes of this disorder, we investigated the results of targeted exome sequencing in 29 patients with WS. **Methods:** Whole exome sequencing (WES) was performed on an Ion Proton™ and variant reporting was restricted to sequences of 620 known epilepsy genes. Diagnostic yield and treatment impact are described for 29 patients with WS. **Results:** A definitely/likely diagnosis was made in 10 patients (34%), which included 10 different genes (*ALG13, PAFAH1B1, SLC35A2, DYNC1H1, ADSL, DEPDC5, ARX, CDKL5, SCN8A, STXBP1*) known to be associated with epilepsy or WS. Most variants were *de novo* dominant (X-linked/autosomal) except for *ARX* (X-linked recessive) and *ADSL* (autosomal recessive). 4 out of 10 (40%) had a genetic diagnosis with potential treatment implications. **Conclusions:** These results emphasize the genetic heterogeneity of WS. The high diagnostic yield, along with the significant genetic variability, and the potential for treatment impact, supports the early use of this testing in patients with unexplained WS.

MS/Neuroinflammatory Disease

P.111

Use of rituximab for pediatric central nervous system inflammatory disorders in Alberta

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Background: Rituximab is a B-cell-depleting monoclonal antibody whose off-label use is funded in Alberta by the Short-Term Exceptional Drug Therapy (STEDT) program. This study describes the use of rituximab for pediatric central nervous system (CNS) inflammatory disorders in Alberta. **Methods:** Rituximab applications for CNS inflammatory indications in patients < 18 years of age were identified from the STEDT database between January 1, 2012 – December 31, 2019. Patient information was linked to other provincial datasets, including the Discharge Abstract Database, Pharmaceutical Information Network, and provincial laboratory data. Analysis was descriptive. **Results:** 51 unique rituximab applications were identified, of which 50 were approved. New applications increased from one in 2012 to a high of 12 in 2018. The most common indication was autoimmune encephalitis (other than anti-NMDA receptor encephalitis; n=20, 39%). Most children were approved for a two-dose (n=33, 66%) or four-dose (n=16, 32%) induction regimen. Physician-reported outcomes were available for 24 patients, of whom 14 (58%) were felt to have fully met outcome targets. **Conclusions:** The use of rituximab for pediatric CNS inflammatory disorders has increased, particularly for the indication of autoimmune encephalitis. This study identified significant heterogeneity in dosing practices and laboratory monitoring, as well as regional disparities in use.

Neuromuscular Disease and EMG

P.112

5q Spinal Muscular Atrophy Canadian Paediatric Surveillance Program - 2020 Results

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Background: Spinal muscular atrophy (SMA) is the leading genetic cause of infant death and the second most common autosomal recessive disorder; the majority of cases are due to homozygous deletion of *SMN1* gene. **Methods:** This study uses the Canadian Paediatric Surveillance Program to determine the minimum annual incidence of 5q-SMA from birth to 18 years of age in Canada. The complete protocol can be accessed at