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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doi: 10.1017/cjn.2021.385

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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doi: 10.1017/cjn.2021.387

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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doi: 10.1017/cjn.2021.388

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

www.cpsp.cps.ca/surveillance. **Results:** Eleven cases were reported in 2020. Five (45%) cases were reported from Ontario and the remaining cases were reported from Atlantic Canada and Western Canada. Their median age was 12 months (IQR 6–21); 64% were male. The most common presenting symptoms were delayed motor milestone and hypotonia in 7 (64%) cases. On average, the diagnosis was delayed after the onset of symptoms by three months for SMA Type 1, by eight months for Type 2, and by 18 months for Type 3. Eight (73%) cases received nusinersen as their first disease-modifying treatment. **Conclusions:** Early recognition and newborn screening are essential to reduce diagnostic delay and enable timely treatment of SMA. Other data sources including the Canadian Neuromuscular Disease Registry and molecular genetic laboratories will be used to estimate the annual incidence of pediatric SMA in Canada.

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A Population-based Study of the Epidemiology, Healthcare Resource Utilization and Costs of Duchenne Muscular Dystrophy in Alberta, Canada

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doi: 10.1017/cjn.2021.389

Background: Duchenne muscular dystrophy (DMD) is a severe progressive neuromuscular disease. This study aimed to estimate the prevalence, healthcare resource utilization (HRU), and medical costs of DMD in Alberta. **Methods:** This retrospective study linked provincial healthcare administrative data to identify patients with DMD utilizing a modified diagnostic code algorithm, including males <30 years of age. Five-year (April 2012 to March 2017) prevalence estimates were calculated and all-cause direct HRU and costs were examined in the first-year post-diagnosis. **Results:** Overall, 111 patients (median age: 12.0 years (IQR 4.7-18.3)) with DMD were identified. The estimated five-year period prevalence was 35.72 (95% CI 31.88-39.91) per 100,000 persons. All-cause HRU in the first-year post-diagnosis included a mean (SD) of 0.48 (1.19) hospitalizations (length of stay: 9.37 days (36.47)), 3.96 (6.16) general practitioner visits, 28.52 (62.98) specialist visits, and 20.14 (16.49) ambulatory care visits. Mean (SD) all-cause direct costs were $18,868 ($29,206) CAD in the first-year post-diagnosis. **Conclusions:** Patients with DMD had multiple interactions with the healthcare system in the year following diagnosis, resulting in substantial direct medical costs. More effective treatment strategies are needed to improve health outcomes and reduce the burden of DMD.

P.114

Benign tumors of peripheral nerves in children at a tertiary-care pediatric hospital

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doi: 10.1017/cjn.2021.390

Background: Tumors affecting peripheral nerves in children are rare. Accurate diagnosis ensures that management is appropriate and timely. **Methods:** We review the clinical presentation and utility of investigations of children with intrinsic tumors affecting peripheral nerves at the Children's Hospital of Eastern Ontario (CHEO). **Results:** From 2009-2019, 14 cases were identified. Mean age of symptom onset was 8.2 years (range 0.3 to 17.3 years). Presenting symptoms included painless muscle wasting (2/14), focal muscle weakness (7/14), contracture (1/14), pain (1/14) or a painless, palpable mass (3/14). MRI was useful at differentiating benign pediatric nerve tumors. Peripheral nerve lipomatosis demonstrated a classic "spaghetti string" appearance. Patients with perineurioma showed evidence of enhancing, nodular lesions while intraneural ganglionic cysts display cystic lesion within the nerve. Neurofibromas appear like a "bag of worms" while schwannomas are more eccentrically positioned around the nerve. Nerve conduction studies (NCS) or electromyography (EMG) were performed in 11/14 patients. Biopsies were performed in 9 patients and surgical management in 4 patients. **Conclusions:** The rare nature of peripheral nerve tumors in children can pose diagnostic challenges. NCS/EMG are important to assist with localization, and MRI important at distinguishing benign tumors. Key MRI, clinical and NCS features can guide management, potentially avoiding invasive procedures.

Neurovascular, Stroke and Neurointerventional

P.115

Vessel Wall Imaging of Unusual Childhood Strokes: a Pediatric Case Series

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doi: 10.1017/cjn.2021.391

Background: MR-based vessel wall imaging (VWI) has gained influence in the clinical investigations, and management of pediatric strokes. Limitations still exist in interpreting it as a singular modality. **Methods:** We present 4 pediatric stroke cases