P.042

Anti-HMG Coenzyme A reductase antibody (anti-HMGCR) myopathy: case review of two pediatric patients from a single center

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Background: Necrotizing anti-HMGCR myopathy is rare in children. Pediatric cases are not typically associated with statin use or malignancy. Methods: Retrospective chart review (January 2009 to December 2023) identified cases of anti-HMGCR myopathy at our hospital. Results: Two patients were identified. Patient A, presented at 8 yo with a 2 year history of proximal muscle weakness. His CK was 4,840 U/L (normal <205 U/L) with a high anti-HMGCR antibody titre. His Childhood Myositis Assessment Scale (CMAS) score was 33/52. Monthly IVIG was started and his muscle strength and CK improved. Two years later, weekly methotrexate was started for persistent mild CK elevation (602 to 869 U/L). At 11 years old, 3 years after diagnosis, his CMAS score was 47 and he could participate in soccer with mild fatigue. Patient B, presented at 8 yo with acute proximal weakness, rash and CMAS 13/52. His CK was 20,185 U/L with elevated anti-HMGCR antibody titre. He received oral corticosteroids, weekly methotrexate and monthly IVIG. At 10 yo, 2 years after diagnosis, he is asymptomatic with CMAS 51. He is maintained on methotrexate monotherapy. Conclusions: Anti-HMGCR antibody myopathy requires prompt diagnosis to obviate muscle necrosis and long-term complications. Our patients showed clinical and CMAS improvement with treatment.

P.043

Developing a brief clinical dataset for Duchenne Muscular Dystrophy

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Background: Duchenne muscular dystrophy (DMD) causes progressive muscle wasting. The Canadian Neuromuscular Disease Registry (CNDR) previously developed a comprehensive DMD dataset in accordance with the International Classification of Functioning, Disability, and Health (ICF). Our objective was to develop a brief ICF core set that best aligns with the priorities of individuals and families affected by DMD. Methods: A literature review of best practices was prepared and reviewed by a multidisciplinary team at the CNDR. The entire process involved patient and parent input and participation and was compliant with the World Health Organization guideline to develop brief ICF core sets. Results: An eight step process was developed. In brief, these included multi-stakeholder consensus meetings, ranking surveys, mapping to international standards, further consensus meeting, evaluation of clinical feasibility in multidisciplinary clinics across Canada, an integrated literature review, and development of a finalized brief ICF core set for DMD. Conclusions: The process of identifying the priorities of those living with DMD using the brief ICF core set will support post-marketing surveillance of novel therapies. The next step in this project will be to identify the specific outcome measures that best align with the brief ICF core set for DMD, for their eventual inclusion in the CNDR registry.

Other Child Neurology

P.044

Ambient air pollution and emergency department presentations for pediatric primary headache and seizure disorders in Calgary, Canada

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Background: Climate change, and fossil fuel combustion threaten the health of children globally through direct and indirect mechanisms,[1] such as the exacerbation of ambient air pollution.[1,2] Increased ambient air pollutant concentrations are associated with emergency department (ED) visits for episodic and paroxysmal neurologic conditions in adults in the Toronto region of Canada.[4,5] We hypothesize that, in Calgary, Alberta, increased ambient air pollutants will be positively associated with the daily burden of pediatric ED presentations for migraine and seizures, and that a greater effect size may be present due to increased regional variability in ambient PM2.5 concentrations.[3,4] Methods: Emergency records from the National Ambulatory Care Reporting System, comprising 17552 primary seizure and headache cases between 0-18 years of age and presenting to Calgary-region emergency departments between January 2012-December 2021, will be included. Quasi-Poisson regression modeling incorporating ambient air pollutants, seasonality and meteorological covariates will estimate relative risk and 95% confidence intervals of ED visit counts relative to increases in air pollutants. Results: Results currently pending and will be available for presentation. Conclusions: Significant results may inform further inquiry into the impact of air pollutants on children with neurological conditions and identify potential contributions of air quality to healthcare service demand in the Calgary region.

P.045

Trofinetide for the treatment of Rett syndrome: long-term safety and efficacy results from the open-label LILAC and LILAC-2 studies

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Background: Trofinetide significantly improved core symptoms of Rett syndrome (RTT) with an acceptable safety profile in

LAVENDER. Here, we report the safety and efficacy results of LILAC and LILAC-2, open-label extension studies of LAVENDER. Methods: Females with RTT, aged 5–21 years, received twice-daily, oral trofinetide in LILAC for 40 weeks. Participants who completed LAVENDER and LILAC continued trofinetide in LILAC-2, a 32-month extension study. Safety assessments included the incidence of adverse events (AEs). Efficacy endpoints included the Rett Syndrome Behaviour Questionnaire (RSBQ) and the Clinical Global Impression–Improvement (CGI-I) scale. Results: Overall, 154 patients were enrolled in LILAC. The most common AEs were diarrhea (74.7%) and vomiting (28.6%). The mean (standard error [SE]) change from the LAVENDER baseline to Week 40 in the LILAC study in RSBQ was -7.3 (1.62) and -7.0 (1.61) for participants treated with trofinetide and placebo in LAVENDER, respectively. Mean (SE) CGI-I scores compared with the LILAC baseline at Week 40 were 3.1 (0.11) and 3.2 (0.14) for patients treated with trofinetide and placebo in LAVENDER, respectively. Similar safety and efficacy trends were observed in LILAC-2. Conclusions: Trofinetide continued to improve symptoms of RTT in LILAC and LILAC-2 with a safety profile consistent with LAVENDER.

P.046

Review of the management of Wernicke encephalopathy in pediatrics

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Background: Wernicke encephalopathy (WE) is a neurological emergency defined by acute encephalopathy, oculomotor dysfunction, and ataxia. Pediatric cases of WE are underdiagnosed despite having a similar incidence to adults. There are no available treatment guidelines for pediatric WE. Prompt treatment with thiamine can prevent devastating consequences. Methods: A rapid review of the literature of the past 20 years with selected relevant older articles was conducted for the research question "How does child and adolescent thiamine therapy management for Wernicke Encephalopathy compare to adult guidelines?" All articles reporting the investigation, management and treatment of Wernicke encephalopathy – both non alcohol related and alcohol-related pediatric cases – were included. Articles not reporting clinical outcomes were excluded. Results: Eleven case studies including one available review article, met the inclusion and exclusion criteria. An algorithm was created for the organization of published reports of the management of WE for children and adolescents. Key considerations were included for the prevention, identification, acute and ongoing management of patients with WE. Conclusions: The recognition of risk factors for thiamine deficiency and symptoms of acute WE should prompt immediate treatment with thiamine – as a routine and safe therapy in the pediatric population.

P.047

Survey of caregivers of individuals with NBIAs to identify relevant quality of life outcomes

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Background: Neurodegeneration with Brain Iron Accumulation (NBIA) is a heterogenous group of disorders with the common theme of iron accumulation in the basal ganglia. These disorders typically present in childhood with progressive neurodegeneration and neuropsychiatric symptoms. Caring for an individual with NBIAs is intensive, however it is unknown what factors impact caregiver well-being and quality of life. Methods: Common themes were obtained via literature review of quality of life surveys in children with neurological and chronic illnesses. Five domains were addressed: Diagnosis, Communication, Symptom Management, Clinical Experience and Resources/Support. The survey was approved by the Family Advisory Committee at the CHEO Research Institute and the CHEO REB. The survey was distributed via the Rare Connect Platform to Canadian caregivers. Results: Survey responses are being analyzed and will be presented at the CNSF. Within each domain, Likert scales will be analyzed. Domains will be ranked according to the caregiver responses. Conclusions: Results of this survey will assist in developing care management guidelines, resources for families and help with future advocacy for patients and families affected by NBIAs. The results will also help guide future NBIA Canada Family Conferences.

P.048

Prevalence, type and risk factors of intracranial hemorrhage in term neonates: a systematic review and meta-analysis

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Background: Intracranial hemorrhage (ICH) in newborns poses a significant challenge to wellbeing and development. In preterm neonates, germinal matrix hemorrhage is most common. In term neonates, prevalence and type of ICH has not been well elucidated. This systematic review aims to assess prevalence, type, and risk factors of ICH in term neonates. Methods: A systematic review was conducted. Inclusion criteria was ICH in neonates born at 37+ weeks gestation. Exclusion criteria was one type of ICH, one risk factor, sample size <20, text not in English, full text not accessible. Eligible studies were evaluated by two authors, data was extracted and analyzed using a predesigned template and MetaXL. Results: A total of 1226 records were initially identified and 20 studies were included in the final analysis. The overall prevalence of ICH was 9.3%. This was subdivided into an asymptomatic subgroup (5.8%) and symptomatic subgroup (29.3%). Analysis showed CT detected ICH most commonly. Extra-axial hemorrhage was most commonly