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