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Leading the Way to the Future: Implementing Novel Therapeutics for Rare Pediatric Neurological Disorders

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Background: Children and Adolescents with rare neurogenetic disorders often have no known cure or disease modifying treatments. Recent advancements in treatments are offering much needed hope to these patients and families. However, these treatments are extremely costly, have complex administration requirements and have many unknown long-term risks and outcomes. **Methods:** In this presentation, we will discuss our experiences with the implementation process, including developing intricate care pathways, collaborating with multiple disciplines and services, supporting and advocating for our patients and families, and interacting with government agencies and pharmaceutical companies. Case studies will highlight the positive impact these treatments are making on the lives of children and adolescents with rare neurological disorders. **Results:** Spinal muscular atrophy and Neuronal Ceroid Lipofuscinosis Type 2 are both rare and devastating neurodegenerative conditions with significant morbidity and mortality. Health Canada and government funding agencies recently approved Nusinersen, Onasemnogene abeparvovec for the treatment of SMA and Cerliponase alfa for the treatment of CLN2, leading us to swiftly integrate these treatments into our standard of care. **Conclusions:** While implementing these novel therapies into clinical practice can be both challenging and rewarding, neuroscience nurses are positioned at the forefront to be leaders in this process at both organizational, national, and international levels.

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The Pediatric Neuroirritability Management Protocol at the Stollery Children's Hospital – Inspired by an Irritable Infant with GM3 Synthase Deficiency

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Background: We describe an infant with a diagnosis of GM3 synthase deficiency, presenting with severe neuroirritability from birth. He required multiple admissions due to extreme agitation and caregiver burnout. Multiple pharmacological agents were tried, and the effect of each medication was modest and short-lasting at best. The literature on the management of neuroirritability in children with progressive genetic and metabolic conditions is sparse, and a neuroirritability management protocol has yet to be developed at our institution. **Methods:** We searched for relevant primary research and articles on PubMed. We reviewed the evidence of each pharmacological agent and added non-pharmacological strategies. We developed management guidelines for neuroirritability at our hospital. This protocol was reviewed by several pediatric neurologists and pediatric palliative care specialists at the Stollery and SickKids Hospitals. **Results:** We present the Pediatric Neuroirritability Management Protocol for the Stollery Children's Hospital. **Conclusions:** Further study is required to assess whether this protocol can be adapted to treat irritability in the context of other neurological conditions such as hypoxic-ischemic encephalopathy and non-accidental injury. In addition, we will expand our guidelines to include other symptoms such as spasticity, dystonia, and autonomic dysfunction.

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Severe DNM1 Encephalopathy with Dysmyelination due to Recurrent Splice Site Pathogenic Variant

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Background: Patients with DNM1-encephalopathy almost exclusively have missense variants, mostly in the GTPase domain of DNM1. Delayed myelination has been reported in at least three patients with DNM1-encephalopathy, all with missense mutations in the DNM1 central domain. Only one *DNM1* splice-site variant has previously been reported, and the authors questioned whether the variant accounted for all aspects of the patient's phenotype. **Methods:** Case-Report. **Results:** Our patient had hypotonia and brief multifocal tonic seizures from age-1-month. He still has profound global developmental delay, daily seizures and microcephaly. MRI-Brain at age-21-months showed T2 hyperintensity in the bilateral periventricular and subcortical white matter; spectroscopy showed a questionable lactate peak and an elevated choline peak relative to N-acetylaspartate. Clinical gene-panel identified a heterozygous de novo pathogenic variant in intron 9 of DNM1 (c.1197-8G > A; IVS9-8G>A). In-silico tools categorized this variant as deleterious secondary to a splicing defect. RT-PCR analysis on peripheral blood was unsuccessful as DNM1 expression is extremely low outside of the brain. **Conclusions:** Our patient carried the same DNM1 variant previously reported, indicating this is a recurrent pathogenic splice-site variant. The spectroscopic abnormalities suggest a possible element of demyelination in *DNM1* variants of the central domain, though the mechanism remains unclear.

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Baseline Assessment of Attention and Executive Function Deficits in Children with Neurodevelopmental Disorders: Data from a Speciality Attention Clinic

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Background: Attention and executive function (EF) deficits in children negatively impact academics, social interactions, and overall quality of life. Children with other brain-based disorders are at high risk for attention and EF concerns, but the effects of these impairments are not well studied in the literature. The Complex Attention and Executive Function Clinic at the Alberta