New Publications from NMD4C Investigators

November 2024

SMN depletion impairs skeletal muscle formation and maturation in a mouse model of SMA

- From Dr Hong Liu with Dr Rashmi Kothary (NMD4C) as a co-author.

Efficacy and safety of maintenance intravenous immunoglobulin in generalized myasthenia gravis patients with acetylcholine receptor antibodies: A multicenter, double-blind, placebo-controlled trial

- From Dr Vera Bril (NMD4C).

Equitable Access to Disease-Modifying Therapies for Canadian Children with SMA and Four SMN2 Copies

- From Dr Hugh McMillan (NMD4C) with Drs Hernan Gonorazky, Craig Campbell, James Dowling, Kristina Joyal, Alex Mackenzie, Jean Mah, Laura McAdam, Maryam Oskoui, Jordan Sheriko, Mark Tarnopolsky and Kathryn Selby (NMD4C) as co-authors.

Ion Mobility QTOF-MS Untargeted Lipidomics of Human Serum Reveals a Metabolic Fingerprint for GNE Myopathy

- From Dr Cristina Manis with Dr Hanns Lochmüller (NMD4C) as a co-author.

Human ITGAV variants are associated with immune dysregulation, brain abnormalities, and colitis

- From Dr Sina Ghasempour with Dr James Dowling (NMD4C) as a co-author.

Assessing corneal dendritic cells in glucose dysregulation small-fibre neuropathy

- From Dr Juan Francisco Idiaquez with Dr Vera Bril (NMD4C) as a co-author.

Late-Onset Mitochondrial Neurogastrointestinal Encephalopathy Presenting With Isolated Ophthalmic Findings

- From Dr Angelica Hanna with Dr Vera Bril (NMD4C) as a co-author.

Long-term safety and tolerability of hyaluronidase-facilitated subcutaneous immunoglobulin 10% as maintenance therapy for chronic inflammatory demyelinating polyradiculoneuropathy: Results from the ADVANCE-CIDP 3 trial

- From Dr Robert D M Hadden with Dr Vera Bril (NMD4C) as a co-author.

Liver SMN restoration rescues the Smn2B/- mouse model of spinal muscular atrophy

- From Dr Emma Sutton with Dr Rashmi Kothary (NMD4C) as a co-author.

Difference in Drinking Times as a Function of Liquid Consistency in Adults With Oculopharyngeal Muscular Dystrophy: A Comparative Study Using Bostwick Consistometer and IDDSI Flow Test Methods

- From Dr Annie Villeneuve-Rhéaume with Dr Cynthia Gagnon (NMD4C) as a co-author.

A Novel De Novo Splice Acceptor Variant in BICD2 Is Associated With Spinal Muscular Atrophy

- From Dr GiuliaDel Gobbo with Dr Hugh McMillan (NMD4C) as a co-author.

C9orf72 expansion creates the unstable folate-sensitive fragile site FRA9A [Preprint]

- From Dr Mila Mirceta with Dr Sandro Abrahao (NMD4C) as a co-author.

Decreased Incidence of Pediatric Neuro-Autoimmune Disorders During COVID-19 Pandemic Restrictions

- From Dr Adam Jaremek with Dr Hugh McMillan (NMD4C) as a co-author.

Assessment of the Clinical Interactions of GAA Repeat Expansions in FGF14 and FXN

- From Dr Brandon Gerhart with Dr Bernard Brais (NMD4C) as a co-author.

Screening for SCA27B, CANVAS and other repeat expansion disorders in Greek patients with late-onset cerebellar ataxia suggests a need to update current diagnostic algorithms

- From Dr Georgios Koutsis with Dr Bernard Brais (NMD4C) as a co-author.

The AMPK allosteric activator MK-8722 improves the histology and spliceopathy in myotonic dystrophy type 1 (DM1) skeletal muscle

- From Dr Aymeric Ravel-Chapuis (NMD4C) with Dr Bernard Jasmin (NMD4C) as a co-author.

Rare Spinocerebellar Ataxia Types in Canada: A Case Series and Review of the Literature

- From Dr Sohaila Alshimemeri with Dr Bernard Brais (NMD4C) as a co-author.

Patient-Relevant Digital-Motor Outcomes for Clinical Trials in Hereditary Spastic Paraplegia Type 7: A Multicenter PROSPAX Study

- From Dr Lukas Beichert with Dr Cynthia Gagnon (NMD4C) as a co-author.