**New Publications from NMD4C Investigators**

**October 2023**

Systematic Literature Review of the Natural History of Spinal Muscular Atrophy: Motor Function, Scoliosis, and Contractures

- From Dr Valerie Aponte Ribero with Dr Maryam Oskoui (NMD4C) as a co-author.

Volume assured pressure support mode use for non-invasive ventilation in pediatrics

- From Dr Nisha Cithiravel with Dr Reshma Amin (NMD4C) as a co-author.

Effect of efgartigimod on muscle group subdomains in participants with generalized myasthenia gravis: post hoc analyses of the phase 3 pivotal ADAPT study

- From Dr Vera Bril (NMD4C)

Characterization of muscle strength and mobility in oculopharyngeal muscular dystrophy

- From Dr Jean-Denis Brisson with Dr Bernard Brais and Dr Cynthia Gagnon (NMD4C) as co-authors.

Caring for children with new medical technology at home: parental perspectives

- From Dr Natalie Pitch with Dr Reshma Amin (NMD4C) as a co-author.

Eplontersen for Hereditary Transthyretin Amyloidosis With Polyneuropathy

- From Dr Teresa Coelho with Dr Vera Bril (NMD4C) as a co-author.

Real-Time Monitoring of Antibiotics in the Critically Ill Using Biosensors

- From Dr Ruvimbo Dephine Mishi with Dr Craig Campbell (NMD4C) as a co-author.

Symptom and Treatment Satisfaction in Members of the US and Canadian GBS/CIDP Foundations with a Diagnosis of Chronic Inflammatory Demyelinating Polyneuropathy

- From Dr Meg Mendoza with Dr Vera Bril (NMD4C) as a co-author.

Evaluation of the diagnostic accuracy of exome sequencing and its impact on diagnostic thinking for rare disease patients in a publicly-funded healthcare system: a prospective cohort study

- From Dr Taila Hartley with Dr Jodi Warman Chardon and Dr James Dowling (NMD4C) as co-authors.

Mutations in PTPN11 could lead to a congenital myasthenic syndrome phenotype: a Noonan syndrome case series

- From Dr Alessia Pugliese with Dr Hanns Lochmüller (NMD4C) as a co-author.

Emerging and established biomarkers of oculopharyngeal muscular dystrophy

- From Dr Ian Smith with Dr Bernard Brais, Dr Hanns Lochmüller and Dr Jodi Warman Chardon (NMD4C) as co-authors.

Introducing the Dysphagiameter: a novel patient-reported outcome measure for evaluating dysphagia in oculopharyngeal muscular dystrophy - from conceptual framework to initial development

- From Dr Claudia Côté with Dr Bernard Brais and Dr Cynthia Gagnon (NMD4C) as co-authors.

Two zebrafish cacna1s loss-of-function variants provide models of mild and severe CACNA1S-related myopathy

- From Dr Yukari Endo with Dr James Dowling (NMD4C) as a co-author.

Prognostic Utility of Cardiovascular Magnetic Resonance-Based Phenotyping in Patients With Muscular Dystrophy

- From Dr Niharika Kashyap with Dr Gerald Pfeffer (NMD4C) as a co-author.