New Publications from NMD4C Investigators

March 2023 - April 2023

Digital health metrics reveal upper limb impairment profiles in ARSACS

- From Dr. Christoph Kanzler, with Dr. Bernard Brais (NMD4C) and Dr. Cynthia Gagnon (NMD4C) as co-authors.

Bi-allelic variants in the ESAM tight-junction gene cause a neurodevelopmental disorder associated with fetal intracranial hemorrhage

- From Dr. Mauro Lecca with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Base editing as a genetic treatment for spinal muscular atrophy

- From Dr. Christiano Alves with Dr. Rashmi Kothary (NMD4C) as a co-author

Trio RNA sequencing in a cohort of medically complex children

- From Dr. Ashish Deshwar with Dr. James Dowling (NMD4C) as a co-author.

Demographics and Clinical Characteristics of Autosomal Dominant Spinocerebellar Ataxia in Canada

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

Variants in ACTC1 underlie distal arthrogryposis accompanied by congenital heart defects

- From Dr. Jessica Chong with Dr. James Dowling (NMD4C) as a co-author.

Concurrent versus terminal feedback: The effect of feedback delivery on lumbar puncture skills in simulation training

- From Dr. Anna Liu, with Dr. Hugh McMillan (NMD4C) as a co-author.

Evolution of sleep complaints in myotonic dystrophy type 1: a 9-year longitudinal study

- From Dr. Luc Laberge, with Dr. Cynthia Gagnon (NMD4C) as a co-author.

Respiratory Management of Patients With Neuromuscular Weakness: An American College of Chest Physicians Clinical Practice Guideline and Expert Panel Report

- From Dr. Akram Khan with Dr. Reshma Amin (NMD4C) as a co-author.

A protocol for single nucleus RNA-seq from frozen skeletal muscle

- From Dr. Tyler Soule, with Dr. Gerald Pfeffer (NMD4C) as a co-author.

Incidence and Mortality of Children Receiving Home Mechanical Ventilation

- From Dr. Reshma Amin (NMD4C).

Periostin as a blood biomarker of muscle cell fibrosis, cardiomyopathy and disease severity in myotonic dystrophy type 1

- From Dr. Chi Nguyen, with Dr. Elise Duchesne, Dr. Cynthia Gagnon and Dr. Hanns Lochmüller (NMD4C), as co-authors.

Variability in newborn screening across Canada: spinal muscular atrophy and beyond

- From Dr. Emilie Groulx-Boivin, with Dr. Homira Osman, Stacey Lintern, Dr. Maryam Oskoui, and Dr. Hugh McMillan (NMD4C) as co-authors.

DNA methylation at the DMPK gene locus is associated with cognitive functions in myotonic dystrophy type 1

- From Dr. Édith Breton, with Dr. Cynthia Gagnon (NMD4C) as a co-author.