New Publications from NMD4C Investigators

December 2023

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.

The emerging spectrum of neurodevelopmental comorbidities in early-onset Spinal Muscular Atrophy

- From Dr Giovanni Baranello with Dr Hanns Lochmüller, Dr Hugh McMillan, Dr Maryam Oskoui, Dr Hernan Gonorazky, and Dr Alberto Aleman (NMD4C) as collaborators as a part of the Neurodevelopment in SMA Working Group

Subcutaneous batoclimab in generalized myasthenia gravis: Results from a Phase 2a trial with an open-label extension

- From Dr Richard Nowak with Dr Vera Bril (NMD4C) as a co-author.

The myotubular and centronuclear myopathy patient registry: a multifunctional tool for translational research

- From Dr Joanne Bullivant with Dr James Dowling (NMD4C) as a co-author.

Long term peripheral AAV9-SMN gene therapy promotes survival in a mouse model of spinal muscular atrophy

- From Aiofe Reilly, with Dr Rashmi Kothary (NMD4C) as a co-author.

Effects of gene replacement therapy with resamirigene bilparvovec (AT132) on skeletal muscle pathology in X-linked myotubular myopathy: results from a substudy of the ASPIRO open-label clinical trial

- From Dr Michael Lawlor, with Dr James Dowling (NMD4C) as a co-author.

Life-Saving Treatments for Spinal Muscular Atrophy: Global Access and Availability

- From Dr Victor Armengol with Dr Gerald Pfeffer (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)

Toward a Better Understanding of Walking Speed in Ataxia of Charlevoix-Saguenay: a Factor Exploratory Study

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

Clinical and genetic keys to cerebellar ataxia due to FGF14 GAA expansions

- From Dr Jean-Loup Méreaux with Dr Bernard Brais (NMD4C) as a co-author.

Pregnancy planning may impact maternal and neonatal outcomes in people with myasthenia gravis

- From Dr Saja Anabusi with Dr Aaron Izenberg (NMD4C) as a co-author.

Safety and Efficacy of Nipocalimab in Patients With Generalized Myasthenia Gravis

- From Dr Carlo Antozzi with Dr Vera Bril (NMD4C) as a co-author.

Bisphosphonates in Glucocorticoid-Treated Patients With Duchenne Muscular Dystrophy

- From Dr Erik Landfeldt with Dr Hanns Lochmüller and Dr Leanne Ward (NMD4C) as co-authors.

Misfolding of fukutin-related protein (FKRP) variants in congenital and limb girdle muscular dystrophies

- From Dr Christopher Esapa with Dr Hanns Lochmüller (NMD4C) as a co-author.

A study on the incidence and prevalence of 5q spinal muscular atrophy in Canada using multiple data sources

- From Dr Tiffany Price with Dr Victoria Hodgkinson, Dr Lawrence Korngut, and Dr Jean Mah (NMD4C) as co-authors.

Quick, Effective Screening Tasks Identify Children With Medical Conditions or Disabilities Needing Physical Literacy Support

- From Dr Patricia Longmuir with Dr Hugh McMillan (NMD4C) as a co-author.

Congenital tremor and myopathy secondary to novel MYBPC1 variant

- From Dr Heather Leduc-Pessah with Dr Jodi Warman Chardon (NMD4C) as a co-author.

The First Decade of Journal of Neuromuscular Diseases: Supporting and Advancing the Rapidly Evolving Field of Translational Research

- From Dr Hanns Lochmüller (NMD4C).

The Clinical Development of Taldefgrobep Alfa: An Anti-Myostatin Adnectin for the Treatment of Duchenne Muscular Dystrophy

- From Dr Francesco Muntonu with Dr Hugh McMillan (NMD4C) as a co-author.