



New Publications from NMD4C Investigators

May 2023 - June 2023

Long-term mechanical ventilation and transitions in care: A narrative review

- From Dr. Lena Xiao, with Dr. Reshma Amin (NMD4C) as a co-author.

Screening for psychosocial risk in caregivers of children with medical complexity during the COVID-19 pandemic: a cross-sectional study

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

Non-invasive ventilation usage and adherence in children and adults with Duchenne muscular dystrophy: A multicenter analysis

- From Dr. Manju Hurvitz, with Dr. Reshma Amin (NMD4C) as a co-author.

A Review of Brain and Pituitary Gland MRI Findings in Patients with Ataxia and Hypogonadism

- From Dr. Alessandra Scaravilli, with Dr. Bernard Brais (NMD4C) as a co-author.

Restoring calcium homeostasis in Purkinje cells arrests neurodegeneration and neuroinflammation in the ARSACS mouse model

- From Dr. Andrea Del Bondio, with Dr. Bernard Brais (NMD4C) as a co-author.

GAA-FGF14 ataxia (SCA27B): phenotypic profile, natural history progression and 4-aminopyridine treatment response

- From Dr. Carlo Wilke, with Dr. Bernard Brais (NMD4C) as a co-author.

A 16-year-old boy with lower extremity muscle wasting and pain [case report]

- From Dr. Mona Hnaini, with Dr. Craig Campbell (NMD4C) as a co-author.



ZnT1 induces a crosstalk between T-type and L-type calcium channels through interactions with Raf-1 kinase and the calcium channel β 2 subunit

- From Dr. Merav Mor, with Dr. Craig Campbell (NMD4C) as a co-author.

Duchenne Muscular Dystrophy Fatigue Trajectories

- From Dr. Yi Sally Wei, with Dr. Craig Campbell (NMD4C) as a co-author.

Optimization of a Series of Novel, Potent and Selective Macrocyclic SYK Inhibitors

- From Dr. Neil Grimster, with Dr. James Dowling (NMD4C) as a co-author.

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

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

Responsiveness of the SARA and natural history in 884 recessive and early onset ataxia patients

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

Do classical and computerized cognitive tests have equal intrarater reliability in myotonic dystrophy type 1?

- From Dr. Julie Fortin, with Dr. Cynthia Gagnon (NMD4C) as a co-author.

The Development of a New Patient-Reported Outcome Measure in Recessive Ataxias: The Person-Reported Ataxia Impact Scale

- From Dr. Marjolaine Tremblay, with Dr. Cynthia Gagnon (NMD4C) as a co-author.

Wheelchair mobility, motor performance and participation of adult wheelchair users with ARSACS: a cross-sectional study

- From Dr. Julie Bourassa, Dr. Cynthia Gagnon and Dr. Bernard Brais (NMD4C) as co-authors.



Expression and subcellular localization of mitochondrial docking protein, syntaphilin, in oligodendrocytes and CNS myelin sheath

- From Dr. Diane Nakamura, with Dr. Rashmi Kothary (NMD4C) as a co-author.

Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects

- From Dr. Andreas Roos , with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Presynaptic Congenital Myasthenic Syndromes: Understanding Clinical Phenotypes through In vivo Models

- From Dr. Alessia Pugliese, with Dr. Hanns Lochmüller as a co-author.

Mitochondrial Mutations Can Alter Neuromuscular Transmission in Congenital Myasthenic Syndrome and Mitochondrial Disease

- From Dr. Kaela O'Connor, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Paper II: thematic framework analysis of registry-based randomized controlled trials provided insights for designing trial ready registries

- From Dr. Karolin Krause, with Dr. Maryam Oskoui (NMD4C) as a co-author.

The emerging spectrum of foetal acetylcholine receptor antibody-associated disorders (FARAD)

- From Dr. Nicholas Allen, with Dr. Maryam Oskoui (NMD4C) as a co-author.

Provisional practice recommendation for the management of myopathy in VCP-associated multisystem proteinopathy

- From Dr. Bhaskar Roy, with Dr. Gerald Pfeffer (NMD4C) as a co-author.

Brachial Plexopathy following Minimally Invasive Coronary Artery Bypass Grafting

- From Dr. Seyed-Mohammad Fereshtehnejad, with Dr. Jodi Warman-Chardon (NMD4C) as a co-author.