



New Publications from NMD4C Investigators

June 2024

[All hands on deck: The multidisciplinary rehabilitation assessment and management of hand function in persons with neuromuscular disorders](#)

- From Dr Colleen O'Connell with Dr Stephanie Plamondon (NMD4C) as a co-author.

[Advancing the Understanding of Vesicle-Associated Membrane Protein 1-Related Congenital Myasthenic Syndrome: Phenotypic Insights, Favorable Response to 3,4-Diaminopyridine, and Clinical Characterization of Five New Cases](#)

- From Dr Daniel Natera-de Benito with Dr Hanns Lochmüller (NMD4C) as a co-author.

[Sex, racial, and ethnic disparities in motor neuron disease: clinical trial enrolment](#)

- From Dr Chia-Chen Tsai with Dr Agessandro Abrahao (NMD4C) as a co-author.

[Chronic glucocorticoid management in neuromuscular disease: A survey of neuromuscular neurologists](#)

- From Dr Lora Stepanian with Dr Aaron Izenberg and Dr Victoria Hodgkinson (NMD4C) as co-authors.

[Whole-body magnetic resonance neurography in patients with chronic inflammatory demyelinating polyneuropathy](#)

- From Dr Davood Fathi with Dr Vera Brill (NMD4C) as a co-author.

[Acute effects of a ketone monoester, whey protein, or their coingestion on mTOR trafficking and protein-protein colocalization in human skeletal muscle](#)

- From Dr Sarkis J Hannaian with Dr Benoit Gentil (NMD4C) as a co-author.

[Digital Gait Outcomes for Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay \(ARSACS\): Discriminative, Convergent, and Ecological Validity in a Multicenter Study \(PROSPAX\)](#)

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

[Exploring Integrin \$\alpha 5 \beta 1\$ as a Potential Therapeutic Target for Pulmonary Arterial Hypertension: Insights from Comprehensive Multicenter Preclinical Studies](#)

- From Dr Sarah-Eve Lemay with Dr James Dowling (NMD4C) as a co-author.

[Identification of a mosaic MTOR variant in purified neuronal DNA in a patient with focal cortical dysplasia using a novel depth electrode harvesting technique](#)

- From Dr Karl Martin Klein with Dr Gerald Pfeffer (NMD4C) as a co-author.

[Office-based respiratory assessment in patients with generalized myasthenia gravis](#)

- From Dr Monica Alcantera with Dr Vera Brill (NMD4C) as a co-author.

[Resistance training in women with myotonic dystrophy type 1: a multisystemic therapeutic avenue](#)



- From Dr Laura Girard-Côté with Dr Cynthia Gagnon and Dr Élise Duchesne (NMD4C) as a co-author.

[Qualitative Analysis of Initial Palliative Care Consultations in Amyotrophic Lateral Sclerosis](#)

- From Christine L Watt with Dr Jocelyn Zwicker (NMD4C) as a co-author.

[Neurological disorders caused by novel non-coding repeat expansions: clinical features and differential diagnosis](#)

- From Dr Elisa Vegezzi with Dr Bernard Brais (NMD4C) as a co-author.

[Safety and efficacy of arimoclomol in patients with early amyotrophic lateral sclerosis \(ORARIALS-01\): a randomised, double-blind, placebo-controlled, multicentre, phase 3 trial](#)

- From Dr Michael Benatar with Dr Christen Shoosmith, Dr Agessandro Abrahao and Dr Lorne Zinman (NMD4C) as co-authors.

[Treatment Options in Spinal Muscular Atrophy: A Pragmatic Approach for Clinicians](#)

- From Dr Sithara Ramdas with Dr Maryam Oskoui (NMD4C) as a co-author.

[Value of multi-modality small fiber assessments in a genotypically diverse cohort of transthyretin-related amyloidosis in the early stages of disease](#)

- From Dr Monica Alcantara with Dr Vera Brill (NMD4C) as a co-author.

[An MRI evaluation of white matter involvement in paradigmatic forms of spastic ataxia: results from the multi-center PROSPAX study](#)

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

[Spinocerebellar ataxia 27B: a frequent and slowly progressive autosomal-dominant cerebellar ataxia-experience from an Italian cohort](#)

- From Dr Sara Satolli with Dr Bernard Brais (NMD4C) as a co-author.

[Valosin-Containing Protein \(VCP\): A Review of Its Diverse Molecular Functions and Clinical Phenotypes](#)

- From Dr Carly S Pontifex with Dr Gerald Pfeffer (NMD4C) as a co-author.

[Association of Gestational Age at Birth and Changes on MRI With Prevalence and Spectrum of Comorbidities in Children With Cerebral Palsy](#)

- From Dr Heather Pেকেles with Dr Maryam Oskoui (NMD4C) as a co-author.

[Temporal and spatial progression of microstructural cerebral degeneration in ALS: A multicentre longitudinal diffusion tensor imaging study](#)

- From Dr Hans-Peter Müller with Dr Agessandro Abrahao, Dr Lawrence Korngut, and Dr Lorne Zinman (NMD4C) as co-authors.



[Pharmacologic Activation of Integrated Stress Response Kinases Inhibits Pathologic Mitochondrial Fragmentation](#)

- From Dr Kelsey R Baron with Dr Gerald Pfeffer (NMD4C) as a co-author.

[A common flanking variant is associated with enhanced stability of the FGF14-SCA27B repeat locus](#)

- From Dr David Pellerin with Dr Bernard Brais (NMD4C) as a co-author.

[AAV gene therapy for hereditary spastic paraplegia type 50: a phase 1 trial in a single patient](#)

- From Dr James Dowling (NMD4C).

[A Systematic Literature Review of the Natural History of Respiratory, Swallowing, Feeding, and Speech Functions in Spinal Muscular Atrophy \(SMA\)](#)

- From Dr Yasmina Martí with Dr Maryam Oskoui (NMD4C) as a co-author.

[The latest on positive airway pressure for pediatric obstructive sleep apnea](#)

- From Dr Natalia S Escobar with Dr Reshma Amin (NMD4C) as a co-author.

[Reduced Age-Dependent Penetrance of a Large FGF14 GAA Repeat Expansion in a 74-Year-Old Woman from a German Family with SCA27B](#)

- From Dr David Pellerin with Dr Bernard Brais (NMD4C) as a co-author.