



New Publications from NMD4C Investigators

November 2023

[Assessing the Measurement Properties of the Self-Administered Amyotrophic Lateral Sclerosis Functional Rating Scale-Revised \(ALSFRS-R\): A Rasch Analysis](#)

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

[Novel RAB39B variant associated intellectual disability and levodopa-responsive young-onset parkinsonism](#)

- From Dr Ali Abusrair with Dr Gerald Pfeffer (NMD4C) as a co-author.

[Differential effect of Fas activation on spinal muscular atrophy motoneuron death and induction of axonal growth](#)

- From Dr Salim Benlefki with Dr Rashmi Kothary (NMD4C) as a co-author.

[Towards the Identification of Biomarkers for Muscle Function Improvement in Myotonic Dystrophy Type 1](#)

- From Dr Amira Aoussim with Dr Elise Duchesne (NMD4C) as a co-author.

[Resistance Exercise Training Rescues Mitochondrial Dysfunction in Skeletal Muscle of Patients with Myotonic Dystrophy Type 1](#)

- From Dr Valeria Di Leo with Dr Elise Duchesne (NMD4C) as a co-author.

[The Medical Action Ontology: A tool for annotating and analyzing treatments and clinical management of human disease](#)

- From Dr Leigh Carmody with Dr Hanns Lochmüller (NMD4C) as a co-author.

[Sustained clinical benefit following systemic gene replacement therapy in Duchenne muscular dystrophy](#)

- From Dr Hugh McMilland with Dr Hanns Lochmüller (NMD4C) as a co-author.

[Safety and efficacy of gene replacement therapy for X-linked myotubular myopathy \(ASPIRO\): a multinational, open-label, dose-escalation trial](#)

- From Dr Perry Shieh with Dr James Dowling (NMD4C) as a co-author.

[French Translation and Cross-cultural Adaptation of the Scale for the Assessment and Rating of Ataxia](#)

- From Dr Dax Bourcier with Dr Cynthia Gagnon and Dr Elise Duchesne (NMD4C) as co-authors.

[Characterization of muscle strength and mobility in oculopharyngeal muscular dystrophy](#)

- From Dr Jean-Denis Brisson with Dr Cynthia Gagnon (NMD4C) as a co-author.



[Reply to letter in response to Rethinking the diagnosis of double-seronegative myasthenia gravis](#)

- From Dr Rodrigo Martinez-Harms with Dr Vera Brill (NMD4C) as a co-author.

[Long term effects of continuous positive airway pressure treatment of obstructive sleep apnea-hypopnea syndrome in multiple sclerosis patients](#)

- From Dr Mylene Mazerolle with Dr Daria Trojan (NMD4C) as a co-author.

[Opening the digital front door for individuals using long-term in-home ventilation \(LIVE\) during a pandemic- implementation, feasibility and acceptability](#)

- From Dr Reshma Amin (NMD4C)