



New Publications from NMD4C Investigators

March 2025

Biallelic variants in GTF3C3 encoding a subunit of the TFIIC2 complex are associated with neurodevelopmental phenotypes in humans and zebrafish

- From Dr Mohamed S Abdel-Hamid with Dr Kessen Patten (NMD4C) as a co-author.

Beyond sialylation: Exploring the multifaceted role of GNE in GNE myopathy

- From Dr Beatriz L Pereira with Dr Hanns Lochmüller (NMD4C) as a co-author.

Thoracic Electric Impedance Tomography Detects Lung Volume Changes in Amyotrophic Lateral Sclerosis

- From Dr Gregory Hansen with Dr Kerri Schellenberg (NMD4C) as a co-author.

Type-2 innate signals are dispensable for skeletal muscle regeneration and pathology linked to Duchenne muscular dystrophy

- From Dr Melina Messing with Dr Fabio Rossi (NMD4C) as a co-author.

Characterization of a novel zebrafish model of MTMR5-associated Charcot-Marie-Tooth disease type 4B3

- From Dr Jordan Lindzon with Dr James Dowling (NMD4C) as a co-author.

Five-Year Results With Patisiran for Hereditary Transthyretin Amyloidosis With Polyneuropathy: A Randomized Clinical Trial With Open-Label Extension

- From Dr David Adams with Dr Michelle Mezei (NMD4C) as a co-author.

Social Participation Restrictions and Explanatory Factors in Adults with Oculopharyngeal Muscular Dystrophy

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

Influence of CTG repeats from the human DM1 locus on murine gut microbiota

- From Dr Manijeh Mahdavi with Dr Elise Duchesne, Dr Cynthia Gagnon and Dr Nicolas Dumont (NMD4C) as co-authors.

A Tailored Home-Based Training Program Improved Ataxia Severity and Participation in Adults With ARSACS

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

Safety and efficacy of chronic weekly rozanolixizumab in generalized myasthenia gravis: the randomized open-label extension MG0004 study

- From Dr Vera Bril with Dr Rami Massie (NMD4C) as a study investigator

Generation of a novel mouse model of nemaline myopathy due to recurrent NEB exon 55 deletion

- From Dr Zachary Coulson with Dr James Dowling (NMD4C) as a co-author.

Efficient Isolation and Ex Vivo Differentiation of Murine Satellite Cells from Healthy and Dystrophic Muscle

- From Alessio A Cusmano with Dr Natasha Chang (NMD4C) as a co-author.

Optimizing electrical field stimulation parameters reveals the maximum contractile function of human skeletal muscle microtissues

- From Dr Yekaterina Tiper with Dr Penney Gilbert (NMD4C) as a co-author.

Reldesemtiv in Amyotrophic Lateral Sclerosis: Results From the COURAGE-ALS Randomized Clinical Trial

- From Dr Jeremy M Shefne with Dr Lorne Zinman, Dr Christen Shoesmith, Dr Colleen O'Connell, Dr Jocelyn Zwicker (NMD4C) as co-authors.

Unraveling undiagnosed rare disease cases by HiFi long-read genome sequencing

- From Dr Wouter Steyaert with Dr Hanns Lochmüller (NMD4C) as a co-author.

Biological sex does not impact intrinsic mitochondrial respiration supported by complexes I and II in human skeletal muscle

- From Dr Emily J Ferguson with Dr Chris Perry (NMD4C) as a co-author.

Transition of patients with Duchenne muscular dystrophy from paediatric to adult care: An international Delphi consensus study

- From Dr Diana Castro with Dr Leanne Ward (NMD4C) as a co-author.

Parental perspectives on family-centered care in pediatric neurology: An explanatory sequential mixed-methods study

- From Dr Ege Sarikaya with Dr Kathryn Selby (NMD4C) as a co-author.

Is Duchenne gene therapy a suitable treatment despite its immunogenic class effect?

- From Dr Annie Tang with Dr Toshifumi Yokota (NMD4C) as a co-author.

The MFN2 Q367H variant reveals a novel pathomechanism connected to mtDNA-mediated inflammation

- From Dr Mashiat Zaman with Dr Gerald Pfeffer (NMD4C) as a co-author.



AOC 1044 induces exon 44 skipping and restores dystrophin protein in preclinical models of Duchenne muscular dystrophy

- From Dr Usue Etxaniz with Dr Toshifumi Yokota (NMD4C) as a co-author.

Lung Volume Recruitment and Quality of Life in Duchenne Muscular Dystrophy: Secondary Analysis of the STEADFAST Randomized Controlled Trial

- From Dr Anne Tsampalieros with Dr Jean Mah, Dr Craig Campbell, Dr Hugh McMillan, Dr Laura McAdam and Dr Mark Tarnopolsky (NMD4C) as co-authors.

FGF14 GAA Intronic Expansion in Unsolved Adult-Onset Ataxia in the Care4Rare Canada Consortium

- From Dr Alexanne Cuillerier with Dr Jodi Warman Chardon and Dr Gerald Pfeffer (NMD4C) as co-authors.

Effect of Rozanolixizumab on Ocular Symptoms in Patients with Generalized Myasthenia Gravis: A Post Hoc Item-level Analysis of Myasthenia Gravis-specific Outcomes in MycarinG (P1-11.006)

- From Dr Zabeen K Mahuwala with Dr Veral Bril (NMD4C) as a co-author.