New Publications from NMD4C Investigators

February 2025

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.

Analysis of Exon Skipping Applicability for Dysferlinopathies

- From Dr Jamie Leckie with Dr Toshifumi Yokota (NMD4C) as a co-author.

Pharmacologic activation of integrated stress response kinases inhibits pathologic mitochondrial fragmentation

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

Longitudinal Outcomes Among Patients With Duchenne Muscular Dystrophy: A Canadian Retrospective Population-Based Study

- From Dr Christina Qian with Dr Jean Mah (NMD4C) as a co-author.

Blood biomarker fingerprints in a cohort of patients with CHRNE-related congenital myasthenic syndrome

- From Dr Adela Della Marina with Dr Hanns Lochmüller (NMD4C) as a co-author.

Antisense oligonucleotide-mediated exon 27 skipping restores dysferlin function in dysferlinopathy patient-derived muscle cells

- From Dr Saeed Anwar with Dr Toshifumi Yokota (NMD4C) as a co-author.

Current biomarkers in inclusion body myositis

- From Dr Eden Daniel with Dr Jodi Warman-Chardon (NMD4C) as a co-author.

Phenotype-driven genomics enhance diagnosis in children with unresolved neuromuscular diseases

- From Dr Berta Estévez-Arias with Dr Alberto Aleman (NMD4C) as a co-author.

International collaboration to improve knowledge on myotonic dystrophy type 2

- From Dr Stojan Peric with Dr Craig Campbell, Dr Victoria Hodgkinson and Dr Lawrence Korngut (NMD4C) as co-authors.

Plasma-derived protein and imaging biomarkers distinguish disease severity in oculopharyngeal muscular dystrophy

- From Dr Ian C Smith with Dr Hanns Lochmüller, Dr Bernard Brais, Dr Mireille Khacho and Dr Jodi Warman-Chardon (NMD4C) as co-authors.

Report on the 6th Ottawa International Conference on Neuromuscular Disease & Biology - September 7-9, 2023, Ottawa, Canada

- From Dr Jodi Warman-Chardon with Dr Bernard Jasmin and Dr Rashmi Kothary (NMD4C) as a co-author.

Peripheral defects precede neuromuscular pathology in the Smn2B/- mouse model of spinal muscular atrophy

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

A Parent Project Muscular Dystrophy-sponsored International Workshop Report on Endocrine and Bone Issues in Patients with Duchenne Muscular Dystrophy: An Ever-changing Landscape

- From Dr Leanne M Ward with Dr Hugh J McMillan (NMD4C) as a co-author.

Dominant rhabdomyolysis linked to a recurrent ATP2A2 variant reducing SERCA2 function in muscle

- From Sivasankar Malaichamy with Dr Hanns Lochmüller (NMD4C) as a co-author.

Clinically Meaningful Improvement in Physical Fatigue and Muscle Weakness Fatigability with Rozanolixizumab: Post-hoc Analysis of MG Symptoms PRO Responder Rate in the MycarinG study (P4-11.001)

- From Dr Ali A Habib with Dr Vera Bril (NMD4C) as a co-author.

Impact of liver-specific survival motor neuron (SMN) depletion on central nervous system and peripheral tissue pathology

- From Dr Monique Marylin Alves de Almeida with Dr Rashmi Kothary (NMD4C) as a co-author.

Genetic characterization of a rare case of pheochromocytoma in a pulmonary transplant patient

- From Dr Stéfanie Parisien-La Salle with Dr Martine Tetreault (NMD4C) as a co-author.

New Horizons in Myotonic Dystrophy Type 1: Cellular Senescence as a Therapeutic Target

- From Dr Cécilia Légaré with Dr Elise Duchesne and Dr Nicolas Dumont (NMD4C) as co-authors.

Involvement of the Superior Cerebellar Peduncles in GAA- FGF14 Ataxia

- From Dr Shihan Chen with Dr Bernard Brais (NMD4C) as a co-author.

X-linked myopathy with excessive autophagy: characterization and therapy testing in a zebrafish model

- From Dr Lily Huang with Dr James Dowling (NMD4C) as a co-author.

Integrating Machine Learning-Based Approaches into the Design of ASO Therapies

- From Dr Jamie Leckie with Dr Toshifumi Yokota (NMD4C) as a co-author.

Late-onset vestibulocerebellar ataxia: clinical and genetic studies in a long follow-up series of 50 patients

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

Loss-of-function mitochondrial DNA polymerase gamma variants cause vascular smooth muscle cells to secrete a diffusible mitogenic factor

- From Dr Samantha Rothwell with Dr Michelle Mezei (NMD4C) as a co-author.

Long-term safety of cyclical rozanolixizumab in patients with generalized myasthenia gravis: Results from the Phase 3 MycarinG study and an open-label extension

- From Dr Ali A Habib with Dr Vera Bril (NMD4C) as a co-author.