New Publications from NMD4C Investigators

December 2024

Rethinking Drug Reimbursement Criteria in Amyotrophic Lateral Sclerosis

- From Dr Lorne Zinman with Dr Agessandro Abrahao (NMD4C) as a co-author.

Oculomotor findings in spinocerebellar ataxia 27B: a case series

- From Dr Guillemette Clément with Dr Bernard Brais (NMD4C) as a co-author.

Assessing corneal dendritic cells in glucose dysregulation small-fibre neuropathy

- From Dr Juan Francisco Idiaquez with Dr Vera Bril (NMD4C) as a co-author.

Long-term safety and tolerability of hyaluronidase-facilitated subcutaneous immunoglobulin 10% as maintenance therapy for chronic inflammatory demyelinating polyradiculoneuropathy: Results from the ADVANCE-CIDP 3 trial

- From Dr Robert D M Hadden with Dr Vera Bril (NMD4C) as a co-author.

Promoting Intensive Transitions for Children and Youth with Medical Complexity from Paediatric to Adult Care: the PITCare study-protocol for a randomised controlled trial

- From Dr Sara Santos with Dr Reshma Amin (NMD4C) as a co-author.

Efficacy and safety of maintenance intravenous immunoglobulin in generalized myasthenia gravis patients with acetylcholine receptor antibodies: A multicenter, double-blind, placebo-controlled trial

- From Dr Vera Bril (NMD4C).

Identification of the Wnt signal peptide that directs secretion on extracellular vesicles

- From Dr Uxia Gurriaran-Rodriguez with Dr Rashmi Kothary and Dr Michael Rudnicki (NMD4C) as co-authors.

Longitudinal analysis of glymphatic function in amyotrophic lateral sclerosis and primary lateral sclerosis

- From Dr Rachel J Sharkey with Dr Lawrence Korngut and Dr Gerald Pfeffer (NMD4C) as a co-author.

Aging and infectious diseases in myasthenia gravis

- From Dr Vera Bril (NMD4C).

C9orf72 repeat expansion creates the unstable folate-sensitive fragile site FRA9A

- From Dr Mila Mirceta with Dr Agessandro Abrahao (NMD4C) as a co-author.

Diagnosing missed cases of spinal muscular atrophy in genome, exome, and panel sequencing datasets

- From Dr Ben Weisburd with Dr Hanns Lochmüller (NMD4C) as a co-author.

Development of a riboflavin-responsive model of riboflavin transporter deficiency in zebrafish

- From Catherine M Choueiri with Dr Alex MacKenzie and Dr Hanns Lochmüller (NMD4C) as co-authors.

Mortality Trends and Causes of Death in Myotonic Dystrophy Type 1 Patients From the UK Clinical Practice Research Datalink

- From Dr Rotana Alsaggaf with Dr Hanns Lochmüller (NMD4C) as a co-author.

A Study on the Incidence and Prevalence of 5q Spinal Muscular Atrophy in Canada Using Multiple Data Sources

- From Dr Tiffany R Price with Dr Victoria Hodgkinson, Dr Lawrence Korngut and Dr Jean Man (NMD4C) as co-authors.

Wnt7a is required for regeneration of dystrophic skeletal muscle

- From Dr Uxia Gurriaran-Rodriguez with Dr Michael Rudnicki (NMD4C) as a co-author.

Exome and Genome Sequencing to Diagnose the Genetic Basis of Neonatal Hypotonia: An International Consortium Study

- From Dr Sarah U Morton with Dr James Dowling (NMD4C) as a co-author.

The First Case of Autosomal Recessive Cerebellar Ataxia with Prominent Paroxysmal Non-kinesigenic Dyskinesia Caused by a Truncating FGF14 Variant in a Turkish Patient

- From Dr Dilşad Türkdoğan with Dr Bernard Brais (NMD4C) as a co-author.

Measurement Properties of the Dysphagiameter for the Assessment of Dysphagia in Oculopharyngeal Muscular Dystrophy

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

Identification of a Founder GLDN Variant Associated With "Lethal" Arthrogryposis in Nunavik Inuit: Implications for Obstetrical and Long-Term Survivors' Management

- From Dr Alexa McAdam with Dr Maryam Oskoui (NMD4C) as a co-author.

Intrinsic Muscle Stem Cell Dysfunction Contributes to Impaired Regeneration in the mdx Mouse

- From Dr Marie E Esper with Dr Michael Rudnicki (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 (NMD4C) as a co-author.

Sensory neuropathy in patients with Pompe disease: a case series in Iran

- From Dr Marzieh Babaee with Dr Hanns Lochmüller (NMD4C) as a co-author.

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.

[Expanding the Molecular Genetic Landscape of Dystrophinopathies and Associated Phenotypes]

- From Dr Katja Neuhoff with Dr Hanns Lochmüller (NMD4C) as a co-author.