New Publications from NMD4C Investigators

January 2024

The FGF14 GAA repeat expansion in Greek patients with late-onset cerebellar ataxia and an overview of the SCA27B phenotype across populations

- From Dr Chrisoula Kartanou with Dr Bernard Brais (NMD4C) as a co-author.

Proposed diagnostic criteria for the diagnosis of hypophosphatasia in children and adolescents: results from the HPP International Working Group

- From Dr Eric Rush with Dr Leanne Ward (NMD4C) as a co-author.

Childhood-Onset Myopathy With Preserved Ambulation Caused by a Recurrent ADSSL1 Missense Variant

- From Dr Dipti Baskar with Dr Hanns Lochmüller (NMD4C) as a co-author.

Subcutaneous batoclimab in generalized myasthenia gravis: Results from a Phase 2a trial with an open-label extension

- From Dr Richard Nowak with Dr Vera Bril (NMD4C) as a co-author.

Biallelic SOX8 Variants Associated With Novel Syndrome With Myopathy, Skeletal Deformities, Intellectual Disability, and Ovarian Dysfunction

- From Dr Jodi Warman-Chardon (NMD4C) with Dr Hanns Lochmüller and Dr Bernard Brais (NMD4C) as co-authors.

Standardization of zebrafish drug testing parameters for muscle diseases

- From Dr Muthukumar Karuppasamy with Dr James Dowling (NMD4C) as a co-author.

Motor Unit Number Index of the Upper Trapezius: A Meta-Analysis and Cross-sectional Study of Its Reliability

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

Evolution of Sleep Complaints in Myotonic Dystrophy Type 1: A 9-Year Longitudinal Study

- From Dr Luc Laberge with Dr Cynthia Gagnon (NMD4C) as a co-author.

Does Spinocerebellar ataxia 27B mimic cerebellar multiple system atrophy?

- From Dr Thomas Wirth with Dr Bernard Brais (NMD4C) as a co-author.

GAA- FGF14-Related Ataxia

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

Spinocerebellar ataxia 27B: A novel, frequent and potentially treatable ataxia

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

Characterization of a novel variant in the HR1 domain of MFN2 in a patient with ataxia, optic atrophy and sensorineural hearing loss

- From Dr Govinda Sharma with Dr Gerald Pfeffer (NMD4C) as a co-author.

Recurring homozygous ACTN2 variant (p.Arg506Gly) causes a recessive myopathy

- From Dr Sandra Donkervoort with Dr Jodi Warman-Chardon (NMD4C) as a co-author.

Long-term safety, tolerability, and efficacy of efgartigimod (ADAPT+): interim results from a phase 3 open-label extension study in participants with generalized myasthenia gravis

- From Dr James Howard Jr, with Dr Vera Bril (NMD4C) as a co-author.