



## New Publications from NMD4C Investigators

January 2025

[The FGF14 GAA repeat expansion is a major cause of ataxia in the Cypriot population](#)

- From Dr Ioannis Livanos with Dr Bernard Brais (NMD4C) as a co-author.

[Aging and infectious diseases in myasthenia gravis](#)

- From Dr Vera Brill (NMD4C).

[Safety and Efficacy of IV Onasemnogene Apeparvovec for Pediatric Patients With Spinal Muscular Atrophy: The Phase 3b SMART Study](#)

- From Dr Hugh J McMillan (NMD4C).

[An exploratory qualitative assessment of patient and clinician perspectives on patient-reported outcome measures and disease-modifying therapies in adults with spinal muscular atrophy](#)

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

[How to break the news in amyotrophic lateral sclerosis/motor neuron disease: practical guidelines from experts](#)

- From Dr Colleen O'Connell (NMD4C).

[Recent Advances in the Genetics of Ataxias: An Update on Novel Autosomal Dominant Repeat Expansions](#)

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

[Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses](#)

- From Dr Steven Laurie with Dr Hanns Lochmüller (NMD4C) as a co-author.

[Associations between water exposure and the development of amyotrophic lateral sclerosis: a matched case-control study](#)

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

[Safety and efficacy of nipocalimab in adults with generalised myasthenia gravis \(Vivacity-MG3\): a phase 3, randomised, double-blind, placebo-controlled study](#)

- From Dr Carlo Antozzi with Dr Vera Brill (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.

[Skeletal Muscle Mitochondrial and Autophagic Dysregulation Are Modifiable in Spinal Muscular Atrophy](#)



- From Dr Andrew I Mikhail with Dr Rashmi Kothary and Dr Mark Tarnopolsky (NMD4C) as co-authors.