New Publications from NMD4C Investigators

September 2024

GNE Myopathy: Genotype - Phenotype Correlation and Disease Progression in an Indian Cohort

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

Using jitter analysis with concentric needle electrodes to assess disease status and treatment responses in myasthenia gravis

- From Dr Vinaya Bhandari with Dr Vera Bril (NMD4C) as a co-author.

Changes in Physiopathological Markers in Myotonic Dystrophy Type 1 Skeletal Muscle: A 3-Year Follow-up Study

- From Dr Marie-Pier Roussel with Drs Aymeric Ravel-Chapuis, Bernard Jasmin, Cynthia Gagnon and Elise Duchesne (NMD4C) as co-authors.

How to break the news in amyotrophic lateral sclerosis/motor neuron disease: practical guidelines from experts

- From Dr Colleen O'Connell (NMD4C).

Clinical, Radiological and Pathological Features of a Large American Cohort of Spinocerebellar Ataxia (SCA27B)

- From Dr Widad Abou Chaar with Dr Bernard Brais (NMD4C) as a co-author.

274th ENMC international workshop: recommendations for optimizing bone strength in neuromuscular disorders. Hoofddorp, The Netherlands, 19-21 January 2024

- From Dr Nicol C Voermans with Dr Leanne Ward (NMD4C) as a co-author.

Biallelic PTPMT1 variants disrupt cardiolipin metabolism and lead to a neurodevelopmental syndrome

- From Dr Micol Falabella with Dr Hanns Lochmüller (NMD4C) as a co-author.

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

- From Dr Samar Muslemani (former NMD4C Knowledge Translation Coordinator), with Drs Bernard Brais and Cynthia Gagnon (NMD4C) as co-authors.

An algorithm for discontinuing mechanical ventilation in boys with x-linked myotubular myopathy after positive response to gene therapy: the ASPIRO experience

- From Dr Robert J Graham with Dr Reshma Amin (NMD4C) as a co-author.

Cognitive outcomes following unilateral magnetic resonance-guided focused ultrasound thalamotomy for essential tremor: findings from two cohorts

- From Dr Julie Petersen with Dr Agessandro Abrahao (NMD4C) as a co-author.

Efficacy and safety of rozanolixizumab in patients with muscle-specific tyrosine kinase autoantibody-positive generalised myasthenia gravis: a subgroup analysis of the randomised, double-blind, placebo-controlled, adaptive phase III MycarinG study

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

Macrostructural Brain Abnormalities in Spinal Muscular Atrophy: A Case-Control Study

- From Dr Emilie Groulx-Boivin with Dr Maryam Oskoui (NMD4C) as a co-author.

Sacsin levels in PBMCs: A diagnostic assay for SACS variants in peripheral blood cells - A PROSPAX study

- From Dr Ceren Tunca with Dr Bernard Brais (NMD4C) as a co-author.

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

- From Dr Berta Estévez-Arias with Drs Alberto Aleman and Hanns Lochmüller (NMD4C) as co-author.

Predictors of cardiac disease in duchenne muscular dystrophy: a systematic review and evidence grading

- From Dr Erik Landfeldt with Drs Alberto Aleman and Hanns Lochmüller (NMD4C) as co-authors.

Somatic instability of the FGF14-SCA27B GAA • TTC repeat reveals a marked expansion bias in the cerebellum

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