New Publications from NMD4C Investigators

March 2024

CHARON: An Imaging-Based Diagnostic Algorithm to Navigate Through the Sea of Hereditary Degenerative Ataxias

- From Dr Alessandra Scaravilli with Dr Bernard Brais (NMD4C) as a co-author.

Biallelic NAA60 variants with impaired n-terminal acetylation capacity cause autosomal recessive primary familial brain calcifications

- From Dr Viorica Chelban with Dr Hanns Lochmüller (NMD4C) as a co-author.

Improving Diagnostic Precision: Phenotype-Driven Analysis Uncovers a Maternal Mosaicism in an Individual with RYR1-Congenital Myopathy

- From Dr Berta Estévez-Arias with Dr Hanns Lochmüller (NMD4C) as a co-author.

Comprehensive phenotypic characterization of an allelic series of zebrafish models of NEB-related nemaline myopathy

- From Dr Lacramioara Fabian with Dr James Dowling (NMD4C) as a co-author.

The Development of a New Patient-Reported Outcome Measure in Recessive Ataxias: The Person-Reported Ataxia Impact Scale

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

Safety and efficacy of givinostat in boys with Duchenne muscular dystrophy (EPIDYS): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial

- From Dr Eugenio Mercuri with Dr Jean Mah (NMD4C) as a co-author.

GAA-FGF14 disease: defining its frequency, molecular basis, and 4-aminopyridine response in a large downbeat nystagmus cohort

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

The genetic landscape and phenotypic spectrum of GAA-FGF14 ataxia in China: a large cohort study

- From Dr Riwei Ouyang with Dr Bernard Brais (NMD4C) as a co-author.

Patient-Reported Outcome Measures in Neuromuscular Diseases: A Scoping Review

- From Dr Nicoline Voet with Dr Cynthia Gagnon (NMD4C) as a co-author.

The emerging spectrum of neurodevelopmental comorbidities in early-onset Spinal Muscular Atrophy

- From Dr Giovanni Baranello and the Neurodevelopment in SMA Working Group, including NMD4C members Dr Alberto Aleman, Dr Hanns Lochmüller, Dr Hugh McMillan, Dr Hernan Gonorazky, and Dr Maryam Oskoui.

Variants in mitochondrial disease genes are common causes of inherited peripheral neuropathies

- From Dr Tomas Ferreira with Dr Hanns Lochmüller (NMD4C) as a co-author.

Comprehensive whole-genome sequence analyses provide insights into the genomic architecture of cerebral palsy

- From Dr Darcy Fehlings with Dr Maryam Oskoui (NMD4C) as a co-author.

Epilepsy in neurofibromatosis type 1: Prevalence, phenotype, and genotype in adults

- From Dr Julien Hébert with Dr Vera Bril (NMD4C) as a co-author.

Reply: Evaluating the Scope and Safety of Bilateral MRgFUS Thalamotomy for Essential Tremor: A Critical Analysis

- From Dr Nadia Scantlebury with Dr Agessandro Abrahao (NMD4C) as a co-author.