New Publications from NMD4C Investigators

May 2024

Expanding the spectrum of LAMB2: Pierson syndrome associated with neuromuscular junction disorder in two patients

- From Dr Freddy Paiz with Dr Hernan Gonorazky (NMD4C) as a co-author.

FcRn Inhibitor Therapies in Neurologic Diseases

- From Dr Nouf Alfaidi with Dr Vera Bril (NMD4C) as a co-author.

"What Services?": Stakeholders' Perceived Unmet Support Needs for Parents With Neurological Disorders

- From Dr Evelina Pituch with Dr Cynthia Gagnon (NMD4C) as a co-author.

E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases. Report from the 3rd eNMD Congress: Pisa, Italy, 29-30 October 2021

- From Dr Erika Schirinzi with Dr Hanns Lochmüller (NMD4C) as a co-author.

Unravelling undiagnosed rare disease cases by HiFi long-read genome sequencing

- From Dr Wouter Steyaert with Dr Hanns Lochmüller (NMD4C) as a co-author.

Teratogenesis, Perinatal, and Neurodevelopmental Outcomes After In Utero Exposure to Antiseizure Medication: Practice Guideline From the AAN, AES, and SMFM

- From Dr Alison Pack with Dr Maryam Oskoui (NMD4C) as a co-author.

Risk Factors for Perinatal Arterial Ischemic Stroke: A Machine Learning Approach

- From Dr Ratika Srivastava with Dr Maryam Oskoui (NMD4C) as a co-author.

Family caregivers of children transitioning hospital to home receiving nasogastric feeding: descriptive qualitative study

- From Dr Samantha Mekhuri with Dr Reshma Amin (NMD4C) as a co-author.

Increased Diagnostic Yield by Reanalysis of Whole Exome Sequencing Data in Mitochondrial Disease

- From Dr Catarina Olimpio with Dr Hanns Lochmüller (NMD4C) as a co-author.

Office-based respiratory assessment in patients with generalized myasthenia gravis

- From Dr Monica Alcantara with Dr Vera Bril (NMD4C) as a co-author.

Electrophysiological grading scale for polyneuropathy severity

- From Dr Alon Abraham with Dr Vera Bril (NMD4C) as a co-author.

Safety and efficacy of arimoclomol in patients with early amyotrophic lateral sclerosis (ORARIALS-01): a randomised, double-blind, placebo-controlled, multicentre, phase 3 trial

- From Dr Michael Benatar with Dr Agessandro Abrahao and Dr. Christen Shoesmith(NMD4C) as a co-author.

Disturbance of the human gut microbiota in patients with Myotonic Dystrophy type 1

- From Dr Manijeh Mahdavi with Dr Élise Duchesne, Dr Cynthia Gagnon and Dr Nicolas Dumont(NMD4C) as co-authors.

Resistance training in women with myotonic dystrophy type 1: a multisystemic therapeutic avenue

- From Dr Laura Girard-Côté with Dr Élise Duchesne and Cynthia Gagnon (NMD4C) as a co-author.