New Publications from NMD4C Investigators

January 2023 - March 2023

Neuromuscular junction involvement in inherited motor neuropathies: genetic heterogeneity and effect of oral salbutamol treatment

- From Dr. Grace McMacken, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

MYTHO is a novel regulator of skeletal muscle autophagy and integrity

- From Dr. Leduc Gaudet (NMD4C early career member), with Dr. Elise Duchesne and Dr. Hanns Lochmüller (NMD4C) as co—authors.

Does e-learning facilitate medical education in pediatric neurology?

- From Dr. Brittany Curry, with Dr. Hugh McMillan (NMD4C) as a co-author.

Biallelic pathogenic variants in the mitochondrial form of phosphoenolpyruvate carboxykinase cause peripheral neuropathy

- From Dr. Neal Sondheimer, with Dr. Alberto Aleman, Dr. Hernan Gonorazky and Dr. James Dowling (NMD4C) as co-authors.

TEFM variants impair mitochondrial transcription causing childhood-onset neurological disease

- From Dr. Lindsey Van Haute, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

The clinical and genetic spectrum of autosomal-recessive TOR1A-related disorders

- From Dr. Afshin Saffari, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Two-year efficacy and safety of risdiplam in patients with type 2 or non-ambulant type 3 spinal muscular atrophy (SMA)

- From Dr. Maryam Oskoui (NMD4C).

In utero Exposure to Maternal Diabetes and the Risk of Cerebral Palsy: A Population-based Cohort Study

- From Dr. Asma Ahmed, with Dr. Maryam Oskoui (NMD4C) as a co-author.

Base editing as a genetic treatment for spinal muscular atrophy

- From Dr. Christiano Alves, with Dr. Rashmi Kothary (NMD4C) as a co-author.

Prevalence of urinary incontinence and other pelvic floor disorders in women with myotonic dystrophy type 1

- From Isabelle Fisette-Paulhus, with Dr. Cynthia Gagnon (NMD4C) as a co-author.

A Homozygous PPP1R21 Splice Variant Associated with Severe Developmental Delay, Absence of Speech, and Muscle Weakness Leads to Activated Proteasome Function

- From Dr. Andreas Hentschel, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

A randomized, cross-over trial comparing the effect of innovative robotic gait training and functional clinical therapy in children with cerebral palsy; a protocol to test feasibility

- From Dr. Anna McCormick, with Dr. Hugh McMillan (NMD4C) as a co-author.

Development of eConsult reflective learning tools for healthcare providers: a pragmatic mixed methods approach

- From Dr. Douglas Archibald, with Dr. Craig Campbell (NMD4C) as a co-author.

Methylenetetrahydrofolate reductase deficiency and high dose FA supplementation disrupt embryonic development of energy balance and metabolic homeostasis in zebrafish

- From Dr. Rebecca Simonian, with Dr. Emanuela Pannia (2022 NMD4C postdoctoral fellow) and Dr. James Dowling (NMD4C) as co-authors.

A novel phenotype of AChR-deficiency syndrome with predominant facial and distal weakness resulting from the inclusion of an evolutionary alternatively-spliced exon in CHRNA1

- From Dr. Pedro Rodríguez Cruz, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Hybrid reflected-ultrasound computed tomography versus B-mode-ultrasound for muscle scoring in spinal muscular atrophy

- From Dr. Vera Danko, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

260th ENMC International Workshop: Congenital myasthenic syndromes 11-13 March 2022, Naarden, The Netherlands

- From Dr. Sally Spendiff, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Empowering Muscle Stem Cells for the Treatment of Duchenne Muscular Dystrophy

- From Dr. Romina Filippelli, with Dr. Natasha Chang (NMD4C) as a co-author.