New Publications from NMD4C Investigators

July - August 2023

Individual transcriptomic response to strength training for patients with myotonic dystrophy type 1

- From Dr Emily Davey with Dr Élise Duchesne (NMD4C) as a co-author.

Resistance Exercise Training Rescues Mitochondrial Dysfunction in Skeletal Muscle of Patients with Myotonic Dystrophy Type 1

- From Dr Valeria Di Leo with Dr Élise Duchesne (NMD4C) as a co-author.

Clearance of defective muscle stem cells by senolytics restores myogenesis in myotonic dystrophy type 1

- From Dr Talita Conte with Drs Élise Duchesne, Cynthia Gagnon and Nicolas Dumont (NMD4C) as a co-authors.

Titin related myopathy with ophthalmoplegia. A novel phenotype

- From Dr Issa Alawneh with Drs James Dowling and Hernan Gonorazky (NMD4C) as co-authors.

Variants in ACTC1 underlie distal arthrogryposis accompanied by congenital heart defects

- From Jessica Chong with Drs James Dowling and Hernan Gonorazky (NMD4C) as co-authors.

Loss of Mtm1 causes cholestatic liver disease in a model of X-linked myotubular myopathy

- From Dr Sophie Karolczak with Dr James Dowling (NMD4C) as a co-author.

Wnt binding to Coatomer proteins directs secretion on exosomes independently of palmitoylation

- From Dr Uxia Gurriaran-Rodriguez with Drs Rashmi Kothary and Michael Rudnicki (NMD4C) as co-authors.

Optimized testing strategy for the diagnosis of GAA-FGF14 ataxia/spinocerebellar ataxia 27B

- From Dr Céline Bonnet with Dr Bernard Brais (NMD4C) as a co-author.

Intronic FGF14 GAA repeat expansions are a common cause of ataxia syndromes with neuropathy and bilateral vestibulopathy

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

Intronic FGF14 GAA repeat expansions are a common cause of downbeat nystagmus syndromes: frequency, phenotypic profile, and 4-aminopyridine treatment response

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

A common flanking variant is associated with enhanced meiotic stability of the FGF14 -SCA27B locus

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

Natural History and Phenotypic Spectrum of GAA-FGF14 Sporadic Late-Onset Cerebellar Ataxia (SCA27B)

- From Dr Thomas Wirth with Dr Bernard Brais (NMD4C) as a co-author.

Non-GAA Repeat Expansions in FGF14 Are Likely Not Pathogenic-Reply to: "Shaking Up Ataxia: FGF14 and RFC1 Repeat Expansions in Affected and Unaffected Members of a Chilean Family"

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

TUFM variants lead to white matter abnormalities mimicking multiple sclerosis

- From Dr Shihan Chen with Dr Bernard Brais (NMD4C) as a co-author.

As Frequent as Polyglutamine Spinocerebellar Ataxias: SCA27B in a Large German Autosomal Dominant Ataxia Cohort

- From Dr Holger Hengel with Dr Bernard Brais (NMD4C) as a co-author.

Frequency of GAA- FGF14 Ataxia in a Large Cohort of Brazilian Patients With Unsolved Adult-Onset Cerebellar Ataxia

- From Dr Luiz Eduardo Novis with Dr Bernard Brais (NMD4C) as a co-author.

Understanding caregiver experiences with disease-modifying therapies for spinal muscular atrophy: a qualitative study

- From Dr Lena Xiao with Dr Reshma Amin (NMD4C) as a co-author.

The Role of Vitamin D in Neuroprotection in Multiple Sclerosis: An Update

- From Dr Amarpreet Sangha with Dr Gerald Pfeffer (NMD4C) as a co-author.

Clinical course, imaging, and pathological features of 45 adult and pediatric cases of myelin oligodendrocyte glycoprotein antibody-associated disease

- From Dr Hayet Boudjani with Dr Maryam Oskoui (NMD4C) as a co-author.

Pre-pregnancy and pregnancy disorders, pre-term birth and the risk of cerebral palsy: a population-based study

- From Dr Neda Razaz with Dr Maryam Oskoui (NMD4C) as a co-author.

The Medical Action Ontology: A Tool for Annotating and Analyzing Treatments and Clinical Management of Human Disease

- From Dr Leigh Carmody with Dr Hanns Lochmüller (NMD4C) as a co-author.

Do classical and computerized cognitive tests have equal intrarater reliability in myotonic dystrophy type 1?

- From Dr Julie Fortin with Dr Cynthia Gagnon (NMD4C) as a co-author.

Duchenne muscular dystrophy respiratory profiles from real world registry data

- From Dr Mona Hnaini with Dr Craig Campbell (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.

SF2Former: Amyotrophic lateral sclerosis identification from multi-center MRI data using spatial and frequency fusion transformer

- From Dr Rafsanjany Kushol with Dr Lawrence Korngut (NMD4C) as a co-author.

White matter abnormalities in 15 subjects with SPG76

- From Dr Abdulrahman Alkhalifa with Dr Bernard Brais (NMD4C) as a co-author.

Analysis of muscle magnetic resonance imaging of a large cohort of patient with VCP-mediated disease reveals characteristic features useful for diagnosis

- From Dr Diana Esteller with Dr Jodi Warman Chardon (NMD4C) as a co-author.

Risk Factors Associated with Incident Vertebral Fractures in Steroid-treated Males with Duchenne Muscular Dystrophy

- From Dr Kim Phung with Drs Laura McAdam, Hugh McMillan and Leanne Ward (NMD4C) as co-authors.

Further characterization of CEP85L-associated lissencephaly type 10: Report of a three-generation family and review of the literature

- From Dr Heather Leduc-Pessah with Dr Hugh McMillan (NMD4C) as a co-author.