New Publications from NMD4C Investigators

September - November 2022

Severe congenital myasthenic syndromes caused by agrin mutations affecting secretion by motoneurons

- Dr. Arnaud Jacquier, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

The White Matter Rounds experience: The importance of a multidisciplinary network to accelerate the diagnostic process for adult patients with rare white matter disorders

- Dr. Yu Tong Huang with Dr. Bernard Brais (NMD4C) as a co-author.

De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders

- Dr. Xiangbin Jia with Dr. Hugh McMillan (NMD4C) as a co-author.

Transcriptome analysis from muscle biopsy tissues in late-onset myopathies identifies potential biomarkers correlating to muscle pathology

- Dr. Matthew Joel with Dr. Gerald Pfeffer (NMD4C) as a co-author.

Suppression of the necroptotic cell death pathways improves survival in Smn 2 B/- mice

- Dr. Lucia Chehade with Dr. Rashmi Kothary (NMD4C) as a co-author.

Egg-laying and locomotory screens with C. elegans yield a nematode-selective small molecule stimulator of neurotransmitter release

- Dr. Sean Harrington with Dr. James Dowling (NMD4C) as a co-author.

'Our time is precious': An exploration of parental feeding behaviours for boys with Duchenne muscular dystrophy

- Dr. Meaghan Walker with Dr. Reshma Amin (NMD4C) as a co-author.

Efficacy and Safety of Vamorolone vs Placebo and Prednisone Among Boys With Duchenne Muscular Dystrophy: A Randomized Clinical Trial

- Dr. Michela Guglieri with Drs. Hugh McMillan and Kathryn Selby (NMD4C) as co-authors.

Cannabis use in patients with Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay

- Dr. William Beauchesne with Dr. Cynthia Gagnon (NMD4C) as a co-author.

Risk Factors for Term-Born Periventricular White Matter Injury in Children With Cerebral Palsy: A Case-Control Study

- Dr. Amaar Marefi with Dr. Maryam Oskoui (NMD4C) as a co-author.

Benefit of 5 years of enzyme replacement therapy in advanced late onset Pompe. A case report of misdiagnosis for three decades with acute respiratory failure at presentation

- Dr. Mandat Maharaj with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Biallelic loss-of-function variants in RABGAP1 cause a novel neurodevelopmental syndrome

- Dr. Rachel Youjin Oh with Dr. James Dowling (NMD4C) as a co-author.

Diagnostic criteria in amyotrophic lateral sclerosis: Time for a change

- Dr. Gord Jewett (NMD4C) with Drs. Victoria Hodgkinson and Lawrence Korngut (NMD4C) as co-authors.

Rate of speech decline in individuals with amyotrophic lateral sclerosis

- Dr. Marziye Eshghi with Dr. Lawrence Korngut (NMD4C) as a co-author.

Participation and Functional Independence in Adults With Recessive Spastic Ataxia of Charlevoix-Saguenay

- Dr. Samar Muslemani with Drs. Cynthia Gagnon and Bernard Brais (NMD4C) as co-authors.

Modulating Myogenesis: An Optimized In Vitro Assay to Pharmacologically Influence Primary Myoblast Differentiation

- Dr. Cordell VanGenderen with Dr. Natasha Chang (NMD4C) as a co-author.

Validation of a Revised Version of the Center for Epidemiologic Depression Scale for Youth with Intellectual Disabilities (CESD-ID-R)

- Dr. Elizabeth Olivier with Dr. Cynthia Gagnon (NMD4C) as a co-author.

Modulating Myogenesis: An Optimized In Vitro Assay to Pharmacologically Influence Primary Myoblast Differentiation

- Dr. Cordell VanGenderen with Dr. Natasha Chang (NMD4C) as a co-author.

Genotype-phenotype correlations in valosin-containing protein disease: a retrospective muticentre study

- Dr. Marianela Schiava with Dr. Jodi Warman Chardon (NMD4C) as a co-author.

Chronic tracheostomy care of ventilator-dependent and -independent children: Clinical practice patterns of pediatric respirologists in a publicly funded (Canadian) healthcare system

- Dr. Aaron St-Laurent with Dr. Reshma Amin (NMD4C) as a co-author.

Documenting manifestations and impacts of autosomal recessive spastic ataxia of Charlevoix-Saguenay to develop patient-reported outcome

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

Respiratory characteristics in children with spinal muscular atrophy type 1 receiving nusinersen

- Dr. Lena Xiao with Drs. Hernan Gonorazky and Reshma Amin (NMD4C) as co-authors.

French Translation and Cross-cultural Adaptation of the Scale for the Assessment and Rating of Ataxia

- Dr. Dax Bourcier with Drs. Bernard Brais, Elise Duchesne and Cynthia Gagnon (NMD4C) as co-authors.

PURA syndrome: neuromuscular junction manifestations with potential therapeutic implications

- Dr. Heba Qashqari (NMD4C) with Drs. Hernan Gonorazky and James Dowling (NMD4C) as co-authors.

Multiple Case Study of Changes in Participation of Adults with Myotonic Dystrophy Type 1: Importance of Redesigning Accomplishment and Resilience

- Dr. Kateri Raymond with Dr. Cynthia Gagnon (NMD4C) as a co-author.

Measurement properties of wheelchair use assessment tools in adults with autosomal recessive spastic ataxia of Charlevoix-Saguenay

- Dr. Julie Bourassa with Drs. Cynthia Gagnon and Bernard Brais (NMD4C) as co-authors.

Improved upper limb function in non-ambulant children with SMA type 2 and 3 during nusinersen treatment: a prospective 3-years SMArtCARE registry study

- Dr. Astrid Pechmann with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Fathers' Experiences in Alberta Family Integrated Care: A Qualitative Study

- Dr. Amy Shafey with Dr. Reshma Amin (NMD4C) as a co-author.

Five patients with spinal muscular atrophy-progressive myoclonic epilepsy (SMA-PME): a novel pathogenic variant, treatment and review of the literature

- Dr. Parvaneh Karimzadeh with Dr. Hanns Lochmüller (NMD4C) as a co-author.

Disease Burden of Spinal Muscular Atrophy: A Comparative Cohort Study Using Insurance Claims Data in the USA

- Dr. Julie Mouchet with Dr. Maryam Oskoui (NMD4C) as a co-author.

A virtual rural medicine self-learning module for preclerkship students

- Dr. Celina DeBiasio with Dr. Craig Campbell (NMD4C) as a co-author.

New SOD1 mutation causing rapid amyotrophic lateral sclerosis with nerve root enhancement

- Dr. Tefani Perera with Dr. Gerald Pfeffer (NMD4C) as a co-author.

A novel MAP3K7 mutation in a child with cardiospondylocarpofacial syndrome and orofacial clefting

- Dr. William Billal Shepherd with Dr. Craig Campbell (NMD4C) as a co-author.

[The clinical spectrum of SMA-PME and in vitro normalization of its cellular ceramide profile]

- Dr. Michelle Lee with Dr. Jodi Warman Chardon (NMD4C) as a co-author.

[Risk factors associated with prevalent vertebral fractures in Duchenne muscular dystrophy]

- Dr. Kim Phung with Dr. Hugh McMillan (NMD4C) as a co-author.

[Bridging clinical care and research in Ontario, Canada: Maximizing diagnoses from reanalysis of clinical exome sequencing data]

- Dr. Taila Hartley with Drs. James Dowling, Hernan Gonorazky and Jodi Warman Chardon (NMD4C) as co-authors.