



New Publications from NMD4C Investigators

November 2022 - December 2022

[Student responses to creative coding in biomedical science education](#)

- Dr. Phillip Gough, with Dr. Craig Campbell (NMD4C) as a co-author.

[A psychometric validation of the motives for physical activity measure for youth with intellectual disabilities \(MPAM-ID\)](#)

- Dr. Christophe Maïano with Dr. Cynthia Gagnon (NMD4C) as a co-author.

[Scoliosis in Spinal Muscular Atrophy Type 1 in the Nusinersen Era](#)

- Dr. Fatima Al Amrani with Dr. Reshma Amin and Dr. Hernan Gonorazky (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.

[Motor cortex functional connectivity is associated with underlying neurochemistry in ALS](#)

- Dr. Avyarthana Dey with Dr. Lawrence Korngut (NMD4C) as a co-author.

[A clustering of heterozygous missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder](#)

- Dr. Lot Snijders Blok, with Dr. Hanns Lochmüller (NMD4C) as a co-author.

[In Utero Exposure to Maternal Injury and the Associated Risk of Cerebral Palsy](#)

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

[Quantitative vs qualitative muscle MRI: Imaging biomarker in patients with Oculopharyngeal Muscular Dystrophy \(OPMD\)](#)

- Dr. Gerd Melkus with Dr. Hanns Lochmüller, Dr. Bernard Brais, and Dr. Jodi Warman-Chardon (NMD4C) as co-authors.

[Variants in CLDN5 cause a syndrome characterized by seizures, microcephaly and brain calcifications](#)

- Dr. Ashish Deshwar with Dr. James Dowling (NMD4C) as a co-author.

[Deep Intronic FGF14 GAA Repeat Expansion in Late-Onset Cerebellar Ataxia](#)

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

[Caregiver Burden of Spinal Muscular Atrophy: A Systematic Review](#)

- Dr. Erik Landfeldt with Dr. Hugh McMillan and Dr. Hanns Lochmüller (NMD4C) as co-authors.



[Prevalence of urinary incontinence and other pelvic floor disorders in women with myotonic dystrophy type 1](#)

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

[The J Domain of Sacsin Disrupts Intermediate Filament Assembly](#)

- Dr. Afroz Dabbaghizadeh with Dr. Bernard Brais (NMD4C) as a co-author.

[Improvements in Walking Distance during Nusinersen Treatment - A Prospective 3-year SMarCARE Registry Study](#)

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

[The discovery of the DNA methylation episinature for Duchenne muscular dystrophy](#)

- Dr. Leighton Schreyer with Dr. Craig Campbell (NMD4C) as a co-author.

[A rehabilitation program to increase balance and mobility in ataxia of Charlevoix-Saguenay: An exploratory study](#)

Dr. Isabelle Lessard with Dr. Cynthia Gagnon and Dr. Élise Duchesne (NMD4C) as co-authors.