**New Publications from NMD4C Investigators**

**September 2023**

Hyaluronidase-facilitated subcutaneous immunoglobulin 10% as maintenance therapy for chronic inflammatory demyelinating polyradiculoneuropathy: The ADVANCE-CIDP 1 randomized controlled trial

- From Dr Vera Bril (NMD4C).

Recessive pathogenic GMPPB Variants Cause a Childhood Onset Myasthenic Syndrome Responsive to Pyridostigmine

- From Dr Gordon Jewett, with Dr Gerald Pfeffer (NMD4C) as a co-author.

Therapeutic developments for valosin-containing protein mediated multisystem proteinopathy

- From Dr Victoria Boock, with Dr Gerald Pfeffer (NMD4C) as a co-author.

Towards the Identification of Biomarkers for Muscle Function Improvement in Myotonic Dystrophy Type 1

- From Dr Amir Aoussim, with Dr Élise Duchesne (NMD4C) as a co-author.

Spinocerebellar ataxia 27B: episodic symptoms and acetazolamide response in 34 patients

- From Dr Catherine Ashton, with Dr Bernard Brais (NMD4C) as a co-author.

Clinical and genetic characterisation of a large Indian congenital myasthenic syndrome cohort

- From Dr Kiran Polavarapu, with Dr Hanns Lochmüller (NMD4C) as a co-author.

Reliability of confocal corneal microscopy for measurement of dendritic cell density in suspected small fiber neuropathy

- From Dr Juan Francisco Idiaquez, with Dr Vera Bril (NMD4C) as a co-author.

Respiratory failure in a patient with VACTERL association and concomitant spinal muscular atrophy

- From Dr Sarah Grace Buttle, with Dr Hugh McMillan (NMD4C) as a co-author.

Symptom and Treatment Satisfaction in Members of the US and Canadian GBS/CIDP Foundations with a Diagnosis of Chronic Inflammatory Demyelinating Polyneuropathy

- From Dr Meg Mendoza, with Dr Vera Bril (NMD4C) as a co-author.

DPAGT1-CDG: Report of Two New Pediatric Patients and Brief Review of the Literature

- From Dr Özlem Özsoy, with Dr Hanns Lochmüller (NMD4C) as a co-author.

Intra-familial variability of oculoleptomeningeal amyloidosis due to the ATTR I107M (c.381T > G) mutation: diagnostic challenges of a rare phenotype

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

Quantitative sonographic assessment of muscle thickness and fasciculations distribution is a sensitive tool for neuromuscular disorders

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

Case report: A case of spinal muscular atrophy in a preterm infant: risks and benefits of treatment

- From Dr Elisa Nigro, with Drs Craig Campbell and Hugh McMillan, Mark Tarnopolsky and Hernan Gonorazky (NMD4C) as co-authors.

Sleep disordered breathing in infants identified through newborn screening with spinal muscular atrophy

- From Dr Jackie Chiang, with Drs Hernan Gonorazky and Reshma Amin (NMD4C) as co-authors.

Palliative care at any stage of amyotrophic lateral sclerosis: a prospective feasibility study

- From Dr Jocelyn Zwicker (NMD4C).

SIGnature Libraries: A roadmap for the formation of special interest group libraries

- From Dr Young-Min Kim, with Dr Maryam Oskoui (NMD4C) as a co-author.