"Neuromuscular Club Meeting 2021 (virtual and in-person meeting in Helsinki, December 3rd)

"Update on treatment options, diagnostics and follow-up of neuromuscular diseases with onset from infancy to adulthood"

Session 1: Early-onset neuromuscular diseases with disease-specific treatments

Chair, Prof Johanna Uusimaa, MD, PhD, Pediatric Neurology Unit, Oulu University Hospital, University of Oulu, Finland
Co-chair, Adjunct prof Tuire Lähdesmäki, MD, PhD, Pediatric Neurology Unit, Turku University Hospital, University of Turku, Finland

9.15-9.25 Welcome and Opening words

09.25-10.05 Spinal muscular atrophy: novel genetic treatments and clinical trial outcome
Dr.Chiara Marini-Bettolo, MD, PhD, Honorary Clinical Senior Lecturer and Consultant Neurologist, Institute of Translational and Clinical Research, Faculty of Medical Sciences, Newcastle University,Newcastle Upon Tyne, UK
10.05-10.15 Discussion
10.15-10.20 Biogen

10.20-11.00 Gene therapy in DMD, paediatric and upcoming trials in adults
Prof Mar Tulinius, MD, PhD, Queen Silvia Childrens´ Hospital and University of Gothenburg, Sweden
11.00-11.10 Discussion
11.10-11.14 Roche

11.15-11.55 Infantile-onset Pompe disease: enzyme replacement therapy and outcome
Dr. Hannerieke van der Hout, MD, PhD, Pediatric neurologist, Center for Lysosomal and Metabolic Diseases, Erasmus MC, University Medical Center Rotterdam, the Netherlands
11.55-12.05 Discussion
12.05-12.10 Sanofi

12.10-12.50 Lunch break

Session 2: Mitochondrial diseases: update on treatment options

Chair, Prof Kari Majamaa, MD, PhD, Neurology Unit, Oulu University Hospital, University of Oulu, Finland

12.50-13.30 Treatment of stroke-like episodes and status epilepticus in mitochondrial diseases
Prof Laurence Bindoff, MD, PhD, Haukeland University Hospital and University of Bergen, Norway

13.40-14.20 Emerging therapies in mitochondrial diseases
Dr Rob Pitceathly, MD, PhD, Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology, London, UK

14.20-14.30 Break and coffee served in the auditorium

Session 3: Small oral presentations selected by abstracts (7 min/presentation)

Chair, Associate prof, Mika H. Martikainen, MD, PhD, Department of Clinical Neurosciences, University of Turku, Finland

14.30-14.40 Manu Jokela, Out-of-Frame Mutations in ACTN2 Last Exon Cause a Dominant Distal Myopathy With Facial Weakness
14.40-14.50 Jaakko Sarparanta, Functional studies on SMPX
14.50-15.00 Marco Savarese, RNAseq characterization of TTN truncating unveils possible disease mechanisms
15.00-15.10 Sandra Harjuhaahto, New mouse and cell models of Spinal muscular atrophy Jokela type
15.10- 15.20 Jaana Lähdetie, Estimation of the prevalence and incidence of neuromuscular diseases in Finland
15.20-15.30 Vilma-Lotta Lehtokari, Food consumption, nutrition and functioning of patients with nemaline myopathy and related disorders in Finland
15.30-15.40 Jenni Laitila, Elucidating myosin dysfunction and the potentially altered energy metabolism in nemaline myopathy
15.40-15.50 Lydia Sagath, Droplet Digital PCR confirms copy number variation in the segmental duplication region of titin
15.50-16.00 Sebastian Kenvin, Threshold of heteroplasmic truncating MT-ATP6 mutation in cellular reprogramming, Notch hyperactivation and motor neuron metabolism

16.00-16.10 Closing remarks including preliminary discussion on the NM club meeting 2022