Finnish Neuromuscular Club meeting 1.12.2023

Scandic Park Hotel, Helsinki

Programme_draft

9.30 Welcome coffee

10.00-12.05 Session I: Ion channel disorders and rimmed vacuolar myopathies

Chair: Johanna Palmio

- 10.00-10.30 Update on multisystem proteinopathies B Udd
- 10.30-11.00 Assessing pathogenicity of ion channel gene variants R Männikkö (UCL)
- 11.00-11.45 Genetics of inclusion body myositis H Houlden (UCL)
- 11.45-11.55 Discussion

- 11.55-12.05 Presentation by silver co-operation partner

12.05-12.50 Lunch

12.50-14.30 Session II (six presentations á 15 min)

Chair: Manu Jokela

- The diagnostic utility of "ion channel EMG" V Periviita
- Update on ACTN2-related disorders J Ranta-aho
- Neuromuscular disorders diagnosed by exome-based gene panel in Turku University Hospital M Haanpää
- What we miss in known disease genes: variant re-evaluation and long read sequencing R Owusu
- Familial inclusion body myositis – more common in Finland? M Jokela
- Pathomechanisms of spinal muscular atrophy-Jokela type (SMAJ) S Harjuhaahto

- 14.30-14.50 Presentation by gold co-operation partner

14.50-15.05 Coffee

15.05-16.35 Session III (14 x á 5 min poster-flash session; the posters are sent beforehand to all participants, 3 slide-presentations and 1-2 questions)

Chair: Carina Wallgren-Pettersson

Rare PMP22 mutations and ultrasound in the assessment of hereditary neuropathies E Palu

Pathomechanisms of ITPR3-mutated CMT1J J Rönkkö

Mosaicism in nemaline myopathy V- L Lehtokari

Dominant nebulin myopathy -an update K Pelin

Variants in tropomyosins TPM2 and TPM3 causing high muscle tone C Wallgren-Pettersson

Unusual cause of acute weakness in an elderly patient A-M Saukkonen

Dominant myopathy due to a HSPB6 mutation J Sarparanta

Alternative splicing of titin in fetal muscle E Nippala

Phenotype-based gene prioritization for fast and accurate analysis of rare muscle diseases V Lillback

Unusual case of SORD neuropathy P Nevalainen

Treatment-refractory CIDP in patients with SLE – a case series M Jokela

The usefulness of routine complement staining of muscle biopsies D Utkan

Anti-HMGCR ab necrotizing autoimmune myopathy in identical twins M Gardberg

Haplotype sharing trees reveal shared ancestry between the C9orf72 hexanucleotide repeat expansion and normal alleles with ≥18-20 repeats – evidence for premutations O Rautila

16.35-17.00

Founding meeting for the Rare Neurological Diseases-section of the Finnish Neurological Society