

Finnish Neuromuscular Club meeting 17.1.2025

Scandic Park Hotel, Helsinki

Programme

9.00-9.25 Welcome coffee and exhibition

9.25-9.30 Opening words, Manu Jokela

9.30-12.10 Session I: Motor neuron disorders and pediatric neurology Chair: Manu Jokela

- 9.30-10.10 Novel neurophysiological methods in motor neuron disorders, Mamede de Carvalho
- 10.10-10.50 Standards of care in pediatric neuromuscular disorders, Tuire Lähdesmäki
- 10.50-11.30 ASO treatments in ALS, Björn Oskarsson
- 11.30-11.40 Discussion

- 11.40-12.10 Presentation by silver co-operation partners (10 min each)

12.10-13.10 Lunch and exhibition

13.10-14.40 Session II (five presentations á 15 min and 15 min for discussion)

Chair: Marco Savarese

- TK2 deficiency and experiences with nucleoside treatment, Pirjo Isohanni
- Infectious myopathies including a case report of acute fatal myositis, Sanna Huovinen
- Charcot-Marie-Tooth type 1J: clinical and mechanistic insights, Julius Rönkkö
- Single fibre studies: Unravelling muscle mysteries one fibre at a time, Fanny Rostedt
- MADD related to sertraline use – a treatable myopathy, Emma Ottela
- Discussion

- 14.40-15.00 Presentation by gold co-operation partner

15.00-15.30 Coffee and exhibition

15.30-16.40 Session III (10 x á 7 min poster-flash session, max 5 slides per presentation and 1-2 questions)

Chair: Jaakko Sarparanta

1. FGF14exp-related SCA27B in Finland, Johanna Palmio
2. Mutational screening of Distal Myopathy gene ACTN2 reveals an aggregation hot-spot in the actin-binding domain, Johanna Ranta-aho
3. Diagnostic value of custom targeted neuromuscular CGH array for CNV detection after negative exome-based CNV analysis, Manu Jokela
4. Exploring the effects of the epi-drug Remodelin on murine myoblast differentiation, Veronica Sian

5. Pathogenetic mechanisms in troponin T-related nemaline myopathy, Jenni Laitila
6. A treatable disorder mimicking distal neuromyopathy, Olli Likitalo
7. Pathomechanisms of HNRNPA1 mutations, Jaakko Sarparanta
8. Nutritional status of Finnish nemaline myopathy patients, Vilma-Lotta Lehtokari
9. A homozygous single-nucleotide variant in TNNT1 causes abnormal troponin T isoform expression in a patient with severe nemaline myopathy: A case report, Milla Laarne

Discussion and closing words 16.40–16.55

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