5th JOINT MEETING BELGIAN-DUTCH NEUROMUSCULAR STUDY GROUP AND GERMAN REFERENCE CENTER FOR NEUROMUSCULAR DISEASES, DGNN

19-20 APRIL, 2024
HOTEL KASTEEL BLOEMENDAL
VAALS, THE NETHERLANDS



PROGRAM Friday 19 April 2024

08:30 - 09:00 Registration (coffee/tea)
09:00 - 09:10 Welcome (J. Weis, K. Claeys)

09:10 - 10:36 SESSION 1: Motor neuron diseases

Chairpersons: Bjarne Udd, Michael Van Es

Therapeutic (r)evolution in SMA: where are we now and what about the future? - Liesbeth De Waele (Belgium)

Novel developments in ALS - Michael Van Es (The Netherlands)

Oral presentations from abstracts

 $\hbox{\it Bi-allelic ATXN2 repeat expansions cause autosomal, recessive ALS-Koen Demaegd}$

The German MND-Net Tissue Bank - an update - Joachim Weis

Risdiplam treatment in adult patients with spinal muscular atrophy: a prospective study investigating clinical, functional, and patient-reported outcome measures over 12 months

- Louise Iterbeke

10:36 - 11:00 Coffee break

11:00 - 12:13 SESSION 2: Neuromuscular junction disorders

Chairpersons: Marc De Baets, Mario Losen

Update on Myasthenia gravis – optimizing treatment for all patients - Martijn Tannemaat (The Netherlands)

Oral presentations from abstracts

Congenital myasthenic syndromes in Belgium: genetic and clinical characterization of pediatric and adult patients - Nathalie Smeets

The relevance of complement activation in myasthenia gravis – a quantification study using the experimental passive transfer MG model - Anja Schöttler

Hock immunization, an update of the experimental autoimmune myasthenia gravis (EAMG) mouse model - Marina Mané Damas

Are thymus-resident plasma cells prognostic factors of thymectomy in patients with antiacetylcholine receptor myasthenia gravis? - Britt Arets

12:13 - 13:00 A tribute to Prof. J. Michael Schröder, 1935-2023 (Joachim Weis. Stephan Züchner, Claudia Sommer, Vincent Timmerman, Hans-Hilmar Goebel)

13:00 - 14:00 Lunch

14:00 - 16:39 **SESSION 3: Peripheral neuropathies I**

Chairpersons: Claudia Sommer, Peter Van den Bergh

New EAN/PNS GBS guideline - Pieter Van Doorn (The Netherlands)

Pathophysiology of demyelinating neuropathies - Ruth Stassart (Germany)

Genomics-To-Therapy: a concept illustrated by CMT-SORD - Stephan Züchner (USA)

Oral presentations from abstracts

Supervised machine learning algorithms for diagnosing chronic inflammatory demyelinating polyradiculoneuropathy - Iris van Doorn

Loss-of-function mutations in the nuclear envelope gene FAM 169A cause autosomal dominant lower and upper motor neuropathy - Maike Dohrn

Recurrent missense variant in ITPR3 causes demyelinating Charcot-Marie-Tooth neuropathy with remarkably variable severity - Danique Beijer

Frequency and clinical characterization of SORD-Related neuropathy in a Belgian cohort -Matthias Opsomer

Dominant OGDH mutations cause peripheral neuropathy with ataxia and optical atrophy - Liedewei Van de Vondel

Let's drop some Fats: The idiosyncrasies of lipid droplets in iPSC-Schwann cell precursor cells from Charcot-Marie-Tooth disease type 1A patients. - Koen Kuipers Inducible dental pulp stem cells-derived Schwann cells for CMT1A modelling - Nathalie Dirkx

16:39 - 17:00 Coffee break

17:00 - 19:14 SESSION 4: Peripheral neuropathies II

Chairpersons: Martin Lammens, Stephan Goedee

Novel therapeutic avenues in hereditary peripheral neuropathies - Davide Pareyson (Italy) Proteostasis regulators in neuromuscular diseases - learning from zebrafish models -Juliane Bremer (Germany)

Oral presentations from abstracts

Comparison of efficacy outcomes with Vutrisiran vs. Patisiran in hATTR amyloidosis with polyneuropathy: post-hoc analysis of the HELIOS-A study - Valérie Dupong

A comparison of neurofilament in serum, cerebrospinal fluid and peripheral nerve in patients undergoing nerve biopsy - Simon Streit

Autophagy phenotypical screening identified new molecules to restore neuronal proteostasis in cellular models of HSPB1 and HSPB8-associated peripheral neuropathies -Angela Sisto

The effects of PMP22 overexpression on cellular stress in Charcot-Marie-Tooth disease type 1A - Hanne Jeurissen

PDE4D inhibition with Gebr32a stimulates Schwann cell differentiation and improves the functional outcome in models for Charcot Marie Tooth disease 1A - Tim Vangansewinkel Foreign body response hampers tissue integration of a poly(E-caprolactone) nanofibrecontaining scaffold for peripheral nerve repair - Pascal Achenbach

Nerve transfers in children with Arthrogryposis Multiplex Congenita - Benedikt Schäfer

19:30 **Dinner**

PROGRAM Saturday 20 April 2024

08:30 - 10:33 SESSION 5: Muscle disorders I

Chairpersons: Werner Stenzel, Anne Schänzer

Neuromuscular imaging: a better eye on nerves and muscles - Nens van Alfen

(The Netherlands)

Clinical and imaging advances in inflammatory myopathies - Anneke van der Kooi

(The Netherlands)

Whats New in Pathogenesis of IIM - Werner Stenzel (Germany)

Oral presentations from abstracts

The adiponectin receptor agonist, ALY688: a promising therapeutic for fibrosis in the dystrophic muscle - Nicolas Dubuisson

Inhibition of KDM5A reverses pathological features in sporadic inclusion body myositislike cell models - Geert de Vries

Automated morphometric analysis of ICAM-1 expression highlights similarities and differences between myositis subtypes - Anne Schänzer

10:33 - 11:00 Coffee break

11:00 - 13:05 SESSION 6: Muscle disorders II

Chairpersons: Ingo Kurth, Anneke Van der Kooi

Genetics in distal muscle diseases - Bjarne Udd (Finland)

Advances and caveats of novel therapies in (neuro)muscular disorders - Benedikt Schoser (Germany)

Oral presentations from abstracts

Comprehensive four-year disease progression assessment of DM1, based on the Dutch MYODRAFT registry - Leandre la Fontaine

Life expectancy and causes of death in patients with Myotonic Dystrophy type 2. -Alide Tieleman

Heterozygous SPTAN1 loss-of-function variants cause early childhood onset distal myopathy - Jonathan De Winter

The exception that proves the rule: a titin truncating variant causing a dominant myopathy with cardiac involvement - Kristl Claeys

Alpha-synuclein as potential biomarker in Inclusion body myositis - a histological and serological study - Alexander Mensch

13:05 - 13:10 **Closure** (J. Weis, K. Claeys)

13:10 Lunch



















