

Program

Day 1 – Tuesday, September 8

Conference language: English

9.30 – 9.50 **Arrival and light breakfast**

9.50 – 10.00 **Welcome and introduction to the meeting**

Charlotte Handberg, Professor, Head of Research, RCFM, Denmark

Moderator for day 1: John Vissing, Professor, MD, Rigshospitalet

10.00 – 11.00 **Physiology and management of respiratory failure in spinal muscular atrophy**

Esther Veldhoen, Pediatric intensivist, PhD, MD, University Medical Center Utrecht, the Netherlands.

11.00 – 11.15 **Break**

11.15 – 12.00 **Psychological care in NMD**

Sabrina Sayah, Clinical Psychologist, Pitié-Salpêtrière Hospital, Paris, France

12.00 – 13.00 Exercise in NMD

12.00 – 12.25 John Vissing, Professor, MD, Rigshospitalet, Copenhagen, Denmark

Exercise from a patient perspective

12.25 – 12.35 Kasper Stokholm Leerskov, Post Doc, Aalborg University, Denmark

12.35 – 12.50 Sofie Skoubo, PhD candidate, RCFM, Denmark

12.50 – 13.00 Discussion

13.00 – 14.00 **Lunch**

14.00 – 15.00 **Health Literacy: Enabling Equity-Driven Communication and Interaction in Healthcare**

Anna Aaby, PhD, MD, Ass. Professor, REACH, Department of Public Health, Aarhus University, Denmark

15.00 – 15.30 **Introduction to workshops including photo session, break, and refreshments**

15.30 – 17.00 PARALLEL SESSIONS:

15.30 – 16.10 **Workshop 1: Weight loss in NMD, including the use of pharmacological treatments (15 minutes presentation and 25 minutes discussion)**

Marianne Nordstrøm, Clinical Nutritionist, PhD, Norwegian Centre for Rare Diseases, Unit Frambu

Workshop 2: Multidisciplinary rehabilitation and follow-up in a life-course perspective (15 minutes presentation and 25 minutes discussion)

Andreas Dybesland Rosenberger, PT, Head of Norwegian Centre for Rare Diseases-unit for neuromuscular disorders, University Hospital of North-Norway, Tromsø

16.10 – 16.20 **Break and change of workshop**

16.20 – 17.00 **Parallel workshops 1 and 2 are repeated**

17.00 – 17.30 **RCFM 50 Years - anniversary lecture**

Ulla Werlauff and Birgit Steffensen, both PhD, PT, Researcher Emerita, RCFM, Denmark

18.30 – 19.15 **Entertainment and drinks (Hall B)**

19.15 **Dinner at Restaurant Evald Krog**

Day 2 – Wednesday, September 9

Conference language: English

(Morning swim in the ocean)

7.30 – 8.30 **Breakfast**

Moderator for day 2: Lone Knudsen, Researcher, PhD, RCFM, Denmark

8.30 – 9.30 **Project presentations**

Denmark:

FORMY: Preventive home visits – a rehabilitation intervention for adults with DM1

Kristin Allergodt, postdoc, RCFM

Investigating needs and challenges for palliative rehabilitation interventions of adults with Duchenne muscular dystrophy in the last years of life

Sanne Okkels Birk Lorenzen, MD, PhD student, RCFM

Norway:

Implementing muscle ultrasound in clinical practice: Challenges and opportunities

Cecilie Aronsen Hæstad, PT, University Hospital of North Norway, National Neuromuscular Center, Tromsø

Mapping and understanding health literacy in adults with rare diagnoses

Una Stenberg, PhD, Social Worker, Norwegian centre for rare disorders, unit Frambu, Oslo University Hospital

9.30-9.40 **Break**

9.40-10.40 *Project presentations continued*

FKRP natural history in Norway - physical tests over a 2-year span

Tine Stokmo, Assistant Professor, Department of Clinical Medicine, University of Tromsø

CMT- postural steadiness through a light finger touch

Fredrik Hellem Stenseth, PT, MSc in neurological conditions, Oslo University Hospital,

Sweden:

COMPIS - COngenital Myopathy Intervention Study

Eva Michael, MD, Child neurologist, Queen Silvia Children's Hospital

Iceland:

Myasthenia Gravis in Iceland: A National Perspective

Ólöf Jóna Elíasdóttir, Assistant Professor of Neurology, Landspítali - University Hospital, and Marianne Elisabeth Klinke, Professor, School of Health Sciences / Faculty of Nursing and Midwifery

10.40 – 11.00 **Refreshments**

11.00 – 12.30 Impact of living with personal care on biopsychosocial health and quality of life when having a neuromuscular disease - Patient reported outcomes from a national survey

11.00 – 11.30 Signe Versterre, PhD student, PT, RCFM, Denmark

11.30 – 11.50 **Patient perspectives on living with a personal assistant**

11.50 – 12.10 Sofie Djurhuus, Qualified hotel receptionist and Host at 'Pærfekt'

Children living with a personal assistant - findings from a master thesis

12.10 – 12.30 Anne Heindorff, MSc education

Panel discussion

12.30 – 12.35 **Closing remarks**

Charlotte Handberg

12.35 **Goodbye and Lunch to go**

Presenters and presentations

Andreas Rosenberger

is a clinical specialist in neurological physiotherapy and Head of the Norwegian Centre for Rare Diseases- unit for neuromuscular disorders. He has extensive experience in both assessment/diagnosis, clinical rehabilitation and research on individualized physiotherapy for persons with neuromuscular conditions. He is co-chair of the "Multidisciplinary management and care group" within ERN Euro-NMD. Diagnoses such as FKRP-related limb-girdle muscular dystrophy, FSHD and Charcot-Marie-Tooth disease are among the main fields of interest. Andreas is also working with pregnancy-related issues in NMDs and different outcome measures such as quantitative neuromuscular ultrasound, gait mats and other clinical measures.

Anna Aaby

is MD specialized in public health, associate professor and daily manager in Research Center for Health Literacy and Equity (REACH) at dep. Of Public Health, Aarhus University.

Anna Aaby's research is centered on the subject area of health literacy – in particular the development of organizations' capacity to respond to the needs of target populations (organizational health literacy responsiveness). Her work focuses on interventions and applied science, and she has also contributed to the translation, development and testing of a range of health literacy instruments.

Anne Heindorff

is trained as a social worker and holds a master's degree in educational psychology.

Anne has always been interested in children and how they develop. During her master's program she had the opportunity to work with children with physical disability. This gave her different perspectives on what it is like to be dependent on a helper during one's school trajectory. She discovered that the definition of the helper's role determines how the child experiences school.

Birgit Steffensen

is one of the founders of the Danish Rehabilitation Centre for Neuromuscular Diseases. Trained as a physiotherapist, she earned her PhD by developing the patient reported outcome scale EK which is an instrument to measure changes – even small ones – in physical function over time in wheelchair users with spinal muscular atrophy and Duchenne muscular dystrophy.

Cecilie Aronsen Hæstad

is a physiotherapist at the Norwegian Centre for Rare Diseases – Unit for Neuromuscular Disorders at the University Hospital of North Norway in Tromsø. She has worked with muscle ultrasound in various projects since 2020, and in 2025 she completed a master's degree in Health Professional Development, with a thesis focused on establishing reference values for quantitative muscle ultrasound.

Together with her team in Norway, and in collaboration with the specialized team at Radboud University Medical Center in the Netherlands, she is currently working to implement quantitative muscle ultrasound as a new and accessible imaging technique within the Norwegian clinical neuromuscular setting.

Esther Veldhoen

is a pediatric intensivist working at the PICU, the Center of Home Mechanical ventilation, the pediatric neuromuscular center and the Netherlands SMA center in the Wilhelmina Children's Hospital (part of University Medical Center). Her research focuses on chronic respiratory failure in neuromuscular diseases.

Eva Michael**Fredrik Hellem Stenseth**

is a specialized physiotherapist within neurology. He took his master from the university of Antwerp; neurological conditions. He has a working background from rehabilitation institutes and hospitals and is today working in the clinical and research field for neuromuscular disorders at the university hospital in Oslo, Unit for rare hereditary neuromuscular disorders (EMAN).

John Vissing

Is a professor of Neurology at the University of Copenhagen, Denmark, and the Director of the Copenhagen Neuromuscular Center at Rigshospitalet in Copenhagen. Main research interests are treatment and pathophysiology of metabolic myopathies, muscular dystrophies, myasthenia gravis and channelopathies, discovery of new neuromuscular diseases, rehabilitation of muscle disease including exercise training, MR imaging of muscle, defining outcome measures for natural history and trials and unravelling genotype-phenotype relations in muscle disease.

Sofie Djurhuus

Sofie was diagnosed with LGMDR9 at the age of nine and was granted a BPA (citizen-led personal assistance) at 18 yrs of age. Growing up, she has had to navigate puberty, adolescence and everyday life while transitioning from walking independently to relying on an electric wheelchair. Her need for support has increased from help from her mother, to 8–16 hours, and today 24-hour personal assistance. As work manager, Sofie manages her own helpers, making her freedom and independence dependent on having people at work.

Kristin Allergodt

is a physiotherapist, holds a master's degree in health science and completed her PhD in 2025. She is currently a postdoc at the National Rehabilitation Center for Neuromuscular Diseases (Denmark). Her work focuses on developing, testing, and evaluating preventive home visits as a rehabilitation intervention for adults with myotonic dystrophy type 1 (DM1). Her research interest lies in neuromuscular rehabilitation, with a special focus on DM1.

Marianne Elisabeth Klinke	is a Professor and Academic Chair of Research and Development in Acute Neurology Nursing and Neuro-Rehabilitation at the University of Iceland. She also holds a position as a Clinical Nurse Specialist in Neurology at Landspítali University Hospital in Iceland. Her work focuses on neurological nursing, neurorehabilitation, patient- and family-centred care, and clinical knowledge translation. She is also part of the Myasthenia Gravis team at Landspítali. Marianne has extensive experience in research, teaching, supervision, and interdisciplinary clinical development.
Marianne Nordstrøm	is a clinical dietitian and senior researcher at the Norwegian Centre for Rare Diseases, Unit Frambu and at the Unit for Inborn and Hereditary Neuromuscular Disorders at Oslo University Hospital. Her research focuses on individuals with intellectual disabilities and hereditary neuromuscular disorders and encompasses various aspects of clinical nutrition, including health-promoting diets and the role of nutrition in managing specific conditions. Nordstrøm currently serves as Chair of the Multidisciplinary Management and Care Working Group within the ERN EURO-NMD.
Ólöf Jóna Elíasdóttir	is a neurologist at Landspítali University Hospital in Iceland and an Assistant Professor at the University of Iceland. Her clinical and academic work focuses primarily on neuromuscular diseases. She is responsible for neuromuscular care at Landspítali and works as part of the Myasthenia Gravis team, contributing to specialist diagnostics, treatment, interdisciplinary follow-up, and clinical development in neuromuscular neurology.
Sabrina Sayah	is a clinical psychologist and neuropsychologist at Pitié-Salpêtrière Hospital in Paris, France. She provides psychological care for individuals living with neuromuscular diseases and their families. Her clinical and research interests focus on the neuropsychology of neurogenetic disorders and the psychological impact of neuromuscular diseases.
Sanne Okkels Birk Lorenzen	is a Specialist in Public Health Medicine with experience in specialized rehabilitation for people with neuromuscular disorders. Her research focuses on symptoms, healthcare utilization, and the needs of adults with Duchenne muscular dystrophy, with particular interest in challenges related to palliative rehabilitation. She is particularly interested in promoting equity in healthcare and addressing cross-sectoral challenges that affect access to and coordination of care.
Signe Versterre	is a physiotherapist and PhD student at the Danish Rehabilitation Centre of Neuromuscular Diseases.

Tine Stokmo

is a doctor at dept. of Neurology and National Neuromuscular Center at the University Hospital of North Norway, Tromsø.

She has recently started a PhD-study on the natural history of FKRP-related Limb-girdle muscular dystrophy, where the overall aim is to evaluate physical tests and ultrasound as measures of disease progression.

Ulla Werlauff

Ulla Werlauff, PhD, MSc, PT, and former Head of Research at the National Rehabilitation Center for Neuromuscular Diseases (RCFM) in Aarhus, Denmark. With nearly three decades of experience, her research has focused on rehabilitation, participation, quality of life, palliative care, and patient-centered approaches for people living with neuromuscular diseases. She has made significant contributions to the development and validation of clinical assessment tools, including the EK-Scale, and has played a key role in advancing evidence-based rehabilitation research and practice internationally.

Una Stenberg

is a trained social worker and researcher specializing in health literacy and coping. She leads several national research projects focusing on the mapping and understanding of health literacy among individuals with rare diagnoses and their relatives. Her work aims to strengthen knowledge, self-management, and support systems within the field of rare diseases.

