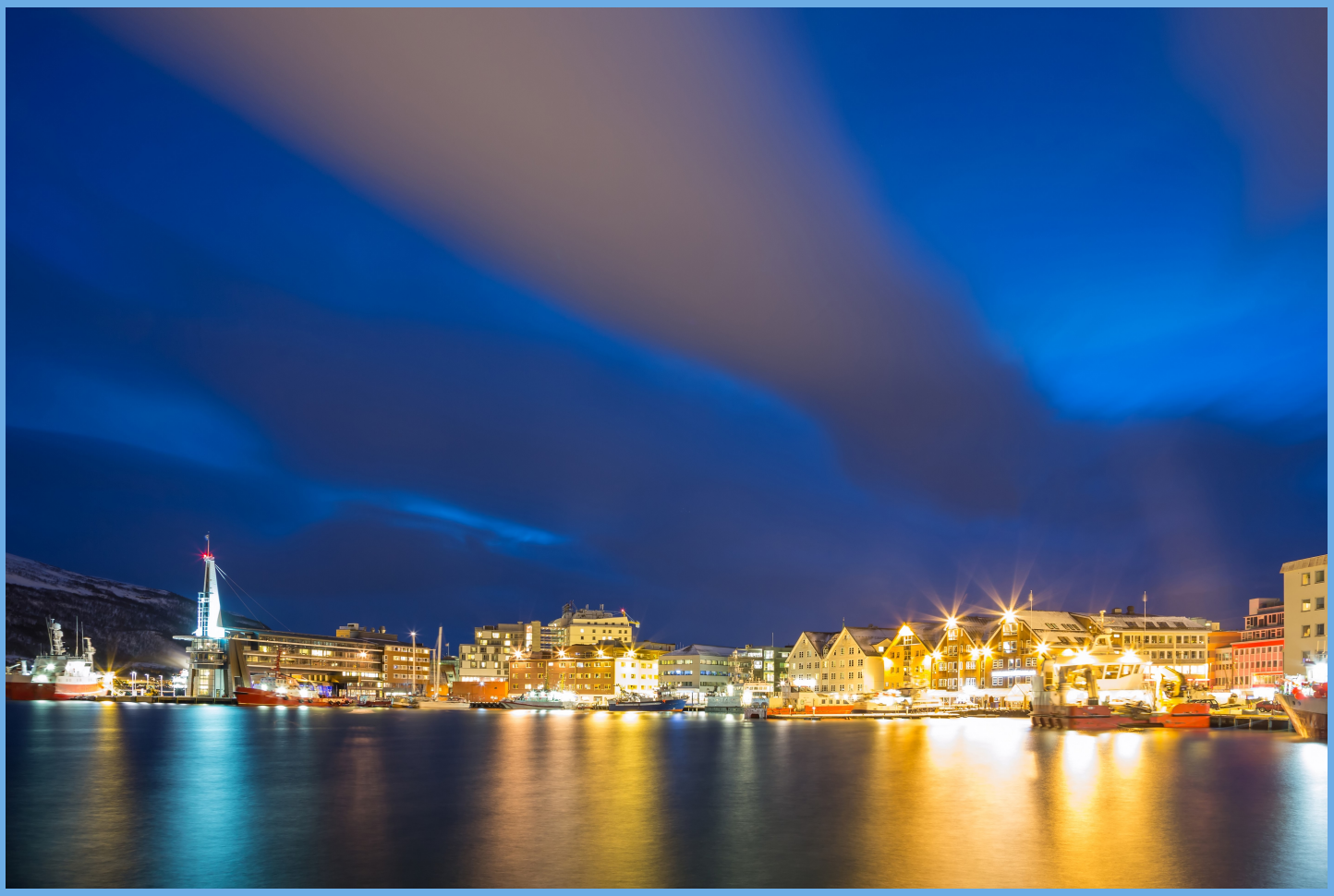
Norwegian Conference for Hereditary Neuromuscular Disorders

Fagkonferansen for arvelige nevromuskulære tilstander

Tromsø 3rd-5th of September 2025



PROGRAM

Scandic Ishavshotellet Tromsø

THE NORWEGIAN NMD - COLLABORATION

NATIONAL NEUROMUSCULAR CENTRE NORWAY

EMAN
Unit for inborn and hereditary neuromuscular disorders

FRAMBU
RESOURCE CENTRE FOR RARE DISORDERS

MEMBER OF
European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)
Member Oslo University Hospital — Norway

Day 1

Program Wednesday September 3rd

Lunch 12:45-13:45

13:45 - 14:00

Welcome!

Introduction

National neuromuscular centre Norway

14:00 - 14:30

Diagnosis as an adult
-a patient story

Tor Egil Roheim, FFM

14:30 - 15:30

Gene therapy in neuromuscular disorders:
what are we learning?

Laurent Servais, University of Oxford and University of Liège

Break with refreshments (15:30 - 16.00)

16:00 - 17:00

«A birds eye view at the neuromuscular patient»

Nicol Voermans, Radboud UMC

17:00 - 17:45

The model of care pathway from trial readiness to standards of care updates. Insights from Newcastle

Meredith James, John Walton MDRC

Short break (17:45-18:00)

18:00 - 19:00

Conversation:

Reflections around the remarkable life as a parent to Ibelin

Robert Steen, Ibelin Foundation

19.30 - ...

«Get-together»

SKIRRI

Day 2

Program before lunch Thursday September 4th

LMS-kurs: «Nye muligheter gir nye behov»

09.00 - 09:30 — LMS-kurs: Oppstart og bli kjent

NMK

09:30 - 10:00 — LMS-kurs (foredrag): *Nye muligheter, nye behov*

Tollef Ladehaug, Foreningen for muskelsyke

Pause (10:00-10:30)

10:30 - 11:30 — LMS-kurs (foredrag): *«Nå får det fanmeg være nok!»*

Einar Angelsen

11:30 - 12;99 — LMS-kurs gruppe-samtale

Parallelles for health care professionals/researchers/others

08:30 - 09:15 — Lecture: *The role of the muscle biopsy in diagnostics of NMDs in 2025 and forward*

Teresinha Evangelista, AP-HP – Pitié-Salpêtrière Hospital and ERN Euro-NMD

08:15 - 09:45 — Workshop: *North Star Assessment for Limb Girdle Type Muscular Dystrophies (NSAD)*

Meredith James, John Walton MDRC

09:15 - 10:00 — Lecture: *Anesthesia and neuro-muscular disorders: what a neurologist needs to know.*

Nicol Voermans, Radboud UMC

09:45 - 10:10 — Lecture: *A motor control and movement quality approach to neuromuscular physiotherapy: Does numerical test data fully capture changes in function? A clinical perspective*

Helge Haestad, National neuromuscular centre Norway

Break with refreshments (10:00 - 10.30)

10:30 - 11:15 — Accelerating clinical development in rare disease. How can we be more efficient?

Laurent Servais, University of Oxford and University of Liège

11:15 - 12:00 — Clinical trials– experiences from Denmark

Nicolai Preisler, Rigshospitalet Copenhagen

Lunch break 12:00—13:00

Day 2

Program after lunch Thursday September 4th

13:00 - 14:30 — Panel discussion: «Start and stop criteria in advanced therapies»

Panel: TBA

Break with refreshments (10:00 - 10.30)

14:45 - 15:15 — Nutrition and neuromuscular disorders (in the age of Wegovy)

Marianne Nordstrøm, Frambu and Oslo University Hospital

15:15 - 16:00 — Pathophysiology and management of respiratory failure in NMD

Tiina Andersen, Western Norway University of Applied Sciences and Haukeland UH

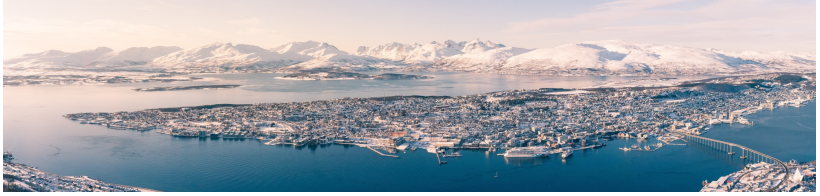
16.00: Academic program finsihed

Approximately from 16:15 to 18:15 — Social activities (separate registration)

- *RIB-tour*
- *City walk*
- *Mack brewery tasting*
- *Afternoon tea*

19.15 - ... — Conference dinner

Scandic Ishavshotel



Day 3

Program Friday September 5th

09:00 - 10:00 — Duchenne muscular dystrophy and other NMDs- associated neurobehavioral difficulties

Sam Geuens, University Hospital Leuven

Break with refreshments (10:00 - 10.15)

10:15 - 11:30 — Projects in the Nordic countries

Denmark, Sofie Skoubo: ***«An investigation of the interaction between children and adolescents with neuromuscular diseases and teachers when using telepresence robots in the Scandinavian educational setting"***

Norway, Kristin Ørstavik: ***«Personalised training for people with rare neuromuscular disorders (PETRA-NMD)»***

Sweden: Jonas Gillenstrand: ***«The prevalence of problematic behaviors in relation to parents' and boys' experiences and management of crises in the context of progressive degeneration in Duchenne Muscular Dystrophy (DMD)»***

Finland, Johanna Ranta-Aho: ***«Molecular disease mechanisms of ACTN2-related myopathies»***

Iceland, Kristjan Dereksson: ***«Stabilize or strengthen? Results from a 4-year follow-up study of respiratory function during sleep and from a 6 month respiratory training program in children with SMA type 2 and 3 on disease modifying treatment"***

Lunch break 11:30—12:30

12:30 - 13:15 — ERN Euro-NMD: what is it, and why do we need it?

Teresinha Evangelista, ERN-Euro NMD

13:15 - 13:45 — National reference network (NRN) in Norway. Answering the needs of tomorrow.

The national consortium on rare neuromuscular disorders in Norway

13:45 - 14:00 — Goodbye and welcome back

National neuromuscular centre Norway

Speakers

Tor Egil Roheim ***-Neuromuscular Disorders Association of Norway (NMD Norway)*** 	**Wednesday September 3rd: *«Diagnosis as an adult- a patient story»*** *Tor Egil Roheim has been the leader of the Neuromuscular Disorders Association of Norway (NMD Norway) since June 2024. He has also been the leader of the local chapter of NMD Norway in Oslo and Akershus from 2015 to 2025. Tor Egil graduated as a civil engineer in structural and construction engineering from the Norwegian Institute of Technology at the University of Trondheim in 1995. He currently works as a consulting engineer.* *Tor Egil has an special interest in universal design and accessibility for people with disabilities so that they can participate in society on an equal footing with everyone else.* *He was diagnosed with FSHD at the age of 42 in 2010 after a period of frequent stumbling and difficulty walking up stairs. Initially he was determined never to end up in a wheelchair, but changed his mind in 2015 (mostly out of need). He was active in various sports during his childhood and youth, but naturally, this has decreased. The diagnosis made him realize that he wasn't lazy, but that there was actually a reason why he couldn't run long distances.*
Laurent Servais ***-University of Oxford, UK, and University of Liège, Belgium*** 	**Wednesday September 3rd: *«Gene therapy in neuromuscular disorders: what are we learning?»*** **Thursday September 4th: *«Accelerating clinical development in rare disease. How can we be more efficient?»*** *Laurent Servais is Professor of Paediatric Neuromuscular Diseases at the University of Oxford in the UK, and invited Professor at the University of Liège in Belgium. He graduated in medicine and pediatrics from the University of Louvain (in Louvain-la-Neuve and Brussels in Belgium) then trained as a child neurologist in the Robert Debré Hospital in Paris, France, and as a myologist at the Institute of Myology, Pitié-Salpêtrière University Hospital, Paris.* *Professor Servais' main research interests cover innovative outcome measures and clinical trials design and newborn screening (NBS). He has been involved as Principal Investigator in several clinical trials in spinal muscular atrophy (SMA), Angelman Syndrome X-linked myotubular myopathy, and Duchenne muscular dystrophy (DMD), and in leading the NBS program for SMA NBS in Belgium and in the UK.* *He is the coordinating investigator of two large natural history studies in Angelman in the UK and in Belgium that aims to identify and validates innovative outcome measures and biomarkers.*

THE NORWEGIAN NMD - COLLABORATION

Speakers

Nicol Voermans ***-Radboud University Medical Center, Netherlands*** 	**Wednesday September 3rd: «A birds eye view on the neuromuscular patient»** **Thursday September 4th: «Anesthesia and neuromuscular disorders: what a neurologist needs to know"** *Nicol C. Voermans is a distinguished neurologist specializing in neuromuscular disorders at Radboud University Medical Center (Radboudumc) in Nijmegen, Netherlands. She completed her studies in Medicine and Philosophy at Radboud University, earning her PhD in 2011 with a thesis on the neuromuscular aspects of Ehlers-Danlos and Marfan syndromes.* *Since joining Radboudumc as a medical specialist in 2008, she has focused on congenital myopathies and facioscapulohumeral muscular dystrophy (FSHD). Her research emphasizes genotype-phenotype correlations, trial readiness, and the development of optimal symptomatic treatments. She has collaborated with esteemed researchers, including Prof. Jungbluth in London and Prof. Treves in Basel.* *In 2021, she launched the FSHD European Trial Network, uniting researchers and clinicians across Europe to advance FSHD research and clinical trials. Her contributions to the field are reflected in her extensive publication record, with over 11,000 citations, underscoring her impact on neuromuscular research. In recognition of her significant contributions, she was appointed Professor of Muscular Diseases at Radboudumc, effective October 1, 2023.* *Prof. Voermans's dedication to bridging the gap between neuromuscular research and clinical practice continues to enhance the understanding and treatment of rare genetic muscle disorders*
Meredith James ***-John Walton Centre for Muscular Dystrophy Research, Newcastle UK*** 	**Wednesday September 3rd: *"Model of care in Newcastle– from trial readiness to standards of care updates"*** **Thursday September 4th: «*Workshop North Star Assessment for Limb Girdle Type Muscular Dystrophies (NSAD)*»** *Dr Meredith James PT PhD is a Clinical Specialist Neuromuscular Physiotherapist and Principal Investigator at the John Walton Centre for Muscular Dystrophy Research at Newcastle, where she is involved in clinical and research activity for children and adults with Neuromuscular disorders.* *Clinically, Meredith is responsible for the physiotherapy management of both children and adults with neuromuscular diseases.* *In her clinical research capacity, Meredith is responsible for the clinical evaluation of children and adults involved in natural history and clinical trials, as well as the development of clinically meaningful, reliable and sensitive outcome measures for Neuromuscular disorders.* *Meredith has been involved in the development of the North Star Ambulatory assessment (NSAA), the Performance of upper limb (PUL) and led the development of the North Star Assessment for Limb Girdle Type Muscular Dystrophies (NSAD) now utilised in clinics and clinical trials in Limb Girdle Muscular Dystrophy, Becker Muscular dystrophy and Late Onset Pompe Disease*

Speakers

Robert Steen

-The Ibelin Foundation, Norway

Wednesday September 3rd: *"Reflections around the remarkable life as a parent to Ibelin"*

Through his gaming character "Ibelin," Mats Steen, who lived with the muscle disease Duchenne muscular dystrophy, became known in the gaming world as a strong and compassionate figure. After Mats' passing, his family shared the story of his life—a story that has now touched millions of people worldwide through the Netflix film "Ibelin." Mats demonstrated that true friendship and meaningful lives can be lived in many ways, including on new platforms and in new arenas.

At the conference, Mats' father, Robert Steen, will be speaking. Robert is also known for his role as Oslo's Commissioner for Health during the COVID-19 pandemic. Robert has previously shared the family's experiences in an interview with NRK in 2019, which became the most-read article in NRK's history, and through the book "Om natten skinner stjernene" ("At Night, the Stars Shine").

In December 2024, the Steen family received VG's "Person of the Year" award for their openness, which has helped break stigmas surrounding both disabilities and gaming culture. Their story reminds us of the value of seeing each other for who we truly are, regardless of the circumstances in which we live.

Teresinha Evangelista

-AP-HP – Pitié-Salpêtrière Hospital and ERN Euro-NMD, Paris, France

Thursday September 4th: *«The role of the muscle biopsy in diagnostics of NMDs in 2025 and forward»*
Friday September 5h: «*ERN Euro-NMD: What is it, and why is it important?*

Dr Teresinha Evangelista has over 20 years' experience working as a Neurologist and later as a Consultant at the Neurosciences Department of the Hospital de Santa Maria in Lisbon. These roles have been completed in conjunction with research work as a member of the Neuromuscular Research Unit at the Institute of Molecular Medicine as well as has an Invited Lecturer at the Faculty of Medicine at Lisbon University in addition to a range of other teaching and advisory posts. She is now the ERN Euro-NMD coordinator, based at AP-HP – Pitié-Salpêtrière Hospital in Paris

After working in Paris, at the Unit 153 of INSERM and having finish a post-graduation in Neuropathology, she set up a fully equipped Neuromuscular Laboratory at the Hospital de Santa Maria in Lisbon. The establishment of this laboratory stimulated the activity within the field of Clinical Neuromuscular research at other facilities in Portugal. The laboratory became a Reference Laboratory for the South of Portugal. Dr Teresinha Evangelista played an important role in the development of a diagnostic network for Neuromuscular Diseases during the years working in Portugal and was appointed as President of the Portuguese Society for The Study of Neuromuscular Diseases in 2009 in recognition of status as a leading reference for Neuromuscular Diseases in Portugal.

Her work currently focuses on clinical and research work on hereditary muscle diseases, FSHD and Congenital Myasthenic Syndromes. Simultaneously as a member of the Joint Actions for rare diseases (EUCERD Joint Action and RD-Action), developed a particular interest on Rare Diseases policies. As a result of the policy work she became responsible for the concept and setting up of the neuromuscular ERN (EURO-NMD) that integrates 61 healthcare providers from across Europe.

Thus far, she has published over 70 peer-reviewed papers and 5 book chapters. She has given many invited lectures at meetings, symposiums and workshops as well as patient/ parent organizations meetings.

Speakers

Helge Hæstad

-National neuromuscular centre, UNN, Norway

Thursday September 4th: *«A motor control and movement quality approach to neuromuscular physiotherapy: Does numerical test data fully capture changes in function? A clinical perspective»*

Helge Aronsen Hæstad is a clinical specialist in neurological physiotherapy at The national neuromuscular Centre of Norway, He has more than 30 years of clinical experience in hands-on work with a mixture of neuromuscular and central nervous system conditions and postgraduate teaching.

He has been involved in several research projects related to physiotherapy interventions at NMK, as well as innovation projects on e-learning courses to physiotherapists and the use of state of the art outcome measures such as gait mats.

A major field of interest is the interaction between the peripheral neuromuscular system and the systems within the brain to activate and sustain functional movements.

Nicolai Preisler

-Copenhagen Neuromuscular Center, Rigshospitalet, Denmark

Thursday September 4th: *«Clinical trials– experiences from Denmark"*

Nicolai Preisler is a Consultant Neurologist and the Head of the Clinical Trial Section at the Copenhagen Neuromuscular Center, Rigshospitalet. He holds an MD and a PhD from the University of Copenhagen and has dedicated his career to advancing clinical research in neuromuscular disorders.

With extensive experience in conducting and overseeing clinical trials, Dr. Preisler has served as Principal Investigator in more than 10 sponsor-driven studies and as a Sub-Investigator in over 25 trials, covering phases I, II, and III. He has significant expertise in gene therapy trials and is ICH-GCP certified, ensuring the highest standards in clinical research.

His work with clinical trials spans various neuromuscular conditions, including mitochondrial disorders, myasthenia gravis, limb-girdle muscular dystrophy, and fatty acid oxidation defects. He has also been involved in studies focusing on Charcot-Marie-Tooth disease, Pompe disease, centronuclear myopathies, dysferlinopathy, facioscapulohumeral muscular dystrophy, spinal muscular atrophy, and dystrophinopathy.

Dr. Preisler has a strong publication record in international peer-reviewed journals. His primary research interests focus on neuromuscular diseases, particularly metabolic myopathies and the role of exercise as a treatment for neuromuscular disorders.

In addition to his clinical and research roles, Dr. Preisler contributes to national healthcare policy as a committee member of the Danish Medicines Council, serving in the specialist group for congenital metabolic disorders, where he has been Deputy Director since 2023.

At the conference, Dr. Preisler will present insights into the ongoing and upcoming clinical trials at the Copenhagen Neuromuscular Center, highlighting their impact on advancing therapies for neuromuscular disorders.

Speakers

Marianne Nordstrøm

-Frambu Resource Centre for Rare Disorders and Oslo University Hospital, Norway

Thursday September 4th «Nutrition and NMDs (in the age of Wegovy)»

Marianne Nordstrøm is a clinical dietitian and researcher at the Frambu Resource Centre for Rare Disorders and at the Unit for Rare Neuromuscular Disorders, Oslo University Hospital. She also holds a part-time position in the Department of Neurohabilitation at Oslo University Hospital.

Her research focuses on nutrition in Spinal Muscular Atrophy and Primary Periodic Paralysis. She currently serves as Chair of the Multidisciplinary Management and Care Working Group and sits on the Executive Committee within the European Reference Network for Neuromuscular Diseases (ERN-NMD).

Tiina Andersen

-Western Norway University of Applied Sciences and Haukeland University Hospital, Norway

Thursday September 4th: *"Patophysiology and management of respiratory failure in NMD"*

Tiina Andersen is a Physiotherapist and Specialist in Cardiopulmonary Physiotherapy with a PhD. She is currently serving as an Associate Professor at the Physiotherapy Education, Faculty of Health and Social Sciences, Western Norway University of Applied Sciences, and has a clinical position at the Advanced Home Hospital at the Thoracic Department of Haukeland University Hospital in Bergen, Norway.

She worked at the Norwegian Advisory Unit for Home Mechanical Ventilation (Bergen, Norway) from its establishment in 2002 until its closure in 2023. This unit played a pivotal role in developing and disseminating expertise on the assessment and treatment of patients requiring long-term mechanical ventilation across Norway.

Tiina was specifically responsible for advancing knowledge and practices related to airway clearance techniques for this patient group. Her research has particularly focused on the role of the upper airways during these techniques, with the goal of enhancing respiratory care for patients. In addition to her research, she has been deeply involved in education, teaching patients, caregivers, students, and healthcare professionals. Today, Tiina is recognized as one of the world's leading physiotherapists in the field of chronic respiratory failure.

Sam Geuens

-University Hospital Leuven, Belgium

Friday September 5th: *«Duchenne muscular dystrophy (and other NMDs)-associated neurobehavioral difficulties»*

Sam Geuens, PhD, is a pediatric neuropsychologist specialized in the neurobehavioral and psychosocial development of children and young adults with neuromuscular or neurological disorders. He works at the University Hospitals in Leuven, Belgium, where he provides psychological counseling to children with neurological disorders and their family.

He combines his clinical work with research to gain a better understanding of neurobehavioral aspects of neuromuscular diseases and has published in international peer-reviewed journals about these topics.

Speakers

Sofie Skoubo ***-National Rehabilitation Center, Denmark*** 	**Friday September 5th*: "An investigation of the interaction between children and adolescents with neuromuscular diseases and teachers when using telepresence robots in the Scandinavian educational setting"*** *Sofie Skoubo is Ph.d student at the National Rehabilitation Center for Neuromuscular Diseases in Denmark. The PhD project, "My Avatar," explores using telepresence robots in the educational setting to reduce school absence for children and adolescents with neuromuscular diseases.* *The project cooperates with the Norwegian company No Isolation, which has developed a telepresence robot. Since 2020, Sofie has worked with telepresence robots for children and adolescents with different illnesses in Denmark.* *Sofie was diagnosed with neuromuscular disease when I was six years old and have experienced the tough years of being in compulsory education and accepting a physical diagnosis*
Kristin Ørstavik **-Section for rare neuromuscular disorders, Oslo University Hospital, Norway** 	**Friday September 5th*: Personalised training for people with rare neuromuscular disorders (PETRA-NMD)»*** *Kristin Ørstavik is a MD and PhD, currently working as a Senior consultant in Neurology and Clinical Neurophysiology. She is the Head of Section for Rare Neuromuscular disorders and EMAN; Department of Neurology, Oslo University Hospital.* *Her research interest covers multiple disorders and thematic fields, including Myotonic dystrophy type 1, pain, effect of exercise in Neuromuscular disorders and Cardiomyopathy in patients with neuromuscular disorders.* *She took the initiative to an informal National Reference network for Rare Neuromuscular disorders in Norway over 10 years ago. This network is a model for future NRNs in Norway. From 2022 she has been the board representative from the Consortium for Rare neuromuscular disorders in Norway to ERN Euro-NMD.*
Johanna Ranta-aho ***-Folkhälsan Research Center and University of Helsinki, Finland*** 	**Friday September 5th*: Molecular disease mechanisms of ACTN2-related myopathies»*** *Johanna Ranta-aho is a PhD student in the Myofin group lead by Dr. Marco Savarese. The group is part of Folkhälsan Research Center and the University of Helsinki, and is based in Helsinki, Finland.* *The group's research is focused on hereditary neuromuscular disease, with an emphasis on titinopathies, other distal myopathies, and progressive muscular dystrophies.* *As part of her PhD project, she aims at characterizing the genotype-phenotype correlations and molecular disease mechanisms of a specific gene known to cause myopathy, ACTN2.*

Day 3

Speakers

Kristjan Dereksson

-Landspitali University Hospital, Reykjavik, Iceland

Friday September 5th*: «Stabilize or strengthen? Results from a 4-year follow-up study of respiratory function during sleep and from a 6 month respiratory training program in children with SMA type 2 and 3 on disease modifying treatment“*

Kristjan Dereksson is a pediatrician since 2014, specialized in pediatric lung medicine. He lives in Reykjavik Iceland with his wife and 3 children and works at Landspitali university hospital, in Reykjavik, with treatment related to sleep and breathing in both children and adults.

He also works at the pediatric unit of Skåne university hospital, in Lund, Sweden where he is responsible for pediatric sleep studies and works extensively with children with breathing difficulties related to neurologic- and neuromuscular-diseases.

His research into SMA is part of doctorate studies at Lunds University, Sweden.

Tollef Ladehaug

-Foreningen for muskelsyke

LMS-kurs torsdag 04.september: *“Nye muligheter, nye behov”*

Tollef Ladehaug er statsviter og ansatt i foreningen for muskelsyke der han blant annet er ansvarlig for søknader om prosjektmidler og som prosjektleder for de mange av foreningens prosjekter.

Han har i den kapasitet ledet ey titalls prosjekter for FFM. 13 av dem har munnet ut i publikasjoner han har skrevet, inkludert "Du ser da frisk ut - Fatigue for muskelsyke og alle andre", Selvforsvar uten muskelstyrke", "Fysiske funksjonshemninger og utmattelse – En kartlegging av elever med muskelsykdommer i grunnskolen" og "Min Kropp, mine grenser" om assistanse og grensesetting.

Einar Johan Angelsen

LMS-kurs torsdag 04.september: “Nå får det fanmeg være nok!”

Einar er mannen ingen synes synd på. Burde vi det? Eller bør vi spørre oss selv hva han kan lære oss? Han ble født uten fingre og høyre fot, noe han har brukt et halvt liv på å lære å mestre.

Det er likevel lite som stopper Einar. Men så - i sommer, hadde han max uflaks på terrengsykkel og pådro seg fem brudd i ryggen, en ryggmargsskade og fire ribbebeinsbrudd. Endelig var det noen som syntes synd på han, men hvor lenge kunne det vare?

Einars mindset, pågangsmot, vilje og humor vil inspirere og berøre oss.

Noen vil kjenne Einar igjen fra NRK-serien «Ingen grenser» med Lars Monsen på tur til Snøhetta i 2012. Mange har sett filmene hans på Youtube, ofte med humoristiske kommentarer, men alltid med fantastiske bilder og god formidling. Einar er høy og lavt – fra kaikanter og asfalt midt i Tromsø by til toppen av Stetind