

# ERN EURO-NMD 8<sup>TH</sup> ANNUAL MEETING

📅 05, 06 and 07 March 2025

📍 Teaching and Learning Center (Lehr- und Lernzentrum),  
Virchowstraße 163a, 45147 Essen, Germany



**European  
Reference  
Network**

for rare or low prevalence  
complex diseases

🌐 **Network**

Neuromuscular  
Diseases (ERN EURO-NMD)

👤 **Coordinator**

Assistance Publique -  
Hôpitaux de Paris,  
Hôpital Pitié-Salpêtrière  
— France

## MEETING PROGRAMME

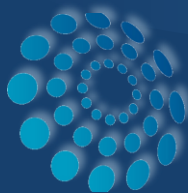
All plenary sessions take place in the Auditorium. Rooms are specified for other meetings.

DAY 1 - WEDNESDAY, MARCH 5 <sup>th</sup>					
Time	Session				
12:45 - 13:30	Registration				
13:30 - 13:40	Welcome note - Ulrike Schara-Schmidt and Teresinha Evangelista				
13:40 - 14:45	Keynote lectures				
13:40 - 14:10	Exercise and Neuromuscular Disorders - Nicole Voet (moderator: Kristin Ørstavik)				
14:10 - 14:20	Discussion				
14:20 - 14:50	Sleep and Neuromuscular Disorders - Matthias Boentert (moderator: Antonio Atalaia)				
14:50 - 15:00	Discussion				
15:00 - 15:30	Coffee Break				
15:30 - 16:00	Panorama of devices for the non-invasive assessment of the neuromuscular system - Jean-Yves Hogrel				
16:00 - 16:30	Patient reported outcome measures: PROMMY the new app in the PROMs' ecosystem - Michelangelo Mancuso				
16:30 - 17:00	Landscape of gene therapy practices across Europe - Kleopas Kleopa				
17:00 - 17:30	Coffee Break - Room change				
17:30 - 19:00	Parallel Working Groups session 1				
	<b>Peripheral nerve diseases</b> <b>Room 1</b> Davide Pareyson and Kleopas Kleopa	<b>Mitochondrial diseases</b> <b>Room 2</b> Michelangelo Mancuso & Cornelia Kornblum	<b>Muscle diseases</b> <b>Auditorium</b> Marianne de Visser	<b>Neuromuscular Junction</b> <b>Room 3</b> Hanns Lochmuller & Lorenzo Maggi	<b>Neuromuscular Imaging</b> <b>Room 4</b> Pierre Carlier
<b>End of the Day 1 plenary programme</b>					

DAY 2 - THURSDAY, MARCH 6 <sup>th</sup>	
Time	Session
08:45 - 10:00	<b>Transition of Care</b>
08:45 - 09:05	Introduction and the Essen use case - Ulrike Schara-Schmidt and Tim Hagenacker
09:05 - 09:20	A French centre experience - Marion Masingue
09:20 - 09:30	EURO-NMD's transition survey results - Teresinha Evangelista
09:30 - 10:00	Round Table "Transition according to patients' needs"
10:00 - 10:30	Coffee Break
10:30 - 12:30	<u>Presentation of Case Reports</u> Moderated by Anna Kostera-Pruszczyk and Tanya Stojkovic
12:30 - 13:30	Lunch
13:30 - 15:00	<b>EURO-NMD outreach activities</b> Moderated by António Atalaia
13:30 - 14:00	NMD4C - interaction between neuromuscular networks - Hanns Lochmüller
14:00 - 14:30	ECTN European Clinical Trials Network - Nicol Voermens and Ria de Haas
14:30 - 15:00	ERDERA - Funding and Clinical Research Network contributions for ERNs development- Gisèle Bonne
15:00 - 15:30	Coffee Break
15:30 - 19:00	<b>Satellite scientific symposium: Newborn Screening</b> <i>This symposium is sponsored by Novartis &amp; UCB.</i> <b>Detailed programme on the last page of this document.</b>
19:30 - 22:00	End of the Day 2 programme - Networking Dinner at Restaurant Hülsmannshof Lehnsgrund 14a, 45149 Essen, <a href="http://www.huelsmannshof.de">www.huelsmannshof.de</a>

DAY 3 - FRIDAY, MARCH 7 <sup>th</sup>					
Time	Session				
08:15 - 09:45	<b>Parallel Working Groups session 2</b>				
	<b>Motor Neuron Disorders</b> Room 1 Jan Kirschner & Vincenzo Silani	<b>Neuropathology</b> Room 2 Edoardo Malfatti	<b>Genetics</b> Auditorium Borut Peterlin	<b>Multidisciplinary Management &amp; Care</b> Room 3 Marianne Nordstrøm	<b>Neurophysiology</b> Room 4 Camiel Verhamme & Shahram Attarian
09:45 - 10:00	Coffee Break - Room change				
10:00 - 12:30	<b>Board meeting (invitation only)</b> <i>Programme sent separately</i>				
12:30 - 13:30	Lunch (invitation only)				
14:00	<b>End of the programme</b>				

Thursday 06 March 2024, 15:30 – 19:00  
Teaching and Learning Center  
(Lehr- und Lernzentrum)  
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# Newborn Screening

Satellite Symposium organized by  
EURO-NMD  
European Reference Network for  
Rare Neuromuscular Disorders

## Agenda

- 15:30 – 15:40** Introduction – *Teresinha Evangelista, AP-HP, Paris, France*
- 15:40 – 16:00** Genetic newborn screening through targeted gene panel and WGS: the Screen4Care approach – *Fernanda Fortunato, University of Ferrara, Italy*
- 16:00 – 16:20** NBS for Spinal Muscular Atrophy – *Jan Kirschner, Uniklinikum Freiburg, Germany*
- 16:20 – 16:40** NBS for SMA seen by patients and carers – *Marie-Christine Ouillade, AFM Téléthon, France*
- 16:40 – 17:00** **Coffee Break**
- 17:00 – 17:20** NBS for Duchenne Muscular Dystrophy – *Elizabeth Vroom, World Duchenne Organization*
- 17:20 – 17:40** NBS for Cystic Fibrosis: The German landscape – *Lutz Nährlich, Justus-Liebig-Universität Gießen, Germany*
- 17:40 – 18:00** NBS for Rare Metabolic Diseases – *Maurizio Scarpa, Coordinator of MetabERN, ERN for Rare Metabolic Diseases*
- 18:00 – 18:20** NBS for Rare Diseases – *Gulcin Gumus, EURORDIS*
- 18:20 – 19:00** Discussion and Closing Remarks

This symposium is sponsored by

