European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

7th NEUROMUSCULAR TRANSLATIONAL TRAINING SCHOOL

Organized by the ERN EURO-NMD and hosted by the Leiden University Medical Center

2 – 5 December 2025

Leiden, Netherlands

7th Neuromuscular Translational Training School

Hosted by LUMC

December 2-5 2025

Leiden University Medical Center, the Netherlands

Programme committee:

Annemieke Aartsma-Rus	Leiden University Medical Center, LUMC, the Netherlands & John Walton Muscular Dystrophy Research Center, Newcastle University, UK
Teresinha Evangelista	APHP - Groupe Hospitalier Pitié-Salpêtrière, Paris, France
Andoni Urtizberea	Institut de Myologie, Paris, France

Target audience:

- MDs
- PhD/Postdoc researchers
- Others working in translational research
- Preferably in the NMD field, but in either case working in the RD field
- Aim for 20-25 participants
- Industry delegates

Aim:

- Facilitate the clinical development of therapies for NMDs

Objectives:

- **Educate clinicians and researchers working in the NMD field on aspects relevant for translational therapy development:**
  - Bench to bedside research
  - Regulatory system
  - Clinical trials
  - Outcome measures
  - Patient communication
  - Registries and biobanks
  - Biomarkers and -omics
- **Outline and showcase how networks like EURO-NMD and TREAT-NMD facilitate therapy development:**
  - Standards of care
  - Clinical trial tools
  - Outcome measure development
  - Interaction with stakeholders

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PROGRAMME

Location: LUMC

Tuesday December 2nd

9.00 – 9.30	**Registration and Coffee**
Session 1	**Introduction and overview of the neuromuscular diseases (NMDs) landscape**
9.30 – 10.00	**Welcome and introduction** *Teresinha Evangelista, Andoni Urtizberea and Annemieke Aartsma-Rus* **Objective:** Introduction of participants and organizers; layout of the program and learning objectives
10.00 – 11.00	**Challenges and needs in RD therapy development** *Teresinha Evangelista and Annemieke Aartsma-Rus*
11.00 – 11.15	**Tea/coffee break**
11.15 – 12.15	**Overview of current state of the art of NMD diseases management** *Andoni Urtizberea* **Objective:** give participants a global overview of the different groups of NMDs, management, practices and currently approved innovative treatments (45' talk, 15' discussion)
12.15 – 13.15	**Innovative therapies for NMDs** *Annemieke Aartsma-Rus* **Objective:** outline how genetic therapies (gene addition, exon skipping and stop codon readthrough) work for Duchenne, and give an overview of approved approaches for NMDs (45' talk, 15' discussion)

13.15 – 14.00	**Lunch**
Session 2	**Preclinical Research**
14.00 – 15.00	**Tools of the trade for preclinical research** *Annemieke Aartsma-Rus* **Objective:** outline different models used in preclinical research, their opportunities and limitations, and the need for standardized tests and the TREAT-NMD advisory committee for therapeutics (TACT) (30' talk, 20' discussion)
Session 3	**Clinical research part 1**
15.00 – 16.00	**Introduction to clinical trials** *Michela Guglieri* **Objective:** introduce how and why clinical trials are conducted, the objective of different trial phases, trial design, primary endpoints, secondary endpoints, clinical significance, ethical concerns and informed consent (45' talk, 15' discussion)
16.00 – 16.30	**Tea/coffee break**
16.30 – 17.15	**Tools to facilitate clinical trials- Registries** *Michela Guglieri* **Objective:** to gain insight in available tools and services for planning and conducting clinical trials (20' talk, 20' discussion)
17.15 – 18.00	**End of day – Q&A with refreshments and nibbles** **Meet the speakers**

Wednesday December 3rd

Session 4	**Clinical research part 2**
09:00 – 10:00	**Unexpected aspects of conducting a clinical trial** *Michela Guglieri* **Objective:** provide insight in the logistics of running a clinical trial (recruitment, treating, dealing with patient expectations, informed consent etc.)
10.00 – 10.15	**Tea/coffee break**
10.15 – 11.45	**How the regulatory system works** *Marta Kollb-Sielecka* **Objective:** explain the centralized system, how it is organized and how regulators decide whether drugs are eligible for marketing authorization, outline patient involvement in committees – focus on rare diseases (60' talk, 30' discussion)
11.45 – 12.45	**Industry perspective on drug development for rare diseases** *Eric van der Veer* **Objective**: provide the perspective of pharmaceutical companies on drug development for rare diseases, which challenges do they face, how do they deal with them (40' talk, 20' discussion)
12.45 – 13.35	**Lunch**
Session 5	**TACT mock review session**
13.35 – 13.45	**Introduction to TACT mock review session**
13.45 – 15.15	**Self-study for TACT mock review session** All students are expected to provide input during the mock review. Study material will be provided the week before the summer school so students can prepare before the meeting or during the self-study time.

15.15 – 16.30	**When to move to a clinical trial? TACT mock review session** *moderated by Annemieke Aartsma-Rus and Teresinha Evangelista* **Objective:** learning to have a critical look at preclinical research. In this mock session, participants will have been provided with a fictitious TACT application from a company planning a clinical trial in the NMD space. Participants will be split into groups to discuss the strengths and limitations and outstanding questions of the application.
16.30 – 17.00	**End of day – Refreshments and snacks**

Faculty Dinner

Thursday December 4th

Session 6	**Focus on outcome measures (Requirements, selection, development, biomarkers, PROMs)**
09.00 – 10.30	**Outcome measures** *Jean-Yves Hogrel* **Objective:** outline of requirements of outcome measures used in clinical trials, how to select the best outcome measure for a pivotal trial; using real life examples of successful and failed trials (e.g. drisapersen 6MWT) (60' talk, 30' discussion)
10.30 – 11.00	**Tea/coffee break**
11.00 – 12.00	**Showcase on outcome measure development** *Jean-Yves Hogrel* **Objective**: outline the steps and stakeholders involved in developing and validating a functional outcome measure (select one – indicate the same applies for all) (45' talk, 15' discussion)
12.00 – 12.45	**Lunch**

12.45 – 13.45	**Showcase: validation of MRI as a biomarker in clinical trials** *Hermien Kan* **Objective:** introduce MRI as an outcome measure in trials, ongoing efforts to validate this biomarker for assessment of muscle quality in neuromuscular diseases (45' talk, 15' discussion)
13.45 – 14.45	**Biomarkers** *Pietro Spitali* **Objective:** explain the different types of biomarkers, how they can be used in trial planning and as outcome measures, the regulatory perspective on biomarkers, highlighting ongoing networking efforts (45' talk, 15' discussion)
14.45 – 15.00	**Tea/coffee Break**
15.00 – 16.00	**Showcase: PROM development** (the questions you ask and why you ask them; practical examples) *Céline Desvignes-Gleizes* **Objective:** explain what is involved in developing a patient reported outcome measure using the DMD PROM development as a showcase (45' talk, 15' discussion)
Session 7	**Patient engagement**
16.00 – 17.00	**How patients can help your research from bench to bedside** *Speaker TBC* **Objective:** Examples are given of how patients can be involved in helping with all steps of therapy development, to underline that patients are not only study objects, but also active participants.
17.00 – 17.30	**Patient participation in clinical trials** *Teresinha Evangelista*
19.00	**Conference Dinner**

Friday December 5th

Session 8	Patient engagement and Post Marketing
9.00 – 11.00	**Translating science to the non-initiated** *Ronald Veldhuizen* **Objective:** Participants will gain insight in how to clearly explain science to people without a scientific background – with a focus on patients where the added challenge is not to overpromise or raise false expectations (60' talk, 30' discussion)
11.00 – 11.20	**Tea/coffee break**
11.20 – 11.30	**Introduction : presenting science to patients** *Annemieke Aartsma-Rus*
11.30 – 13.30	**Preparation for the Symposium on presenting science to patients** *Annemieke Aartsma-Rus* **Objective:** participants are divided into groups of 4 that will each be provided with a scientific paper. They are required to prepare a presentation (~10 minutes with 5 minutes discussion time) to inform patients of the scientific findings in a clear and objective manner; other groups will listen as patients/families. Lunch will be served at 13.00
13.30 – 15.00	**Symposium:** Presentations from each of the groups *facilitated by Teresinha Evangelista, Annemieke Aartsma-Rus and Maaike van Putten* **Objective:** each of the groups presents their presentation, the other participants are the audience and listen as if they were a patient
15.00 – 16.00	**Feedback and Evaluation** *facilitated by Teresinha Evangelista and Annemieke Aartsma-Rus* **Objective:** participants explain what they learned, how they will apply this in their daily work, aspects that should be kept in the programme, aspects that could be removed, things that are missing.
16.00	**End of the programme/departure**