European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

TREAT-NMD Neuromuscular Network

# 6th NEUROMUSCULAR TRANSLATIONAL SUMMER SCHOOL

***Under auspices of ERN EURO-NMD and TREAT-NMD***


**9 – 12 July 2024**

**Leiden, Netherlands**

# 6th Neuromuscular Translational Summer School

***Under auspices of EURO-NMD and TREAT-NMD***

**July 9-12 2024**

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 |
| Silvere van der Maarel | Leiden University Medical Center, LUMC, the Netherlands |
| 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

# 6th Neuromuscular Translational Summer School 2024

## Leiden, Netherlands

## PRELIMINARY PROGRAMME

## Tuesday July 9th LUMC

| | |
|---|---|
| **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**<br>*Teresinha Evangelista, Silvere van der Maarel, Andoni Urtizberea and Annemieke Aartsma-Rus*<br><br>**Objective:** Introduction of participants and organizers; layout of the program and learning objectives |
| **10.00 – 11.00** | **Overview of current state of the art of NMD therapies and management** (overview of genetics and standards of care)<br>*Andoni Urtizberea*<br><br>**Objective:** give participants a global overview of the different groups of NMDs (genetic and acquired) and the current care and management practices (45' talk, 15' discussion) |
| **11.00 – 12.00** | **Innovative therapies for NMDs** *Annemieke Aartsma-Rus*<br><br>**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) |
| **12.00 – 12.45** | **Introduction to TREAT-NMD and the ERN EURO-NMD**<br>*Annemieke Aartsma-Rus and Teresinha Evangelista*<br><br>**Objective:** Introducing the problematic of RD and introduce the networks (goals, achievements and partners etc.; two 15' talks, 15' discussion) |

| | |
|---|---|
| **12.45 – 13.45** | **Lunch** |
| **Session 2** | **Preclinical Research** |
| **13.45 – 14.45** | **Tools of the trade for preclinical research** *Annemieke Aartsma-Rus*<br><br>**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** |
| **14.45 – 15.45** | **Introduction to clinical trials** *Michela Guglieri*<br><br>**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 |
| **15.45 – 16.15** | **Tea/coffee break** |
| **16.15 – 17.00** | **Tools to facilitate clinical trials- Registries** *Michela Guglieri*<br><br>**Objective:** to gain insight in available tools and services for planning and conducting clinical trials (20 ' talk; 20' discussion) |
| **17.00 – 17.10** | **Introduction to TACT mock up session** *Annemieke Aartsma-Rus* |
| **17.10 – 18.00** | **End of day – Q&A with refreshments and nibbles**<br>**Meet the speakers** |

## Wednesday July 10th LUMC

| | |
|---|---|
| **Session 4** | **Clinical research part 2** |
| **09:00 – 10:00** | **Unexpected aspects of conducting a clinical trial**<br>*Teresinha Evangelista and Michela Guglieri*<br><br>**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*<br><br>**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 time) |
| **11.45 – 12.45** | **Industry perspective on drug development for rare diseases**<br>*Eric van der Veer*<br><br>**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 time) |
| **12.45 – 13.45** | **Lunch** |
| **Session 5** | **TACT mock review session** |
| **13.45 – 15.15** | **Self-study for TACT mock review session**<br><br>*All students are expected to provide input during the mock review. Study material will be provided the week before the summerschool 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**<br>*moderated by Annemieke Aartsma-Rus and Teresinha Evangelista*<br><br>**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 – Drinks with nibbles** |

**Faculty Dinner**

## Thursday July 11th LUMC

| **Session 6** | **Focus on outcome measures (Requirements, selection, development, biomarkers, PROMs)** |
|---|---|
| **09.00 – 10.30** | **Outcome measures** *Jean-Yves Hogrel*<br><br>**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 (PUL)**<br>*Jean-Yves Hogrel on behalf of Anna Mayhew*<br><br>**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 – 13.00** | **Lunch** |

| | |
|---|---|
| **13.00 – 14.00** | **Showcase: validation of MRI as a biomarker in clinical trials** *Hermien Kan*<br><br>**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) |
| **14.00 – 15.00** | **Biomarkers** *Pietro Spitali*<br><br>**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<br>(45’ talk, 15’ discussion) |
| **15.00 – 15.15** | **Tea/coffee Break** |
| **15.15 – 16.15** | **Showcase: PROM development** (the questions you ask and why you ask them; practical examples) *Céline Desvignes-Gleizes*<br><br>**Objective:** explain what is involved in developing a patient reported outcome measure using the DMD PROM development as a showcase<br>(45’ talk, 15’ discussion) |
| **Session 7** | **Patient engagement** |
| **16.15 – 17.30** | **How patients can help your research from bench to bedside** *Speaker to be confirmed*<br><br>**Objective:** Examples are given of how patients can be involved in helping with all steps of therapy development, to underline that patients or not only study objects, but also active participants |
| **19.00** | **Conference Dinner** |

## Friday July 12th LUMC

| Session 8 | Patient engagement and Post Marketing |
|---|---|
| 9.00 – 11.00 | **Translating science to the non-initiated** *Ronald Veldhuizen*<br><br>**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<br>(60' lecture, 30' discussion) |
| 11.00 – 11.30 | **Tea/Coffee break** |
| 11.30 – 11.45 | **Introduction : presenting science to patients**<br>*Annemieke Aartsma-Rus* |
| 11.45 – 13.30 | **Lunch and preparing for presenting science to patients**<br>*Annemieke Aartsma-Rus*<br><br>**Objective:** participants are split in groups of 4 and will be provided with a scientific paper. It is their task to generate 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 |
| 13.30 – 15.00 | **Symposium:** Presentations from each of the groups *facilitated by Teresinha Evangelista, Annemieke Aartsma-Rus and Maaike van Putten*<br><br>**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, Annemieke Aartsma-Rus and Maaike van Putten*<br><br>**Objective:** everyone explains what they learned, how they will apply this in their daily work, aspects we should keep in the summerschool, aspects we might considered removing, things that are missing |
| 16.00 | **End of the programme/departure** |