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

TREAT-NMD Neuromuscular Network

5th NEUROMUSCULAR TRANSLATIONAL SCHOOL

Under auspices of ERN EURO-NMD and TREAT-NMD

July 10-14 2023

Leiden, Netherlands

5th Neuromuscular Translational School

Under auspices of EURO-NMD and TREAT-NMD

July 10-14 2023

Leiden University Medical Center, the Netherlands

Building 3 of the LUMC, room V3-28/30 (third floor)

Programme committee:

Annemieke Aartsma-Rus	(Leiden University Medical Center, LUMC, the Netherlands & John Walton Muscular Dystrophy Research Center, Newcastle University, UK)
Teresinha Evangelista	(Institut de Myologie, 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 16-20 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

5th Neuromuscular Translational School – Leiden 2023

Monday July 10th

12.00 – 12.30	**Registration and Lunch**
Session 1	**Introduction** **Overview of current state of the art of NMD therapies and management** (A. Urtizberea) **Introduction to TREAT-NMD and EURO-NMD** (A. Aartsma-Rus and T. Evangelista) **Genetic therapies for NMDs** (A. Aartsma-Rus)
12.30 – 13.00	**Welcome and introduction** (T. Evangelista, A. Urtizberea & A. Aartsma-Rus) ***Objective:*** *Introduction of participants and organizers; layout of the program and learning objectives*
13.00 – 14.00	**Overview of current state of the art of NMD therapies and management** (A. Urtizberea) (overview of genetics and standards of care) ***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)*
14.00 – 15.00	**Genetic therapies for NMDs** (A. Aartsma-Rus) ***Objective:*** *outline how genetic therapies (gene addition, exon skipping and stop codon readthrough) work, and give an overview of approved approaches for NMDs* *(45' talk, 15' discussion)*
15.00 – 15.15	**Tea/coffee break**
15.15 – 16.00	**Introduction to TREAT-NMD and EURO-NMD** (A. Aartsma-Rus and T. Evangelista) ***Objective:*** *Introducing the problematic of RD and introduce the networks (the goals, achievements and partners etc.; two 15' talks, 15' discussion)*

Session 2:	**Preclinical Research** **Tools of the trade for preclinical research** (A. Aartsma-Rus) **Introduction to TACT mock up session** (A. Aartsma-Rus)
16.00 – 16.50	**Tools of the trade for preclinical research** (A. 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)*
16.50 – 17.00	**Introduction to TACT mock up session** (A. Aartsma-Rus)
17.00	**End of day – Q&A with refreshments and nibbles; MEET THE SPEAKERS**

Tuesday July 11th

09.00 – 10.00	**Self-study for TACT mock review session** **When to move to a clinical trial? TACT mock review session** (moderated by A. Aartsma-Rus and Teresinha Evangelista)
10.00 –11.00	**When to move to a clinical trial? TACT mock review session** (moderated by A. Aartsma-Rus with active involvement of Teresinha Evangelista and Andoni Urtizberea) ***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.*
11.00 –11.30	**Tea/Coffee break**
Session 3	**Clinical research** **Introduction to clinical trials** (M. Guglieri) **Ethical discussion role play** (moderated by Annemieke Aartsma-Rus) **How the regulatory system works** (Elena Hernandez Martinez De Lapiscina) **Industry perspective on drug development for rare diseases** (Eric Van der Veer) **Clinical trial practicality forum** (T. Evangelista and T.Gomes)

11.30 – 12.30	**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*
12.30 – 13.30	**Lunch**
13.30 – 14.30	**Ethical discussion role play** (moderated by Annemieke Aartsma-Rus, with active participation of the organisers) *Participants will be provided with a scenario for a clinical trial plan for a drug to be tested in children. Different roles will be given to different participants.* ***Objective:*** *gain insight in ethical discussions related to clinical trials and the perspectives of different stakeholders. Reading of provided material ~5'; discussion ~40', evaluation (leaving roles behind) ~15'*
14.30–15.30	**How the regulatory system works** (Elena Hernandez Martinez De Lapiscina) ***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 (30' talk, 30' discussion time)*
15.30 –16.00	**Tea/Coffee break**
16.00 –17.00	**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 time)*
17.00 – 18.00	**Clinical trial practicality forum** (Teresinha Evangelista and Tiago Gomes) ***Objective:*** *provide insight in the logistics of running a clinical trial (recruitment, treating, dealing with patient expectations, informed consent etc.)*
18.00	**End of day – Q&A with refreshments; MEET THE SPEAKERS**

Faculty Dinner

Wednesday July 12th

Session 4	**Outcome measures** (Jean Yves Hogrel) **Showcase on outcome measure development (PULL)** (Jean-Yves Hogrel) **Showcase: validation of MRI as a biomarker in clinical trials** (Melissa Hooijmans) **Tools to facilitate clinical trials** (Michela Guglieri) **Biomarkers** (Pietro Spitali) **Showcase: PROM development** (Elizabeth Vroom)
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 (PULL)** (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 – 13.00	**Lunch**
13.00 – 14.00	**Showcase: validation of MRI as a biomarker in clinical trials** (Melissa Hooijmans) ***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 – 14.40	**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)*

14.40 – 15.40	**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)*
15.40 – 16.10	**Tea/Coffee Break**
16.10 – 17.10	**Showcase: PROM development** (Elizabeth Vroom) (the questions you ask and why you ask them; practical examples) ***Objective:*** *explain what is involved in developing a patient reported outcome measure using the DMD PROM development as a showcase* *(45' talk, 15' discussion)*
17.10	**End of day – Q&A with refreshments; MEET THE SPEAKERS**

Thursday July 13th

Session 5	**Patient engagement and Post Marketing**
09.00 – 10.30	**How patients can help your research from bench to bedside** (Elizabeth Vroom) ***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*
10.30 – 10.50	**Tea/Coffee break**
10.50 – 11.00	**Introduction : presenting science to patients** (Maaike van Putten)
11.00 – 12.30	**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' lecture, 30' discussion)*
12.30 – 13.30	**Lunch**

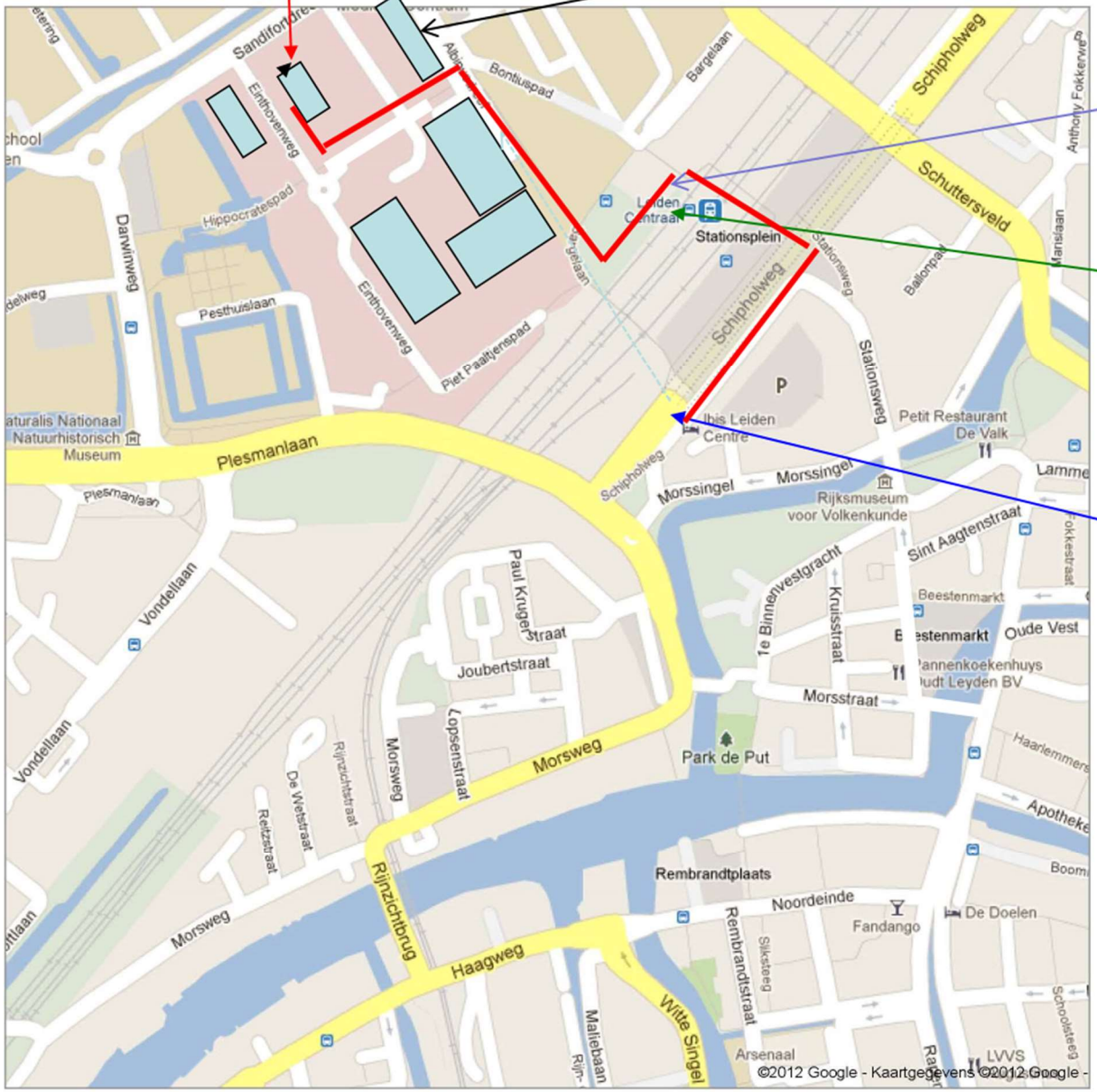
13.30 – 14.30	**Presenting science to patients** (Maaike van Putten) ***Objective:*** *participants are split in groups of 4 and will be provided with a scientific paper. It is their task to generate a presentation (~10-15 minutes) to inform patients of the scientific findings in a clear and objective manner*
14.30 – 16.00	**Symposium:** Presentations from each of the groups (facilitated by Teresinha Evangelista 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*
16.00 – 18.30	**Translating science to inform patients (BLOG)** ***Objective:*** *participants are asked to bring a scientific publication on a potential therapy of their choice. They will now have to summarize this into a 500-word blog understandable by patients and their families*
19.00	**Network dinner**

Friday July 14th

09.00 – 10.30	**Participants work on preparing their final presentation**
10.30 – 12.00	**Participant presentations** (Chaired by Teresinha Evangelista) We ask groups of 4 participants to prepare a 10-15' talk - Who they are and what they expected from the summer school - The things they learnt - How this will influence their daily work - What we should keep in future summer schools - What we should drop/improve - What they were missing ***Online questionnaire for eligibility for EACCME attendance certificates***
12.00 – 12.30	Feedback and general discussion (Chaired by Teresinha Evangelista)
12.30	**Lunch and departure**

LUMC, bldg. 3 – Educational Building / Onderwijsgebouw LUMC.

Parking LUMC



Route by foot

Fletcher Hotel
Bargelaan 180

Railway station

Ibis Hotel
Stationsplein 240