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

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Leiden University Medical Center, the Netherlands

Building 3 of the LUMC

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

PROGRAMME

Monday July 10th Room V-02-010/014

12.00 – 12.30	**Registration and Lunch**
Session 1	**Introduction** **Overview of current state of the art of NMD therapies and management** *Andoni Urtizberea* **Introduction to TREAT-NMD and EURO-NMD** *Annemieke Aartsma-Rus and Teresinha Evangelista* **Genetic therapies for NMDs** *Annemieke Aartsma-Rus*
12.30 – 13.00	**Welcome and introduction** *Teresinha Evangelista, Andoni Urtizberea and Annemieke Aartsma-Rus* **Objective:** Introduction of participants and organizers; layout of the program and learning objectives
13.00 – 14.00	**Overview of current state of the art of NMD therapies and management** (overview of genetics and standards of care) *Andoni Urtizberea* **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** *Annemieke 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** *Annemieke Aartsma-Rus and Teresinha Evangelista* **Objective:** Introducing the problematic of RD and introduce the networks (goals, achievements and partners etc.; two 15' talks, 15' discussion)
Session 2	**Preclinical Research** **Tools of the trade for preclinical research** *Annemieke Aartsma-Rus* **Introduction to TACT mock up session** *Annemieke Aartsma-Rus*
16.00 – 16.50	**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)
16.50 – 17.00	**Introduction to TACT mock up session** *Annemieke Aartsma-Rus*
17.00	**End of day – Q&A with refreshments and nibbles; MEET THE SPEAKERS**

Tuesday July 11th Room V-02-010/014

09.00 – 10.00	**Self-study for TACT mock review session** **When to move to a clinical trial? TACT mock review session** *Annemieke Aartsma-Rus and Teresinha Evangelista*
10.00 –11.00	**When to move to a clinical trial? TACT mock review session** *moderated by Annemieke 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** *Michela Guglieri* **Ethical discussion role play** *moderated by Annemieke Aartsma-Rus*

	How the regulatory system works *Elena Hernandez Martinez De Lapiscina* **Tools to facilitate clinical trials** *Michela Guglieri* **Clinical trial practicality forum** *Teresinha Evangelista and Tiago 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* **Objective:** gain insight in ethical discussions related to clinical trials and the perspectives of different stakeholders. 5' Reading of provided material; 40' discussion, 15' evaluation (leaving roles behind) 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.
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 –16.40	**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)
16.40 – 17.40	**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.)
17.40 – 18.00	**End of day – Q&A with refreshments; MEET THE SPEAKERS**

Faculty Dinner

Wednesday July 12th Room V-04-018/022

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* **Industry perspective on drug development for rare diseases** *Eric Van der Veer* **Biomarkers** *Pietro Spitali* **Showcase: PROM development** *Céline Desvignes-Gleizes, 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 – 15.00	**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)
15.00 – 15.15	**Tea/Coffee Break**
15.15 – 16.15	**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)

16.15 – 17.15	**Showcase: PROM development** (the questions you ask and why you ask them; practical examples) *Céline Desvignes-Gleizes, Elizabeth Vroom* **Objective:** explain what is involved in developing a patient reported outcome measure using the DMD PROM development as a showcase (45' talk, 15' discussion)
17.15	**End of day – Q&A with refreshments; MEET THE SPEAKERS**

Thursday July 13th Room V-03-026/030

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**

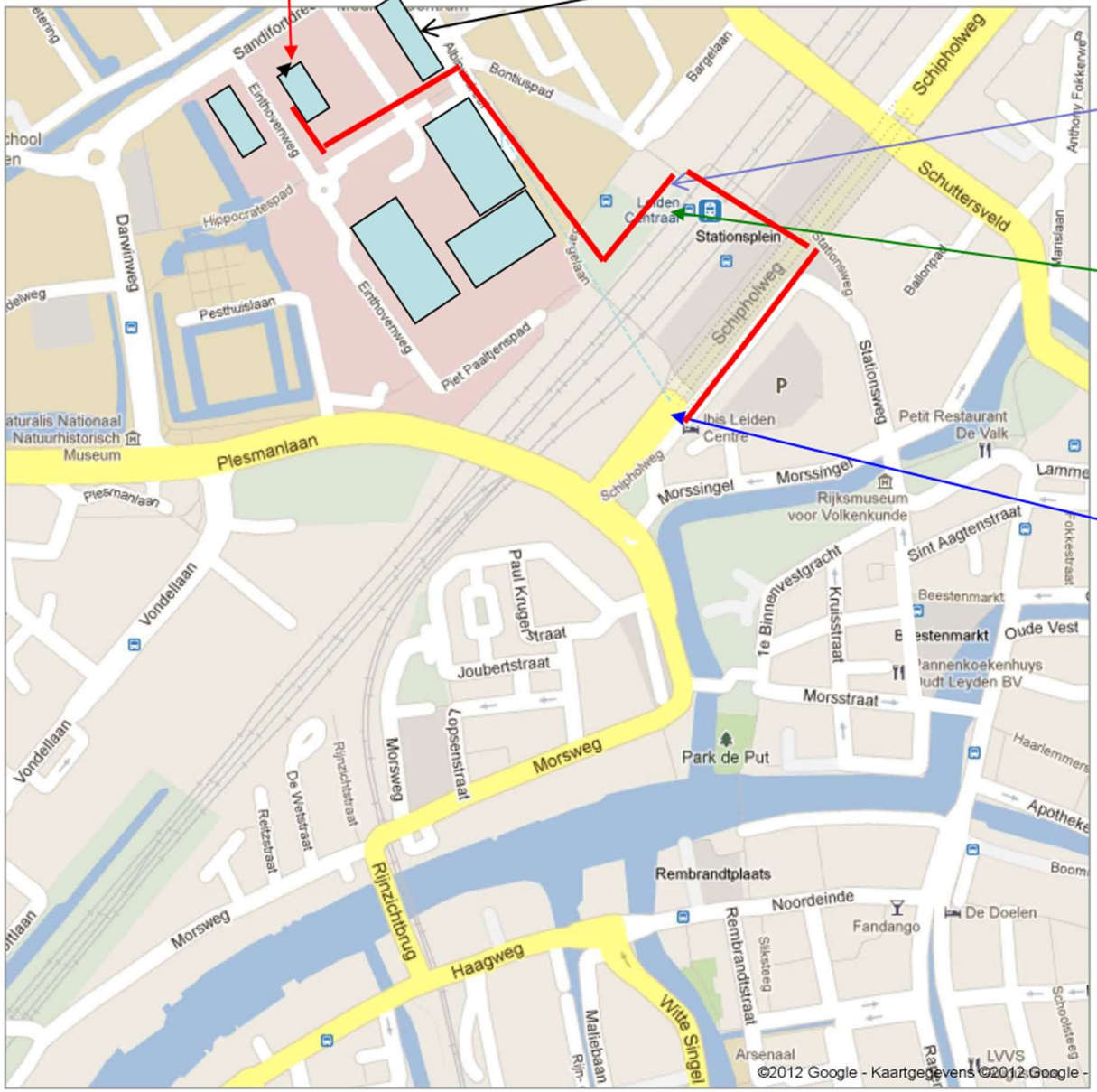
Friday July 14th Room V-02-018/022

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**

"The **5th NEUROMUSCULAR TRANSLATIONAL SCHOOL, Leiden, Netherlands, 10/07/2023- 14/07/2023** has been accredited by the European Accreditation Council for Continuing MedicalEducation (EACCME®) with **26** European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity."

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

Parking LUMC



Route by foot

Fletcher Hotel
Bargelaan 180

Railway station

Ibis Hotel
Stationsplein 240